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Associations Between Concepts of the Family Management Style Framework, and Measures of Child Adherence to Treatment for Heterozygous Familial Hypercholesterolemia

Heather Harker Ryan
University of Massachusetts Boston

ASSOCIATIONS BETWEEN CONCEPTS OF THE FAMILY MANAGEMENT STYLE FRAMEWORK, AND MEASURES OF CHILD ADHERENCE TO TREATMENT FOR HETEROZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA

A Dissertation Presented

by

HEATHER HARKER RYAN

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Approved as to style and content by:

_____
Laura L. Hayman, Professor
Chairperson of Committee

_____
Teri Aronowitz, Professor
Tan Chingfen Graduate School of Nursing
UMass Chan Medical School
Member

_____
Philimon Nyakauru Gona, Associate Professor
Member

_____
Sarah de Ferranti, Associate Professor
Harvard Medical School
Member

_____
Ling Shi, Director
Nursing PhD Program

_____
Jacqueline Fawcett, Chair
Department of Nursing

ABSTRACT

ASSOCIATIONS BETWEEN CONCEPTS OF THE FAMILY MANAGEMENT STYLE FRAMEWORK, AND MEASURES OF CHILD ADHERENCE TO TREATMENT FOR HETEROZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA

May 2022

Heather Harker Ryan, B.A., Barnard College
B.S., Columbia University School of Nursing
M.S., University of Massachusetts Boston
Ph.D., University of Massachusetts Boston

Directed by Professor Laura L. Hayman

Background: Heterozygous Familial Hypercholesterolemia (HeFH) is an underdiagnosed, autosomal dominant, monogenic condition affecting ~1:250 individuals in the United States (U.S.), resulting in cardiovascular events 10-20 years earlier than in unaffected peers.

Sample: Fifty-one parents of youth aged 2-18 years followed for HeFH in a pediatric specialty clinic.

Purpose: Assess parental perceptions of HeFH, child adherence to treatment, and parenting in HeFH-affected households.

Methods: A cross-sectional, descriptive, and correlational survey study congruent with elements of the Family Management Style Framework (FMSF). Pearson's and Spearman's correlations assessed linear relationships between parentally observed HeFH treatment adherence measures, parenting style, and parental perceptions of high cholesterol and risk for heart disease as applied to themselves or their families, and their children.

Results: Participating parents were largely middle aged (mean 46.1 years, SD 5.6), mothers (78.4%), Caucasian (86.4%), highly educated (Bachelor's or higher, 86.3%), and had personally received a diagnosis of genetically elevated high cholesterol (70%). Reference children were mostly adolescents (mean 13.4 years, SD 3.4), diagnosed with HeFH while school-aged (age 6.8 years, SD 4.1), and treated with a statin (80%). Median reported adherence to statins over past month was 94% (IQR 90-100). Missed doses were associated with forgetfulness (56.4%), carelessness (29%), or other reasons (41%). Illness perceptions differed between HeFH sub-concepts (high cholesterol and heart disease risk), respondent cholesterol status (+/-), and family position (parent/child). Patterns of association between illness perceptions and child treatment adherence, and illness perceptions and parenting styles, emerged along the same parameters. Parenting style generally did not directly correlate with observed child adherence, nor did child or family history with HeFH diagnosis and management.

Conclusions: Parents largely reported adequate to excellent adherence to HeFH treatments among their children. High cholesterol and risk for heart disease were perceived differently and may be valuable individually in future research. Consistent with proposed relationships within the FMSF, adherence was correlated with many aspects of illness perceptions and varied by family position. Parenting styles were not directly correlated with adherence but were associated with illness perceptions within family positions, suggesting an avenue for moderation of the illness perceptions/adherence relationships deserving of further research.

DEDICATION

As with so many other works of doctoral accomplishment, this dissertation is dedicated first and foremost to my mother, Paula Wilcox Harker. In a different world you would have been the first of our family to attain a doctoral degree; you have always seen my potential and have kept me motivated for the many years it has taken to complete it myself. I submit this final work for you, my role model and lifetime exemplar - no amount of love or thanks could ever fully convey the extent of my feelings for you.

This, too, is for my dedicated and loving husband, Zachary. You have encouraged and promoted me, my faithful “dissertation support” for almost a decade. Thank you for your patience.

And finally, for my daughters Helen and Madeleine – that you will someday see this as your example of academic passion, and dedication to the betterment of others, that I so blessedly inherited from your Nana.

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CHAPTER I

INTRODUCTION

Hereditary Familial Hyperlipidemia (HeFH) is the most common monogenic disorder in humankind, affecting anywhere from 1:200 to 1:313 individuals worldwide (de Ferranti et al., 2016; Kastelein et al., 2020). In the United States (U.S.), where cardiovascular disease (CVD) remains the leading cause of death, individuals with HeFH have a 2- to 27-times greater risk of CVD and an atherosclerotic process accelerated by 10 to 30 years compared to unaffected peers (Ahmad & Anderson, 2021; Kastelein et al., 2020; Perak et al., 2016). HeFH causes low-density lipoprotein cholesterol (LDL-C) elevations above the 95th percentile for age and sex, identifiable as young as two years of age, which is only minimally responsive to diet and lifestyle behaviors alone (de Ferranti et al., 2016; Hopkins et al., 2011). Management in childhood relies heavily on optimization of overall cardiovascular health until age 8-10 years, when HMG Co-A Reductase (statin) therapy can be incorporated (NHLBI Panel, 2011). The Centers for Disease Control and Prevention (CDC) has classified HeFH as a tier-one genetic condition, denoting its clinical significance for CVD risk and the strength of the evidence that this risk may be mitigated with early identification and treatment (Tier 1 Genomics Applications and Their Importance to Public Health | CDC, 2014). For these reasons the National Heart, Lung, and Blood Institute (NHLBI) Expert Panel recommends universal screening for cholesterol abnormalities starting at 9-11 years of

age to identify hypercholesterolemia among those whose family histories may not indicate other reasons to screen, or who may have been missed by selective screening based on family history (NHLBI Expert Panel, 2011).

Despite the ramifications that an HeFH diagnosis holds for children and families, research focusing on affected family units is minimal. The transition to parenthood appears to hold particular significance for affected individuals, manifesting as a moment of uncertainty, anxiety, guilt, or denial regarding the affected status of their children (de Jongh et al., 2003; Frich et al., 2007a; Senior et al., 1999). Once a child receives an HeFH diagnosis, an affected parent's beliefs around mitigating their own cardiovascular risk, the risk to their child, and their values regarding health behaviors may change. These may be conveyed to that child and subsequently internalized, thus informing adherence to treatment. Research suggests that a narrative around the importance of treatment and adherence may arise in these families (Mackie et al., 2015). Moreover, as socializing agents parents may reinforce these beliefs through their approach to parenting. Thus, understanding the interactions and associations between these intra-familial elements may shed light on how to better address adherence behaviors in pediatric populations.

Diagnosis and management of childhood health conditions depends on parent/guardian investment and frequently has implications for all members of a household. Parenting styles, intra-familial relationships, and family functioning have all been identified as significant elements in the overall effectiveness of the in-home care provided to children across a variety of health conditions (Leeman et al., 2016; Pinquart, 2013). Interventions based on these concepts have been found to effectively increase treatment adherence (Jones et al., 2014; Knafl et al., 2012; Knafl & Deatrick, 1990, 2003). Continued investigation into

the strength of these theories and their practical application to childhood disease management may prove beneficial for affected families.

Researchers are only beginning to investigate the intra-familial repercussions of genetic conditions on family dynamics and disease outcomes. Most of the literature pertaining to family management of genetic diseases in childhood focuses on autosomal recessive conditions, wherein parents carry the gene(s) for a condition, but do not present with symptoms themselves (Knafl et al., 2012; Knafl & Deatrick, 1990, 2003). Factors thought to be relevant to condition management, such as beliefs or perceptions regarding a child's condition, may be affected by personal experience when a parent has an identical diagnosis (Mackie et al., 2015).

The applicability of condition management theories where treatment primarily aims to minimize long-term health risks, rather than to address short-term health maintenance goals or prevention of disease complications, also warrants investigation. Conditions with delayed health implications necessitate vigilance in daily behavior without the biofeedback of perceptible change in wellness, posing a different type of challenge than conditions with immediate health consequences for ineffective management (Urke et al., 2019). Although researchers have considered the applicability of management theory to continued wellness, such as with maintained breastfeeding, to date there have been no investigation of chronic conditions that present in childhood but pose minimal risk before adulthood (Knafl et al., 2012; Knafl & Deatrick, 1990, 2003). Application of family management theory to an autosomal dominant condition such as HeFH, which is chronic and asymptomatic prior to a cardiovascular event, may provide insight into the multigenerational effect of illness. Investigation into the intra-familial implications of HeFH on management outcomes may

provide a basis for interventions on HeFH youth adherence. The purposes of this study were to assess parental perceptions of HeFH as it affects themselves or their families, gain knowledge pertaining to child adherence to HeFH treatment in children 2-18 years of age, and describe how parents in households affected by HeFH approach parenting in general. Finally, this study sought to identify relationships between these concepts to provide insights into areas of potential family intervention for the improvement of HeFH treatment adherence among youth. Understanding these relationships will help clinicians provide more holistic and informed care for families with HeFH.

Theoretical Framework

The Family Management Style Framework (Knafl et al., 2012; Knafl & Deatrick, 1990, 2003) guiding this study identifies one indirect and three direct concepts influencing management (see Figure 1). The sociocultural context is the indirect concept, and the direct concepts are definition of the situation, management behaviors, and perceived consequences related to condition management (Knafl & Deatrick, 2003).

Sociocultural Context

The sociocultural context influences management and includes social and support networks (including health care and educational professionals) and available resources (Knafl et al., 2012). Other influences may include disease-specific influences, characteristics of the affected child (i.e., sex, age at diagnosis, stage of psychosocial development, birth order rank), family structure (such as number of other children in the family, divorced parents, or other characteristics that influence social relations and resource availability/constraint), the race or ethnicity of the family, and educational status (Gibson-Young et al., 2014; Jang &

Whittemore, 2015; Kim et al., 2016). All these factors act unidirectionally to affect how the situation is defined, management behaviors, and perceived consequences related to condition management (Knafl & Deatrick, 2003).

Major Components

Definition of the Situation. The definition of the situation relates to parental perceptions of their affected child and that child's condition, the work of managing that condition, and how well parents' perceptions of this work agree with one another (presuming a two-parent/caregiver household). Parents perceive their children across a spectrum of normal, and as individuals apart from their condition (Knafl et al., 2012; Knafl & Deatrick, 2003). Parents also have conceptualizations and expectations of the medical condition itself, particularly regarding its cause, seriousness, predictability, and course (Knafl & Deatrick, 2003). Each parent has their own responsibilities in traditional two-parent households. The greater the extent to which parents agree about what these responsibilities are, and the equity of their distribution, the more effective their adaptation to managing their child's condition (Knafl & Deatrick, 1990, 2003).

This concept has been articulated in other disciplines, particularly psychology, as relating to illness perceptions, defined as thoughts, feelings, beliefs, and personal knowledge pertaining to a condition. The domains proposed in the Common Sense Model of Self-Regulation (CSM) (Leventhal et al., 2003)parsimoniously correspond with those proposed by the FMSF (Knafl & Deatrick, 2003) and a measure of the CSM was used to measure the definition of the situation within this study. The CSM posits that the impetus for coping with a condition stem from ones' biological, emotional, and cognitive experience with that condition (Leventhal et al., 2003).

Management Behaviors. Management behaviors relate to the philosophy that underpins the parental approach to condition management and perceptions of how well parents feel they institute measures necessary to treatment. Philosophical perspectives on management encapsulate parental beliefs, values, goals, and priorities towards parenting and the child's condition. Parental approaches to management, particularly the ability to develop and implement a management routine, demonstrate the capacity to make inroads in the direction that the family's philosophy directs. Although behaviors necessitated by a health condition vary, management behaviors provide insight into the gestalt of the family's management approach (Knafl & Deatrick, 2003). Management behaviors was defined as parenting style (Baumrind, 1966, 1967) within this study, which is how parents transmit values and encourage children to meet socialization goals (Darling & Steinberg, 1993).

Baumrind (1966) conceived of three archetypes through which parents socialize their children to the norms and values of the family and larger society, specifically authoritarian, permissive, and authoritative. Authoritarian parents demand obedience and respect for authority, do not discuss rules with children prior to their implementation, and place no value on ensuring the child understands their rationale. Authoritarian parents expect rules to be followed without question and punish the child who does not (Maccoby & Martin, 1983). In contrast, permissive parents set minimal demands on their children, and are highly responsive to the child's demands through emphasis on and reinforcement of the child's impulses (Baumrind, 1966). Rules, structure, and discipline may be minimal in these families (Maccoby & Martin, 1983). Authoritative parents seek to strike a balance between permissive and authoritarian approaches. These parents value their children's opinions and

needs, while maintaining final authority and expecting compliance. Negotiation may be acceptable but conformity to final decisions is expected (Maccoby & Martin, 1983).

Parenting styles encapsulate parental expectations for obedience, respect for the child as another autonomous individual, and willingness to engage in socialization. They provide the context for parenting practices, and the behaviors used by parents to socialize children by imbuing them with emotional and relational meaning (Knafl & Deatrick, 2003). Finally, having been studied in the context of both wellness and illness outcomes, parenting style is ideally suited for application to the FH population given its overlap with both states of health.

Perceived Consequences. The third major concept of FMSF pertains to how parents perceive the impact of the child's condition on the family unit. This includes both the centrality of the disease in daily functioning and the long-term implications of the diagnosis. Ideally, as parents become more proficient in condition management in the home, the condition becomes less central and other elements of family life can take precedence. Where a condition confers lower long-term quantity or quality of life, parents may elect not to consider these implications (Knafl & Deatrick, 2003).

Nursing Implications

Employing the FMSF in relation to HeFH-affected families had two implications for the theory itself. First, it offered insights into its applicability to families affected by an autosomal dominant condition with children at risk for poor health and adverse events in the distant - rather than immediate – future. Second, HeFH had yet to be approached as an intergenerational condition with potential for parental experience to directly impacting a child's adherence to treatment. Identifying patterns in parental perceptions of HeFH, feelings

towards management, and parenting styles that may be associated with greater youth adherence may prove useful to clinicians working with HeFH-affected families. Third and finally, not only has research been scant in HeFH family experience, but studies have been primarily undertaken among adults in northern European populations. This study added to the current literature on HeFH in the U.S., as well as offered the first quantification of daily adherence behaviors in a specific population of American HeFH youth.

This study also had direct implications for nursing care and research. First, the use of the FMSF promotes both the theory itself and the utility of nursing theory more generally. Secondly, although nurses have lent their voices to the HeFH literature (de Ferranti et al., 2012; Gidding et al., 2015; Hayman, 2000; Muir et al., 2012; Zawacki et al., 2018), the nursing perspective remains in the minority of that represented. The result is minimal evidence relating to holistic family care and general family life experience, which has been identified as a major topic for ongoing research in a recent agenda statement by the American Heart Association (Gidding et al., 2015). This study was designed to further this body of literature, through which it may encourage more family-focused HeFH care, and perhaps eventually substantiate a transition to a more nursing-centric care model.

CHAPTER TWO

REVIEW OF THE LITERATURE

Each aspect of family management of pediatric chronic illness goes through every individual family member prior to translation into a "style" or level of management effectiveness (see Figure 1). The various perspectives of each family member are valuable, as experiences or values might translate into improved adherence or alternatively contribute to adherence that is sub-optimal. Thus, understanding parents with a diagnosis of HeFH and how the diagnosis affects family dynamics is integral to appropriate application of the FMSF in this population. The following review of the literature summarizes the current understanding of how the HeFH diagnosis affects youth and their families, as well as current knowledge pertaining to parenting style as it relates to adherence outcomes among youth affected by similar conditions. The general literature on the adult experience with HeFH is used as proxy for that of parents more specifically.

Parental Definition of the Situation

View of the Condition: Perceptions of HeFH Among Affected Adults

Individual Illness Perceptions of HeFH. Much of the literature on perceptions of HeFH relate to the identification of a silent disease, and subsequent rationalizations around cardiovascular risk. The majority of adults interviewed regarding their perceptions of HeFH verbalize a pragmatic acceptance of the diagnosis, particularly the benefits of identification –

specifically initiation of pharmacotherapy – being preferable to ignorance (Agård et al., 2005; Hardcastle et al., 2015). This frequently occurs in tandem with a variety of social comparisons that act to either limit or heighten perceived risks. For example, an individual may compare themselves to an 'average' person, emphasizing their own greater awareness of and adherence to heart-healthy behaviors resulting from their HeFH diagnosis (Agård et al., 2005; Gooding et al., 2016; Senior et al., 2002). Others may compare themselves to earlier generations of affected family members, highlighting improvements in medical care or a lack of early cardiovascular events as a pre-hoc explanation of their state of wellness (Senior et al., 2002). Yet others still compare themselves to the HeFH "other" who receives no treatment, and to those with other inheritable conditions with a greater perceived impact on health and daily function (Jenkins et al., 2012; Senior et al., 2002; Urke et al., 2019). All serve to rationalize a lower personal risk for heart disease than has been shown for those with HeFH in general.

Alternatively, comparisons may serve to heighten perceived risk, particularly when the patient identifies similarities between themselves and those family members who have suffered from HeFH-related coronary artery disease (Frich et al., 2006; Hallowell et al., 2017; Jenkins et al., 2012; Keenan et al., 2019; Kools et al., 2008; Mackie et al., 2015; Senior et al., 2002; Urke et al., 2019). These individuals may perceive the morbidity or mortality of relatives, particularly at specific ages, as proxies for their own events (Frich et al., 2006; Jenkins et al., 2012). This tendency appears to be an effective cue to screening and/or treatment implementation for some individuals (Agård et al., 2005; Hallowell et al., 2017; Keenan et al., 2019; Mackie et al., 2015). It may produce vigilance towards possible CAD symptoms, although the relative benefit of this is unclear (Senior et al., 2002). These

individuals may also take actions on a family level, disclosing the diagnosis to other at-risk relatives for the purposes of receiving emotional support, and unburdening themselves of potential guilt or responsibility if a family member experiences a poor CAD outcome that might have been prevented from knowledge of HeFH (Hallowell et al., 2017; van den Nieuwenhoff et al., 2007).

An important but rarely heard voice in this literature comes from affected individuals who elect not to persist with routine FH follow-up or avoid formal identification entirely. From those who have been formally interviewed, some voice a 'carpe diem' philosophy, and view genetic inheritance through a deterministic lens (Hardcastle et al., 2015; van den Nieuwenhoff et al., 2007). These views appear most frequently in the context of frustrating treatment failures, treatment non-adherence, and refusal to partake in HeFH screening (Agård et al., 2005; Hardcastle et al., 2015; van den Nieuwenhoff et al., 2007). Until recently, all of these accounts come second hand in interviews where those receiving treatment describe their relatives. These interviewees often describe efforts to change their family member's perspective, thus suggesting that the experiences of other family members may inform one's own approach to diagnosis and treatment (Hardcastle et al., 2015). No literature to date has investigated HeFH from the perspective of individuals that know the possibility of their having the diagnosis exists, but intentionally elect not to pursue screening.

The only study to include "lost to follow-up" patients with HeFH was published in 2019, comparing statements from 11 young adults who had elected not to return to their specialist for at least two years to those from 13 similarly aged youth who had (Urke et al., 2019). It found that the defining difference in perspective between these individuals pertained to whether they consider the consequences of their condition. Those who

maintained their FH care expressed greater salience in the potential risk for long-term heart disease, and likewise verbalized a high trust in professional management, a commitment to treatment, and greater motivation towards maintaining a heart-healthy lifestyle (Urke et al., 2019). Unlike the fatalistic rationales hypothesized in earlier literature by second-hand sources, participants who let their care lapse had not done so because they felt their genetic fate had been determined; rather, they "postponed" its consideration. These individuals expressed an inability to be concerned about an imperceptible condition, rationalized the prioritization more pressing concerns, and felt close management would medicalize them in unnecessary or unwanted ways (Urke et al., 2019). Those who considered the consequences of the condition also found its management more challenging, perhaps expectedly; those who did not consider the consequences of FH instead belittled its management as not needing to be strictly followed, or not caring to do so (Urke et al., 2019). Interestingly, both groups of interviewees took mental steps to "normalize" the condition, but the definition of "normal" differed substantially. Consequence-focused individuals sought normalization through habituation of lifestyle and medications, describing them as "something we're used to…Like, I've just thought that…this is the way it is" (Keenan et al., 2019; Meulenkamp et al., 2008; Smets et al., 2008; Urke et al., 2019). Non-consequence focused individuals emphasized the inability to decern themselves from unaffected peers, seeing it as "easy to live with" and consequently easy to disregard, particularly in comparison to the daily management of other chronic conditions (Urke et al., 2019). This has implications for the application of the FMSF, wherein normalization has been postulated as a major goal of families well-adapted to condition management, as well as future research on maintained patient engagement.

Intrafamilial and Life Course Experiences with HeFH. As individuals affected by HeFH transition through social and familial roles across their lifetime, the condition changes in relevance and meaning. In marriage, HeFH takes on new meaning to unaffected spouses; cohabitation often results in shared diet, and unaffected spouses frequently arise as both adopters and implementers of the affected spouse's recommended restrictions (Mackie et al., 2015; Tonstad, 1996). Pregnancy can increase an individual's perceived vulnerability to HeFH and heightens awareness around the importance of keeping oneself and one's child well. It may trigger screening in those who had yet to be tested, and affected women have described concerns about the implications of lifetime HeFH treatment on their fertility and the health of a fetus (Frich et al., 2006, 2007a; van den Nieuwenhoff et al., 2007). Unlike those affected by other genetic conditions, patients with HeFH do not perceive a mandate to be screened for the purposes of family planning; its treatability and relative lack of impact compared to conditions such as Huntington's Disease appears to alleviate affected individuals of the moral burden of reproductive decision-making relative to their diagnosis (Hallowell et al., 2017; Weiner, 2011). However, parents may worry about the prospect of having potentially passed "bad genes" to their children, and express feelings of pain or guilt when a child is diagnosed (de Jongh et al., 2003; Frich et al., 2007a). In some families, parental diagnosis may result from the diagnosis of a child; in such cases, a poor understanding of the condition may result in feelings of fatalism regarding their child's long-term prospects (Senior et al., 1999). The social and developmental transition from adolescence to adulthood carries the additional responsibility of condition management for HeFH+ youth: previously active and engaged families become a separate entity from the emerging adult, peers take on approximately the same significance as family, and support

from the healthcare system becomes something they must assume full responsibility for themselves (Sliwinski et al., 2017). Young adults express uncertainty around how to take over their disease management, particularly pertaining to navigation of the healthcare system (securing health insurance, making appointments, obtaining scripts, etc.), and how to maintain a heart-healthy lifestyle in the context of changing priorities and increasing responsibilities in other life spheres (Sliwinski et al., 2017). The degree of success an individual has in navigating these challenges may influence their overall perceptions of the condition, and the iterative process of forming a heuristic around its relevance and management (Mackie et al., 2015).

Illness Perceptions and Adherence Behaviors among HeFH Adults. Four research groups have sought to quantify the HeFH experience using illness perceptions, all relying on elements of the Common Sense Model of Self-Regulation (CSM) as their conceptual basis. As mentioned previously, the CSM theorizes that effective coping with a condition stems from ones' biological, emotional, and cognitive experiences with that condition. Adherence to management recommendations represents one of many possible outcomes related to coping (Leventhal et al., 2003). The CSM consists of six standard domains of experience. These include: 1.) how one identifies with a condition (commonly related to experience with symptoms); 2.) how long one expects to be affected by a condition, and/or whether one is affected continuously or intermittently; 3.) the perceived cause of the condition; 4.) one's understanding of the potential consequences of the condition; 5.) how able one feels in controlling the condition, either personally or through treatment recommendations; and 6.) how well one feels they understand their condition (Leventhal et al., 2003; Moss-Morris et al., 2002). Researchers have taken different approaches to defining HeFH and its most salient

perceptible aspects for their cohorts, framing measures of perception in terms of high cholesterol (HC), increased risk for CVD, fatalism around an HeFH diagnosis, and "familial hypercholesterolemia" more generally (Brewer et al., 2002; Claassen et al., 2010; Hagger et al., 2016; Hagger et al., 2018, 2019). Findings have likewise varied, particularly relative to their associations with medication adherence.

The first such investigation sought to determine if perceptions of HC among hyperlipidemic individuals (n = 169, Mean age = 67, Mean education 15 years), including those with HeFH, were associated with lipid lowering therapy adherence or achievement of LDL goal (Brewer et al., 2002). The CSM measures used were developed in conjunction with an original CSM theorist and tested for validity using confirmatory factor analysis; no preliminary reliability measures were reported and means for the individual aspects of illness perceptions were not provided. Participants frequently identified genetics as a primary cause of their condition (Brewer et al., 2002). In regression analysis, self-reported adherence was statistically significantly associated with ones' perceptions of the consequences of HC (β = 0.28 [$p < 0.05$]). Meeting LDL goal was associated with identification of HC with specific symptoms (examples included 'fatigue' or 'tiredness'; $\beta = -0.16$, $p < 0.05$), belief that one's cholesterol fluctuates greatly regardless of medication adherence (akin to a cyclical timeline; $\beta = -0.17$, $p < 0.05$), and understanding of consequences ($\beta = 0.23$ [$p < 0.005$]) (Brewer et al., 2002). Illness perception domains explained 0.15% and 0.17% of the total variance in the LDL goal and treatment adherence outcome models respectively.

Alternatively, Claassen and colleagues (2010) investigated the relationships between perceptions of CVD, risk for CVD, and self-reported engagement in treatment recommendations within a cohort of genetically identified HeFH+ individuals in the

Netherlands (n = 81, Mean age = 48, 36% higher education/vocational training). The IPQ-R – a widely used and well-validated instrument for measurement of CSM concepts – was adapted for application to perceptions of CVD and showed acceptable to strong reliability across most domains. Most respondents had two or more first-degree family members diagnosed with CVD and acknowledged both genetic and lifestyle components to their risk for CVD. Interestingly, the average responses to perceived duration of CVD, consequences of having CVD, and controllability of CVD risk through medication and lifestyle all fell in the "neither agree nor disagree" range on 1-5 Likert scales, suggesting possible uncertainty or ambivalence in these areas (Claassen et al., 2010). Having strong family history of CVD (identity) and greater confidence in the efficacy of lifestyle (treatment control) both weakly but statistically significantly correlated with greater self-reported adherence to lifestyle recommendations ($r = 0.29$ & $r = 0.36$, respectively, $p < 0.05$ each). Of note, the outcome measures related to diet and lifestyle showed suboptimal reliability in this population ($\alpha = 0.54$), and adherence to lipid lowering therapy was near-universally reported as perfect, making assessment of variation based on illness perceptions not possible (Claassen et al., 2010).

The Genetic Risk Assessment for FH Trial (GRAFT) Study Group used the Revised Illness Perceptions Questionnaire to specifically investigate prospective relationships between concepts of personal control across multiple concepts pertaining to HeFH, and causal attributions to perceived treatment effectiveness and actual adherence outcomes (Marteau et al., 2004; Senior et al., 2004, 2005; Senior & Marteau, 2007). They measured perceived personal control over HC, heart disease, HeFH, and HeFH-related fatalism, as well as effectiveness of diet and medication management. Participants (n = 340 at initiation) were

identified through a network of associated lipid clinics in Southeast Britain and included clinically diagnosed probands and eligible family members; all participants were randomized into receiving detailed clinical diagnosis (n = 196 at completion) or detailed clinical diagnosis plus genetic testing (n = 79 at completion).

Overall, on a scale of 0-5 participants in the GRAFT study perceived themselves as having control over their HC (4.09/5) and HDR (3.93/5), and their treatment as effective (4.08/5) (Senior et al., 2005). Six months following the receipt of diagnostic testing results, genetic testing did not result in lower perceived personal control over cholesterol, heart disease, or HeFH, nor did it increase perceived fatalism (Marteau et al., 2004). Self-reported adherence did not vary by diagnostic type, nor was it predicted by perceived control over cholesterol, control over heart disease, or effectiveness of treatment six months post diagnosis (Marteau et al., 2004; Senior et al., 2004). Multiple correlational and predictive associations were identified between perceptions of personal control over cholesterol, heart disease risk, and perceived causative influences, as well as between diagnostic type and perceived effectiveness of the treatment types (Senior et al., 2005; Senior & Marteau, 2007).

Contemporary research in this area has been undertaken by Hagger, Hardcastle, Watts and colleagues (2016, 2018, & 2019). Unlike the previous studies, these investigations have used *intention* to adhere to medication, diet, and exercise – rather than self-reported adherence – as the primary outcome of interest. Moreover, they have utilized the full IPQ-R, and adapted it to refer to HeFH rather than one of its component parts, reporting composite reliabilities for each domain in the excellent range apart from treatment control (ρ range = 0.49 – 0.93). Finally, Hagger, Hardcastle, Watts and colleagues (2016, 2019) have compared the relative predictive strengths of the components of the CSM to those of two other highly

recognized theories: the Theory of Planned Behavior, and Social Cognitive Theory (self-efficacy).

Initially piloting their survey on a cohort of 110 genetically-diagnosed HeFH patients in Perth, Australia (Mean age 50.7, Mean time since diagnosis 4.6 years, 20% with CAD diagnosis), Hagger et al. (2016) concluded that the domains of the CSM provided little predictive value on adherence intentions after controlling for past behavior, gender, age, and medical history; the only statistically significant relationship related to treatment control and physical activity (PA) (β = -0.201, 95% CI [-0.38, -0.02]) (Hagger et al., 2016). These hypotheses were subsequently retested in 2019 using a sample consisting of HeFH patients from ten nations of varying cultural backgrounds and socioeconomic conditions (n = 762), identifying direct relationships between personal control and intention to participate in PA (β = 0.071, 95% CI [0, 0.142]), and between understanding of consequences and intention to adhere to dietary recommendations (β = 0.106, 95% CI [0.035, 0.177]), even after controlling for reported previous adherence. Previous adherence, however, was statistically significantly associated with virtually every domain of the CSM across all three treatment types (Hagger et al., 2019). Finally, Hagger et al. (2018) proposed and tested a pathway model to predict intention to take lipid lowering therapy using an international sample (n = 551), wherein the relationship between historical medication adherence and future intention to adhere are mediated in stepwise fashion, first by general perceptions of pharmacotherapy (beliefs about medications, side effects, and the treatment control domain of the CSM), then by the more behavior-specific elements of the Theory of Planned Behavior (Hagger et al., 2018). While treatment control did not directly predict intention to adhere to pharmacotherapy, it acted as a significant mediator of reported past adherence (SEM effect -0.12, p = 0.05) through

treatment attitudes (SEM effect 0.192, $p = 0.001$) and subjective norms towards therapy (SEM effect 0.151, $p = 0.001$).

In all three investigations the Theory of Planned Behavior had stronger and more consistently predictive relationships with intention to adhere to treatment in this population. The researchers hypothesized that the domains of the CSM may hold more salience in conditions that elicit emotions, fears, or anxieties, which they maintain HeFH does not evoke in most affected individuals, although 25-33% of HeFH+ individuals do endorse such feelings (Andersen et al., 1997; Hagger et al., 2019). Notably for the interpretation of these findings, the outcome variable – intention to engage in a specified behavior – is an explicit component of the theory of planned behavior (Hagger et al., 2016). Additionally, in the analyses where self-reported previous adherence was included, the measure employed limited responses to "always" or "never;" this may not accurately reflect the actual habits of respondents. Third and lastly, the partial least squares analyses undertaken for their 2016 and 2019 publications depend on the researcher to explicitly specify the direction of theoretical relationships, thus it is still possible that other untested relationships may explain adherence intention as well (i.e., other aspects of the theory of planned behavior could moderate the relationships between the domains of the CSM and adherence intentions).

Child Identity: Parent's Perceptions of How HeFH Affects Their Children

Although often implicit in the literature on HeFH in childhood, parents do not generally perceive their children as different than their unaffected peers. Rather, parents in HeFH-affected families inevitably face the decisions of whether and when HeFH should be identified and subsequently treated in their children, as doing so can be perceived by some parents as causing worry and medicalizing an otherwise "normal" child (Keenan et al., 2019;

Mackie et al., 2015; Meulenkamp et al., 2008). Other parents emphasize the importance of screening children to establish HeFH behaviors early, ascribing to an alternative definition of "normal" through making those behaviors habitual (Frich et al., 2006; Keenan et al., 2019; Mackie et al., 2015; Meulenkamp et al., 2008; Weiner, 2011).

Children with HeFH also overwhelmingly emphasize how "normal" they are in relation to their peers (Kools et al., 2008; Meulenkamp et al., 2008). In general, children diagnosed with HeFH have not been found to experience more psychosocial challenges or poorer quality of life than their unaffected peers, although the development of disordered eating patterns and anxiety has been anecdotally noted (de Jongh et al., 2003; Smets et al., 2008; Tonstad, 1996; Tonstad et al., 1996). This overwhelming agreement that children and youth with HeFH are "normal" is particularly relevant to observe in the context of the FMSF, wherein achievement of a beneficial management style has historically relied in part on whether parents believe the family is achieving some definition of "normalization" (Knafl et al., 2010). In HeFH populations, parents may use differing definitions of normal to rationalize both adherence and nonadherence to condition management (Urke et al., 2019).

Management Mindset

Attitudes Towards Youth Adherence to Treatment. The literature on families with HeFH affected children has considered attitudes towards treatment as frequently as actual adherence to it. To date, only four studies have considered children's attitudes towards treatment with medication and diet; exercise has not previously been examined.

Attitudes Towards Medication. Using a non-validated survey, de Jongh et al. (2003) assessed HeFH family attitudes towards medication and general administration habits, but not specific adherence rates. Children generally felt safer on medications (n = 37, or 61%),

and only a minority of parents (n = 6, or 7%) found pharmacotherapy to "cause" intrafamily conflict (de Jongh et al., 2003). Notably, while 70% (n = 43) of youth reported often or always taking their medication "regularly," substantially more parents (n = 78, or 90%) believed this of their children. Unfortunately, the term "regularly" does not appear to have been defined by the researchers. Seven percent (n = 4) of children acknowledged "often" or "always" forgetting their medications, and 26% (n = 23) of parents stated often or always having to remind their children to take their medications (de Jongh et al., 2003).

Attitudes Towards Diet Modification. In two surveys totaling 205 families from Norway and Amsterdam regarding general attitudes and perceptions around recommended HeFH diet, responses were remarkably consistent (de Jongh et al., 2003; Tonstad, 1996). Almost (53%) stated keeping to their diet while at school (de Jongh et al., 2003). Far fewer (n=23, or 33%) disclosed their dietary restrictions to friends and peers. More than half of parents found keeping the HeFH diet "easy" (n = 92/154), while 10% (n = 9/87) found monitoring their child's diet "hard," and 31% (n = 48/154) found motivating themselves to keep to the diet difficult in the face of many healthy years before the potential onset of cardiovascular events (de Jongh et al., 2003; Tonstad, 1996). A minority of parents (n = 5, 5.7%) found that instituting the recommended diet caused "trouble" for them with their children (de Jongh et al., 2003). More recently, a study conducted in Spain intending on engaging in a stepwise dietary adherence intervention trial incidentally found themselves also measuring attitudes towards their chosen interventions; more than 50% of families not meeting recommended dietary guidelines after receiving a qualitative dietary intervention encouraging Mediterranean-style intake outright refused to participate in the subsequent

quantitative intervention (food weighing, daily energy intake counts) due to its perceived "management difficulties" (Cicero et al., 2019).

More generally and importantly in the context of the FMSF, the behaviors of parents were identified as important influencers on the attitudes of HeFH-diagnosed children in an American cohort (Kools et al., 2008). Parents who expressed fatalistic attitudes towards their disease or role modeled poor dietary adherence behaviors produced children who generally held similar perspectives, although a few adopted an opposite perspective. Parents that took a hyper-involved stance, acting overly vigilant in their attempts to make their children conform to a diet, cultivated spite, and drove their children further away from adherence (Kools et al., 2008).

Parental Adherence to Treatment. The expertise of parents in managing a condition has been explicitly identified as a quality that a family's overall management mindset may be sensitive to; observing a parent's own adherence to HeFH recommendations may provide further insights into this expertise. Moreover, parental perspectives on the ease of managing HeFH may be reflected in personal adherence; indeed, for many affected individuals the treatment of HeFH defines the condition itself. Only a handful of studies have sought to measure adult adherence to the three pillars of HeFH treatment – pharmacotherapy, dietary management, and regular PA – and are summarized below.

Adult Pharmacotherapy Adherence. Healthcare providers with prescriptive powers in nations with well-documented HeFH populations appear to consistently prescribe statins to HeFH patients, particularly after genetic diagnosis (Marteau et al., 2004; Umans-Eckenhausen et al., 2002; van Maarle et al., 2001, 2003). In cross-sectional studies performed in England, the Netherlands, and Sweden, self-reported statin adherence rates

have been consistently above 85% (Claassen et al., 2010; Galema-Boers et al., 2014; Hollman et al., 2006; Senior et al., 2004; Senior & Marteau, 2007). More recent investigations using prescription-coverage data has not substantiated these reports, finding Medication Possession Ratios (# days covered by prescription/observation period) ranging between 64-78% (Casula et al., 2016; Jones et al., 2020). In interviews, HeFH patients in these nations appear unconcerned about using pharmacotherapies; cholesterol management by this means seems natural and straightforward given the minimal effectiveness of lifestyle and diet interventions (Frich et al., 2007b; Hardcastle et al., 2015; Weiner, 2009).

Only one investigation was identified with adherence data pertaining to a U.S. sample of HeFH+ individuals, which found a substantially lower rate of adherence among women with FH (n = 200) compared to international samples (Benson et al., 2016). Using the Morisky-Green-Levine 4-item adherence scale, only 58% (n = 116) reported never forgetting or intentionally skipping doses of their lipid lowering therapy (Benson et al., 2016). Of note, more recent use of this adherence measure in an FH-diagnosed population in Russia has resulted in similar findings (Korneva et al., 2019). Participants in Benson et al.'s US population are unlikely to be representative of those with FH generally, as they solicited women who self-registered for the FH Foundation CASCADE Registry. Compared to participants from the National Coalition for Women with Heart Disease database subsample, individuals from the FH Foundation registry were more likely to be white, married, and have high incomes (Benson et al., 2016). However, no substantial difference in adherence to lipid lowering therapy was identified between those with FH and those without.

Overall, the literature on statin adherence among HeFH patients suggests tolerability of this form of treatment but has relied heavily upon self-report measures. Those employing

secondary data sources have identified significantly lower adherence rates, suggesting an inflation of self-reported adherence measures. Thus, reason exists to suspect bias and possibly cultural differences within and between available studies.

Adult Diet and Physical Activity Adherence. Descriptions of PA and diet among those with HeFH have received considerably less attention by researchers than pharmacotherapy. Among studies measuring general dietary "adherence" or saturated/polyunsaturated fat intake, investigators have found the diet of most HeFH+ patients acceptable, sometimes even prior to having a formal diagnosis. For example, in a randomized trial of the effects of molecular HeFH diagnosis versus clinical diagnosis alone in an English population, Marteau and associates found 77-92% of participants consumed diets "low" in total fat at baseline as measured by DINE method, and 73-88% consumed "high" amounts of unsaturated fats; "low" and "high" were left undefined (Marteau et al., 2004). These findings were mirrored more generally in a report from the Netherlands, finding 70% adherence at time of HeFH diagnosis (n = 215), improving to 75% at follow-up (van Maarle et al., 2002). In their 2007 publication, Senior & Martineau reported all participants (n = 317) met the recommendation of < 25g total fat daily at the time of enrollment, thus determining that the variable held little value as an outcome measure (Senior & Marteau, 2007). A recent population-level comparison of FH+ and FH- individuals in Spain found significant differences in diet, finding FH+ individuals to consume less energy overall, less energy from fat, less saturated fat, and to have a greater propensity towards a Mediterranean-style diet than their FH- relatives (Arroyo-Olivares et al., 2019). The applicability of these Europe-based investigations to FH+ adults in the US is questionable, however, substantiated by the only US-based cohort of FH+ individuals with published measures of diet. This

randomized controlled trial showed higher than recommended saturated fat intake at baseline (Women mean 25.6-28.8g/day, recommendation ≤ 22g; men mean 32.6-33.4g/day, recommendation ≤ 28g), as well as low fruit and vegetable intake, neither of which improved significantly after a structured dietary intervention (Broekhuizen et al., 2012).

Estimates of PA adherence have varied substantially between populations and have not been as consistently acceptable as measures of diet. In their previously mentioned randomized trial of differing diagnosis types, Marteau et al. (2004) found 24%-55% of participants rarely or never engaged in moderate-to-vigorous activities. Six months after randomization to either clinical or genetic means of HeFH diagnosis, participant exercise had not changed significantly nor differed across groups. Van Maarle et al. (2002) also found low rates of general "lifestyle" adherence at baseline (33%, n = 41), but reported an increase to 86% (n = 107) after a HeFH diagnosis. Recent SAFEHEART population-level data from Spain again found statistically significantly higher rates of reported moderate and vigorous PA in the preceding 7-day period among FH+ individuals than their unaffected family members, although the specific self-reported frequency or duration of PA cannot be deduced from their report (Arroyo-Olivares et al., 2019).

Studies pertaining to HeFH+ individuals in the US have found better PA engagement than studies conducted in Europe; for example, Broekhuizen and colleagues (2012) reported baseline levels of Moderate-to-Vigorous PA at 363 and 422 minutes weekly in two separate treatment groups (Broekhuizen et al., 2012). Benson et al.'s (2016) survey of US women also found relatively high levels of PA, with 69% (n = 138) of HeFH+ persons surveyed reporting meeting the target of ≥150 minutes of PA weekly, approximately the same percentage as those surveyed that had been diagnosed with genetic cholesterol elevation.

Overall, the current available literature accessible to us suggests acceptable adherence to both diet and PA recommendations among FH+ adults despite the widely acknowledged ineffectiveness of these interventions at substantially lowering cholesterol in this population (Senior et al., 2002; Weiner, 2009). However, overall research remains minimal in this area, and findings can vary by methodology, nationality, and cultural region. Additional research, particularly in US cohorts, could provide further information and insights.

Parent's Perceived Consequences of HeFH

Family Focus: How HeFH Affects the Collective Family Unit

Impacts on the entire family system, regardless of whether one personally possesses an HeFH diagnosis, have been documented. Just as unaffected spouses often adopt the HeFH diet and lifestyle, so too do parents often apply them to all their children; this has been identified as a means of normalizing dietary management and improving overall adherence (Kools et al., 2008; Mackie et al., 2015). The occurrence of early cardiovascular events in a parent or other close family member emotionally impacts all family members (Froese et al., 1980). Affected children may try to use their diagnosis as an leverage over unaffected siblings to garner sympathy or favor from parents or caregivers, potentially causing intra-sibling strife (Tonstad, 1996). Interparental or interfamilial discrepancies in perceived value of HeFH treatment in children can cause discord as well, a particular challenge for families where parents have separated or divorced (Kools et al., 2008; Tonstad, 1996; Urke et al., 2019).

A major theme in this literature is parental responsibility for both identification and treatment decisions, as well as controlling the messaging around HeFH risk. It manifests as

parental protectiveness in the management decisions articulated by parents in Meulenkamp et al.'s (2008) investigation of the effects of hereditary cardiovascular disease screening, particularly among the minority that enacted this principle by attempting to control as much of their child's life as possible to ensure health, safety, and treatment adherence. Parental responsibility is described through an emphasis on autonomy and ultimate control over their child's awareness of HeFH, the messages delivered by clinicians, and the timeline along which these are relayed as articulated by Keenan et al. (2019). Finally, it is the primary focus of theory of decision-making outlined by Mackie et al. (2015), wherein parents stated feeling responsible for balancing the opportunity to optimizing their children's long-term cardiovascular outcomes with protecting them from potential unnecessary complications. These perceived parental responsibilities can be seen as a means of actively managing how HeFH is incorporated into family life.

Future Expectations

Future expectations are an inherent aspect of how HeFH is perceived by affected adults, specifically as it relates to perceived risk for future heart disease. A full discussion can be found within the previous section "Individual Illness Perceptions of FH."

Parental Management Behaviors

Family Management Approach: Treatment Adherence Among HeFH Diagnosed Youth

Just as a family's management mindset may be partially predicated on the adherence of parents to their own HeFH treatment, so too may a family's management approach be reflected in the adherence of their children. Treatment recommendations for the primary prevention of CVD are the same for youth and adults with HeFH, although pharmacotherapy

is often less aggressive among youth possibly related to fewer approved lipid-lowering agents and concern for short- and long-term side effects (de Ferranti et al., 2016; Gillman & Daniels, 2012; Panel, 2011).

Child dietary adherence. When asked to report perceived adherence to recommended diet, parents participating in one study of Norwegian children were less likely to perceive their children as adherent than their child themselves (42.5% versus 52-60%); however, research attempting to quantify dietary adherence among children with FH have had varied findings (de Jongh et al., 2003). Tonstad and Siversten (1997) found that slightly more than 75% of their sample of 172 treated FH children aged 9-17 met the recommendation of <30% energy from total fat, and Molven and associates (2013) found that 65-88% of their 28 children with FH aged 12-14 years consumed low or no-fat dairy products, lean meats, and substituted butter for very low saturated fat margarine. In these categories, these HeFH children appeared to make better dietary choices than their non-FH peers as measured by a summative index developed by the researchers. Moreover, dietary patterns of consumption did not appear to vary between HeFH children aged 12-14 years and HeFH young adults aged 18 – 28 years, suggesting good dietary habits learned in childhood may correspond with good habits in adulthood (Molven et al., 2013). More recently, a self-report macronutrient survey among a small cohort of HeFH children (5-18 years) in Norway found them to consume an average of 12% of energy from saturated fats, well above the 7% recommended; reassuringly, this was subsequently responsive to targeted dietary counseling on fatty acid and cholesterol intake (Torvik et al., 2016). Of note, Nordic diets include higher quantities of polyunsaturated fats from rapeseed oil, low-fat margarine, and whole grains

than other regionally quantified diets; thus, dietary adherence data from these regions may not be generalizable to FH populations elsewhere (Rodríguez-Borjabad et al., 2021).

Child physical activity adherence. Thavendiranathan and associates (2007) characterized engagement in physical and sedentary activities, and attitudes towards PA, in a sample of 147 5-18 years old HeFH patients from an established pediatric lipid clinic in Toronto, Ontario. Patients responded to clinician-developed survey regarding types of PA engaged in, the amount of time devoted to physical activities per week, the availability of exercise equipment in the home, and any symptoms experienced while exercising; anthropometric measurements were obtained via chart review. Patients generally tolerated exercise well, with 86% of patients reporting no difficulties with exercise; the difficulty most frequently reported was shortness of breath (24%). Patients also expressed positive attitudes towards PA, with 80% calling it "fun." On average, patients participated in physical activities for 10 hours weekly (range 0-88 hours), and sedentary activities for 20 hours weekly (range 0-73 hours) (Thavendiranathan et al., 2007). Despite receiving regular diet and exercise counseling, this sample had a higher proportion of obesity than the general Canadian population (21% compared to 11.8-13.5%). Of note, only age ($r = 0.2$, $p = 0.02$) and time spent by other family members in PA ($r = 0.24$, $p = 0.003$) were associated with time spent in PA by the patients (Thavendiranathan et al., 2007).

Child adherence to statin treatment. Quantification of adherence using the pill count method has been published in three safety and efficacy trials of statins in youths, finding rates of adherence ranging from ≥70% in 85% of enrollees, to an average of 93% across all enrollees (Gandelman et al., 2011; Knipscheer et al., 1996; Lambert & Lupien, 1996). The largest trial enrolled 72 HeFH youths; all trials monitored compliance over 8-12

weeks. The short monitoring periods, small sample sizes, and treatment monitoring protocols specifically intended to assess statin efficacy raises questions regarding the generalizability of their findings to HeFH youth monitored bi-yearly by providers in the wider population.

Population-level adherence to lipid lowering treatment among youth with hypercholesterolemia has recently been evaluated through the analysis of medication disbursement data in an employer-based health insurance claims database. Joyce and associates (2016) reviewed trends in prescription filling, refilling, and lapses among 8,710 children aged 8-20 years prescribed statins or other lipid lowering medications from 2005-2013. Defining an episode of non-adherence as the passage of 90 days or more from the anticipated end of the last filled prescription, researchers found that 87% of youths prescribed a lipid lowering treatment experienced at least one episode over the 1st 30 months after treatment initiation (Joyce et al., 2016). Of those who filled an initial statin prescription, 76% (n = 3,366) also filled their second prescription (Joyce et al., 2016). Notable trends include a greater likelihood of non-adherence among older patients (Hazard Ratio = 1.21 [95% CI 1.07–1.38]), and a lower likelihood of non-adherence among patients treated with statins compared to other lipid-lowering therapies (Hazard Ratio = 0.58 in the 1st 12 months {95% CI 0.56–0.61}; Hazard Ratio = 0.81 after the 1st 12 months [95% CI 0.68–0.96]) or having a diagnosis of dyslipidemia compared to other diagnoses where cholesterol may require clinical lowering (Hazard Ratio = 0.61 [95% CI 0.57–0.65]). A major limitation of this study is the lack of specificity regarding youths with HeFH, likely in part related to the lack of an ICD-9 code for HeFH prior to 2016. However, given the relative infrequency of lipid lowering prescriptions among youths in general, it is likely that this analysis encompasses some significant proportion of those with HeFH (Cox et al., 2008). Additionally, this study

cannot account for prescriptions written by healthcare providers but not filled by families; thus, adherence may be incompletely estimated.

Parenting Philosophy

Parenting Style and Obesity in Youth. Parenting style has been previously identified by PMSF researchers as potentially relevant in preventive-disease populations. In their concept analysis and theoretical synthesis of FMSF with the management of pediatric obesity, Jang and Whittemore (2015) explicitly cite the literature on parenting style, weight, and dietary and lifestyle behaviors in an argument for theoretical consistency between parenting style and the FMSF concept of parenting philosophy (Jang & Whittemore, 2015). Given the overlap in behavioral recommendations for youth with obesity and youth with hypercholesterolemia, and the lack of literature on parenting style as it relates to children with hypercholesterolemia, it is relevant to examine the current conceptual relevance of parenting styles in the obesity context.

In their seminal work, Rhee, Lumeng, Appugliese, Kaciroi, and Bradley (2006) used national data from the Study of Early Child Care and Youth Development to investigate the association between parenting style and obesity status (BMI >95th%tile) among 872 families raising 1st graders. Participating families were primarily white (82.8%), mothers had an average of 14.6 years of education, and the mean family income/need ratio was 3.6 (poverty level ≤1) (Rhee et al., 2006). Parenting style was defined as the interaction between self-control expectations and maternal sensitivity to children's needs, and styles were assigned by dichotomizing each measure at the mean, then combining them in a manner consistent with Baumrind's styles. First graders with authoritarian mothers had substantially greater odds of being obese than those with authoritative mothers (OR 4.88; 95% CI 2.15-11.1); children

with permissive or negligent mothers also experienced greater odds of obesity, although not to the same extent (permissive OR 2.84, 95% CI 1.1-7.35; negligent OR 2.67, 95% CI 1.12-6.38). Unfortunately, Rhee et al. (2006) do not specify what control variables were included in the final regression models, making the strength of these findings difficult to assess.

More recently, research has suggested that the associations between weight status and parenting style may be more complex; the work of Fuemmeler et al. (2012) provides a strong example. Although no general associations between parenting style and BMI were identified among 15-year-olds participating in the National Longitudinal Study of Adolescent Health, adolescents from families with authoritative and neglectful parents showed an increased slope for weight gain between ages 15-22 and positive parameter estimates in quadratic models, suggesting slower rates of weight gain but less weight stabilization with age (Fuemmeler et al., 2012). Separate models of weight trajectory accounting for parental education and single/dual parent households identified clear differences across demographics: parenting style did not appear to predict weight trajectory among Hispanic adolescents of either sex or black males. However, Hispanic males with authoritarian parents and Black males with negligent parents both had lower BMI than those with authoritative parents at age 15 (Multivariable-adjusted linear regression coefficient Hispanic: $\beta = -2.04$, SE = 0.69; Black: $\beta = -2.69$, SE = 1.05; all p-values < 0.05), and Hispanic males with authoritarian parents had a significantly lower BMI intercept than White males with parents utilizing any style (Fuemmeler et al., 2012). Having an authoritarian parent was associated with an unhealthy weight trajectory among both black and white adolescent girls (Linear model: Black $\beta = -0.49$, SE = 0.2; White $\beta = -0.27$, SE =0.1; Quadratic model: Black $\beta = 0.04$, SE = 0.2; White $\beta = 0.03$, SE = 0.01; all p-values < 0.05) and neglectful parents was

associated with unhealthy weight trajectory among white boys (Linear model: $\beta = -0.65$, SE $=0.25$; Quadratic model: $\beta = 0.04$, SE $=0.02$, $p < 0.05$). The ethnic variations and potential longitudinal effects of parenting style are corroborated by the findings of Olvera and Power (2010), who found obesity increased at a slower rate among school aged Mexican-American children with either authoritative ($F [1, 27] = 4.32$; $p < 0.05$) or authoritarian ($F [1, 25] = 6.87$; $p < 0.05$) mothers compared to indulgent mothers, with no difference between indulgent and neglectful parenting (Olvera & Power, 2010). Researchers have also identified income as a moderator of the relationship between parenting style and obesity (Kakinami et al., 2015; Lane et al., 2013). These findings highlight potential variations in the effects of parenting style across demographic factors, its implications across the lifespan, and its relevance as a moderating influence on weight outcomes.

Parenting Style and Weight-Related Behaviors in Youth. Although the association between parenting style and weight has been relatively consistent, the potential mechanisms underlying the relationship are less clear. Kremers, Brug, de Vries, and Engles (2003) published the seminal investigation into the relationship between quantifiable dietary intake and parenting style, hypothesizing that general parenting atmosphere could shape child receptiveness to direct dietary related parenting practices, evidenced through varying levels of intake. Accounting for the effects of gender, age, and religiosity, they determined that the average Dutch adolescent raised by an authoritative parent ate an average of 76 grams more fruit per day than those raised by an authoritarian or neglectful parent, and 45 grams more fruit daily than those with permissive parents (Tukey's Range method, $p < 0.05$). Adolescents with authoritative parents also scored statistically significantly higher on measures of attitudes towards fruit, fruit eating self-efficacy, and social support for eating

fruit, than those raised by any other parenting style (Kremers et al., 2003). These findings were subsequently contradicted by De Bourdeaudhuij and colleagues (2009) in a survey of 3760 parents of 11-year-old children from the four European Union nations (Spain, Portugal, Belgium, the Netherlands) on fruit and vegetable intake, in-home accessibility, parental modeling and encouragement, and family food rules. No significant differences were identified across parenting styles pertaining to daily portions of fruits or vegetables, but they were found to have a significant effect on parental encouragement to eat fruit, demands to eat fruit, and availability of fruits and vegetables in the home (F-value range 3.45-5.15; $p < 0.01$). The effect of fruit consumption varied by parenting style as well; however, no other interactions between nation and parenting style arose. In general, nation of origin appeared to have a substantially larger effect on all variables (de Bourdeaudhuij et al., 2009). Multiple studies have since considered fruit and vegetable intake, as well as sugar-sweetened beverages, snacking, non-core food consumption (sweets, fatty foods), and habitual breakfast eating, all with inconsistent results (Parletta et al., 2012; Rodenburg et al., 2012; Vereecken et al., 2009; Xu et al., 2013).

Parenting styles have also been implicated in the exercise and other lifestyle-related activities children engage in. Schmitz and colleagues (2002) determined that authoritative parenting predicted the likelihood that adolescents in Minnesota ($n = 3798$) reported participating in strenuous PA for at least 20 minutes three times weekly; however, such predictions varied by the sex of the parent and the child. Having a non-authoritative mother predicted a greater likelihood of reporting higher intensity and/or more frequent PA for both boys and girls (odds not reported, $p < 0.05$ and <0.001, respectively). Paternal parenting style was not significantly predictive of PA in daughters or sons (Schmitz et al., 2002). Saunders,

Hume, Timperio, & Salmon (2012) also identified variation between parenting styles and the types of physical activities adolescent girls engaged in (n = 222). Controlling for family status (single versus dual-parent households), girls of authoritative parents more frequently engaged in organized athletics (β = 0.27, [95%CI 0.02, 0.52]), and children of authoritative and indulgent parents less frequently engaged in freestyle walks and bicycle riding (β = -0.45 [95%CI -0.88, -0.02], β = -0.56 [95%CI -0.92, -0.20] respectively). In bivariate regressions, statistically significant associations were not observed between accelerometry-measured Moderate or Vigorous PA and any parenting style (Saunders et al., 2012). However, family status moderated the relationships between parenting style and PA, particularly for single-parent households. Girls of parents with more authoritative parents in these homes engaged in significantly more MVPA daily, while girls of parents with less authoritarian and neglectful tendencies also engaged in more MVPA daily, and more frequently engaged in walks and bicycle outings. Girls of single parents who were more permissive took part in significantly less MVPA daily (Saunders et al., 2012).

The relevance of parenting styles on both weight and weight-related health behaviors was recently confirmed by meta-analysis. Pinquart (2014) located 156 publications seeking associations between parenting styles and weight. Studies looking at the outcomes of fruits and vegetable consumption, snack intake, drinking sugar sweetened soft drinks, and fast foods; global healthy eating; and PA (self-report or accelerometer versus sedentary behavior or time) were also included. The definition of parenting style varied by publication, with the three most frequently used instruments being employed in 7-9 studies each; some using an estimation of Baumrind's styles (authoritative, authoritarian, etc.), others measuring the theoretical concepts underpinning these styles (ex. warmth, responsiveness, demandingness,

psychological control) (Pinquart, 2014). Overall, effect sizes were small but statistically significant, with neglectful parenting having the greatest negative impact on weight among the Baumrind-based concepts (Hedge's g = 0.12 [95%CI 0.02, 0.21]; Z = 2.38, p < 0.05). Authoritative parents displayed the greatest positive impact on weight (Hedge's g range -0.08 to -0.11; Z range -2.81 to -4.02; p <0.01). Similar relationships were reported for dietary and PA outcomes (Pinquart, 2014). High heterogeneity between studies suggested the need for moderation analyses, which identified several factors independently affecting the strength of the parenting style/weight relationship. Effect sizes were generally larger in studies using longitudinal designs, employing dichotomous weight cutoffs (obese/non-obese), enrolling preschool or school-aged children, and non-diverse samples (Pinquart, 2014).

Parenting Style and Treatment Adherence in Chronically Ill Youth. Parenting style also affects adherence to treatment among children and adolescents with chronic illnesses, although relationships tend to vary in relation to several methodological and measurement differences. In general, parents of children with chronic physical conditions appear to have lower quality parent/child relationships than the parents of well children (Pinquart, 2013). In a recent meta-analysis of 325 papers comparing parenting dimensions between families with healthy and chronically ill youth, the parents of chronically ill children appeared to show a particular tendency toward lower levels of responsiveness (Hedge's g = -0.22, p <0.001) and authoritative parenting (Hedge's g = -0.13; p < 0.01), and greater demandingness (Hedge's g = 0.18, p <0.001), authoritarianism (Hedge's g = 0.24, <0.05), overprotection (Hedge's g = 0.39, p <0.001), and neglectful parenting (Hedge's g = 0.51, p <0.05) (Pinquart, 2013). These findings validate the need for continued investigations into

sub-optimal parent/child relationships among families affected by various chronic illnesses such as in the current study.

The literature also suggests potentially significant relationships between parenting styles and treatment adherence among chronically ill children. Alone and compared to children with authoritarian and permissive parents, chronically ill children with authoritative parents or parents scoring highly on measures of warmth, caring, and acceptance consistently report higher adherence and better levels of physiologic disease control (Butler et al., 2007; Davis et al., 2001; Gau et al., 2006; Greene et al., 2010; Monaghan et al., 2012; Saletsky et al., 2014; Sheehan et al., 2012; Shorer et al., 2011; Stein et al., 2005). Authoritarian parenting was correlated with lower adherence to treatment, particularly when applied specifically to child adherence behaviors, and authoritarian tactics have predicted lower direct-measured medication adherence (Davis et al., 2001; Ievers-Landis et al., 2005; Saletsky et al., 2014). Children of permissive parents appear to generally adhere more poorly to disease management, however. Collective results suggest age interacts with this style such that greater authoritarianism in youth and greater permissiveness in adolescence may in fact predict better adherence (Greene et al., 2010; Saletsky et al., 2014; Shorer et al., 2011). This suggests a potential temporal change component to effective parenting style. Other personal factors such as family race and ethnicity, education, child developmental stage, and socioeconomic status have also been associated with parenting style tendencies, and in some cases moderate the direction of the relationship between style and adherence outcome (Davis et al., 2001; Ievers-Landis et al., 2005; Lane et al., 2013; Saletsky et al., 2014).

As with obesity, parallels exist between the experiences of youths with HeFH and those managing more immediate chronic diseases, such as diabetes. Children and adolescents

with these conditions generally feel well day-to-day and, in this context, they are asked to monitor their dietary intake, engage in PA, and take medication for the prevention of disease-related consequences. Long-term CVD may result from non-adherence for all these affected youths; however, those with Type I Diabetes may also experience potentially life-threatening episodes of ketoacidosis, adding urgency to adequate treatment. Davis and associates (2001) first investigated the parenting style/diabetic adherence relationship in a sample of preschool and school aged children (n = 55) and their parents. Parental warmth showed a moderate to large correlation with self-care adherence ($r = 0.56$, $p < 0.001$), and parental restrictiveness showed a moderate correlation with HgbA1c ($r = 0.36$, $p < 0.05$). In hierarchical regression models accounting for demographic, socioeconomic, and parenting style variables, only parental warmth predicted self-care adherence (adjusted $R^2 = 0.28$; $b_{warmth} = 0.32$, $p < 0.001$); however, no parenting style variables predicted adequate glycemic control (Davis et al., 2001). As with the literature on obesity and wellness behaviors, such results have not been consistent across studies; the results of Butler et al. (2007) are frequently cited in contradiction. Applying the same diabetes self-care adherence instrument as Davis et al. (2001) in a sample of 78 adolescents, they identified no significant associations with measures of maternal psychological control, firm control, or acceptance (Butler et al., 2007).

In the only study of its kind, Saletsky and colleagues (2014) measured the effects of parenting style on nurse-observed medication adherence (percent of tabs taken) in a sample of racial and ethnically diverse adolescents from baseline diagnosis with Type II Diabetes through their first year of treatment (n = 63 at 12 months). Whereas adolescents perceived their parents becoming less authoritarian and more permissive over time, parents perceived their parenting style as stable (Saletsky et al., 2014). In regression analysis of adolescent

perception of parental authoritarianism related to diabetes self-care tasks, accounting for the effects of gender and race/ethnicity, baseline perception predicted lower adherence from months 8-12 (β = -15.27 [95% CI -29.64, -0.90]; p = 0.036), and permissiveness in the relationship at baseline more generally predicted increased adherence from months 8-12 (β = 25.4 [95% CI 6.74, 44.07]; p = 0.008); however, parental perception of parenting style did not predict adolescent adherence in any model. As Type II Diabetes is perhaps the best direct parallel with HeFH, these findings may be particularly informative.

Summary

The purpose of this descriptive study was to examine the relationship between parental illness perceptions, parenting style, and adherence to treatment recommendations among families with youth aged 2-18 years diagnosed with HeFH. It supplemented the literature on a variety of under-investigated areas related to HeFH: in addition to supplementing the body of descriptive knowledge on dietary, lifestyle, and medication adherence behaviors, it examined the relationships between parental perceptions of HeFH and their perceptions of their child's treatment adherence. Moreover, the results of this study quantified parental illness perceptions in a fashion consistent with the views expressed in the qualitative literature on the HeFH experience by conjoining perceptions of HC and CVD risk. Consistent with the FMSF, parenting style was investigated as a potential means through which parents convey their attitudes and beliefs regarding HeFH to the next generation. The following questions were proposed to descriptively examine the associations between these FMSF constructs and parentally observed adherence to HeFH treatments among youth:

Research Questions

1) How well do parents in HeFH-affected households believe their children with HeFH adhere to their medication and lifestyle recommendations?

2) How do parents in HeFH-affected households perceive high cholesterol (HC) and risk for heart disease (HDR) as it affects themselves or their child/family members?

3) In HeFH-affected households, what are the relationships between parental perceptions of HC and HDR and parents' observed child's adherence to treatment recommendations?

4) In HeFH-affected households, what are the relationships between parenting styles and parents' perception of HC and HDR?

5) In HeFH-affected households, what are the relationships between parenting styles and parents' observed child's adherence to treatment recommendations?

CHAPTER THREE

METHODOLOGY

Study Design

This was a descriptive study using the administration of the validated instruments described below to assess for associations between parentally observed adherence to treatment among HeFH diagnosed youth and constructs of the FMSF. Surveys were administered cross-sectionally via weblink or paper hard copy based on family preference.

Sample

Sampling frame. The recruiting clinic is the oldest and most established pediatric lipid specialty program in the Northeastern U.S., providing approximately 1200 patient visits annually. Work on a recent PICORI-funded study informed by HeFH community members suggested interest in HeFH research within this particular clinic (Mackie et al., 2015). Therefore, those recruited represent a convenient, non-probability sample.

Inclusion & exclusion criteria. Parents were eligible for inclusion if a) at least one child had received a diagnosis of HeFH (LDL >190 mg/dL, or LDL >160 mg/dL with known family history of heart disease in a 1st or 2nd degree relative), b) the child was between 2 and 18 years old, c) the child had been diagnosed with HeFH for at least 6 months, and evaluated by the recruiting specialty clinic on at least two occasions, and d) their child's second clinic visit had occurred within the past three years.

The 2-18 year age range was selected to maximize statistical power and the potential for subsamples related to developmental stage. Parental diagnosis status was gathered, as parents with HeFH may have stronger perceptions regarding their condition secondary to family narratives around CVD, and how children receive messaging from this parent may differ from the messaging from the unaffected parent (Kools et al., 2008; Mackie et al., 2015). No predefined types of treatments for HeFH, nor minimum or maximum duration between child HeFH follow up visits were applied, as differences between individualized treatment plans and clinic engagement were thought to potentially provide useful variation in adherence and/or parenting style. Restriction on minimum timeframe between diagnosis and enrollment, and minimum number of encounters at the recruiting clinic, allowed for family adjustment to new knowledge and time to integrate recommended management plans. Initial exclusion of all other chronic conditions in childhood was found to be overly restrictive of eligibility and was relaxed to allow for a few select and common well-controlled conditions (Table 1); exclusion of all potentially impactful acute conditions, such as anaphylactic allergy, allowed for conclusions related primarily to HeFH to be isolated. Fluency in reading and writing English was essential for the survey mode of administration (see Study Implementation).

Participant screening. The inclusion and exclusion criteria described above were applied to multiple research databases and clinical data pulls to ensure all eligible families from the recruitment clinic received one, and not more than one, invitation to participate, and that the particularly eligible child was specified.

Initial records were obtained for screening from those patients at the recruitment site identified as eligible to enroll in a national prospective FH registry. Additionally, a database

designed for quality improvement initiatives was utilized to identify patients meeting age criteria at time of last visit, and either a.) taking lipid lowering medication at that visit or b.) have had at least one LDL-C ≥160 mg/dL. Finally, Medical Record Numbers (MRNs) were requested from the clinical research informatics group at the recruitment site for all patients seen at the clinic on or after Dec 1, 2017 and assigned an FH-associated ICD 10 diagnosis code (E78. 00/E78.01/E78. 5/Z83.42). With each additional database or data collection for screening, duplicate/prescreened MRNs were removed.

All unique MRNs underwent chart review by the PI to determine eligibility; those patients whose eligibility was unclear due to a.) a lack of reference to HeFH in their record or b.) the presence of an additional and potentially excluding condition underwent secondary review by a recruitment site clinician. In the event of multiple HeFH+ children within one household, the eldest eligible child was referenced in recruitment materials and all other children within the household excluded; parents of eligible patients were subsequently contacted. When providing survey responses, participating parents were instructed to consider their eldest child, or in the event of multiples, their child with the highest cholesterol levels.

Study Implementation

The survey instruments selected for this project were self-administered at locations of participants choosing, either by web-based or mailed paper option based on participant preference. A computer assisted personal interview (CAPI) method was employed to administer the web survey using the secure, HIPPA-compliant, and cost-effective database. Multiple means of contact were utilized as described in Table Two. In total, potential

participants received a total of seven contacts over an 8-week period, including 3-4 mailings, one email where addresses were available, and three telephone follow-up calls. This schedule allowed for reminders without overburdening those who elected not to opt-in, provided non-electronic options to account for privacy and/or technology literacy concerns, and maximized cost efficiency. Due to social distancing restrictions imposed on research during the time of COVID-19, no in-person recruitment took place for this study.

Scientific Approvals and Human Subjects Protections

This study received approval for scientific validity from the Cardiology Department at Boston Children's Hospital, and Internal Review Board approval via expedited review by Boston Children's Hospital (BCH IRB-P00027929). The University of Massachusetts Boston entered into an agreement to rely on the Boston Children's Hospital IRB for this project (BCH IRB-RL00027929-1).

Informed Consent and Incentives

Implied consent was approved for this study; thus, participants provided consent to participate by way of participation itself. Prior to initiation of the research questions, participants reviewed a synopsis of risks and benefits to participation and were asked to confirm their understanding of this information prior to proceeding by way of a check mark. Participants received a $25 gift card to Amazon.com for participating once the entire survey was completed, sent by way of USPS to preserve privacy.

Addressing Non-Response

A variety of metrics were maintained as a guide for estimating nonresponse. Emails returned with the text "STOP" in the subject line or body, returned letters marked with "I am not interested in participating," and telephone conversations wherein the respondent stated

that the family did not wish to participate were designated “refusal to participate;” emails that “bounced”, letters that were marked “return to sender,” and telephone numbers without voicemail were designated “noncontact,” as were emails, letters, and telephone voice messages never returned nor acted upon. Ultimately, whether the unreturned and unacted upon invitations are refusals or noncontacts is less relevant than having made the attempt to contact the case in the first place; given the nonrandom sample, participants are not representative of the overall FH population. Thus, attempting to include the largest possible sample for statistical power was of great importance.

Addressing Data Safety and Privacy

The data collection platform was hosted by parent hospital of the recruitment clinic, and was password protected, on an encrypted network, and met HIPPA privacy requirements. Identifiers provided to participants were for the purposes of providing remuneration only and were randomly assigned without relation to the screening or recruitment process. Participants were advised that assigned identifiers were not required to be entered after participation if participants preferred to avoid any risk of possible identification. Participant screening logs and recruitment trackers were password protected and stored on a HIPPA-compliant server in a folder requiring administrative approval for access. Letters and postcards with information pertaining to eligibility based on diagnosis were sent in opaque envelopes. Participant identifiers were removed from downloaded data, which was subsequently saved on an encrypted thumb drive for backup preservation.

Administered Measures

Demographics, Social History, Medical History, and Family History

Data on sociodemographics were collected via self-report (See questionnaire in Appendix, Figure 5). Participants were asked regarding race and ethnicity, education, gender, child age, and child gender. Respondents were also asked about their family structure: marital status, and total number of children in household.

Items pertaining to history and management of HeFH included diagnosis status of the respondent or child's other biological, family history of HC and/or cardiovascular event in the child's first and second degree relatives, child's age at diagnosis with HC, and age at start of treatment for HC. Where a family had multiple children followed by the recruitment clinic, the responding parent was instructed to answer all questions relative to the oldest child, or the child with the highest cholesterol in the event of multiples. To allow for categorization and control by exposure to FH counseling, parents were asked how many visits their child has had with a specialist physician or nurse practitioner, and a Registered Dietitian or Nutritionist, in the past two years.

The Brief Illness Perception Questionnaire

The Brief Illness Perception Questionnaire (B-IPQ) is a generic 9-item tool composed of 8 separate Likert scales and one rank order item intended to measure the major concepts of the Common Sense Model (timeline, personal control, treatment control, identity, consequences, emotional response, causative factors, and understanding; see Chapter 2 for full discussion). Responses can be individually interpreted, or partially reverse coded to provide a summary score. Originally developed in 2006, the instrument has since been applied across at least seventeen condition and illness types, psychometrically validated in

multiple languages, given to adult and pediatric populations, and used in both cross-sectional and longitudinal investigations (Broadbent et al., 2006, 2015). Initial test-retest reliabilities at three and six weeks among renal patients attending outpatient clinic visits showed moderate to strong correlations (r = 0.42-0.73, p = 0.01-0.001); of note, correlations between the personal control measures appeared to decrease, and treatment control and understanding appeared to increase (Broadbent et al., 2006). Internal reliability has been found to be consistently acceptable across studies (Cronbach's α range 0.62-0.85), although this varies substantially between populations given the reliance of the B-IPQ on single-item measures (de Raaija, et al., 2012; Hallegraeff et al., 2013; Karatas et al., 2017; Zhang et al., 2017). Convergent validity with the IPQ-Revised – an 80-item instrument of the same CSM concepts – has been shown to be acceptable; all correlations between like concepts were significant and ranged from moderate to strong (r = 0.33-0.63, p = <0.001) in patients affected by asthma, diabetes, and chronic renal disease (Broadbent et al., 2006). Finally, the B-IPQ has been found to have both concurrent and predictive validity across a wide range of outcomes, including HgbA1c in diabetic patients, anxiety in cardiac patients after myocardial infarction, and quality of life in CHD (Broadbent et al., 2015). In a "think aloud" validation study assessing difficulties and variations in the considerations patients applied to B-IPQ items in a small Dutch study, a few items were found to require modifications to increase response accuracy; these were adopted in the current study (van Oort et al., 2011).

In the current investigation, the B-IPQ has been administered to parents relative to four separate scenarios dependent on their own reported proximity to the condition (See Figures 5 & 6). Parents reporting a personal diagnosis with HeFH received versions asking about how one's own HC and HDR was perceived; the versions received by parents without

a personal diagnosis of HeFH focused on how they perceived HC and HDR impacting the family globally. All parents regardless of their personal diagnosis status were then asked regarding how they perceive HC and future HDR as they relate to their eligible affected child. This distinction between cholesterol levels and HDR is apparent in the FH literature: behaviors may be motivated by either an immediate gratification from concrete lipid lowering (HC), or knowledge of long-term consequences (HDR) (Gooding et al., 2016; Sliwinski et al., 2017; Urke et al., 2019). Additionally, although illness perceptions of HC and HDR have each been measured in HeFH populations previously, they have not been measured concurrently, making elimination of illness perceptions as contributors to adherence challenging in the event of null findings (Brewer et al., 2002; Claassen et al., 2010; Hagger et al., 2016). Finally, in a population where diagnosis with HeFH (and use of this specific diagnostic term) is inconsistent, separation of the concepts could have more relevance than the term HeFH itself. Assessing for perceived distinctions or alignments between parental perceptions of their own condition and that of their child will allow for a fuller understanding of how parents view the impacts and consequences of HeFH on the family unit.

The Parenting Styles and Dimensions Questionnaire – Short Form

The Parenting Styles and Dimensions Questionnaire (PSDQ) was originally developed as a 62-item instrument composed of three scales measuring the concepts constructs of parenting style: authoritative, authoritarian, and permissive parenting (Robinson et al., 1995). Parents were asked to separately assess the frequency with which they and their spouse or parenting partner (if applicable) employ a variety of parenting behaviors and approaches in their interactions with their child, on a scale of 1 (never) to 5 (always); items

previously determined to assess each type of parenting style were then summed and averaged for an overall score ranging from 1-5 in each respective area. In 2001, the authors published a revised instrument that has been utilized in its full length, as a 32-item short form, and multiple variations focusing on specific subdimensions or parenting styles (Robinson et al., 2001). In their 2012 review of the psychometric properties of the PSDQ, Olivari and colleagues found only 18% of researchers used the original 62-item instrument; at least 30% used the short form (Olivari et al., 2013).

The short form has been shown to retain acceptable reliability across international populations, parents of children ranging in age from toddlerhood through adolescence, and in both maternal and paternal samples, although the permissive scale consistently shows lower reliability than the scales for authoritarian and authoritative behaviors (Olivari et al., 2013). Most researchers employing the PSDQ have focused on self-assessment of parenting behaviors, with reliability scores generally lower on scales where parents have been asked to rate a partner (Olivari et al., 2013). Psychometric validity has been confirmed through the translation of the PSDQ short form into Spanish, Chinese, Lithuanian, and Portuguese versions (Batool & Mumtaz, 2015; Kern & Jonyniene, 2012; Monge-Rojas et al., 2021; Oliveira et al., 2018; Porter et al., 2005; Wu et al., 2002). In recent years the PSDQ short form has been employed in multiple investigations into relationships between parenting style, health behaviors such as fruit and vegetable intake, screen time, and healthy eating practices, and healthy childhood weight (Detnakarintra et al., 2020; Goodman et al., 2020; Howe et al., 2017; Monge-Rojas et al., 2021; Sutter et al., 2019).

Adherence Measures 1 & 2: Shea's Adaptation of the Morisky-Green-Levine Medication Adherence Test, and 30-Day Recall Visual Analog Scale.

The Morisky-Green-Levine (MGL) test is a widely used, public domain, general adherence measure consisting of four "yes/no" answer types regarding willful and accidental skipping of medication doses (Morisky et al., 1986). Initially conceptualized for categorical scoring where zero "yes" responses equated to high adherence, one to two equating to moderate adherence, and three or more equating to poor adherence; it has also been used as a dichotomous (zero "yes" responses correspond with perfect adherence, one or more corresponds to imperfect adherence) and a continuous measure. In its original development, applied to antihypertensive medications adherence in a diverse patient population, it was found to have acceptable internal reliability (α = 0.61) and predictive validity in identifying individuals with controlled blood pressure over time (sensitivity = 0.81, specificity = 0.44; +PV = 0.75, -PV = 0.47) (Morisky et al., 1986). Researchers have utilized the MLG test to assess pharmacotherapy adherence among FH individuals, although to date its psychometric properties within this population have not been published (Benson et al., 2016; Korneva et al., 2019; Oñatibia-Astibia et al., 2020). In studies of more general atherosclerosis prevention and statin adherence, its reliability and validity has varied; Beyhaghi and associates (2016) calculated a Cronbach's α = 0.49 among a nationwide sample of elderly individuals at risk for atherosclerosis (Beyhaghi et al., 2016). Bermingham et al. (2011) found the MGL test positively identified greater adherence among primary care patients meeting LDL-lowering goal compared to those who did not (OR 1.9, CI 0.8-4.7, p = 0.036), however Dunbar-Jacob et al. (2012) found the tool better able to identify nonadherence than adherence to treatment

among those who had minimal cholesterol change while randomized to lovastatin (sensitivity 34.2, specificity 0.80) (Bermingham et al., 2011; Dunbar-Jacob et al., 2012).

One advantage of the MGL test is its adaptability, both in wording and applicability across patient populations, while recognizing the potential for such modifications to result in variations in the reliability of the tool (Nobles & Erickson, 2018). Shea and colleagues (1992) made small wording modifications to the original four instrument items and added one additional item encapsulating missed doses "for any reason," and found improvement of the internal reliability (α = 0.71 vs 0.61); Dunbar-Jacob et al. (2012) found these changes to offer substantial improvements in associations between LDL change and lovastatin use compared to the original MGL test (Dunbar-Jacob et al., 2012; Shea et al., 1992). While the Shea et al. (1992) adaptation and MGL test were highly correlated (r = 0.809, p = < 0.001), the Shea adaptations predicted change in LDL (OR 2.68 [CI 1.06-6.78], p = 0.035), and had improved sensitivity (73.7%) and specificity (48.9%) comparatively (Dunbar-Jacob et al., 2012). Only electronic adherence measurement showed better predictive validity. Thus, the Shea adaptations were employed here. While having been shown to increase the likelihood of greater adherence reporting, the timeframe of reporting has also been lowered from the original three-months to thirty days, to be consistent with inquiries regarding adherence made in the clinical setting.

In addition to the adapted MGL test, a visual analog scale (VAS) of adherence was also administered. Families were asked to mark on a scale of 0-100 approximately what percentage of medication doses were taken in the past thirty days, with 100% equaling perfect adherence. VAS provide a global assessment of adherence on a continuous scale, found to correlate well with the Shea adaptations, and offering additional statistical

possibilities in conjunction with it (Dunbar-Jacob et al., 2012; Finitsis et al., 2016). These two outcome measures are observational, and thus are open to error through deliberate and unintentional misreporting. However, in the context of the time and resource restrictions of the project, these means of reporting were most feasible.

Adherence Measures 3 & 4: The Preventive Cardiology Lifestyle Survey.

The Preventive Cardiology Lifestyle Survey (PCLS) combines dietary recall of food items containing relevant macronutrients (saturated fats in particular) with questions regarding regular PA, sedentary habits, and sleep hygiene. Developed by a Registered Dietitian in the PCP, it has been validated against the Block FFQ and the Physical Activity Block Plus Rapid Eating Assessment for Participants (Griggs et al., 2018), and was chosen for its familiarity to families in this sample. The PCLS asks families to consider hours per week participating in a variety of different activities (ex. Walking, gym class, sports). A single item then guides families to answer item on total PA weekly, with responses ranging from "never" to "10+ hours per week." The current study used this item to quantify PA as a continuous variable.

Frequency of servings of foods high in saturated fat, cholesterol, or fried foods (for example, "regular fat ice cream," "Fresh red meats including beef, lamb, pork") were also tallied for the assessment of diet-driven LDL elevation. Families were asked, "Thinking back over the past thirty days, please check the response that best represents how often your child ate a serving of the following foods" on a scale of "Never/<1 day/1-2 days per week/3-4 days per week/5-6 days per week/1 time per day/2 or more times per day." Responses were coded from 0-6, with higher numbers equating with greater numbers of saturated fat servings and totaled into one overall continuous score of weekly saturated fat servings, then added to

produce a score of 0 to 42, where lower scores equate to lesser frequencies of saturated fat intake.

Post-Survey Qualitative Input.

At the completion of the survey, participants were given the opportunity to provide additional open-ended information to the researchers, specifically: "Is there anything else you feel is important, or would like us to know about your or your child's experience with genetically inherited high cholesterol?" Response was not required.

Conceptual-Theoretical-Empirical Structure

Figure 2 depicts the relationships between the FMSF concepts and their relationships with the empirical measures selected for their measurement to be explicated below. This investigation was based in the grand theory of Socioecological Systems, which conceptualizes the social environment similarly to a biological system wherein each social unit is influenced by concentric layers of societal environment with increasingly nonspecific affects (Brofenbrenner, 1979). It focused specifically on the smallest sphere of influence in an individual's life, termed the microsystem, wherein the most concrete relationships between social units exist. The FMSF provided the theory of how one microsystem unit – the family – approaches the management of illnesses and disease processes in their offspring. The "sociocultural context" postulated by Knafl et al. (2003) represents the incorporation of other Socioecological Systems spheres of influence into the theory, which then affect the interworking of the family itself. For this study, measurement of these influences included the child's age, age at first diagnosis, and length of treatment; parent described race and ethnicity, education, and family relationship structure; and the history of heart disease among

the child's parents and second-degree relatives. Elements of the parent's "definition of the situation" was assessed through their perceptions of how affected the child is by their condition (both HC and future HDR) using the B-IPQ, as were the "perceived consequences" of HeFH. "Management behaviors" focused on the parenting style employed, as measured by the PSDQ. Finally, addressing the role of the individual parent in the FMSM, the B-IPQ was also employed to characterize the parent's perception of their own hypercholesterolemia and HDR if the respondent endorsed the diagnosis themself, or their perceptions of how it affected the family unit when respondents did not.

Data Management & Statistical Methods

Sample Size

Given that the absolute standardized effect size |ES| based on correlational analyses for variables of illness perception on outcome measures of adherence behaviors commonly range from 0.15-0.30, identifying a similar effect with a two-sided $\alpha = 0.05$ and $\beta = 0.20$ required a sample size ranging from 88 – 349 families respectively. Our study sample consisted of 51 individuals; using the same alpha and beta criterion, this study was powered for effect sizes $r = 0.39$ or greater (Hully et al., 2013; Machin et al., 1997; Rosner, 2011).

Data Entry and Cleaning

Ten responses obtained via paper were manually entered into the RedCap database. Where written responses included a range of responses (ex. 7-9 hours weekly exercise), the midpoint of the range was entered into the database. Data was then exported from the data collection platform into an Excel document and uploaded into STATA/BE 17 (College Station, Tx). Once uploaded, data was initially cleaned through entry of "99" into fields

where skip logic was employed or marital responses corresponded with a lack of parenting partner for skipped PSDQ responses, thus designating these fields not applicable. All missing data not designated as not applicable were denoted with a period. Responses were assessed for logical consistency, and deleted where inconsistencies were found (ex., both answers were removed where a respondent endorsed early heart disease in a first-degree family member, but then listed '0' for the number of affected first-degree family members). One respondent record was excluded from analysis secondary to reporting on a child not meeting criteria for age. A summary on missing data was generated for each variable, finding missingness ranging from 2% (equivalent of one response) to 8%; responses to the PSDQ partner scales contained the most missing data.

Data Analysis

Research question 1.) How well do parents in HeFH-affected households believe their children with HeFH adhere to their medication and lifestyle recommendations? Research question one was addressed using responses to the adapted MGL Medication Adherence Test ("adapted MGL"), the 30-Day Recall VAS ("VAS"), the PCLS saturated fat recall items, and the PCLS physical activity item. The adapted MGL Test was summed by percentage for each item, then tallied for a potential score of 0 – 5, with lower scores indicating more adherent behavior. A composite score for weekly average servings of saturated fat captured by the PCLS were summed into an overall score with a range of 0 – 36. All instruments and items were summarized using descriptive statistics including means, medians, interquartile ranges, standard deviations, and percentages as appropriate.

Research question 2.) How do parents in households affected by HeFH perceive high cholesterol and risk for heart disease as it affects themselves or their child/family members? Research question two was addressed using responses to the B-IPQ. Respondents were classified as having hypercholesterolemia or not, i.e., either HC+ or HC-. Responses to the B-IPQ items pertaining to reference children were not divided by the cholesterol classification of their participating parent. B-IPQ items were first tabulated into medians and interquartile ranges. In view of small sample sizes and lack of normality in the distribution of data, intra-respondent responses to the HC and HDR illness perception scales were then compared using the Wilcoxon's Sign Rank test; differences between items of parental perception of self/family and parental perception of children were similarly analyzed. B-IPQ scales of HC+ versus HC- respondents for each concept (HC and HDR) were compared using the two sample Student's t-tests. Also in view of small sample sizes and lack of normality in the distribution of B-IPQ data, Spearman's correlation coefficient matrices were next calculated correlating concepts of interest by family member (ex. perceptions of HC by perceptions of HDR within HC+ respondents, HC- respondents, and respondent perceptions of their children), and within concepts between family members (ex. perceptions of HDR among HC+ respondents vs. HC- respondents) for further elucidation of intrafamilial and intra-conceptual associations. The goal of the correlation analyses was to elucidate intrafamilial and intra-conceptual associations.

Research question 3.) In HeFH-affected households, what are the relationships between parental perceptions of high cholesterol and risk for heart disease and parents' observed child's adherence to treatment recommendations? Research question three was addressed using responses to the B-IPQ, the adapted MGL, the VAS, the PCLS saturated fat

recall items, and the PCLS physical activity item. Spearman's correlation coefficient matrices were calculated to assess the magnitude, direction, and significance of relationships between each scale of the six adaptations of the B-IPQ (HC+ B-IPQ of HC, HC+ B-IPQ of HDR, HC- B-IPQ of family HC, HC- B-IPQ of family HDR, Respondent perceptions of child HC B-IPQ, Respondent perceptions of child HDR B-IPQ) and the four adherence outcomes (the adapted MGL, the VAS, the overall saturated fat frequency score, and the reported average weekly hours of PA).

Research question 4.) In HeFH-affected households, what are the relationships between parenting styles and parents' perception of high cholesterol and risk for heart disease? Research question four was addressed using responses to the PSDQ and the B-IPQ. The PSDQ was scored for each individual and parenting partner (as applicable) along the parameters of authoritarian, authoritative, and permissive tendences resulting in a score range of 1 – 5, where higher scores indicated greater frequency of use of representative behaviors (Robinson et al., 2001). Scores for overall household parenting environment were calculated by summing the score for each parameter within a respondent and respondent-reported parenting-partner dyad and dividing by two; in the case of single parents, scores for the responding parents were included whole as representative of all parenting in the home. Medians and interquartile ranges were reported for survey respondents, respondents' parenting partners, and intrahousehold averages. Spearman's correlation matrices were then computed to assess the magnitude, direction, and significance of relationships between each scale of the six adaptations of the B-IPQ (HC+ B-IPQ of HC, HC+ B-IPQ of HDR, HC- B-IPQ of family HC, HC- B-IPQ of family HDR, Respondent perceptions of child HC B-IPQ,

Respondent perceptions of child HDR B-IPQ) and the overall household parenting style score for each parameter (authoritarian, authoritative, and permissive).

Research question 5.) In HeFH-affected households, what are the relationships between parenting styles and parents' observed child's adherence to treatment recommendations? Research question five was addressed using responses to the PSDQ, the adapted MGL, the VAS, the PCLS saturated fat recall items, and the PCLS physical activity item. Spearman's correlation coefficient matrices were calculated to assess the magnitude, direction, and significance of relationships between the three household averages of parenting styles (authoritarian, authoritative, and permissive) and the four adherence outcomes (VAS, adapted MGL, overall saturated fat frequency score, and reported average weekly hours of physical activity).

CHAPTER FOUR

RESULTS

Screening & Participation

As shown in Figures 3 & 4, a total of 1940 individual MRNs were screened, of whom 297 met initial inclusion criteria. After secondary review for exclusions, and tertiary review by a second clinician where indicated, a total eligible sample of 136 individuals representing 110 families were identified as eligible. No families responded opted out of further contact by email or USPS, 10 families declined further contact by telephone. No response to phone, email, or USPS contacts were received from 27 families, another 33 were able to be reached by phone but are not known to have submitted a completed response (two responses were submitted anonymously). Three secure emails "bounced;" 15 received "read" confirmation (13.9% of total emails sent). Thirteen surveys (25%) were completed and returned via USPS. Forty-nine surveys were initiated via REDCap, with 39 completed, resulting in a total of 52 surveys (participation rate 47.3%). One survey was removed from analysis due to having responded relative to a child >18 years old, resulting in a total of 51 survey responses included.

Descriptive Findings – Participant & Family Characteristics

Demographic characteristics

Respondents to this survey primarily consisted of 42 non-Hispanic white (82.4%), middle-aged mothers (mean age 46.1, 78.4% female) (Table 3). Over 84% of respondents had obtained a bachelor's degree or higher. Most households consisted of married couples (78.4%) and two children (median 2); two respondents reported having been widowed, at least one as a direct result of FH-associated myocardial infarction (Table 26). The FH-diagnosed children described by their parents (henceforth "index child/children") ranged from 3-18 years old (mean 13.4) with more daughters than sons (29 girls [58%] vs. 21 boys [42%]). There were 36 HC+ parents (70.6%) and 15 HC- parents (29.4%), and no significant differences in age, sex, race, education, marital status, age or gender of index child, or number of children in the household by cholesterol diagnosis status of the respondent (Table 3).

Family Histories with Hypercholesterolemia & Heart Disease

Table 4 summarizes the family experience with HC diagnoses. HC+ parents had generally been aware of their diagnosis for more than a decade (84.3%), and most had received a formal diagnosis with HeFH (70%). Index children had been treated for their HeFH diagnosis for a mean of 6 years, with the mean age at diagnosis of 6.8 years. Nine families had experienced atherosclerotic heart disease in at least one parent, and more than two-in-three (66%) had experience with it through a second-degree relative (aunts, uncles, grandparents), with a total of 71 extended family members reportedly having experienced some type of event or intervention (Table 4).

HeFH Management & Treatments

Children attended visits with medical providers (MDs, NPs) approximately once yearly over the past two years (mean 2.4 visits per child); this schedule was consistent with the standard recommendations of the recruitment clinic (Table 5). Registered Dieticians were consulted less frequently (median 1 visit over past 2 years, mean 1.6 visits). Pharmacotherapy for hypercholesterolemia had been recommended to 92.2% of the index children; of those for whom it had not been recommended, two had not yet reached the consensus age for statin initiation. A total of 80% (n = 40) of the index children were taking an HMG Co-A Reductase inhibitor (i.e., statin), with 8.5% additionally taking Ezetimibe. No families endorsed using over-the-counter supplements implicated in lipid lowering (examples: DHA/EPA, Phytosterols/Phytostanols, psyllium, Red Yeast Rice).

Main Results by Study Questions

Question 1.) How well do parents in HeFH-affected households believe their children with HeFH adhere to their medication and lifestyle recommendations?

Descriptions of Parent-Reported Patient Adherence. The adapted MGL Test had acceptable internal reliability in this sample (See Table 6 with summary of internal reliability scores for all instruments). Perfect adherence, defined as no endorsement of any type of non-adherence, was reported by 35.9% of parents. No parent reported their child as intentionally skipping pharmacotherapy doses in the past 30 days due to perceived state of health; missed doses were instead attributed to forgetfulness (56.4%), carelessness (29%), or other reasons

(41%). Using the VAS, parents estimated their index child taking an average of 85.6% of total prescribed doses taken in the prior 30 days, with a median adherence of 93.5%.

No families endorsed adherence to a diet without any saturated fat. However, the majority considered their child's intake of full fat dairy, red & processed meats, and other foods high in saturated fats (innately or due to preparation) to be only occasional (two or fewer servings weekly within each category) in any given week. Only 19.6% of respondents reported more than occasional intake of red and processed meats or other sources of saturated fat; index children consumed full fat dairy and ice cream with more frequency, with 31.4% observing servings 3-6 times a week, and nearly 6% endorsing one or more servings a day. The Cronbach's α for the six utilized items of the PCLS was 0.53.

Pertaining to PA, parents observed a mean of 8.4 hours in their child's average week, with a range of 0 – 20 hours. Almost three-quarters (73.5%) of parents reported their child to obtain at least the amount of PA currently recommended for health maintenance by the American Academy of Pediatrics (5 or more hours weekly).

Question 2.) How do parents in households affected by HeFH perceive high cholesterol and risk for heart disease as it affects themselves or their child/family members?

Analyses to address research question two were comprised of 51 parents from HeFH-affected households, of which 36 were HC+ and 15 were HC-; responses varied by +/- n = 1 based on missing data. Tests for internal reliability were undertaken on each adaptation of the B-IPQ used to measure perceptions of hypercholesterolemia and HDR in this sample and were at acceptable levels or better (Table 6).

Perceptions of High Cholesterol and Heart Disease Risk Among HC+ Parents. Parents diagnosed with HC perceived their understanding of cholesterol as high (Median 9

[IQR 7, 10]), reflected in their accurate understanding of the lifetime persistence of HC (Median 10 [IQR 10, 10]). They ranked their general affectedness by, emotionality about, and concern for HC as low (see Table 7). Personal ability to control their HC was ranked on the lower/mid-range of the 11-item Likert scale (Median 4.5 [IQR 2, 6.5]), but pharmacotherapy was perceived to be a highly effective means of control (Median 10 [IQR 8, 10]). Diet and exercise were not ranked as highly (Medians 6.5 [IQR 3, 8] & 7 [4, 8] respectively). When ranking their top three causes of their HC, all but three (92%) HC+ parents identified genes, genetics, family history, or heredity as the primary cause, and many did not provide any a second or third explanation or cause. Other causes listed included diet, lack of regular exercise, weight, stress, and liver malfunction. HC+ respondents perceived their HDR very similarly to HC (Table 8), although overall understanding of HDR was perceived to be slightly lower (HDR Understanding Median 8 [IQR 6, 9] versus HC Understanding 9 [IQR 7, 10], $p < 0.001$) and medication perceived as slightly less able to control HDR than HC (HDR medication control Median 9 [IQR 7, 10] versus HC medication control 10 [IQR 8, 10], $p = 0.003$).

Perceptions of High Cholesterol and Heart Disease Risk Among HC- Parents. Parents without hypercholesterolemia in FH-affected households perceived the effect of HC on themselves and their family similarly to that of their HC+ counterparts. The primary exceptions related to general affectedness and emotionality around hypercholesterolemia (HC- affectedness median 6 [IQR 4, 8] versus HC+ affectedness median 3 [1, 5.5], $p = <0.05$; HC- emotionality median 8 [4, 8] versus HC+ emotionality median 2 [0, 5], $p = <0.05$) and HDR (HC- affectedness median 7 [IQR 5, 8] versus HC+ affectedness median 4.5 [1.5, 7], $p = <0.05$; HC- emotionality median 8 [3, 8] versus HC+ emotionality median 3 [0,

6], p = <0.05). Additionally, HC- parents in this sample perceived their personal ability to control their family members' hypercholesterolemia more highly than that of their HDR (Personal control HC median 5 [IQR 3, 7] versus Personal control HDR median 3 [2, 5], p = 0.04).

Respondents' Perceptions of Hypercholesterolemia and Heart Disease Risk Relative to Children. Respondents perceived their children as having somewhat greater concern for HDR than hypercholesterolemia (Median 5 [IQR 3, 8] versus 6 [IQR 3, 9], p = 0.03) (Tables 7 & 8). Additionally, respondents perceived their children as having slightly more personal control over cholesterol than HDR (Median 4 [IQR 2, 7] versus 3 [IQR 3, 5], p = 0.05), and perceived more controllability of HDR through diet and exercise than for cholesterol. Regardless of diagnosis status, parents perceived their children as having poorer understanding of HC and HDR than themselves (Tables 7 & 8). HC+ parents perceived their children as being more concerned about both HC and HDR than themselves, whereas HC- parents perceived their children as experiencing less general affectedness and emotionality about HC and HDR than themselves (Tables 7 & 8).

Question 3.) In HeFH-affected households, what are the relationships between parental perceptions of high cholesterol and risk for heart disease and parents' observed child's adherence to treatment recommendations?

Analyses to address research question three were comprised of 38 parents from HeFH-affected households, of which 24 were HC+ and 14 were HC-; responses varied by +/- n = 1 based on missing data. Tables nine through 14 present the relationships between adaptations of the B-IPQ (HC+ parents on HC, HC+ parents on HDR, HC- parents on HC, HC- parents on HDR, parents' perceptions of HeFH+ children relative to HC, & parents'

perceptions of HeFH+ children relative to HDR) with each adherence item (medication adherence on VAS, medication adherence on adapted MGL test, overall frequency of saturated fat intake, and average hours of weekly PA).

Overall, saturated fat intake and adherence by adapted MGL were correlated to $p \leq 0.05$ level with three items of perception (out of 96 correlations across the six B-IPQ adaptations). Coefficient strength was moderate. Medication adherence measured by VAS, and to lesser degree average weekly hours of PA, were statistically significantly correlated with multiple aspects of condition perception across family unit subsamples (11 items $p \leq 0.05$ out of 96 correlations across the six B-IPQ adaptations). Coefficient strength ranged from moderate to strong (Tables 9 – 14). The frequency of statistical significance, strength, and directionality of Spearman correlations varied by family unit subsample (HC+ parent, HC- parent, parental perception HeFH+ child) and HeFH component (HC, HDR).

The perceptions of illness among HC+ respondents were not statistically significantly associated with adherence outcomes within the two HeFH concepts with two exceptions: HC+ parents perceiving more personal control over their HC also perceived more child forgetfulness on the adapted MGL test ($r = 0.43$, $p \leq 0.05$), and those perceiving more general affectedness by their HDR also perceived more child hours of weekly PA ($r = 0.46$, $p \leq 0.05$) (Tables 9 & 10).

Multiple perceptions of HC- respondents pertaining to their family members' hypercholesterolemia were associated with child medication adherence on VAS (r dietary control = -0.063 [$p \leq 0.05$]; r exercise control = -0.61 [$p \leq 0.05$]; r understanding = 0.77 [$p \leq 0.01$]) and associated with average weekly hours of PA (r medication control = 0.59, $p \leq$

0.05) (Table 11). In contrast, only perceived duration correlated with average weekly hours of PA ($r = 0.66$, $p \leq 0.05$) across B-IPQ items related to family members' HDR (Table 12).

Perceptions of children's experience with HC & HDR produced statistically significant correlations with perceptions of children's adherence more often than either HC+ or HC- parental perceptions (Tables 13 & 14). Correlations between perceptions of children's experience with HC and adherence ranged in strength from 0.34-0.37, whereas those relative to perceptions of children's experience with HDR and adherence ranged in strength from 0.36-0.55. VAS-measured medication adherence was associated with the greatest number of B-IPQ items: respondent perception of the duration of child's HC ($r = 0.34$, $p \leq 0.05$), medication control over HDR ($r = 0.55$, $p \leq 0.001$), and child understanding of HDR ($r = 0.48$, $p \leq 0.01$) were all associated with greater adherence, whereas perceived exercise control over child's HC was associated with lower adherence ($r = -0.35$, $p \leq 0.05$). Higher perceived child understanding of HDR was inversely associated with forgetfulness ($r = -0.36$, $p \leq 0.05$), and perceived duration of child's HDR was positively associated with average hours of PA ($r = 0.37$, $p \leq 0.05$). Perceived affectedness by HC was inversely correlated with frequency of saturated fat intake ($r = -0.37$, $p \leq 0.05$).

Based on the inter- and intra-correlative patterns noted across family unit subsamples described above (Question #2, *Associations Within and Between Concepts of Perception*), aspects of perception in this sample appear to fall into three "categories": associations related to social/emotional experience (affect, concern, and emotion), associations related to control (overall personal, and medication-, diet-, and exercise-specific control), and associations related to understanding and duration across the HeFH subcomponents. Keeping these categories in mind, additional patterns of relationships become evident.

All items of perceived control, particularly related to control over HC, were statistically significantly associated with one or more adherence measure excepting the item for frequency of saturated fat intake. The item for personal control of HC+ parents over their HC was positively and statistically significantly correlated with higher scores on the adapted MGL test ($r = 0.43$, $p \leq 0.05$); HC- parents' perceived treatment control of family members' HC by medication directly correlated with average weekly hours PA ($r = 0.59$, $p \leq 0.05$); HC- parents' perceived control over family members' HC by diet & exercise was inversely correlated with VAS medication adherence ($r = -0.63$, $p \leq 0.05$ & $r = -0.61$, $p \leq 0.05$ respectively). Children's perceived ability to control HC by exercise was also inversely correlated with VAS medication adherence ($r = -0.35$, $p \leq 0.05$). Pertaining to HDR, only perceived control of children's HDR by medication was associated with any adherence measure (r VAS $= 0.55$, $p \leq 0.001$).

Perceived understanding of family members' HC among HC- parents was directly correlated with higher reported VAS medication adherence ($r = 0.77$, $p \leq 0.01$), and perceived duration of family member's HDR was positively correlated with average weekly hours of PA ($r = 0.37$, $p \leq 0.05$). Perceived child understanding of HDR was associated with more ideal adherence to medication by both VAS and adapted MGL test ($r = 0.48$, $p \leq 0.01$ & $r = -0.36$, $p \leq 0.05$ respectively). Perceived duration of child's HC and VAS medication adherence tracked in the same direction; likewise, perceived duration of child's HDR was also positively correlated with average weekly hours of PA ($r = 0.66$, $p \leq 0.01$). Perceived understanding and duration of the HeFH components reported by HC+ parents were not correlated with any child adherence outcome.

Items of perceived social/emotional experience were least frequently correlated with perceived child adherence. General affectedness of children by HC and frequency of saturated fat intake tracked weakly in opposite directions (r = -0.37, $p \leq 0.05$). General affectedness of HC+ parents by their HDR was positively associated with average weekly hours of PA (r = 0.46, $p \leq 0.05$). No items related to concern or emotionality proved to be significantly associated with any adherence measure, nor were any social/emotional perception items reported by HC- parents correlated with any adherence measure.

Question 4.) In HeFH-affected households, what are the relationships between parenting styles and parents' perceptions of high cholesterol and risk for heart disease?

Analyses to address research question four were comprised of approximately 46 parents from HeFH-affected households, of which 32 were HC+ and 14 were HC-; responses varied by +/- n = 1 based on missing data. Reliability of the PSDQ was adequate for respondent self-assessment and household averages of parenting styles, but low for respondent assessment of parenting partners (Table 6). As summarized in Table 15, respondents saw themselves as highly authoritative overall (Median 4.3 [IQR 3.7, 4.7]), with lesser authoritarian (Median 1.4 [IQR 1.2, 1.6]) and permissive (Median 1.8 [IQR 1.5, 2.3]) parenting tendencies. Respondents judged themselves as more authoritative than their partners (Parenting Partner Median 3.9 [IQR 3.4, 4.3], p = 0.001), but no differently in authoritarian or permissive parenting behaviors.

The handful of statistically significant correlative relationships emerged between overall household parenting styles and parent's perceptions of illness were generally weak or moderate in strength (Tables 16-21). Household permissiveness was only associated with social/emotional responses to HC and HDR among HC+ parents (r HDR Affect = 0.42, $p \leq$

0.05; *r* HC Emotion = 0.42, $p \leq 0.05$; *r* HDR Emotion = 0.38, $p \leq 0.05$) (Tables 15 & 16). Household permissiveness was not correlated with any B-IPQ item relative to HC- parents or respondent's perceptions of children.

Social/emotional responses of HC- parents to family members' HC & HDR were directly associated with household authoritarian parenting (*r* HC Affect = 0.56, $p \leq 0.05$; *r* HC Concern = 0.56, $p \leq 0.05$; *r* HDR Emotion = 0.54, $p \leq 0.05$) (Tables 18 & 19). No B-IPQ items answered by HC- parents produced statistically significant correlations with authoritative or permissive parenting styles.

Household authoritarian parenting frequently correlated with control-type B-IPQ items, particularly parental perceptions of their children's HC & HDR (Tables 20 & 21). Both perceived control of children's HC through diet, and perceived control of children's HDR through exercise, were positively associated with household authoritarian parenting ($r = 0.32$, $p \leq 0.05$ for both); perceived control of children's HC through medication tracked with household authoritarian parenting in opposite directions ($r = -0.31$, $p \leq 0.05$). HC+ parent personal control over HDR positively tracked with household authoritarian parenting ($r = 0.36$, $p \leq 0.05$).

Within the understanding/duration category of B-IPQ items, only duration statistically significantly associated any parenting style, tracking in the opposite direction as household authoritarian behaviors among HC+ parents relative to HDR ($r = -0.38$, $p \leq 0.05$)

Question 5.) In HeFH-affected households, what are the relationships between parenting styles and parents' observed child adherence to treatment recommendations?

Analyses to address research question five were comprised of 37 respondents from HeFH-affected households, with 32 providing responses for partners and a total of 35

contributing data for overall households. Very few statistically significant correlative relationships emerged in matrices of parenting style and parentally observed adherence of children to HeFH treatment recommendations (three statistically significant correlations out of 36 across three respondent subsamples [respondent, parenting partner, and household]) (Tables 22-24). Authoritarian behaviors among parenting partners tracked negatively with VAS adherence and positively with the adapted MGL (r = -0.52, $p \leq 0.01$; r = 0.39, $p \leq 0.05$ respectively) (Table 23); this relationship persisted for VAS adherence at the household level (r = 0.40, $p \leq 0.05$) (Table 24). When broken down by respondent cholesterol diagnosis, authoritative behaviors among HC- respondents and their parenting partners statistically significantly correlated negatively with VAS adherence (r = -0.59, p = 0.03 & r = -0.79, p = 0.006 respectively, n = 12-14; data not shown). Respondent parenting style alone did not statistically significantly correlate with any adherence measure, nor did the parenting styles of HC+ respondents or their parenting partners. Saturated fat intake and PA measures did not statistically significantly correlate with parenting style across any respondent subsamples (Tables 22-24).

Supplementary Results

Associations Within and Between Concepts of Perception

Spearman correlations within B-IPQ adaptations for each HeFH concept and subsample (HC+ respondent, HC- respondent, respondent's perceptions of children) were undertaken to assess for collinearity. Additionally, Spearman correlations between the HC and HDR B-IPQ adaptations were calculated to assess whether the concepts were perceived similarly within each subsample. Finally, comparisons between the subsamples were

assessed by way of Spearman correlations within each HeFH concept to determine if parents perceive their children's experiences as like or different from their own.

Correlations Within B-IPQ Adaptations Among HC+ Parents. Few significant correlations in perception were identified related to hypercholesterolemia among affected parents. The strongest relationship was between perceived control of cholesterol through lifestyle; parents endorsing higher perceived control from diet also perceived higher perceived control from exercise ($r = 0.92$, $p = <0.0001$). Additionally, perceiving hypercholesterolemia to last forever was weakly associated with perceiving medications as having greater effectiveness, and exercise to have lesser effectiveness in cholesterol control. Finally, relationships were identified between the concepts of general affectedness, concern, and emotionality; greater perceived affectedness by hypercholesterolemia was moderately correlated with greater perceived emotionality, while greater perceived concern about hypercholesterolemia was weakly and negatively correlated with general affectedness and emotionality.

In contrast, multiple perceptual concepts were statistically significantly correlated within the B-IPQ adaptation pertaining to HDR. Strong, positive relationships were again identified between perceived control of HDR through diet and exercise ($r = 0.89$, $p = <0.0001$), and perceiving prolonged duration of HDR was again weakly associated with greater perceived control through pharmacotherapy ($r = 0.46$, $p = 0.005$). The relationships between the concepts of general affectedness, concern, and emotionality identified relative to hypercholesterolemia were mirrored pertaining to HDR; additionally, higher emotionality about risk, and greater perceived affectedness by risk, were weakly correlated to increased perceived effectiveness of lifestyle interventions ($r = 0.46$ & $r = 0.42$ respectively; $p <0.01$).

Perceived effectiveness of neither lifestyle intervention related to perceived personal control over HDR; indeed, lower perceived control over HDR was weakly correlated with higher perceived understanding of HDR (r = -0.48, p = 0.004) and greater perceived effectiveness of pharmacotherapy (r = -0.48, p = 0.003). Interestingly, the concepts of HDR duration and HDR understanding were not correlated.

Correlations Between HC & HDR B-IPQ Adaptations Among HC+ Parents. Correlations between HC+ parent perceptions of HC and HDR displayed three distinct patterns of relationships reflective of the intra-concept associations discussed above. First, each aspect of perception positively correlated with itself across concepts; for example, greater perceived control over HC was associated with greater perceived control over HDR (r = 0.42, p = 0.01). Most of these associations were weak to moderate, with the only strong correlation being between perceived duration of each concept (r = 0.79, p = <0.0001).

Second, perceived control of both HC and HDR by lifestyle interventions were moderately positively correlated, e.g., higher perceived effectiveness of diet in controlling HC was associated with higher perceived effectiveness of exercise in controlling HDR (r = 0.55, p = 0.0005), and vice versa (r = 0.61, p = <0.0001).

Third, greater perceived general affectedness by HC was correlated with greater perceived emotionality around HDR (p = 0.47, r = 0.004), and vice versa (r = 0.61, p = 0.0001). Moreover, greater perceived general affectedness and emotionality around HC were both associated with lower perceived concern about HDR.

Additional relationships of interest included correlations between HC understanding and perceived control over HDR; between lower perceived understanding of HDR and higher

perceived general affectedness and emotionality around HC; and between greater understanding of HDR and greater perceived control of HC through pharmacotherapy.

Correlations Within B-IPQ Adaptations Among HC- Parents. Items adapted to HC- parents were framed to focus on the agency, understanding, and affectedness of the individual as it pertained to their HeFH+ family unit (examples: "How much does your family members' diagnosis of high cholesterol affect you?", "How much control do you think you have over your family members' high cholesterol?"). Many fewer significant correlations were identified between B-IPQ items within the concepts of HC and HDR in this context. Related to HC, only general affectedness and emotionality (r = 0.78, p = <0.0001) and control by diet and exercise (r = 0.94, p = <0.0001) were positively associated. These relationships persisted in the HDR matrix; additionally, concern about HDR was correlated with control by diet (r = -0.51, p = 0.05) and control by exercise (r = -0.56, p = 0.03), and emotionality was correlated with control by diet (r = 0.59, p = 0.019) and control by exercise (r = 0.59, p = 0.02).

Correlations Between B-IPQ Adaptations Among HC- Parents. Placing perceptions of both HC and HDR into one matrix resulted in relationships mirroring the withing-concept matrices for HC- parents described above, plus correlations between overlapping B-IPQ item types as seen in the corresponding matrix for HC+ parents (affect with affect, concern with concern, etc.). Among these overlapping items, only perceived timeline of HC failed to correlate with the corresponding perception of HDR timeline. The only relationships new to HC- parents identified within this matrix were those between dietary control of cholesterol and exercise control of HDR (r = 0.91, p = <0.0001), and between exercise control of cholesterol and dietary control of HDR (r = 0.92, p = <0.0001);

however, this correlational pattern was consistent with that identified between lifestyle effectiveness items among HC+ parents.

Perceptual Correlations Between HC+ Parents & Children Related to HC. In comparing HC+ parent perceptions of their own HC diagnosis with their perceptions of their child's HC diagnosis, no strongly significant correlations were identified. Weak to moderate correlations were identified along similar patterns as those described above, specifically overlapping in elements of experience (parent's general affectedness by HC versus their perception of their child's general affectedness by HC, for example, excepting parental versus child's perceived concern); effectiveness of lifestyle interventions; and general affectedness versus emotionality. Of note, relationships between parental general affectedness and emotionality around their own HC diagnosis were positively correlated with perceived dietary control (r = 0.37 [p = 0.03]; r = 0.42 [p = 0.01], respectively) and exercise control (r = 0.42 [p = 0.01]; r = 0.38 [p = 0.02], respectively) of their child's HC. Similarly, how greatly a parent believed in the effectiveness of exercise was positively associated with how generally affected by HC they perceived their child to be (r = 0.36, p = 0.03).

Perceptual Correlations Between HC+ Parents & Children Related to HDR. HC+ parent perceptions of their own HDR compared very differently from their perceptions of their child's HDR. Most notably, none of the aspects of parents' perception of their own HDR were statistically significantly correlated with perceptions of children's understanding of or concern about HDR. In contrast to the findings within and between HC and HDR among HC+ parents described previously, multiple aspects of how HC+ parents perceived their HDR were negatively correlated with perceptions of children's control over HDR: parent affectedness (r = -0.48, p = 0.004), perceived duration (r = -0.42, p = 0.01), parent's

personal control (r = 0.52, p = 0.002), medication effectiveness (r = -0.44, p = 0.01), dietary effectiveness (r = -0.51, p = 0.002), exercise effectiveness (r = -0.53, p = 0.001), and parent's emotionality (r = -0.47, p = 0.006).

Perceptual Correlations Between HC- Parents & Children Related to HC. A small number of statistically significant relationships were observed between HC- parents' perceptions of HC and their perceptions of children's experience with HC compared to the number of statistically significant associations in the equivalent model for HC+ parents. No clear pattern of overlapping perception items materialized (ex. parental perception of their general affectedness by HC did not correlate with how they perceived their children's general affectedness). No aspect of how HC- parents perceived HC were statistically significantly correlated with how they perceived children to understand or be concerned by HC, nor was their perceived understanding of HC correlated with any aspect of how they perceived the experience of children with HC.

HC- parents perceived general affectedness by their family's HC were statistically significantly correlated with their perceptions of dietary (r = 0.56, p = 0.04) and exercise (r = 0.49, p = 0.07) control of child's HC; dietary (r = 0.56, p = 0.03) and exercise control (r = 0.56, p = 0.03) of children's HC also were statistically significantly correlated with HC- parents' perceived emotionality about HC. Likewise, HC- parents' perceptions of dietary and exercise control of their family's HC were related to how generally affected by HC they perceived children to be (r = 0.71, p = 0.003; & r = 0.74, p = 0.002 respectively), as well as children's emotionality (exercise r = 0.57, p = 0.03). Parental concern for HC was statistically significantly correlated with parents' perceptions of children's control over HC (r = 0.75, p = 0.001). Interestingly, HC- parents' perceptions of dietary and exercise control

over HC were also negatively with how much control over HC they perceived their children to have (r = -0.58, p = 0.02; & r = -0.62, p = 0.01 respectively). The intercorrelations of lifestyle-type interventions (dietary and exercise control) again persisted.

Perceptual Correlations Between HC- Parents & Children Related to HDR. The most common relationships between HC- parents' perceptions of their family HDR, and children's HDR, were related to perceived dietary and exercise control over family members' risk. Higher perceived effectiveness of lifestyle-type interventions were statistically significantly correlated with perceived higher general affectedness (diet r = 0.72, p = 0.004; exercise r = 0.68, p = 0.01) and higher emotionality (diet r = 0.70, p = 0.005; exercise r = 0.69, p = 0.007) in children; there was also an inverse association between lifestyle-type interventions and perceived child control over HDR. HC- parents' emotionality around their family HDR also correlated with higher perceived dietary and exercise control over children's HDR (r = 0.59, p = 0.03; & r = 0.73, p = 0.003 respectively). Finally, unlike the relationships found pertaining to perceptions of HC, HC- parent's perception of their family HDR duration was negatively associated with children's emotionality around HDR (r = -0.56 p = 0.04).

Relationships Between Family History with HeFH, Child History of HeFH, and Child History of Treatment for HeFH and Observed Child Adherence

This analysis was undertaken to address contextual aspects identified as influencing illness management by the FMSF, and assess for similarities between the current sample and those in the published literature. The analysis comprised of 35 respondents from HeFH-affected households. Residing in a household where an affected parent had been formally diagnosed with HeFH was positively and statistically significantly associated with VAS

adherence to medication ($r = 0.38$, $p = 0.02$) and negatively and statistically significantly associated with weekly frequency of saturated fat intake ($r = 0.54$, $p = 0.001$) (Table 25). Additionally, VAS-reported medication adherence was positively and statistically significantly correlated with MD/NP specialist visits over the previous two years ($r = 0.35$, $p = 0.04$). The presence of heart disease in a child's first-degree or second-degree family member did not statistically significantly correlate with any adherence measure. Likewise, neither adapted MGL-measured medication adherence nor average weekly PA were statistically significantly correlated with any item pertaining to family history, child medical history, or child treatment history for HeFH.

Qualitative Post-Survey Remarks by Respondents

An informal analysis was undertaken on the post-survey remarks by the PI through an iterative process of multiple reviews, first for general content, then for topics mentioned by more than one respondent. Eighteen parents (7 HC-, 3 fathers) offered thoughts pertaining to four general themes: childhood HeFH treatment (medications, 8 mentions; diet, 6 mentions; and exercise, 4 mentions), perceptions of the condition (8 mentions), family history of heart disease (4 mentions), and treatment team (3 mentions) (Table 26).

Parents described pharmacotherapy as something inherent to the "journey" of HeFH, and something to be avoided, with diet and exercise cited as both minimally helpful and the means of postponing or avoiding medication entirely. Challenges to administering medications and adhering to lifestyle based on age and developmental stage were also mentioned. A few respondents focused on the importance of finding the appropriate frame for discussing HeFH treatment - diet and lifestyle in particular - and two expressed discontentedness over the methods employed with their own children and families. In

contrast, two parents praised their specialist and/or care team. Exercise, it should be noted, was explicitly identified by two parents as challenging in the context of the co-occurring COVID-19 pandemic.

The perceptions of HeFH mentioned most frequently were awareness and understanding, particularly that parents have these; children are expected to improve in them as they age and therefore hopefully improve in their self-care behaviors. Emotions like anxiety, angst, and care were also mentioned relative to implementing HeFH management. One mother mentions the poignant and traumatic experience of witnessing the consequences of heart disease in a first-degree family member; family history was additionally noted an impetus for screening and adhering to treatment.

CHAPTER FIVE

DISCUSSION

This was a cross-sectional, descriptive, correlational study guided by the FMSF and aimed at investigating relationships between illness perceptions, parenting styles, and parentally observed adherence to treatment within families affected by HeFH. Parents of children with HeFH in this sample overwhelmingly reported their children as being adherent to the medication, diet, and physical activity recommendations of their cholesterol specialists. Characteristics of the respondents were relatively homogeneous, as were their self-reported parenting behaviors. Perceptions of genetic hypercholesterolemia and its associated risk for heart disease, as well as their relationships with adherence measures, varied by the cholesterol diagnosis status of the parent and parental perceptions of their child's HeFH experience. Parenting styles were very infrequently correlated with the adherence measures; however, parenting styles were associated with multiple elements of parental illness perceptions.

Descriptive Findings: Sample Characteristics

Results of this study must be interpreted in the context of the sample characteristics. Participating parents were overwhelmingly middle-aged, highly educated, Caucasian, and mothers. Most households had experienced an atherosclerotic event of some type in at least one first- or second-degree relative, and respondents predominantly had a longstanding

awareness of their or their spouse/child's other biological parent's hypercholesterolemia and its genetic etiology; many of the parents in these households had received a formal diagnosis with HeFH. Parents also appeared very receptive to treatment of their children with HeFH; in addition to routine attendance at pediatric preventive specialist visits (Median two visits/two years) and dietician consults (Median one visit/two years), only six families declined or otherwise reported not starting their child on an HMG CoA-reductase inhibitor after it had been recommended (13%).

These findings likely reflect a variety of social and diagnostic trends not necessarily reflective of those impacting the greater U.S. HeFH population. Pediatric research findings often derive from the participation of mothers, and educated individuals may be more likely to participate in research in some settings (Costigan & Cox, 2001; Scanlon et al., 2021). People identifying as Caucasian are more heavily represented in research; however, in the context of HeFH, the most well-documented mutations resulting in this phenotype frequently present in Northern European founder populations (examples: Dutch Afrikaners and Appalachians, French Canadians, and Christian Lebanese) (Austin, 2004). Thus, Caucasian decent is genetically common among those with HeFH, although this may not entirely explain the lack of diversity in this sample; indeed, questions remain regarding adequate identification of HeFH in the larger U.S. non-Caucasian population (Dixon et al., 2014).

Likewise, the preponderance of parental awareness of their or their spouse/child's other biological parent having genetically elevated cholesterol – and HeFH specifically, in contrast to international estimates of HeFH diagnosis – suggests a sample wherein parents were the index case of HeFH for their family, with children diagnosed secondary to this. However, this may not be the case among younger parents or in populations receiving less

medical care generally. Different routes to identification and treatment could impact illness perceptions, relate differently to parenting styles, and result in variations in child adherence to HeFH treatments.

Child & Youth Adherence to HeFH Treatment Recommendations (Question #1)

To our knowledge, this research provides the first assessment of pediatric HeFH treatment adherence within a HeFH-specific sample. Respondents generally perceived HeFH children in this sample as adherent to both medication and lifestyle interventions. Parents reported that children took 90% or more of their monthly doses of statins and never intentionally skipped doses. Although the numeric and behavioral measures of medication adherence were highly correlated, only 35.7% of respondents reported perfect behavioral adherence, with forgetfulness being the primary driver (54%); this suggests possible parental intervention to ensure adherence. Regarding lifestyle measures, 73.5% of children reportedly met AAP guidelines for adequate amounts of PA, and while no family endorsed entire avoidance of saturated fat intake, the majority expressed clear consideration of it as evidenced by most families reporting two or fewer weekly servings of foods in each saturated fat food category. Interestingly, PA and frequency of saturated fat intake adherence measures were not intercorrelated in this sample, despite the frequent associations between perceived effectiveness of dietary and physical activity by responding parents; nor was lifestyle adherence correlated with either medication adherence measure. This suggests that the factors influencing adherence to each aspect of HeFH treatment may vary as well and may require differing types of intervention to improve. Overall, these findings of high adherence are consistent with the self-reports published among adults with HeFH described previously

(Claassen et al., 2010; Galema-Boers et al., 2014; Hollman et al., 2006; Senior et al., 2004; Senior & Marteau, 2007).

What *is* FH? Parental Illness Perceptions and the Family Experience of HC and HDR (Question #2)

Perceptions of HeFH were operationally defined by the most salient subcomponents of the condition, specifically high cholesterol (HC) and risk for heart disease (HDR). While correlations between the perception parameters for each of these subcomponents show significant overlap, differences in perspective – that is, whether the respondent had the diagnosis themselves, or assisted in its management as a partner or parent alone – illuminated varied associations. For HC- respondents, strong and significant correlations were predominantly intracorrelative (within the same perception category); thus, the concepts of HC and HDR appear largely overlapping, suggesting little cognitive or experiential difference between them. Among HC+ parents, however, the associations between perceptions of HC and HDR appeared more complex; in addition to moderate-to-strong intracorrelations, a variety of other significant weak-to-moderate correlations between perception categories also emerged. This may reflect more nuanced perceptions of the HeFH subcomponents among those having personally received a genetic HC diagnosis: while clearly related concepts, HC and HDR do not appear to be perceived identically by these respondents.

These differences between HC+ and HC- respondents suggests variation in the familial experience of parenting pairs where only one parent has a genetic hypercholesterolemia diagnosis, as the FMSF would predict and the correlative relationships

between parental perceptions of the HeFH subcomponents and child experience substantiate. Perceptions of HC and HDR reported by HC- respondents were widely intracorrelated with their perceptions of how these HeFH subcomponents are experienced by HeFH+ children, with the notable exceptions of perceived concern or understanding. These findings support the inference that parents without HC see their experience with the HeFH subcomponents as largely the same as that of their HeFH+ children except in these areas; alternatively, it may suggest that HC- parents do not largely perceive a difference between their experiences with HC & HDR with their child, and that with the whole family unit.

Respondents with genetic HC, however, appear to perceive their children's experience with HC and HDR through the lens of their own diagnosis, as evidenced by multiple significant weak-to-moderate associations across a multitude of perception parameters. Notable here were multiple positive associations between HC+ parents' general affectedness and emotionality about HC correlating with perceptions of childs' HC. Additionally, multiple HC+ parent perceptions of HDR correlated with perceptions of childs' personal control of HDR, suggesting that many aspects of HC+ parents own experience tells them that their children will not be able to control their own HDR.

These relationships support the continued use of HeFH subcomponents in this population and highlight important intrafamilial variations in the HeFH experience. Globally, it is important to note that both HC and HDR were not generally perceived as very concerning, affective of parent's daily lives, or emotionally affective for parents who have a diagnosis of FH/genetic high cholesterol. These parents felt that they understood HC and HDR, and although they did not believe they could personally control them, they did believe in the effectiveness of medications, while perceptions of lifestyle effectiveness are middling.

HC- parents generally perceived HC and HDR similarly to HC+ parents, although they endorsed feeling more general affectedness and emotionality than affected parents did. Parents both with and without HC reported their children as being minimally affected by HC and HDR, and lacking in understanding of them compared to themselves; however, children were reported as feeling more concern about their HC and HDR than their parents expressed for themselves or their families generally. While these findings are overall consistent with the previous literature on adults with HeFH, this is the first study incorporating the perceptions of unaffected parents, as well as parental perceptions of their children's experience.

Parenting the FH Family: Intersections Between Perceptions and Parenting Styles (Question #4)

Somewhat unexpectedly, multiple relationships emerged between parental perception of HC & HDR and parenting behaviors of particular styles; unexpected because parents presumably gravitate towards or consciously elect a parenting philosophy prior to their knowledge of their child's diagnosis, and in some cases prior to knowledge of their own. HC+ parental perceptions of illness most often correlated with household parenting behaviors, particularly those related to HDR. In this sample more general affectedness or emotionality among HC+ parents around their HDR moderately correlated with greater permissiveness, and higher reported perceptions of HDR duration and control weakly correlated with fewer authoritarian behaviors. Presumably, these relationships are driven by parent's illness perceptions; that is, one chooses how to parent secondary to how one feels about a personal or familial diagnosis, rather than those feelings stemming from how one

engages in parenting. Thus, our findings suggest that parents with genetic HC may consciously or unconsciously adopt childrearing styles relative to their illness experiences.

Authoritarian parenting was associated with a variety of illness perception parameters among parents without genetic HC. In contrast to the HC+ findings, where HC- parents endorsed more general affectedness, concern, or emotionality around their family members' HC and HDR, they also reported engaging more frequently in authoritarian parenting behaviors. Thus, whereas greater emotive-type perceptions correlated with greater parental permissiveness and lower control among HC+ parents, these types of perceptions correlated with increased control behaviors in HC- parents. Notably, illness perceptions of HC- parents were not associated with either of the other parenting style behaviors. Parental perceptions of medication effectiveness in controlling children's HC, and perceptions of exercise effectiveness in controlling children's HDR, also correlated with household authoritarian parenting. One possible explanation may be the utilization of authoritarian behaviors as a tactic in enforcing compliance with HeFH treatment recommendations, however the inconsistent directionality of these relationships across or within perceptions of treatment effectiveness does not fully support this hypothesis.

The relationships between parenting style based on parental experience with chronic physical medical conditions has not been well explored in the literature. Where correlations between parenting style and parental diagnoses or illness symptoms have been included in publication, they have not been significant (Jones et al., 2008; Poppert Cordts et al., 2020). Although addressing family-based obesogenic environments has long been a recognized aspect of pediatric weight management, research to date pertaining to parenting style and childhood obesity has focused on parental perceptions of their children's weight rather than

their own (Kitzmann et al., 2008). Developmental psychology has empirically supported a multi-factor model of parenting practice determinants, but personal illness management has not historically been included (Taraban & Shaw, 2018). Thus, with increasing acknowledgement of the family environment as influential on cardiovascular health, future research should seek replication of these findings in this and other chronic condition populations (Bell & Belsky, 2008; Byrne et al., 2017; Vedanthan et al., 2016).

The FH Experience and Associations with Observed Child Treatment Adherence (Questions # 3 & 5)

This research provides preliminary evidence for the presence of relationships between parentally observed adherence to treatment recommendations and the independent variables included in this survey, particularly perceptions of HC and HDR. Of the four adherence variables included, independent variable associations were predominantly with VAS-measured medication adherence and average weekly PA. Both medication adherence by Adapted MGL test and weekly saturated fat intake were virtually unassociated with the included variables, each only weakly correlating with two variables of history or perception.

Previous research on the relationships between illness perception and treatment adherence in HeFH-affected individuals has varied widely in their targeted samples, selected outcome measures, aspects of the condition considered relevant to perceptions, and methodologies (Brewer et al., 2002; Claassen et al., 2010; Hagger et al., 2016; Hagger et al., 2018, 2019; Marteau et al., 2004; Senior et al., 2004, 2005; Senior & Marteau, 2007). Unsurprisingly, results have varied substantially across studies, making direct comparison of findings challenging. Our research represents the first investigation into relationships

between parental (i.e., adult) perceptions of illness and observed adherence among offspring, and the only research that defines HeFH by both its clinically relevant components and provides comparisons of the two. Additionally, it also is the only research in this population to date that considers the relevance of the perceptions of unaffected family members and household context more generally to HeFH treatment adherence.

Associations Between Family, Diagnostic, and Treatment Histories and Child Treatment Adherence

Few associations were observed between treatment adherence and family history with HeFH, child HeFH diagnostic history, or child HeFH treatment history in this sample. Formal diagnosis of HeFH in one parent within a household was weakly associated with greater VAS-reported medication adherence, and moderately associated with lower reported frequency of weekly saturated fat intake. Likewise, greater frequency of medical specialist visits (MD or NP) over the previous two-year period was weakly associated with higher VAS-reported medication adherence. Length of parental knowledge regarding their own genetically elevated high cholesterol, family history of heart disease in either a first or second degree relative, child age at diagnosis with HeFH, length of child treatment for HeFH, and frequency of Registered Dietician counseling in the previous two years were not associated with parentally observed adherence to their child's HeFH treatments.

These findings contrast with the literature to date pertaining to influences on HeFH treatment adherence. Witnessing the progression of heart disease and associated atherosclerotic events in first- or second-degree family members has been frequently cited in the qualitative literature as both the trigger to identify HeFH, and the impetus for maintaining HeFH treatment over the life course (Frich et al., 2006; Hallowell et al., 2017; Jenkins et al.,

2012; Keenan et al., 2019; Kools et al., 2008; Mackie et al., 2015; Senior et al., 2002; Urke et al., 2019), but lacked association with any form of adherence reported by respondents to this survey. Similarly, developmental stage at diagnosis and early integration of HeFH-related lifestyle habits have been hypothesized as relevant to long-term success in treating HeFH and have provided rationale for early screening and identification (Frich et al., 2006; Keenan et al., 2019; Mackie et al., 2015; Meulenkamp et al., 2008; Weiner, 2011); however, neither attained statistical significance in the present analysis. These findings may be related to the overall homogeneity of the sample and lack of overall adherence variability reported; alternatively, it may be secondary to the developmental stage of most children referenced by parents. With a mean age of 13 years, most children in this survey have already entered adolescence, a period of development recognized for its emphasis on peer relationships, autonomy without parental direction, new and changing personal interests, and greater emphasis on immediate concerns than longer-term considerations. Habits of earlier years may be less visible to parents, or temporarily lose relevance for affected children. Thus, these findings must be interpreted in context. Future research focusing on associations between lifestyle adherence behaviors among preschool and school-aged children and HeFH family history could be valuable for further substantiation of these early screening arguments.

The correlation between observed child adherence and formal diagnosis of a parent with HeFH is particularly notable. Diagnosis of HeFH has long been underdiagnosed, and substantial attention has been paid recently to theorized relationships between molecular diagnosis and treatment adherence among adults with HeFH (Lee et al., 2019; Nordestgaard et al., 2013). Our findings suggest that phenotypic diagnosis alone, when detected before or during childbearing and childrearing years, may have a positive impact on treatment

adherence of the whole family unit. As parental genotype was not ascertained, the possibility that parents with a formal diagnosis know the mutation they carry cannot be entirely excluded; however, current practice in the U.S. does not emphasize molecular means of identification, making this likely the exception rather than the rule. Thus, this would seem to add support for the ongoing effort to improve both identification and diagnostic labeling of HeFH among younger adults regardless of the use of molecular means. There may also be an element of desirability bias here that cannot be excluded; parents with HeFH may feel compelled to report better adherence among their offspring given this knowledge.

Relationships Between Parental Illness Perceptions and Child Treatment Adherence

Two patterns emerged from these correlational relationships: one pertaining to the perception categories, the other pertaining to the family member and their diagnostic status. Few aspects of HC+ parent illness perception correlated with observed child adherence, in contrast to those of HC- parents and parental perceptions of their child's experience with HC & HDR. Similarly, emotive-type perceptions – those of general affectedness, emotionality, and concern – did not generally correlate with adherence, whereas multiple associations were identified related to illness controllability, understanding, and duration.

It is interesting that few statistically significant correlations were observed between treatment adherence and the illness perceptions of parents with their own diagnosis of genetically elevated cholesterol. It might be expected that HC+ parents could have stronger perceptions of having HC or HDR, or that their experience with these could more directly or indirectly influence the adherence of their affected offspring; however, this does not appear to be the case in this sample. We found that the perceptions of parents without genetically elevated cholesterol were more frequently and more strongly associated with observed child

adherence than those of affected parents, particularly related to medication adherence and perceptions of HC. Additionally, parents' perceptions of their children's experience with HC appeared much more salient to adherence than those of HC+ or HC- parents, producing multiple significant relationships, if generally weak to moderate in strength. These findings correspond with the assumptions of the FMSF, wherein the perspectives of every family member individually and differentially contribute to overall family-unit adaptation to condition management. As the number of parents without HC in this sample is small (n =15) and entirely female, it is not possible to determine whether the maternal family role of these respondents is confounded with diagnosis status but should be considered.

Additionally, correlative relationships between the items of the B-IPQ in this sample suggest three types or categories of perception that subsequently present themselves in the associative relationships with observed child adherence: emotive-type (affectedness, emotionality, and concern), cognitive-type (understanding and duration), and controllability-type (general controllability, controllability with medication, controllability with diet, and controllability with exercise). Our analyses found very few significant associations between adherence and emotive-type perceptions, with only general affectedness by HC among children being weak-to-moderately correlated with weekly frequency of saturated fat intake, and general affectedness by HDR among HC+ parents being correlated with PA.

It should be noted methodologically that the B-IPQ items pertaining to affectedness and emotional affectedness were highly intercorrelated among these respondents. Operationally, these are single items encapsulating concepts of the Illness Perceptions Questionnaire-Revised and the Common Sense Model of Illness Representation, specifically the perceived consequences of a concern or condition on the life of an individual (general

affectedness) and the emotional representations that an individual may derive from their diagnoses (emotionality). However, the intercorrelation between these items in this population suggests overlapping interpretations by respondents, i.e., respondents may not perceive them as meaningfully different. Future research should include further qualitative investigation into respondents' personal definitions of these items, to ensure appropriate use in this population going forward.

Relationships Between Parenting Styles and Child Treatment Adherence

Statistically significant relationships between parenting styles and observed child treatment adherence parameters were few, and the larger clinical relevance for this sample is questionable. Respondent parenting behaviors were not correlated with any adherence outcomes overall; higher authoritarian behaviors observed of parenting partners correlated with lower adherence to medications on both VAS and the Adapted MGL and persisted for VAS-adherence when parenting styles were averaged by household. When considering respondent cholesterol diagnosis, the reported parenting styles of HC+ parents and their parenting partners did not significantly correlate with any adherence outcome. Lower VAS-measured medication adherence was associated with higher reported authoritarian parenting among HC- respondents and HC- respondent's parenting partners; however, it must be noted that these subsamples consisted of only 14 and 12 observations respectively. Thus, while this sample provides some evidence of poorer medication adherence when parents – particularly those without genetically elevated high cholesterol themselves – engage in more frequent authoritarian parenting behaviors, its relevance could be overstated.

It is interesting that the findings in this sample are more consistent with that of families with chronic illness than those promoting wellness. Although the literature on

parenting style and obesogenic behaviors – which are largely wellness-based – has consistently found greater or improved adherence in the context of authoritative parenting, this was not borne out in this sample (Kiefner-Burmeister & Hinman, 2020). Parental authoritarianism, however, has been found to correlate with poorer medication adherence in chronically ill youth (Davis et al., 2001; Ievers-Landis et al., 2005; Saletsky et al., 2014). Individuals with HeFH consistently reject the notion of being chronically ill, as evidenced by both published interview studies and the overall perceptions of the condition captured in this survey (Frich et al., 2006; Keenan et al., 2019; Mackie et al., 2015; Meulenkamp et al., 2008; Weiner, 2011), thus making these findings surprising. However, it has been noted by multiple researchers that consistency in the parenting styles literature pertaining to wellness adherence outcomes may be predicated on multiple factors, including sample size, outcomes of interest, and survey instruments (Kiefner-Burmeister & Hinman, 2020; Pinquart, 2014). Future research in this area pertaining to families affected by HeFH should attempt to replicate our findings in larger samples, with more diverse participants, ideally employing measurement tools tailored to narrower age ranges for greater precision.

Implications & Areas for Future Research: The Family Management Style Framework

In the years since conceptualization of this study, researchers have continued to expand the assumptions, definitions, and outcomes of the FMSF. Most notably, the family management pattern has been hypothesized as an intermediate to multiple clinical outcomes (rather than itself the terminus of the theoretical model), and the perspective of affected child or adolescent has been acknowledged as an integral aspect of the family system separate from that of parents with its own distinct impact on the Family Management Pattern

(Beacham & Deatrick, 2015, 2019; Knafl et al., 2012; Wollenhaupt et al., 2012). Additionally, the applicability of the FMFS has been confirmed across a broader array of sociocultural contexts, family stages, and chronic conditions (Knafl et al., 2021). Our study has been the first to utilize multiple aspects of the FMSF as correlates with measures of treatment adherence, which has recently been highlighted as an area where the FMSF may be applied in the clinical setting (Knafl et al., 2021). Additionally, this research remains novel in its application of the FMSF to families affected by a disorder that crosses multiple generations, causes minimal perceptible illness prior to cardiovascular event, and relies predominantly on universally accepted wellness behaviors along with pharmacotherapy in its management (Knafl et al., 2021).

The distinct roles and perspectives held by individuals within a family unit seem particularly relevant to observed child treatment adherence in this sample. Only two aspects of HC+ parental illness perception correlated with observed child treatment adherence, suggesting that personal experience with diagnosis and management may not directly influence the adherence of one's affected offspring. In contrast, multiple aspects of HC- parent's perceptions of HC & HDR moderately or strongly correlated with observed child medication and exercise adherence, particularly their understanding, duration, and perception of controllability by various treatments. Parental perceptions of children's experience with HC and HDR most frequently correlated with observed treatment adherence, potentially indicating that parents see their children as the primary drivers of their own treatment adherence. This may in part be a consequence of the average age of those children considered here, but also supports the most recent recommendations for updating the FMSF to specifically include the perspective of the affected youth; future research should consider

directly surveying affected individuals of school age or older regarding their perceptions of HC and HDR in addition to each parental figure (Beacham & Deatrick, 2019).

While the perceptions of parents affected by genetic hypercholesterolemia did not strongly or frequently correlate with child adherence in this sample, they did correlate with household parenting styles; HC- parents' perceptions correlated with household parenting styles as well, although less frequently and in a distinctly different pattern. The measurement of parenting styles was included here as a measure of parenting philosophy, a subdimension of management behaviors in the FMSF that encapsulates that which "guides the overall approach and specific strategies for condition management" (Beacham & Deatrick, 2019); the generalized nature of parenting styles was hypothesized to have relevance in the production of an environment supportive of the wellness behavior components of HeFH treatment. Parenting styles, when assessed overall and by respondent diagnosis status, also were not frequently or strongly associated directly with observed child treatment adherence. However, the correlations between parental illness perceptions and parenting styles suggest a possible pathway of influence on affected offspring that may result in differences in adherence; that is, personal experience managing HC and HDR (or the lack thereof) may influence the tone or environment set by parents, which may moderate the adherence behaviors of their children. Future research should consider methodologies and pursue sufficient sample sizes to test these potential relationships.

Parenting styles were the only aspect of management behaviors from the FMSF included in this study; likewise, contextual influences were theorized and measured, but due to sample size constraints were not utilized for more complex modeling. Additionally, global family management styles were not defined nor measured here. The findings of this study

suggest that these omitted or under-addressed aspects of the FMSF may be of particular importance to the HeFH population. Illness perceptions across family perspectives most frequently correlated with VAS-measured medication adherence but were often of only weak or moderate strength; less so with measures of lifestyle adherence, and virtually not at all with the measure of medication behavior adherence. Daily pill taking is a habit formed later in treatment when HeFH children have well-informed and previously diagnosed parents; lifestyle habits may be entered into from toddlerhood, whereas HMG CoA-reductase inhibitors are unlikely to be engaged prior to the minimum FDA-approved age of 8 years and thus may require more conscious habit-formation on the part of families. This habit formation results from routines, strategies, and other daily behaviors specifically targeted at treatment; these are addressed in the management approach subdimension of management behaviors in the FMSF, which this study did not measure. It may be that these aspects of management more directly impact adherence, perhaps in manner such as postulated by Hagger and colleagues, and their inclusion should be considered in future research applying the FMSF to the HeFH population (Hagger et al., 2016; Hagger et al., 2018, 2019). Additionally, future research should consider replicating our findings in samples of HeFH+ families where a child represented the proband of condition identification, thereby testing whether our lifestyle adherence findings were indeed related to the context of this sample.

Another potential explanation for our findings may be that family management styles, here represented by adherence as proxy, in fact play a separate moderating role in overall adherence. Family management styles have previously been defined through thematic analysis of the FMSF dimensions as a collective whole, resulting in a description of the overall gestalt of a family's approach to condition management and the perceived

successfulness thereof. Five initial categories elicited from families facing T1DM placed families on a management scale from floundering to thriving; more recent investigation into T2DM – the interventions and consequences of which more closely resemble that of HeFH – described families as thriving, accommodating, indulging, or indifferent (Knafl et al., 1996; Wirattanapoki et al., 2013). Both these sets of descriptors rely in part on a notion of "normal" and whether/to what extent an affected child is perceived to diverge from this (Knafl et al., 2010). However, the literature on HeFH clearly shows that affected individuals across the lifespan perceive themselves as normal and use the concept to support both adherence and nonadherence to treatment (Urke et al., 2019; van Maarle et al., 2001, 2003; Weiner, 2009; Weiner & Durrington, 2008). Indeed, daily pill-taking often defines the condition for those prescribed statins; whereas the lifestyle behaviors indicated for HeFH treatment are also recommended for the general population, medication administration represents a clear diversion from childhood norms, offering yet another potential explanation for why our research found the most frequent associations with VAS-measured medication adherence. This underlying assumption of families seeking "normal" in the FMSF may need reconsideration in the context of a wellness-focused condition, the integration of which may provide further insights into how to best measure the underlying FMSF concepts. Future research should include qualitative or mixed-methodology investigations to further elucidate how illness perceptions relate to management behaviors and management style in the HeFH population.

Implications for Clinical Practice: Intrafamilial HeFH Management

The findings of this study provide initial support for establishing a family-centered model of care for HeFH patients. Application of the FMSF has exposed variations in perspective held by parents affected and unaffected by genetic hypercholesterolemia, their views on the HeFH experience of their children, and differences in how these perceptions directly correlate with parenting styles and observed child adherence. Including all members of a household in consultation and routine care may provide a forum for assessing intrafamilial affectedness by the HeFH through collective discussion of individual experiences, opinions, and perspectives, and through which provide support for adherence through promotion of healthy family affectedness and concern about the diagnosis. Additionally, such a program could provide the framework for education and messaging tailored to each individual's role. For example, our findings suggest initial education for HC-parents may be most supportive of child adherence if targeted to HC understanding, the lifetime nature of HDR, and the efficacy of each treatment modality in its ability to manage HC and HDR both immediately and cross the lifespan. Directly educating children on these topics in a developmentally appropriate manner appears potentially supportive of improved adherence as well. Finally, such a model of care may support continued adherence through the transitional phase of emerging adulthood into adult preventive cardiovascular care by providing an infrastructure to address socio-contextual challenges previously documented as particularly pertinent over that stage of life (Sliwinski et al., 2017).

Study Limitations

As previously noted, the findings of this study must be interpreted in the context of the sample, which was recruited from a single specialty program in the Northeast U.S. and thus is convenient in nature. Families with less education, from diverse racial and ethnic backgrounds, and from varied family structures were not well represented here, nor were families with lesser knowledge of genetic hypercholesterolemia in an affected parent. Additionally, the provision of all surveys in English-written format excluded the participation of HeFH families where parents spoke other primary languages. Finally, while the overall response and participation rates were acceptable, 43.6% of eligible families were unable to be contacted or elected not to participate after contact; differences between participants and nonparticipants cannot be determined. Thus, caution is warranted in generalizing our results to other HeFH samples or the larger HeFH population.

Additional methodological limitations should be considered in interpretation of our findings as well. The PSDQ, while having reasonable reliability among the self-report of the respondent and the household overall, had poor reliability where respondents were asked regarding their parenting partners, highlighting the challenges of self-report measures even when designed for observationally reporting on others. Additionally, the PSDQ was designed with parents of school-aged children or younger in mind; the sample ultimately recruited skewed much more towards parents of adolescents, potentially compromising some of the validity of the tool. Additionally, youth adherence was not directly measured, but rather measured vis-a-vis parental observation. Finally, the small sample size prevented sufficient power to control for cofounders such as sociodemographic characteristics and increased the possibility of Type I and Type II statistical errors, particularly relevant to the analyses of

diagnosis-based subsamples. Future research should consider improving on this work by asking each parent within a household to report their own parenting behaviors, applying different parenting style tools across developmental stages if possible, and surveying youth directly when using self-reported adherence measures (or alternatively, utilizing direct adherence measures). A multi-site study should be considered to obtain a larger sample.

Conclusions

This work provides the first quantitative data on treatment adherence among children affected by HeFH, provides insights into the perceptions of parents in HeFH affected households on how the salient aspects of the condition affects themselves and/or their families, and provides initial information on how these perceptions might directly relate to their children's adherence and their stylistic approach to parenting. As with the self-reported data on HeFH treatment adherence among adults, parents in this sample generally reported that their children adhere well to the three pillars of HeFH management in childhood. They also saw their children as unaffected generally or emotionally by their HeFH despite perceiving them to have little control over their HC or HDR. They expressed concern about children's HC and HDR and believe their children to be concerned too, but that children do not have the same understanding of the condition as themselves which some believed may explain some part of imperfect adherence in childhood. Parents' perceptions of children's experience with HC & HDR correlated more frequently with observed adherence than their own. Future research should seek to confirm that these observations of youth adherence are accurate, and employ qualitative or mixed methodologies with the youth themselves to further assess their experience with HeFH relative to adherence-related factors.

Consistent with the hypothesized intrafamilial influences postulated within the FMSF, differences in perception across parent diagnosis status produced different patterns of relationships with parenting styles and observed youth adherence. While few aspects of illness perception reported of HC+ parents were directly associated with observed adherence, aspects related to understanding, duration, and condition controllability by the various treatment modalities were strongly correlated by report of HC- parents. Variations in parental experience relative to diagnosis status may contribute to the differences observed; HC+ parents' perceptions of their children's condition appeared to be influenced by their perceptions of their own HC & HDR, whereas HC- parents' perceptions of their children's HC & HDR to largely correlated with their perceptions of the family's experience with HC & HDR overall. Additionally, correlations between HC & HDR perceptions suggested that HC+ parents have a nuanced view of how these two concepts relate to each other, whereas HC- parents saw them more similarly. Researchers should consider the differences in parental perspective by diagnosis status when designing further studies into familial experiences with HeFH, ideally including all parental units in a family to investigate these differences and their impact on family adherence.

Interestingly, illness perceptions correlated with parenting style, again varying by parental HC diagnosis status. Authoritarian parenting correlated with higher general affectedness, emotionality, and concern among HC- parents; these same aspects of illness perception correlated with permissiveness in HC+ parents. Both authoritative and authoritarian parenting behaviors correlated with parents' perceptions of child's treatment control by medication and exercise. Although parenting style alone was not directly associated with the majority of measures of observed youth adherence regardless of family

unit or diagnosis status perspective, the relationships between illness perceptions and parenting styles found in this study suggest a potential indirect avenue for the impact of parenting style on youth adherence. Parents with HeFH may consciously or unconsciously modify their approach to parenting based on their experiences managing their own disease. Future investigations should probe this hypothesis further with HeFH diagnosed parents, and if correct, further incorporate it into models of youth and family adherence.

The findings of this study support continued research into the applicability of the FMSF in populations where childhood management is intended to promote long-term health rather than minimize short-term illness, as well as additional use of the FMSF as a tool to elicit additional insights into HeFH families specifically. Our findings show that translation of this midrange theory into nonspecific quantitative measures produces positive correlative relationships with the treatment outcome of adherence, suggesting promise for future research on applications of the FMSF in the clinical setting. Moreover, this research identified aspects of the FMSF that require further elucidation in an HeFH population specifically and highlighted the potential relevance of subdimensions not included in this study. Finally, our findings provide initial support for the theory that parents in HeFH households may promote optimal adherence through the establishment of a household climate relative to their personal experiences with HC & HDR management. The distinctions in perceptions reported by parents across family roles and diagnosis status may be relevant to clinical practice, supporting the integration of education and messaging targeted to each member of a household to support ideal adherence.

Finally, further research will be needed to replicate the findings herein in samples with greater cultural diversity and a broader range of HeFH experiences, ideally using more

direct forms of adherence measure, and including additional considerations for the daily behaviors engaged in for treatment adherence across the three pillars of management. Additional qualitative or mixed-methodology investigation into the assumptions inherent to the FMSF and their applicability or divergence from the HeFH experience would also be illuminating. Family-based interventions and family-centered management of HeFH may also stem from additional research into specific family-based pathways into improved adherence.

APPENDICIES

Figure 1. *The Family Management Style Framework*

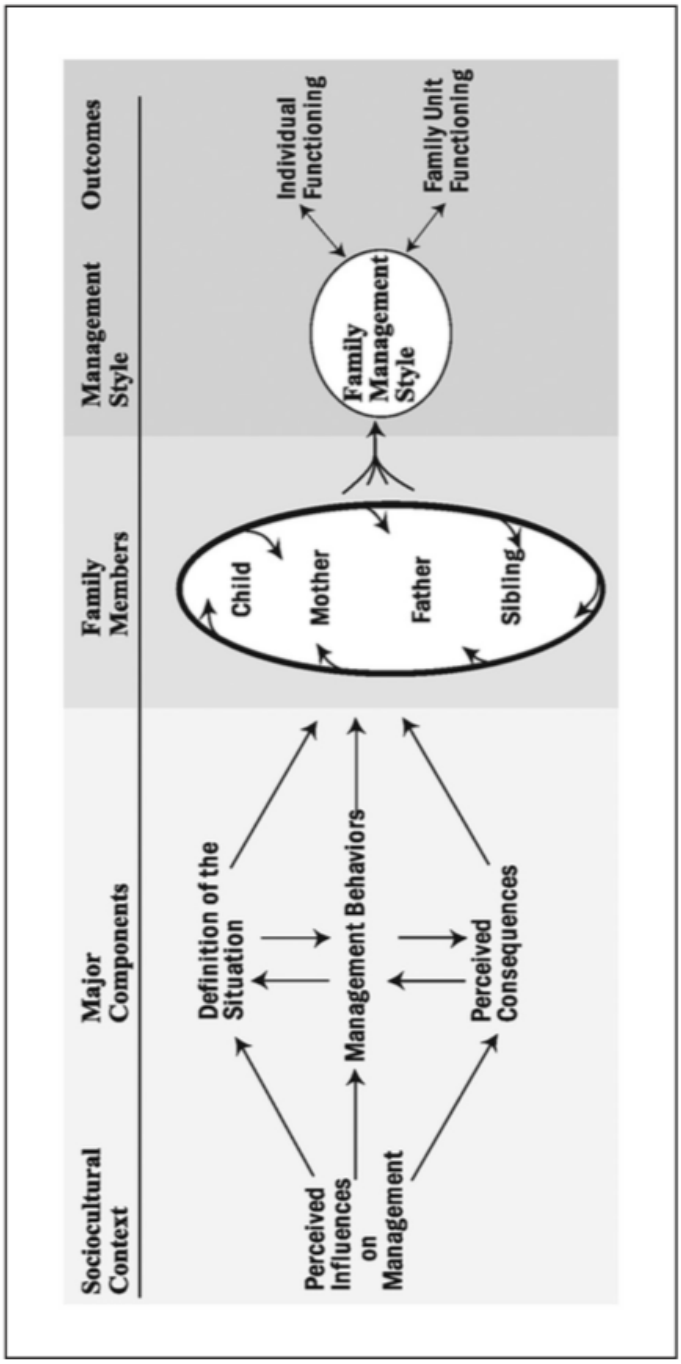


Image From: Knafl, K.; Deatrick, J.; & Havill, N. 2012. Continued Development of the Family Management Style Framework. *Journal of Family Nursing*, 18 (1), p. 25

Figure 2. *Conceptual-Theoretical-Emperical Structure*

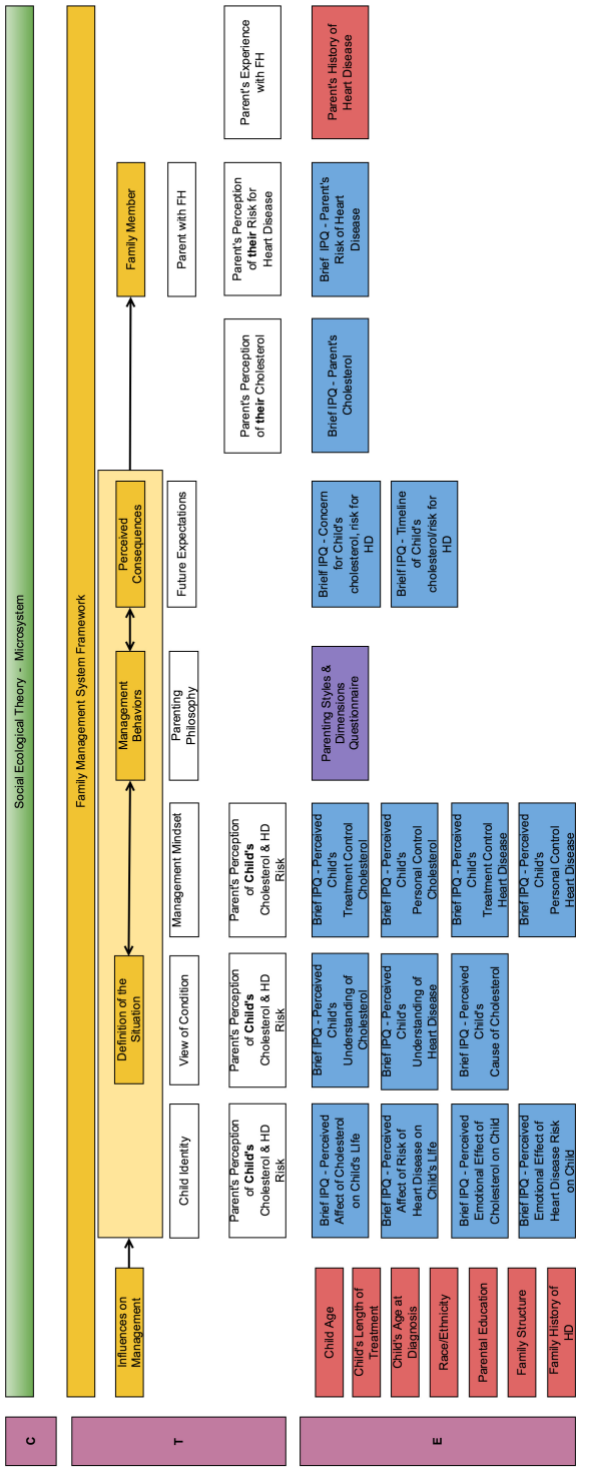


Table 1.

*Delimited Exclusion Criteria: Approved Diagnoses and Treatments**

Asthma	Albuterol PRN Budesonide/Flovent
PCOS	Metformin Oral OCP (or OCP alone)
Acne	Topical medications (Retin-A, antibiotic, etc.)
Environmental allergies	Claritin, Zyrtec, ketotifen, Flonase, Epinephrine IM
Constipation	Miralax PRN
HSV	Valtrex PRN
Migraines	Triptans PRN
Amblyopia	Ocular Atropine
Anxiety	Escitalopram
Depression	Fluoxetine
ADD/ADHD	Adderall, Concerta
Other OTC Treatments	Pain relievers, sleep assistants, vitamins, acid reducers

*Eligibility required two or fewer concurrent conditions, well controlled or in remission with medications as indicated by clinical documentation.

Table 2.

Schedule of Contacts Over Study Period

Week 1	Week 2	Week 3	Week 4	Week 5	Week 6	Week 7
Introduction Letter with weblink-Mail, and email where address was available	Confirmation Phone Call	Email or Postcard with weblink	Follow-up Phone Call	Letter containing paper survey and self-addressed stamped envelope	Follow-up Phone Call	Final Reminder Postcard

Figure 3. *Sample Identification Process, Inclusion Screening*

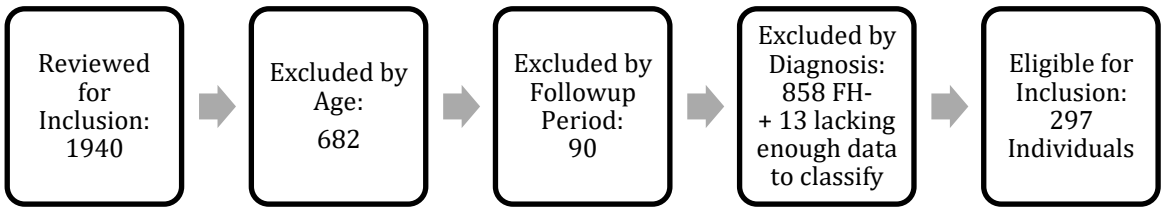


Figure 4. *Sample Identification Process, Exclusion Screening*

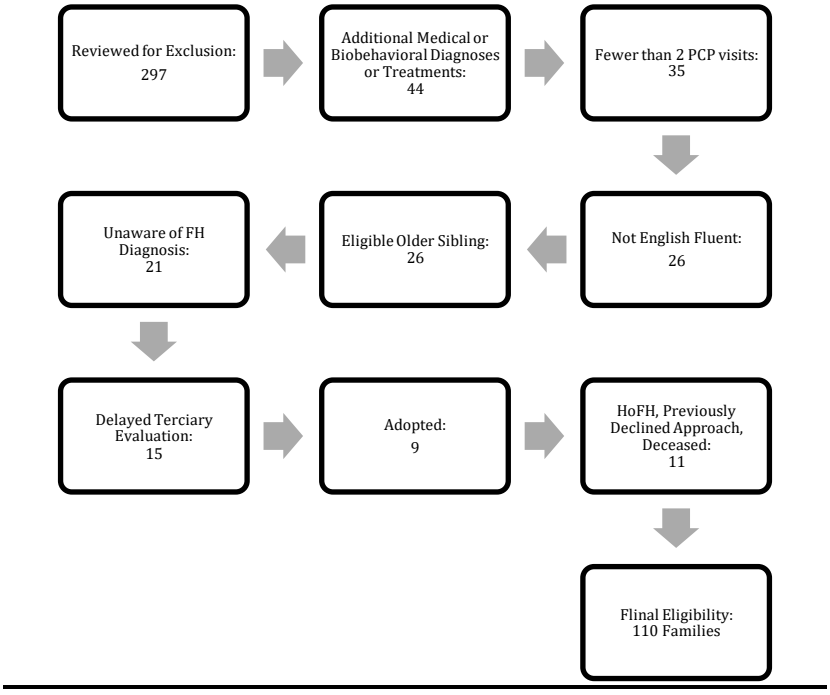


Table 3.

Sociodemographic Characteristics of Study Participants by Cholesterol Diagnosis Status

		Overall	Parent w/ High Chol (n = 36)	Parent w/o High Chol (n = 15)	T-Test P-value
Parent Age (Mean + [SD])	Years	46.1 (5.6)	46.2 (6.6)	46.1 (5.3)	0.9
Parent Gender (n, [%])	Mothers	40 (78.4)	26 (72.2)	14 (93.3)	<0.1
	Fathers	11 (21.6)	10 (27.8)	1 (6.7)	
Child Age (Mean + [SD])	Years	13.4 (3.4)	13.3 (3.3)	13.7 (3.8)	0.7
Child Gender (n, [%])	Daughters	29 (58)	21 (60)	7 (46.7)	0.7
	Sons	21 (42)	14 (40)	8 (53.3)	
Race (n, [%])	White or Caucasian	44 (86.3)	31 (86.1)	13 (86.7)	0.7
	Asian	3 (5.9)	1 (2.8)	2 (13.3)	
	Black or African American	2 (3.9)	2 (5.6)	0	
	Other or None of the Above	2 (3.9)	2 (5.6)	0	
Ethnicity (n, [%])	Hispanic/Latino	3 (5.9)	3 (8.3)	0	0.2
Level of Education (n, [%])	High school diploma or GED	4 (7.8)	3 (8.3)	1 (6.7)	0.9
	Technical/Vocational/Occupational	1 (2)	1 (2.8)	0	
	Associates Degree	2 (3.9)	2 (5.6)	0	
	Bachelors Degree	22 (43.1)	14 (38.9)	8 (53.3)	
	Masters Degree	16 (31.4)	12 (33.3)	4 (26.7)	
	Professional/Doctoral Degree	5 (9.8)	3 (8.3)	2 (13.3)	
	Other	1 (2)	1 (2.8)	0	
Relationship Status (n, [%])	Married	40 (78.4)	30 (83.3)	10 (66.7)	0.3
	Living with Partner	1 (2)	0	1 (6.7)	
	Separated	1 (2)	1 (2.8)	0	
	Divorced	4 (7.8)	2 (5.6)	2 (13.3)	
	Never Married	3 (5.9)	3 (8.3)	0	
	Widowed	1 (2)	0	1 (6.7)	
	Other (Widowed & Remarried)	1 (2)	0	1 (6.7)	
Children in Household (Median + [IQR])		2 [2, 3]	2 [2, 3]	2 [2, 3]	0.3

Table 4.

Family History of Study Participants Pertaining to HeFH

Respondents with HC (n/participants [%])		36/51 (70.6%)
Respondents with HeFH Diagnosis (n/responses [%])	*Yes*	25/36 (69.4%)
	No	7/36 (19.4%)
	No Response	4/36 (11.1%)
Households with Parental HeFH Diagnosis (n/responses [%])		35/51 (68.6%)
Household Time Since Parental HC Diagnosis (n/responses [%])	*> 1 year, < 5 years*	5/51 (9.8%)
	> 5 years, < 10 years	2/51 (3.9%)
	≥10 years	43/51 (84.3%)
	Unknown	1/51 (1.9%)
1st Degree Relatives with history of CVD Event* (n/responses [%])	*Yes*	10/50 (20)
Any Relatives with history of CVD Event (1st or 2nd Degree)* (n/responses[%])	*Yes*	36/51 (70.6)
Number of Family Members with Any Reported History of CVD Event* (Median, [IQR])		1 (0, 2)

HeFH: Heterozygous Familial Hypercholesterolemia
HC: High Cholesterol
CVD: Cardiovascular Disease
**Relative to the diagnosed child*

Table 5.

Child History with HeFH Diagnosis & Adherence to Standard HeFH Treatment Recommendations (n = 51)

Child Age at Diagnosis (Mean [SD])		6.8 years (4.1)
Length of Treatment (Mean [SD])		6 years (3.9)
Visits with Specialist (MD/NP) in Last Two Years (Median [IQR])		2 (2, 3)
Visits with Dietitian (RD) in Last Two Years (Median [IQR])		1 (1, 2.3)
Prescribed Medications (n [%])	Overall	40 (80.0)
	Atorvastatin	30 (76.9)
	Simvastatin	6 (15.4)
	Rosuvastatin	2 (5.1)
	Pravastatin	1 (2.6)
	Zetia	4 (8.5)
Adapted Morisky-Green-Levine Test for Adherence (n [%])	*In the past 30 days, has your child…*	*"Yes"*
	Ever forgotten?	22 (56.4)
	Ever been careless?	11 (29.0)
	Stopped taking d/t feeling better?	0
	Stopped taking d/t feeling poorly?	0
	Missed a dose for any reason?	16 (41.0)
	Perfect Adherence Behavior (0 "Yes" Responses)	14 (35.9)
Medication Adherence (VAS) (Median [IQR])	Estimated % doses taken	94 (90, 100)
Estimated Weekly Physical Activity	Hours/week (Mean [SD])	8.4 (5.1)
	≥ 5 hours weekly (n [%])	36 (73.5)
Servings of Full Fat Dairy & Ice Cream (n [%])	Occasional (≤ 2 servings/week) Regular (3-6 days/week) Frequent (≥1 serving/day)	32 (62.8) 16 (31.4) 3 (5.9)
Servings of Red & Processed Meats (n [%])	Occasional (≤ 2 servings/week) Regular (3-6 days/week) Frequent (≥1 serving/day)	41 (80.4) 10 (19.6) 0
Servings of Other Unhealthy Fats (n [%])	Occasional (≤ 2 servings/week) Regular (3-6 days/week) Frequent (≥1 serving/day)	40 (78.4) 10 (19.6) 1 (2)
PCLS Overall Score (Mean [SD]) Total possible range: 0-36		9.2 (3.6)

Table 6.

Summary of Reliability Coefficients by Instrument and Subsampled Divisions

Instrument	**Subsample**	**N**	**Cronbach's ∝**
Adapted Morisky Green Levine Test		49	0.65
Preventive Cardiology Lifestyle Questionnaire	Saturated Fat Intake	49	0.53
Brief Illness Perceptions Questionnaire	HC+ Parent, HC	35	0.65
	HC+ Parent, HDR	35	0.83
	HC- Parent, HC	14	0.65
	HC- Parent, HDR	15	0.78
	HeFH+ Child, HC	51	0.74
	HeFH+ Child, HDR	50	0.72
Parenting Styles and Dimensions Questionnaire	Respondent	49	0.69
	Parenting Partner	43	0.35
	Overall Household	47	0.68

Table 7.

Comparisons of Perception of High Cholesterol (Median [IQR]) Across Parental Cholesterol Diagnosis Status and Family Unit as Measured by B-IPQ

	Parents with HC (n = 36)	**Parents without HC (n = 15)**	**Children (n = 51)**
Affect (Higher = More affected)	3 (1, 5.5)	6 (4, 8)^	3 (1, 4)‡
Timeline (Higher = Longer)	10 (10, 10)	10 (9, 10)	10 (9, 10)
Personal Control (Higher = More control)	4.5 (2, 6.5)	5 (3, 7)	4 (2, 7)
Treatment Control: Medication (Higher = More control)	10 (8, 10)	10 (8, 10)	10 (8, 10)
Treatment Control: Diet (Higher = More control)	6.5 (3, 8)	5 (3, 10)	5 (3, 8)
Treatment Control: Exercise (Higher = More control)	7 (4, 8)	5 (4, 10)	6 (3, 8)
Concerns (Higher = More concerned)	4 (2, 6.5)	1 (0, 4)	5 (3, 8)†
Understanding (Higher = Greater understanding)	9 (7, 10)	9 (8, 9)	6 (4, 9)†‡
Emotions (Higher = More Emotional)	2 (0, 5)	8 (4, 8)^	2 (1, 5)‡
Parental concern for child's high cholesterol	8 (5, 8)	8 (5, 9)	N/A

Comparisons of similarity = Wilcoxon signed-rank
^ $p < 0.05$, difference between parents with HC and parents without HC
† $p < 0.05$, difference between children and parents with HC
‡ $p < 0.05$, difference between children and parents without HC

HC: High Cholesterol

Table 8.

Comparisons of Perception of Risk for CVD (Median [IQR]) Across Parental Cholesterol Diagnosis Status and Family Unit as Measured by B-IPQ

	Parents with HC (n = 36)	**Parents without HC (n = 15)**	**Children (n = 51)**
Affect (Higher = More affected)	4.5 (1.5, 7)	7 (5, 8)^	3 (1, 5) †‡
Timeline (Higher = Longer)	10 (8.5, 10)*	10 (8, 10)	10 (9, 10)
Personal Control (Higher = More control)	3 (2.5, 5)	3 (2, 5)*	3 (3, 5)*
Treatment Control: Medication (Higher = More control)	9 (7, 10)*	10 (8, 10)	10 (8, 10)
Treatment Control: Diet (Higher = More control)	7 (4.5, 8)	7 (3, 10)	7 (3, 8)*
Treatment Control: Exercise (Higher = More control)	7 (4.5, 8)	7 (3, 10)	7 (4, 8)*
Concerns (Higher = More concerned)	4 (2, 6)	2 (0, 4)	6 (3, 9)*†
Understanding (Higher = Greater understanding)	8 (6, 9)*	9 (7, 9)	5 (3, 7)†‡
Emotions (Higher = More Emotional)	3 (0, 6)	8 (3, 8)^	3 (0, 5) ‡
Parental concern for child's heart disease risk	8 (5, 8)	9 (6, 9)	N/A

Comparisons of similarity = Wilcoxon signed-rank
* $p < 0.05$, within group difference between perceptions of HC and HDR
^ $p < 0.05$, difference between parents with HC and parents without HC
† $p < 0.05$, difference between children and parents with HC
‡ $p < 0.05$, difference between children and parents without HC

HC: High Cholesterol
CVD: Cardiovascular Disease

Table 9. *Correlations Between Items Related to Parents' Perceptions of Hypercholesterolemia and Child's Treatment Adherence Among Parents with High Cholesterol Diagnosis (n = 23)*

	Parent's Perceptions of Personal Hypercholesterolemia									Perceptions of Child Treatment Adherence			
Parent's Perceptions of Personal Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	0.13	1											
Personal Control	0.18	0.25	1										
Treatment Control: Medication	0.13	0.45*	-0.11	1									
Treatment Control: Diet	0.09	-0.23	-0.16	0.31	1								
Treatment Control: Exercise	0.17	-0.23	-0.04	0.28	0.88****	1							
Concerns	-0.48*	-0.25	-0.04	-0.22	0.25	0.28	1						
Understanding	-0.22	0.07	-0.25	0.53**	0.17	0.23	0.01	1					
Emotions	0.60**	-0.02	0.08	0.16	0.21	0.13	-0.53**	-0.07	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	0.04	-0.16	-0.34	0.26	0.02	-0.1	-0.1	0.025	0.04	1			
Medication – Adapted Morisky	0.03	0.03	0.43*	-0.03	-0.06	0.08	-0.19	-0.26	0.12	-0.59**	1		
Saturated Fat Frequency	-0.02	-0.39	0.09	-0.37	-0.07	0.02	0.26	-0.11	0.03	0.02	0.07	1	
Average Weekly Physical Activity	0.33	0.06	0.25	0.04	-0.05	0.1	0.002	0.22	0.19	-0.08	0.19	0.03	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 10. *Correlations Between Items Related to Parents' Perceptions of Heart Disease Risk and Child's Treatment Adherence Among Parents with High Cholesterol Diagnosis (n = 22)*

	Parent's Perceptions of Personal Heart Disease Risk									Perceptions of Child Treatment Adherence			
Parent's Perceptions of Personal Heart Disease Risk	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	0.31	1											
Personal Control	-0.01	0.1	1										
Treatment Control: Medication	0.34	0.61**	-0.43*	1									
Treatment Control: Diet	0.33	0.06	0.14	0.26	1								
Treatment Control: Exercise	0.34	0.03	0.01	0.26	0.93****	1							
Concerns	-0.72***	-0.33	0.22	-0.47*	-0.21	-0.17	1						
Understanding	-0.12	0.33	-0.49*	0.47*	0.14	0.19	0.12	1					
Emotions	0.90****	0.15	0.17	0.22	0.49*	0.44*	-0.56**	-0.23	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	-0.09	0.29	-0.27	0.31	0.1	0.13	0.05	0.2	-0.11	1			
Medication – Adapted Morisky	0.19	0.15	-0.07	0.26	0.2	0.11	-0.39	0.18	0.09	-0.53*	1		
Saturated Fat Frequency	-0.12	-0.29	0.01	-0.32	-0.14	-0.2	0.17	-0.08	0.05	0.12	0.001	1	
Average Weekly Physical Activity	0.46*	0.28	-0.09	0.12	0.18	0.28	-0.06	0.07	0.36	-0.03	0.16	0.16	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 11. *Correlation Between Items Related to Parents' Perceptions of Family Members' Hypercholesterolemia and Child's Treatment Adherence Among Parents Without a High Cholesterol Diagnosis (n = 11)*

	Parent's Perceptions of Family Members' Hypercholesterolemia									Perceptions of Child Treatment Adherence			
Parent's Perceptions of Family Members' Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	0.05	1											
Personal Control	-0.08	0.03	1										
Treatment Control: Medication	0.48	0.1	0.02	1									
Treatment Control: Diet	0.39	-0.45	-0.61*	-0.09	1								
Treatment Control: Exercise	0.39	-0.46	-0.70**	0.07	0.92****	1							
Concerns	-0.09	-0.05	0.57*	-0.03	-0.33	-0.4	1						
Understanding	-0.1	0.49	0.43	0.41	-0.71**	-0.62*	-0.14	1					
Emotions	0.76**	-0.14	0.03	0.52	0.32	0.45	0.11	-0.08	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	-0.06	0.36	0.51	0.39	-0.63*	-0.61*	0.09	0.77**	-0.003	1			
Medication – Adapted Morisky	0.24	-0.17	-0.44	-0.27	0.55	0.51	-0.48	-0.43	-0.01	-0.79***	1		
Saturated Fat Frequency	0.01	-0.14	0.09	0.19	-0.14	0.05	0.33	0.02	0.24	-0.16	-0.22	1	
Average Weekly Physical Activity	0.55	0.1	0.15	0.59*	0	0.02	0.32	-0.02	0.32	-0.12	-0.17	0.19	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 12. *Correlation Between Items Related to Parents' Perceptions of Family Members' Heart Disease Risk and Child's Treatment Adherence Among Parents Without a High Cholesterol Diagnosis (n = 14)*

	Parent's Perceptions of Family Members' Heart Disease Risk									**Perceptions of Child Treatment Adherence**			
Parent's Perceptions of Family Members' Heart Disease Risk	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	0.04	1											
Personal Control	-0.09	0.24	1										
Treatment Control: Medication	0.41	0.13	0.03	1									
Treatment Control: Diet	0.31	-0.27	-0.44	0.27	1								
Treatment Control: Exercise	0.3	-0.27	-0.43	0.31	0.99****	1							
Concerns	0.01	-0.001	-0.01	-0.15	-0.47	-0.54*	1						
Understanding	-0.2	0.3	0.18	0.27	-0.41	-0.38	0.08	1					
Emotions	0.78**	0.11	-0.51	0.32	0.54*	0.55*	-0.09	-0.3	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	0.29	0.24	0.33	0.29	-0.37	-0.37	-0.06	0.06	-0.14	1			
Medication – Adapted Morisky	-0.28	0.08	-0.15	-0.19	0.28	0.31	-0.21	-0.15	0.14	-0.82***	1		
Saturated Fat Frequency	-0.11	-0.39	-0.34	-0.12	-0.22	-0.2	0.48	0.06	-0.06	-0.29	-0.01	1	
Average Weekly Physical Activity	0.29	0.66**	0.09	0.42	0.02	0.04	0.28	0.08	0.41	-0.01	0.01	-0.01	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 13. *Correlation Between Items Related to Parents' Perceptions of Their Child's Hypercholesterolemia Diagnosis and Treatment Adherence (n = 37)*

	Parent's Perceptions of Child's Hypercholesterolemia									**Perceptions of Child Treatment Adherence**			
Parent's Perceptions of Child's Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	-0.08	1											
Personal Control	-0.47**	-0.14	1										
Treatment Control: Medication	-0.05	0.14	-0.25	1									
Treatment Control: Diet	0.55***	-0.1	-0.56***	-0.11	1								
Treatment Control: Exercise	0.50**	-0.22	-0.58***	0.13	0.86****	1							
Concerns	-0.54***	0.13	-0.02	-0.02	-0.31	-0.29	1						
Understanding	-0.21	-0.01	-0.03	0.45**	-0.05	-0.01	-0.07	1					
Emotions	0.76****	-0.2	-0.40**	-0.1	0.51**	0.49**	-0.58***	-0.13	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	0.002	0.34*	-0.05	0.18	-0.19	-0.35*	-0.01	0.21	-0.09	1			
Medication – Adapted Morisky	-0.02	-0.13	-0.09	-0.11	0.14	0.21	0.04	-0.29	0.06	-0.65****	1		
Saturated Fat Frequency	-0.37*	-0.16	0.16	-0.08	-0.28	-0.26	0.2	0.06	-0.09	-0.07	0.02	1	
Average Weekly Physical Activity	0.05	0.23	-0.03	0.28	0.03	0.07	-0.23	0.07	-0.05	-0.06	0.12	-0.01	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 14. *Correlation Between Items Related to Parents' Perceptions of Their Child's Heart Disease Risk and Treatment Adherence (n = 35)*

	Parent's Perceptions of Child's Heart Disease Risk									Perceptions of Child Treatment Adherence			
Parent's Perceptions of Child's Heart Disease Risk	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Affect	1												
Timeline	0.07	1											
Personal Control	-0.22	-0.2	1										
Treatment Control: Medication	-0.1	0.43*	-0.46**	1									
Treatment Control: Diet	0.47**	-0.14	-0.48**	-0.11	1								
Treatment Control: Exercise	0.37*	0.01	-0.43**	0.24	0.72	1							
Concerns	-0.51**	0.05	0.1	-0.06	-0.23	-0.40*	1						
Understanding	-0.06	0.22	-0.41*	0.47**	0.04	0.18	-0.14	1					
Emotions	0.69****	-0.29	-0.08	-0.14	0.40*	0.29	-0.67****	-0.13	1				
Perceptions of Child Treatment Adherence													
Medication – VAS	0.02	0.28	-0.22	0.55***	-0.1	-0.06	-0.16	0.48**	-0.1	1			
Medication – Adapted Morisky	-0.01	0.18	-0.05	-0.06	0.17	0.17	0.19	-0.36*	-0.2	-0.62****	1		
Saturated Fat Frequency	-0.31	-0.22	0.16	-0.08	-0.23	-0.12	0.22	0.13	-0.18	-0.04	0.004	1	
Average Weekly Physical Activity	-0.14	0.37*	-0.16	0.17	-0.05	0.25	-0.11	0.11	-0.17	-0.16	0.23	0.03	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 15.

Summary Scores of Parenting Style Behavior Types as Measured by the PSDQ (Median [IQR])

	Authoritative	*Authoritarian*	*Permissive*
Respondent (n = 50)	4.3 (3.7, 4.7)	1.4 (1.2, 1.6)	1.8 (1.5, 2.3)
Parenting Partner (n = 44)	3.9 (3.4, 4.3)*	1.4 (1.2, 1.7)	2 (1.5, 2.3)
Household (n = 48)	3.9 (2.9, 4.3)**	1.5 (1.2, 2)	1.9 (1.4, 2.3)

1 = Never, 5 = Always

* Difference between perceived self- vs partner authoritative behaviors P = 0.001 (Sign Rank)

** Difference between perceived self- vs household authoritative behaviors P < 0.001 (Sign Rank)

Table 16. *Correlation Between Perceptions of High Cholesterol and Household Average Parenting Behavior Styles Among Parents Diagnosed with High Cholesterol (n = 32)*

	Parent's Perceptions of Personal Hypercholesterolemia									**Household Average Parenting Styles**		
Parent's Perceptions of Personal Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	0.22	1										
Personal Control	0.13	0.17	1									
Treatment Control: Medication	0.21	0.51**	0.01	1								
Treatment Control: Diet	0.14	-0.28	-0.31	0.17	1							
Treatment Control: Exercise	0.21	-0.28	-0.25	0.13	0.92****	1						
Concerns	-0.39*	-0.3	0.11	-0.09	0.27	0.27	1					
Understanding	-0.3	0.03	-0.37*	0.25	0.11	0.14	0.002	1				
Emotions	0.57***	-0.1	0.19	0.03	0.16	0.13	-0.48**	-0.22	1			
Household Average Parenting Styles												
Authoritative	-0.21	-0.01	-0.14	-0.06	-0.09	-0.03	0.24	0.1	-0.37*	1		
Authoritarian	0.12	-0.16	0.14	-0.2	0.08	0.09	0.06	-0.15	0.19	-0.65*	1	
Permissive	0.08	-0.19	0.05	-0.09	-0.001	0.05	-0.05	0.02	0.42*	-0.22	0.52*	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 17. *Correlation Between Perceptions of Heart Disease Risk and Household Average Parenting Behavior Styles Among Parents Diagnosed with High Cholesterol (n = 32)*

	Parent's Perceptions of Personal Heart Disease Risk									**Household Average Parenting Styles**		
Parent's Perceptions of Personal Heart Disease Risk	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	0.33	1										
Personal Control	0.02	-0.09	1									
Treatment Control: Medication	0.09	0.44*	-0.42	1								
Treatment Control: Diet	0.42*	0.12	0.04	0.19	1							
Treatment Control: Exercise	0.45**	0.12	-0.003	0.16	0.88****	1						
Concerns	-0.71****	-0.2	0.06	-0.18	-0.11	-0.13	1					
Understanding	-0.26	0.21	-0.47**	0.45**	-0.1	0.07	0.15	1				
Emotions	0.88****	0.2	0.15	-0.05	0.45**	0.40*	-0.57***	-0.33	1			
Household Average Parenting Styles												
Authoritative	-0.05	0.19	-0.11	-0.09	-0.07	-0.07	0.18	0.04	-0.07	1		
Authoritarian	-0.02	-0.38*	0.36*	-0.2	0.15	0.13	0.03	-0.13	0.08	-0.66****	1	
Permissive	0.42*	-0.16	-0.05	0.02	0.35	0.33	-0.21	-0.2	0.38*	-0.23	0.51**	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 18. *Correlation Between Perceptions of High Cholesterol and Household Average Parenting Behavior Styles Among Parents without High Cholesterol (n = 13)*

	Parent's Perceptions of Family Members' Hypercholesterolemia									**Household Average Parenting Styles**		
Parent's Perceptions of Family Members' Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	-0.16	1										
Personal Control	0.22	-0.05	1									
Treatment Control: Medication	0.32	0.1	-0.01	1								
Treatment Control: Diet	0.43	-0.66*	-0.27	-0.24	1							
Treatment Control: Exercise	0.45	-0.65*	-0.33	-0.09	0.94****	1						
Concerns	-0.05	0.06	0.44	0.03	-0.19	-0.26	1					
Understanding	0.08	0.39	0.53	0.36	-0.58*	-0.48	-0.14	1				
Emotions	0.78**	-0.27	0.18	0.44	0.4	0.52	0.09	0.05	1			
Household Average Parenting Styles												
Authoritative	-0.39	-0.07	-0.49	0.14	0.002	0.08	-0.46	-0.19	-0.02	1		
Authoritarian	0.56*	-0.21	0.1	0.05	0.45	0.42	0.56*	-0.41	0.48	-0.54	1	
Permissive	-0.33	-0.29	-0.19	0.09	0.21	0.22	0.02	-0.35	-0.16	0.35	0.07	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 19. *Correlation Between Perceptions of Risk for Heart Disease and Household Average Parenting Behavior Styles Among Parents without High Cholesterol (n = 14)*

	Parent's Perceptions of Family Members' Heart Disease Risk									**Household Average Parenting Styles**		
Parent's Perceptions of Family Members' Heart Disease Risk	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	-0.01	1										
Personal Control	-0.01	0.22	1									
Treatment Control: Medication	0.37	0.13	0.003	1								
Treatment Control: Diet	0.46	-0.44	-0.19	0.15	1							
Treatment Control: Exercise	0.45	-0.44	-0.18	0.19	0.99****	1						
Concerns	-0.05	0.12	-0.16	-0.06	-0.45	-0.55*	1					
Understanding	-0.12	0.28	0.25	0.25	-0.41	-0.38	0.06	1				
Emotions	0.81***	-0.03	-0.25	0.23	0.60*	0.60*	-0.14	-0.23	1			
Household Average Parenting Styles												
Authoritative	-0.03	-0.4	-0.29	0.41	0.24	0.28	-0.29	-0.3	-0.06	1		
Authoritarian	0.37	0.16	-0.34	-0.14	0.21	0.2	0.42	-0.22	0.54*	-0.49	1	
Permissive	-0.02	-0.15	-0.01	0.15	0.06	0.08	0.2	-0.2	-0.02	0.31	0.15	1

Spearman's Rho selected d/t non-normal distribution of some response categories
* p </=0.05
** p </= 0.01
*** p </= 0.001
****p </= 0.0001

Table 20. *Correlation Between Parental Perceptions of Child's High Cholesterol and Household Average Parenting Behavior Styles (n = 47)*

Parent's Perceptions of Child's Hypercholesterolemia	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	-0.26	1										
Personal Control	-0.32*	0.05	1									
Treatment Control: Medication	0.01	0.13	-0.22	1								
Treatment Control: Diet	0.58****	-0.23	-0.51***	-0.09	1							
Treatment Control: Exercise	0.58****	-0.27	-0.56****	0.02	0.91****	1						
Concerns	-0.57****	0.11	0.04	0.08	-0.41**	-0.39**	1					
Understanding	-0.11	0.13	-0.14	0.25	0.09	0.1	-0.25	1				
Emotions	0.72****	-0.27	-0.22	-0.19	0.49***	0.57****	-0.62****	0.05	1			
Household Average Parenting Styles												
Authoritative	-0.13	0.21	0.01	0.31*	-0.25	-0.17	0.34*	-0.01	-0.15	1		
Authoritarian	0.15	-0.16	-0.004	-0.31*	0.32*	0.24	-0.25	-0.07	0.17	-0.63****	1	
Permissive	0.24	-0.17	-0.01	-0.04	0.25	0.17	-0.15	-0.06	0.24	-0.15	0.45**	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 21. *Correlation Between Parental Perceptions of Child's Risk for Heart Disease and Household Average Parenting Behavior Styles (n = 45)*

	Parent's Perceptions of Child's Risk for Heart Disease									Household Average Parenting Styles		
Parent's Perceptions of Child's Risk for Heart Disease	Affect	Timeline	Personal Control	Treatment Control: Medication	Treatment Control: Diet	Treatment Control: Exercise	Concerns	Understanding	Emotions	Authoritative	Authoritarian	Permissive
Affect	1											
Timeline	-0.04	1										
Personal Control	-0.2	-0.17	1									
Treatment Control: Medication	-0.2	0.28	-0.31*	1								
Treatment Control: Diet	0.43**	-0.16	-0.37**	0.02	1							
Treatment Control: Exercise	0.22	-0.08	-0.30*	0.13	0.69****	1						
Concerns	-0.56***	0.12	-0.04	0.11	-0.24	-0.28	1					
Understanding	-0.01	0.09	-0.25	0.21	0.07	0.12	-0.34*	1				
Emotions	0.79****	-0.16	-0.1	-0.2	0.32*	0.2	-0.73****	0.15	1			
Household Average Parenting Styles												
Authoritative	-0.27	0.21	-0.17	0.25	-0.27	-0.21	0.23	-0.02	-0.15	1		
Authoritarian	0.13	-0.17	0.21	-0.23	0.22	0.32*	-0.23	0.01	0.17	-0.65****	1	
Permissive	0.1	-0.07	-0.09	0.02	0.25	0.15	-0.17	0.01	0.28	-0.16	0.45**	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 22.

Correlations Between Respondent's Parenting Styles and Perceived Child Adherence to Treatment (n = 37)

Respondents' Parenting Style				**Perceptions of Child Treatment Adherence**			
Respondents' Parenting Style	Authoritative	Authoritarian	Permissive	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Authoritative	1						
Authoritarian	-0.50**	1					
Permissive	-0.32	0.63****	1				
Perceptions of Child Treatment Adherence							
Medication – VAS	0.02	-0.25	-0.14	1			
Medication – Adapted Morisky	-0.12	0.32	0.2	-0.65****	1		
Saturated Fat Frequency	0.08	0.03	-0.005	-0.07	0.02	1	
Average Weekly Physical Activity	0.13	0.04	-0.04	-0.06	0.12	-0.01	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 23.

Correlations Between Parenting Partners' Parenting Styles and Perceived Child Adherence to Treatment (n = 32)

Parenting Partners' Parenting Style				**Perceptions of Child Treatment Adherence**			
Parenting Partners' Parenting Style	Authoritative	Authoritarian	Permissive	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Authoritative	1						
Authoritarian	-0.27	1					
Permissive	-0.13	0.45**	1				
Perceptions of Child Treatment Adherence							
Medication – VAS	0.07	-0.52**	-0.21	1			
Medication – Adapted Morisky	-0.28	0.39*	0.19	-0.74****	1		
Saturated Fat Frequency	0.23	-0.05	-0.06	-0.09	-0.08	1	
Average Weekly Physical Activity	0.28	0.22	0.11	-0.11	0.02	-0.05	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 24.

Correlations Between Average Household Parenting Styles and Perceived Child Adherence to Treatment (n = 35)

Household Average Parenting Styles				**Perceptions of Child Treatment Adherence**			
Household Average Parenting Styles	Authoritative	Authoritarian	Permissive	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical
Authoritative	1						
Authoritarian	-0.59***	1					
Permissive	-0.27	0.48**	1				
Perceptions of Child Treatment Adherence							
Medication – VAS	0.15	-0.40*	0.2	1			
Medication – Adapted Morisky	-0.19	0.26	0.23	-0.72****	1		
Saturated Fat Frequency	0.05	0.12	0.03	-0.1	-0.04	1	
Average Weekly Physical Activity	-0.06	0.32	0.06	-0.09	0.06	-0.06	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 25. *Correlations Between Aspects of Clinical Family History, Child Diagnostic and Treatment History, and Perceived Child Adherence to Treatment (n = 35)*

	Parental History with FH		Child History with HeFH						Perceptions of Child Treatment Adherence			
Parental History with HeFH	Knowledge of HC, Duration	Formal Diagnosis with HeFH	First Degree Relative with HD	Any Relative with HD	Age at HC Diagnosis	Length of HeFH Treatment	Average # Specialist Visits	Average # RD Visits	Medication – VAS	Medication – Adapted Morisky	Saturated Fat Frequency	Average Weekly Physical Activity
Knowledge of HC, Duration	1											
Formal Diagnosis with HeFH	0.11	1										
Child History with HeFH												
First Degree Relative with Heart Disease	0.11	-0.11	1									
Any Relative with Heart Disease	0.43**	-0.04	0.29	1								
Age at HC Diagnosis	-0.19	-0.16	-0.18	-0.2	1							
Length of HeFH Treatment	0.23	0.14	0.17	0.22	-0.78****	1						
Average # Specialist Visits	-0.09	0.07	0.16	-0.25	-0.26	-0.4	1					
Average # Registered Dietician Visits	-0.25	0.07	0.03	-0.06	0.2	-0.40*	0.36*	1				
Perceptions of Child Treatment Adherence												
Medication – VAS	0.1	0.38*	0.29	-0.05	-0.27	0.15	0.35*	-0.02	1			
Medication – Adapted Morisky	0.07	-0.19	-0.17	-0.01	0.1	-0.1	-0.33	0.03	-0.71****	1		
Saturated Fat Frequency	0.01	-0.54***	0.05	0.04	0.15	0.12	0.26	0.28	-0.15	0.002	1	
Average Weekly Physical Activity	-0.11	-0.07	-0.14	0.08	-0.04	0.03	-0.04	0.09	-0.16	0.14	-0.08	1

Spearman's Rho selected d/t non-normal distribution of some response categories

* p </=0.05

** p </= 0.01

*** p </= 0.001

****p </= 0.0001

Table 26.

Qualitative Post-Survey Remarks by Respondents

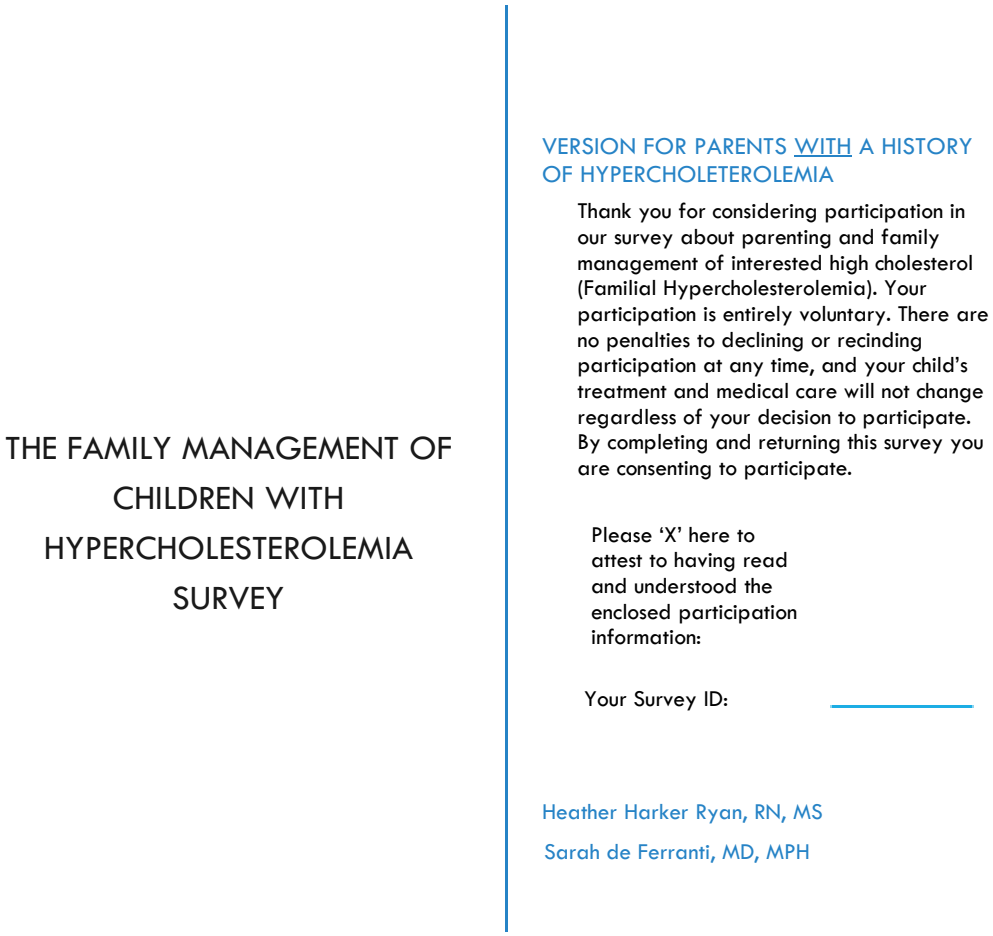
Mother, Age 43, HC+	The medication that they provide is difficult to swallow for young children just learning to swallow pills, other alternatives should be available.
Mother, Age 43, HC-	I would say that physical activity has been far more difficult to incorporate with COVID-19. We have made the decision to allow team sports for his mental and physical wellness, though there's some level of angst with that.
Father, Age 44, HC-	My child's biological mother (my wife at the time) died at age 39 from a heart attack due to her genetic cholesterol issue. After this happened, I tested the children and discovered that the younger child [had] the same genetic issue. We have been mitigating her cholesterol levels with diet, exercise, and by medication (after she reached the age of 10). Exercise has been recently limited due to coronavirus. We now test every 6 months and have been maintaining below the threshold level of LDL. We would be happy to share our methodology with anyone interested.
Mother, Age 52, HC+	She is one of three children and 2 of them have high cholesterol. My sister's heart attack was at 35-years-old with quadruple bypass surgery.
Mother, Age 44, HC+	She was anxious about it when she first found out; since she's been on medication, she has not expressed anxiety about this.
Mother, Age 45, HC+	Care and understand the importance but really do not stick to taking medicine consistently. I don't have a good answer for why.
Mother, Age 31, HC-	This is still very new for us! So we have only had 2 months of medication- so far so good.
Father, Age 47, HC+	Diet is becoming more challenging as she is getting older. She makes poor choices if she is not with her parents.
Mother, Age 49, HC+	The nutritional counseling was poor. In my opinion the advice give around diet and exercise shouldn't have been grounded in cholesterol management, but more towards mitigation of other CV risks
Mother, Age 50, HC+	Thanks to the team at [the clinic] we feel both are children have the knowledge, skills and support to successfully manage their cholesterol and overall health. Staying and keeping healthy connects our family and we have fun doing it!
Mother, Age 53, HC-	[Our doctor] has been excellent!
Mother, Age 48, HC+	I am intrigued by the study that says that familial hypercholesterolemia does better with a dietary limited in gluten as opposed to focusing solely on saturated fat. At one point we eliminated all but 15 grams of sat fat a day and his cholesterol was still sky high. We were doing everything right dietarily to no affect. Short of becoming vegan, there wasn't anything left for us to do.
Father, Age 45, HC+	She is still very early in her healthcare journey. I expect her awareness and understanding will grow over time as mine did. I also expect she will start on statins around 11 years old, whereas I started at 18.
Mother, Age 47, HC+	We hope as does he, that he'll have a firmer understanding of the role of diet in cholesterol, when he gets a little older. Our goal is to keep him off medicine and hope he as able to combined his high level of activity with better diet.

Table 26.

Qualitative Post-Survey Remarks by Respondents (Con'd)

Mother, Age 51, HC-	I have three girls who have been followed for high cholesterol since they were toddlers. While I understand the importance of diet, I have found that there is some insensitivity to body image issues and the difficulty controlling FHC with diet.
Mother, Age 51, HC-	I and his father are both physicians. I am an adult cardiologist so perhaps am more aware of the biologic basis and treatment. He also is small for his age and is undergoing growth hormone therapy. Having him eat his meals is important for growths and it is challenging to meet this with a low fat cardiac diet at his age.
Mother, Age 46, HC-	We lost my husband due to a heart attack about five years ago. My kids and I were present and it was a pretty traumatic experience.
Mother, Age 51, HC+	Because my younger sister had a heart attack at 35 and quadruple bypass surgery and lives with a pacemaker, we have taken diet and exercise seriously. We have good food/lifestyle choices that I hope will continue into adulthood.

Figure 5. *Survey for HC+ Respondents*



THE FAMILY MANAGEMENT OF CHILDREN WITH HYPERCHOLESTEROLEMIA SURVEY

VERSION FOR PARENTS WITH A HISTORY OF HYPERCHOLETEROLEMIA

Thank you for considering participation in our survey about parenting and family management of interested high cholesterol (Familial Hypercholesterolemia). Your participation is entirely voluntary. There are no penalties to declining or recinding participation at any time, and your child's treatment and medical care will not change regardless of your decision to participate. By completing and returning this survey you are consenting to participate.

Please 'X' here to attest to having read and understood the enclosed participation information:

Your Survey ID: _____

Heather Harker Ryan, RN, MS

Sarah de Ferranti, MD, MPH

Please read the questions carefully, as some questions look similar but refer to different members of the family. If you have more than one child with a diagnosis of FH, please answer questions as they apply to your oldest child. If you have multiples affected by high cholesterol (ie., twins, triplets, etc.), please answer questions as they relate to the child whose cholesterol is highest. Some questions may not apply to you; bold and italicized directions will instruct you to which question to proceed to when this is the case.

Personal & Family History of High Cholesterol and Heart Disease

To start, we would like to know a bit about your cardiovascular health history, and the cardiovascular health history of some of your family members. Some of these questions are worded from the perspective of your child, so please read carefully.

1. How long have you been aware that your cholesterol is elevated?

   ☐ Less than 6 months ago
   ☐ 6 months – 1 year ago
   ☐ >1 year but <5 years ago
   ☐ >5 years but <10 years ago
   ☐ >10 years ago

2. Have you ever been told that your cholesterol elevation is likely genetic or inherited in nature?

   ☐ Yes ☐ No → ***Skip to Q5***

3. How long have you been aware that your cholesterol elevation is likely genetic or inherited in nature?

   ☐ Less than 6 months ago
   ☐ 6 months – 1 year ago
   ☐ >1 year but <5 years ago
   ☐ >5 years but <10 years ago
   ☐ >10 years ago

4. Have you ever received a diagnosis of Familial Hypercholesterolemia?

   ☐ Yes ☐ No

5. Does your child's other biological parent have high cholesterol?

   ☐ Yes ☐ No → ***Skip to Q10***

6. How long has your child's other biological parent been aware that their cholesterol is elevated?

   ☐ Less than 6 months ago
   ☐ 6 months – 1 year ago
   ☐ >1 year but <5 years ago
   ☐ >5 years but <10 years ago
   ☐ >10 years ago

7. Has your child's other biological parent ever been told that their cholesterol elevation is likely genetic or inherited in nature?

☐ Yes ☐ No → ***Skip to Q10***

8. How long has your child's other biological parent been aware that their cholesterol elevation is likely genetic or inherited in nature?

☐ Less than 6 months ago
☐ 6 months – 1 year ago
☐ >1 year but <5 years ago
☐ >5 years but <10 years ago
☐ >10 years ago

9. Has your child's other biological parent been ever received a diagnosis of Familial Hypercholesterolemia?

☐ Yes ☐ No

10. How many of <u>your child's</u> **first-degree family members (parents, siblings)** have been diagnosed with high cholesterol?

_____ Family Members

11. How many of <u>your child's</u> **second-degree family members (grandparents, aunts, uncles)** have been diagnosed with high cholesterol?

_____ Family Members

12. Have any of <u>your child's</u> **first-degree family members (parents, siblings)** ever had a myocardial infarction (also known as a heart attack), stroke, a positive exercise or stress test, open heart surgery, coronary artery stenting, or have a diagnosis of coronary artery disease or peripheral vascular disease?

☐ Yes ☐ No → ***Skip to Q14***

13. How many of <u>your child's</u> **first-degree family members (parents, siblings)** have ever had a myocardial infarction (also known as a heart attack), stroke, a positive exercise or stress test, open heart surgery, coronary artery stenting, or a diagnosis of coronary artery disease or peripheral vascular disease?

_____ Family Members

14. Have any of <u>your child's</u> **second-degree family members (grandparents, aunts, uncles)** ever had a myocardial infarction (also known as a heart attack), stroke, a positive exercise or stress test, open heart surgery, coronary artery stenting, or have a diagnosis of coronary artery disease or peripheral vascular disease?

☐ Yes ☐ No → ***Skip to Q16***

15. How many of <u>your child's</u> **second-degree family members (grandparents, aunts, uncles)** have ever had a myocardial infarction (also known as a heart attack), stroke, a positive exercise or stress test, open heart surgery, coronary artery stenting, or a diagnosis of coronary artery disease or peripheral vascular disease?

_____ Family Members

Illness Perceptions Questionnaires – Personal History of High Cholesterol & Risk of Heart Disease

*The following questions ask for your thoughts about **your** diagnosis of high cholesterol. There are no right or wrong answers, we just want to know what you think. Please circle the number that best corresponds to your views:*

16. How much does *your* high cholesterol affect your life?

0	1	2	3	4	5	6	7	8	9	10
No Affect										*Severely Affects*

17. How long do you think *your* high cholesterol will continue?

0	1	2	3	4	5	6	7	8	9	10
A Very Short Time										*Forever*

18. How much control do you think *you* have over your high cholesterol? (That is, to what extent do you think you can personally do things to improve your cholesterol?)

0	1	2	3	4	5	6	7	8	9	10
Absolutely No Control										*Completely Control*

19. How much do you think medications can help lower *your* high cholesterol?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

20. How much do you think dietary interventions can help lower *your* high cholesterol?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

21. How much do you think exercise can help lower *your* high cholesterol?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

22. How concerned are you about *your* high cholesterol?

0	1	2	3	4	5	6	7	8	9	10
Not Concerned At All										*Extremely Concerned*

23. How well do you feel you understand *your* high cholesterol?

0	1	2	3	4	5	6	7	8	9	10
Don't Understand At All										*Completely Understand*

24. How much does *your* high cholesterol affect *you* emotionally?

0	1	2	3	4	5	6	7	8	9	10
Not At All										*Extremely*

25. What do you believe causes your high cholesterol? Please list anything you believe caused your high cholesterol in order of most causative to least causative.

I. _____.

II. _____.

III. _____.

The following questions ask for your thoughts about ***your*** *risk for heart disease.* ***These questions may look very similar to those you have previously answered; please pay close attention to changes in the concept of interest*** *and circle the number that best corresponds to your views:*

26. How much does *your* risk for heart disease affect your life?

0	1	2	3	4	5	6	7	8	9	10
No Affect										*Severely Affects*

27. How long do you think *your* risk for heart disease will continue?

0	1	2	3	4	5	6	7	8	9	10
A Very Short Time										*Forever*

28. How much control do you think *you* have over *your* risk for heart disease? (i.e., to what extent do you think you can personally do things to improve your risk for heart disease?)

0	1	2	3	4	5	6	7	8	9	10
Absolutely No Control										*Completely Control*

29. How much do you think medications can help lower *your* risk for heart disease?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

30. How much do you think dietary interventions can help lower *your* risk for heart disease?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

31. How much do you think exercise can help lower *your* risk for heart disease?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

32. How concerned are you about *your* risk for heart disease?

0	1	2	3	4	5	6	7	8	9	10
Not Concerned At All										*Extremely Concerned*

33. How well do you feel you understand *your* risk for heart disease?

0	1	2	3	4	5	6	7	8	9	10
Don't Understand At All										*Very Clearly Understand*

34. How much does *your* risk for heart disease affect *you* emotionally?

0	1	2	3	4	5	6	7	8	9	10
Not At All Affected										*Extremely Affected*

35. What do you believe causes your risk for heart disease? Please list anything you believe caused your risk for heart disease in order of most causative to least causative.

I. _____.

II. _____.

III. _____.

Your Child's History of High Cholesterol

*In this section we would like you to tell us about **your child** with high cholesterol and their management. If you have multiple children affected by high cholesterol, please answer the questions as they relate to your oldest child. If you have multiples affected by high cholesterol (ie., twins, triplets, etc.), please answer questions as they relate to the child whose cholesterol is highest.*

36. How old is your child?

_____ Years Old

37. How old was your child when they were diagnosed with high cholesterol?

_____ Years Old

38. How old was your child when they first received counseling from a cholesterol specialist (MD/NP) about their cholesterol?

_____ Years Old

39. Approximately how many times has your child seen a cholesterol specialist (MD/NP) to address their cholesterol in the past two years?

_____ Visits

40. Has your child ever seen a nutritionist or dietitian to address their cholesterol?

☐ Yes ☐ No → ***Skip to Q42***

41. Approximately how many times has your child seen a nutritionist or dietitian to address their cholesterol in the past two years?

_____ Visits

42. Has your child's cholesterol specialist (MD/NP) ever recommended a prescription medication to lower their cholesterol level?

☐ Yes ☐ No → ***Skip to Q51***

43. Has your child's cholesterol specialist (MD/NP) ever written a medication prescription to lower their cholesterol?

☐ Yes ☐ No → ***Skip to Q51***

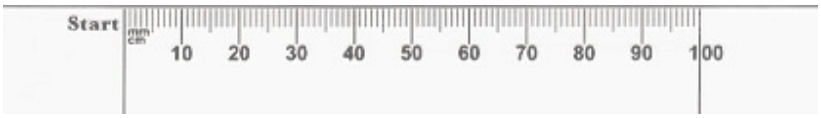
44. What cholesterol lowering medication does your child's cholesterol specialist currently prescribe?

☐ Simvastatin
☐ Pravastatin
☐ Atorvastatin
☐ Rosuvastatin
☐ Questran
☐ Welchol
☐ Supplements: _____
☐ Prescribed, but not taking → ***Skip to Q51***

The next questions are about how regularly your child takes their cholesterol lowering medications. Please answer them to the best of your ability.

Over the Past Thirty (30) Days…	Yes	No
45. … has your child ever forgotten to take their cholesterol lowering medication?	☐	☐
46. … has your child at times been careless about taking their cholesterol lowering medication?	☐	☐
47. … has your child ever stopped taking their cholesterol lowering medication because they felt better?	☐	☐
48. … has your child ever stopped taking their cholesterol lowering medication because they felt poorly?	☐	☐
49. … has your child ever missed taking their cholesterol lowering medication for any reason?	☐	☐

50. Over the past thirty (30) days, what percentage of your child's cholesterol lowering medication doses would you estimate he/she took? *Please mark on the corresponding number on the ruler below.*



The following questions are about your child's usual diet and physical activity habits over the past month. Please answer them to the best of your ability.

Thinking back over the past month, please check the response that best represents how often your child ate a serving of each food or drink.	Never	<1 day per WEEK	1-2 days per WEEK	3-4 days per WEEK	5-6 days per WEEK	1 time per DAY	≥ 2 times per DAY
51. Regular fat ice cream	☐	☐	☐	☐	☐	☐	☐
52. Butter, regular fat dairy (milk, cheese, yogurt, cottage cheese)	☐	☐	☐	☐	☐	☐	☐
53. Fresh red meats including beef, lamb, pork	☐	☐	☐	☐	☐	☐	☐
54. Processed meets including deli meats, bacon, salami	☐	☐	☐	☐	☐	☐	☐
55. Egg yolks, shrimp, squid, liver/pate	☐	☐	☐	☐	☐	☐	☐
56. Pizza	☐	☐	☐	☐	☐	☐	☐
57. Fried foods (fries, nuggets, chicken wings, fried fish)	☐	☐	☐	☐	☐	☐	☐

Next, over the past month, on average how much time per week did your child spend…	Never	<1 day per WEEK	1-2 days per WEEK	3-4 days per WEEK	5-6 days per WEEK	1 time per DAY	≥ 2 times per DAY
58. Walking for exercise or transport	☐	☐	☐	☐	☐	☐	☐
59. In gym/PE at school	☐	☐	☐	☐	☐	☐	☐
60. Playing on a competitive sports team	☐	☐	☐	☐	☐	☐	☐
61. Other activity: _____	☐	☐	☐	☐	☐	☐	☐
62. Now indicate the total hours of activity your child gets per week:							

High Cholesterol, Heart Disease, and Your Child

The following questions ask for your thoughts about how <u>you and your child</u> think and feel about <u>their high cholesterol</u>. ***These questions may look very similar to those you have previously answered; please pay close attention to changes in the individual and concept of interest****. If you have multiple children affected by high cholesterol, please answer the questions as they relate to your oldest child. If you have multiples affected by high cholesterol, (i.e., twins, triplets, etc.), please answer as they related to the child whose cholesterol is highest.*

Please circle the number that best corresponds to your, or your understanding of your child's, views:

63. How much does *high cholesterol* affect <u>your child's</u> life?

0	1	2	3	4	5	6	7	8	9	10
No Affect										*Severely Affects*

64. How long do you think <u>your child's *high cholesterol*</u> will continue?

0	1	2	3	4	5	6	7	8	9	10
A Very Short Time										*Forever*

65. How much control do you think <u>your child</u> has over their *high cholesterol*? (That is, to what extent do you feel your child can personally do things to improve their cholesterol?)

0	1	2	3	4	5	6	7	8	9	10
Absolutely No Control										*Completely Control*

66. How much do you think medications can help lower <u>your child's *high cholesterol*</u>?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

67. How much do you think dietary interventions can help lower <u>your child's *high cholesterol*</u>?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

68. How much do you think exercise can help lower <u>your child's *high cholesterol*</u>?

0	1	2	3	4	5	6	7	8	9	10
Not At All Helpful										*Extremely Helpful*

69. How concerned are you about <u>your child's *high cholesterol*</u>?

0	1	2	3	4	5	6	7	8	9	10
Not Concerned At All										*Extremely Concerned*

70. How concerned are <u>you</u> about <u>your child's *high cholesterol*</u>?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

71. How concerned is <u>your child</u> about their *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

72. How well do you feel <u>your child</u> understands their *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Don't Understand At All *Very Clearly Understand*

73. How much does having *high cholesterol* affect <u>your child</u> emotionally?

0 1 2 3 4 5 6 7 8 9 10

Not At All Affected *Extremely Affected*

74. What do you believe causes <u>your child's *high cholesterol*</u>? Please list anything you believe caused your child's high cholesterol in order of most causative to least causative.

I. _____.

II. _____.

III. _____.

The following questions ask for your thoughts about how <u>you and your child</u> think and feel about <u>their risk for future heart disease</u>. ***These questions may look very similar to those you have previously answered; please pay close attention to changes in the individual and concept of interest.***

Please circle the number that best corresponds to your, or your understanding of your child's, views:

75. How much does having a *risk for future heart disease* affect <u>your child's</u> life?

0 1 2 3 4 5 6 7 8 9 10

No Affect *Severely Affects*

76. How long do you think <u>your child's *risk for future heart disease*</u> will continue?

0 1 2 3 4 5 6 7 8 9 10

A Very Short Time *Forever*

77. How much control do you feel your child has over their *risk for future heart disease*? (i.e., to what extent do you think your child can personally do things to improve their risk for heart disease?)

0 1 2 3 4 5 6 7 8 9 10

Absolutely No Control *Complete Control*

78. How much do you think medications can help lower your child's *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

79. How much do you think dietary interventions can help lower your child's *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

80. How much do you think exercise can help lower your child's *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

81. How concerned are you about your child's *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

82. How concerned is your child about their *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

83. How well do you feel your child understand their *risk for future heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Don't Understand At All *Very Clearly Understand*

84. How much does having a *risk for future heart disease* affect your child emotionally?

0 1 2 3 4 5 6 7 8 9 10

Not At All Affected *Extremely Affected*

General Parenting Styles and Behaviors Used in the Family

These questions relate to how you and your child's other biological parent approach parenting in general. Please read all statements carefully. There may be some questions you think are not applicable to your family or child; please try to answer these questions to the best of your ability. At times, there may be questions you might think: "I would like to act this way, but in reality I am not doing this". Please answer these questions by indicating what you are actually doing.

In the text boxes next to each item, please note the frequency of each behavior (Never, Once in a While, About Half of the Time, Very Often, Always). ***First answer about yourself, and then your child's other biological parent.*** *If your child does not have another biological parent active in their care, please answer for any other parental figure, or leave blank.*

1 = Never 2 = Once in a While 3 = About Half of the Time 4 = Very Often 5 = Always

	I…	*He/ She/ They…*	*Parenting Behaviors*
85.			… encourage our child to talk about their troubles.
86.			… find it difficult to discipline our child.
87.			… give praise when our child is good.
88.			… spank when our child is disobedient.
89.			… punish by taking privileges away from our child with little if any explanation.
90.			… spoil our child.
91.			… give comfort and understanding when our child is upset.
92.			… yell or shout when our child misbehaves.
93.			… scold and criticize to make our child improve.
94.			… grab our child when being disobedient.
95.			… state punishments to our child but do not actually do them.
96.			… am/are responsive to our child's feelings or needs.
97.			… allow our child to give input into family rules.
98.			… give our child reasons why rules should be obeyed.
99.			… help our child to understand the impact of behavior by encouraging our child to talk about the consequences of own actions.
100.			… take our child's desires into account before asking the child to do something.
101.			… explode in anger towards our child.
102.			… threaten our child with punishment more often than actually giving it.
103.			… use physical punishment as a way of disciplining our child.
104.			… have warm and intimate times together with our child.

	I…	He/ She/ They…	Parenting Behaviors
105.			… encourage our child to freely express themselves even when disagreeing with us.
106.			… scold or criticize when our child's behavior does not meet expectations.
107.			… show respect for our child's opinions by encouraging them to express themselves.
108.			… explain to our child how we feel about the child's good and bad behavior.
109.			… use threats as punishment with little or no justification.
110.			… take into account our child's preferences in making plans for the family.
111.			When our child asks why [he/she] has to conform, [I/they] state: because I said so, or [I am/they are] the parent and want you to.
112.			… explain the consequences of the child's behavior.
113.			… emphasize the reasons for rules.

Demographic Information

114. How old are you?

_____ Years Old

115. How many children do you have?

_____ Children

116. What is your gender?

☐ Male ☐ Female ☐ Other

117. What is the gender of your child?

☐ Male ☐ Female ☐ Other

118. Do you consider yourself to be Hispanic, Latino, or of Spanish origins?

☐ Yes ☐ No ☐ Don't Know

119. What races or ethnicities do you consider yourself to be? *Please select all that apply.*

☐ American Indian or Alaska Native ☐ Asian

☐ Black or African American ☐ Native Hawaiian or Pacific Islander

☐ White or Caucasian ☐ Other or None of the Above

☐ Don't Know or Unsure

120. What best describes your current relationship status?

☐ Never Married ☐ Living with Partner

☐ Married ☐ Widowed

☐ Separated ☐ Divorced

121. What is the highest level of school you have completed, or the highest degree you have obtained?

☐ Less than High School ☐ High school diploma or GED

☐ Some College, No Degree ☐ Associates Degree

☐ Bachelor's Degree ☐ Technical, Vocational, or Occupational Degree

☐ Master's Degree ☐ Professional or Doctoral Degree (ex.: MD, JD, PhD)

☐ Other: _____

Is there anything else you feel is important, or would like us to know about your or your child's experience with genetically inherited high cholesterol?

_____

_____

_____

_____

_____

_____

_____

_____

_____

May we contact you in the future about any additional research pertaining to you or your child's experience with genetically inherited high cholesterol (for example, focus groups, family interviews, or individual interviews)?

☐ Yes ☐ No

Figure 6. *B-IPQ Items Adapted for HC- Respondents*

Illness Perceptions Questionnaires – Familial High Cholesterol & Risk of Heart Disease

The following questions ask for your thoughts about how the diagnoses of high cholesterol affects you and your family generally. Please consider how your child's/children's high cholesterol - and if you believe it applicable, the high cholesterol of any other members of your household - makes ***you*** *feel. There are no right or wrong answers, we just want to know what you think. Please select the number that best corresponds to your views.*

11. How much does your family members' diagnosis of *high cholesterol* affect you?

0 1 2 3 4 5 6 7 8 9 10

No Affect *Severely Affects*

12. How long do you think your family members' will continue to have *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

A Very Short Time *Forever*

13. How much control do you think you have over your family members' *high cholesterol*? (That is, to what extent do you think you can personally do things to help improve their cholesterol?)

0 1 2 3 4 5 6 7 8 9 10

Absolutely No Control *Completely Control*

14. How helpful do you think medications can be in lowering your family members' *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

15. How much do you think dietary interventions can help lower your family members' *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

16. How much do you think exercising can help lower your family members' *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

17. How concerned are you about your family members' *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

18. How well do you feel you understand your family members' *high cholesterol*?

0 1 2 3 4 5 6 7 8 9 10

Don't Understand At All *Completely Understand*

19. How much does your family members' *high cholesterol* affect you emotionally?

0 1 2 3 4 5 6 7 8 9 10

Not At All Affected *Extremely Affected*

The next questions ask for your thoughts about how the risk for heart disease affects you and your family generally. Please consider how your child's risk for heart disease makes you feel, and if you believe it applicable, the risk faced by any other members of your household. ***These questions may look very similar to those you have previously answered; please pay close attention to changes in the concept of interest****. Please circle the number that best corresponds to your views:*

20. How much does your family members' *risk for heart disease* affect your life?

0 1 2 3 4 5 6 7 8 9 10

No Affect *Severely Affects*

21. How long do you think your family members' *risk for heart disease* will continue?

0 1 2 3 4 5 6 7 8 9 10

A Very Short Time *Forever*

22. How much control do you think you have over your family members' *risk for heart disease*? (i.e., to what extent do you think you can personally do things to improve their risk for heart disease?)

0 1 2 3 4 5 6 7 8 9 10

Absolutely No Control *Completely Control*

23. How much do you think medications can help lower your family members' *risk for heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

24. How much do you think dietary interventions can help lower your family members' *risk for heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

25. How much do you think exercise can help lower your family members' *risk for heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not At All Helpful *Extremely Helpful*

26. How concerned are you about your family members' *risk for heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Not Concerned At All *Extremely Concerned*

27. How well do you feel you understand your family members' *risk for heart disease*?

0 1 2 3 4 5 6 7 8 9 10

Don't Understand At All *Very Clearly Understand*

28. How much does your family members' *risk for heart disease* affect you emotionally?

0 1 2 3 4 5 6 7 8 9 10

Not At All Affected *Extremely Affected*

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