



The role of causal knowledge in stigma considerations in African genomics research: Views of South African Xhosa people

Olivia P. Matshabane, PhD^{a,*}, Megan M. Campbell, PhD^{b,c}, Marlyn C. Faure, MSc^a, Paul S. Appelbaum, MD^{d,e}, Patricia A. Marshall, PhD^f, Dan J. Stein, MD, PhD^{g,h}, Jantina de Vries, PhD^a

^a Department of Medicine, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa

^b Department of Psychiatry and Mental Health, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa

^c Department of Psychology, Rhodes University, Grahamstown, South Africa

^d Department of Psychiatry, Columbia University, Irving Medical Center, New York, United States

^e New York State Psychiatric Institute, New York, United States

^f Department of Anthropology, Case Western Reserve University, Cleveland, United States

^g Department of Psychiatry & Neuroscience Institute, Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa

^h SA MRC Unit on Risk & Resilience in Mental Disorders, Cape Town, South Africa

ARTICLE INFO

Keywords:

Causal beliefs
Stigma
Xhosa people
South Africa
Genomics
Genetic attribution

ABSTRACT

Introduction: Advances in genomics research have raised several ethical concerns. One concern is the potential impact of genomics research on stigma experienced by people affected by a disease. Studies have found that the type of illness as well as disease causal beliefs impact on the relation between genetic attribution and stigma. This study explored the potential impact of genetic attribution of disease on stigma among Xhosa people with Rheumatic Heart Disease (RHD). **Methods:** Study participants were 46 Xhosa people with RHD living in the Western Cape Province of South Africa. Using video vignettes in 7 focus group discussions we explored whether and how genetic attribution may impact on disease-stigma. Vignettes introduced participants to non-genetic and genetic causal explanations and were followed-up with a series of open-ended questions eliciting their perceptions of non-genetic disease causes as well as genetic causation and its impact on internalised stigma. **Results:** This study found that Xhosa people with RHD have a general understanding of genetics and genetic attribution for disease. Additionally, and not withstanding their genetic knowledge, these participants hold multiple disease causal beliefs including genetic, infectious disease, psychosocial, behavioural and cultural explanations. While there was evidence of internalised stigma experiences among participants, these appeared not to be related to a genetic attribution to the disease. **Discussion:** The findings of this study provide clues as to why it is unlikely that a genetic conceptualisation of disease impacts internalised stigma experiences of Xhosa people. The causal explanations provided by participants reflect their cultural understandings and their context, namely, living in low-income and poverty-stricken environments. Divergence in these findings from much of the evidence from high-income countries emphasises that context matters when considering the impact of genetic attribution on stigma and caution against generalising findings from one part of the globe to another.

1. Introduction

Globally, genomics research continues to grow at an extraordinary pace, and with it, its predicted role in clinical medicine (Parens and

Appelbaum, 2019). The increase in genomics research has raised concerns about potential negative ethical consequences for individuals and communities, particularly its impact on stigma (Ramsay et al., 2014). The literature suggests that this phenomenon could play out in various

* Corresponding author. Office J52-16, UCT Centre for Clinical Research, Old Main Building, Groote Schuur Hospital, Department of Medicine, University of Cape Town, Observatory, 7925, Cape Town, South Africa.

E-mail addresses: olivia.matshabane@uct.ac.za, preciousmatshabane@gmail.com (O.P. Matshabane), m.campbell@ru.ac.za (M.M. Campbell), marlyn.faure@gmail.com (M.C. Faure), paul.appelbaum@nyspi.columbia.edu (P.S. Appelbaum), pam20@case.edu (P.A. Marshall), dan.stein@uct.ac.za (D.J. Stein), jantina.devries@uct.ac.za (J. de Vries).

<https://doi.org/10.1016/j.socscimed.2021.113902>

Received in revised form 16 February 2021; Accepted 31 March 2021

Available online 7 April 2021

0277-9536/© 2021 The Author(s).

Published by Elsevier Ltd.

This is an open access article under the CC BY-NC-ND license

(<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

ways and the evidence to support this concern is inconsistent. On the one hand, authors have expressed the hope that genetic information could reduce stigma by reducing some degree of personal responsibility for developing a condition (Angermeyer et al., 2011; Mehta and Farina, 1997; Phelan, 2005; Phelan et al., 2002). On the other hand, authors have cautioned that genetic attribution could increase stigma by placing emphasis on conditions perceived to be fundamental and unchangeable, with little possibility for recovery or treatment (Angermeyer et al., 2011; Kong et al., 2017). This explanation is arguably rooted in the negative history of eugenics (Savulescu and Kerin, 1999) and in early experiences with genetic screening for conditions such as sickle cell disease (Duster, 2004; Phelan et al., 2002). Yet both of these possibilities require a view that persons are genetic essentialists – meaning that they are prone to thinking of genetic attributions as being immutable, natural, of a specific aetiology, as well as resulting in people being distinctively divided into homogeneous and discrete groups (Heine et al., 2017). Countering that assumption, Condit proposes that the available evidence suggests this to be a false premise and suggests that rather, people are ‘strategic essentialists’, meaning that they tend to hold multiple causal beliefs for human traits and conditions simultaneously, and that these are deployed strategically (Condit, 2019; Heine et al., 2017; Jayaratne et al., 2009). This was evident in Sanderson et al.’s study which found that people who had received information of a genetic influence on cancer and heart disease were more, not less likely to recognise that lifestyle causal beliefs impact these chronic diseases (Sanderson et al., 2011). Similarly, Jayaratne and colleagues’ found that people not only held multiple causal beliefs, but they were not deterministic about any of the three causal accounts (i.e., genetics, environment and choice) investigated in their study (Jayaratne et al., 2009). In light of such findings, a growing number of scholars observe that the impact of genomics information on individuals is strongly influenced by context and that people from different contexts interpret and use genomic information differently (Condit, 2019; Parens and Appelbaum, 2019; Wade, 2019).

And yet the majority of evidence available on questions about stigma and genetic attribution, emanates from North American and European countries, with only few studies specifically focusing on this relation from Africa. Two of these studies (de Vries et al., 2012; Tekola et al., 2009) caution about the potential for increased ethnic stigmatisation as a result of genomics research, particularly for individuals from already stigmatised or marginalised population groups. In Ethiopia, Tekola et al.’s study focusing on Podoconiosis – a highly stigmatised disease – found that participants were afraid to participate in genomics research due to fear of receiving genetic information about the origin of the disease, which could contribute to increased social stigma (Tekola et al., 2009). In Kenya, Marsh et al. found evidence of associative stigma among mothers of children with sickle cell disease (Marsh et al., 2011). In South Africa, Faure et al. found no evidence of an effect (Faure et al., 2019). In research with schizophrenia, Matshabane et al. found that a genetic attribution may reduce personal blame (Matshabane et al., 2020). Notwithstanding this work, the nature of this possible relationship remains largely understudied among African populations, particularly those from marginalised poverty-stricken communities which may have unique implications for stigma.

In this study, we set out to examine the relation between genetic attribution of disease, and a condition which has both environmental and genetic causation. Rheumatic Heart Disease (RHD) is a chronic heart condition caused by recurrent untreated infections with group A streptococcus (GAS) (Carapetis et al., 2005), an individual’s genetic make-up, and poverty-related environmental factors (Mocumbi, 2015). Non-genetic factors such as GAS infection and the socioeconomic status of the patient and community contribute to the onset of RHD (Engel et al., 2011). The disease runs parallel with a host of variables indicating poverty, such as crowded housing, poor nutrition, low levels of education and limited access to adequate health care (Barth et al., 2015). Whilst there is a strong heritable component (Engel et al., 2011, 2017), little is known about the host of genetic factors that increase the risk of

developing RHD (Muhamed et al., 2020). Sadly, whilst the prevention of RHD is both affordable and effective, the burden of disease remains high in some parts of the world (Irlam et al., 2013). As a disease of poverty, RHD disproportionately affects marginalised populations in Low and Middle-Income Countries (LMICs) (French and Poppas, 2018; Robertson and Mayosi, 2008; Watkins et al., 2016).

People with RHD often manifest physical symptoms that inhibit their physical functioning, such as shortness of breath, tiredness and chest pains, which result in challenges in completing tasks that require physical strength (Petricca et al., 2009). Because of these physical limitations, people with RHD are sometimes stigmatised by others as weak and vulnerable (Sliwa et al., 2018). The primary consequences of stigma include status loss and discrimination, which often result in long-term social and economic inequality between those who stigmatise and those who are stigmatised (Link and Phelan, 2014). In this study we focused on exploring internalised/self-stigma. Internalised stigma involves an individual experiencing shame and expecting discrimination when others know about their disease (Corrigan and Watson, 2002; Gray, 2002; Livingston and Boyd, 2010). In the literature internalised stigma has been defined in relation to stereotypes (negative connotations), prejudice (ignorance or misinformation) and discrimination (Corrigan and Watson, 2002; Thornicroft et al., 2007). Given the dearth of literature on the ethical implications of genetic attribution on the lives of African people, this study investigates how a (partial) genetic attribution for RHD relates to the internalised stigma experiences of Xhosa people living with RHD.

2. Methods

2.1. Participants and recruitment

We enrolled 46 Xhosa people with RHD from the Western Cape Province of South Africa in 7 Focus Group Discussions (FGDs). All enrolled participants had previously participated in a parent genomics of RHD study, and some had been involved in RHD patient awareness events where they received information on genetics and RHD (Zühlke et al., 2018). Following their participation in the parent study, they were re-contacted to voluntarily participate in this study. We recruited 39 females (84.78%) and seven males (15.21%). Their mean age was 43.05 years (range 23–75 years). Thirty-seven (80.43%) had secondary schooling or above, yet 42 (91.30%) reported being unemployed. The higher rate of females recruited in our study is reflective of the sample in the larger parent study, which enrolled 71% females, and other previous RHD research (Faure et al., 2019). The gender differences correspond with evidence from other LMICs which found a significantly higher rate of females presenting with RHD (Padmavati, 2001; Rizvi et al., 2004), possibly because RHD is often diagnosed during pregnancy due to the physiological changes of pregnancy, which include an increase in stroke volume, heart rate, and cardiac output, often resulting in clinical deterioration in patients with severe valve conditions (French and Poppas, 2018). This suggests that it may be under-diagnosed in males (Otto et al., 2011). Another reason may be because women more readily access primary healthcare services in South Africa (Nteta et al., 2010; Otto et al., 2011).

2.2. Focus groups and discussion guides

FGDs are ideal for exploring people’s experiences, opinions, concerns and beliefs about causes of disease, while allowing for interaction amongst people and the emergence of shared views (Kitzinger, 2005; Wong, 2008). We chose what is sometimes called “reactance format FGDs” which have been used in research with minority communities (predominantly African American) in the US, where participants are provided with some information about the subject at the start of the FGD, following which responses are structured in relation to that information (Condit et al., 2003). This is different from FGDs where no

material is shared with participants prior to engaging in questions. We found this format appropriate to allow participants to discuss and reflect on a range of issues which they were unlikely to have considered previously. Additionally, we used vignettes in the FGDs because they explore a potentially difficult topic of inquiry (stigma) and we thought the use of a storyline with a leading character who is diagnosed with RHD may help participants not feel pressure to respond from their own experiences but could allow them to respond from the perspective of the vignette character (Holloway, 2005; Hughes and Huby, 2002; Wong, 2008). During the FGDs however, out of their own accord, participants felt comfortable enough to share their own experiences and we allowed for that to occur organically without being driven by the researchers.

We identified suitable characters and with the help of a professional film-maker we turned the vignettes into short videos. Each vignette was told using a voiceover, and the questions at the end of each story stage appeared on screen. The facilitator played the video and stopped the recording at each question for discussion. The decision to using a video instead of reading out the text was made to make this process livelier and more understandable, in addition to ensuring that the same information is provided in each group. The short video-vignettes were based on a 26-year-old Xhosa male character who has RHD (see Appendix A). We selected a male protagonist because the tools used in this research were created simultaneously for use among patients participating in genomics research on another disease (Matshabane et al., 2020), and the gender balance in that group was reversed (including mostly males). Furthermore, at the time the study was designed, the primary study had only just started recruitment and we were not aware that the gender-division of patients receiving treatment for RHD in the hospital, was this heavily skewed towards females. The vignettes and questions were forward-translated into isiXhosa by a bilingual team which included four first-language isiXhosa speaking health professionals – two of whom have extensive experience in conducting research with Xhosa people who have RHD. The translations were collated by the first author (who is first-language isiXhosa speaking) in a table and discussed and finalized at a committee meeting with the translation team, including the first, second and senior authors. The vignettes were identical other than the cause of RHD being explained as either genetic, environmental or a combination of genetic and environmental causes. Each group watched one of the three vignettes and ultimately, we conducted three FGDs with the genetic explanation vignette, and two each for the environmental and the mixed (genetic and environmental) explanations. Each vignette had three segments embedded with questions exploring: 1) participants' general understanding of RHD and genetics; 2) participants' perceptions of how one's life may change after being diagnosed with RHD and knowing that the cause is genetic, non-genetic or a combination; and 3) how participants' view of the cause of the disease as related to genetic or non-genetic explanations may impact on internalised stigma experiences. Questions were structured around key concepts such as a desire to maintain social distance as well as anticipated and associative stigma.

2.3. Analysis

FGDs were conducted in isiXhosa by the lead researcher and supported by a bilingual co-facilitator. All FGDs were tape-recorded, transcribed verbatim and translated from isiXhosa into English for thematic analysis (Braun and Clarke, 2012). English versions of transcripts were imported into NVivo12 software (Siccamo and Penna, 2008) for data management. Notes taken during the FGDs by the facilitator and co-facilitator, including the sequence of responses, were used to allocate quotes to participants. No differences were observed in the data based on which vignette was watched by participants – therefore all data were analysed as one dataset. Data were analysed inductively (Braun and Clarke, 2012) through multiple rounds of coding by OPM and JdV. Thematic domains were then developed by OPM, JdV and MMC which were applied to all the transcripts. Throughout data analysis, emerging

insights were discussed primarily between OPM, JdV and MMC, and later with all the co-authors. Data analysis occurred alongside data collection.

2.4. Ethics approval

The participants in this study provided written consent before participation. The study was approved by the University of Cape Town (FHS204-2015). Permission to conduct this research was obtained from the local hospital head nurses in the relevant units and the South African Department of Health.

3. Findings

3.1. Knowledge of genetics

A critical first component of this study was to understand what participants know about genetics and heredity, as well as how they relate genetics to their disease. For that reason, we began the FGDs by asking participants what they know about genetics. Some participants described genetics as 'something passed down in the blood' or 'something inherited from generation to generation'. Our participants mostly described genetics in relation to disease rather than phenotypic traits. For instance, one participant described it as follows:

"I think heredity ... I think it is something that is in the family. Let us say if my grandmother had a heart disease, her children and grandchildren can also have the heart disease." (P.6: FGD 7)

Participants reported being aware that a disease may be passed down to some offspring, while other children may not have the disease. Additionally, they noted that even if genes were passed down to offspring, the disease may not affect that generation but could affect future generations. However, interestingly, participants seemed to describe this process as being skewed towards the genes of the parent that the person most closely resembles or the parent that has the dominant genes.

"It would depend on the genes. If your genes are different from your mother's but are similar to your father's, you might have it, if your father had it. It depends on whose genes of your parents is stronger." (P.6: FGD 1).

3.2. RHD and genetic attribution

Although some participants recognized that a genetic predisposition could have played a role in the development of their illness, most had not thought about their disease at all in relation to genetics. This was despite having previously participated in a RHD genomics study which described RHD as potentially linked to a genetic predisposition and having experiences of being asked by doctors whether they knew anyone in their family who had RHD. With the exception of a few participants, many described being the first in their family to have the disease.

"I'm the only one who has this heart disease in my family. I haven't heard of anyone who had it before me, and so I wouldn't say it's inherited." (P.3: FGD 2).

Five out of six participants in the first FGD echoed the same sentiments as participant 3 above, all stating that they are the first people in their family to have a heart disease and therefore they do not think a genetic predisposition could have been the cause of them developing RHD. In all FGDs there was at least one participant who held the same perception. Yet there was also at least one participant in all FGDs who considered their heart disease to be linked to a genetic predisposition. Most participants, however, were able to identify other diseases which they considered to have a (partial) genetic causation like cancer, asthma

and hypertension.

Some participants described encountering the genetic explanation in relation to RHD susceptibility for the first time in the FGDs. For example:

“For me I do not have any knowledge about genetics and heart disease. I was never informed about genetics or inheritance. I just heard about it now, but I did not inherit this disease from anyone. But maybe my child will inherit it though as the time goes on but for now she does not have it.” (P. 4: FGD 4).

Age also played a role in whether people considered genetics to play a role in the onset of their condition.

“No. It is only now that I am old when I realised that I inherited heart disease from my mother ... Otherwise back then, nobody said anything about inheritance.” (P.1: FGD 4).

It is important to note here that participants often conflated general heart disease and RHD. This may not be altogether surprising considering that the term RHD does not exist in the Xhosa language, therefore generally people with RHD refer to themselves as having a ‘heart disease’ (*isifo sentliziyo*) and it was translated as such in the vignettes.

3.3. Alternative explanations

The questions embedded in the RHD vignettes specifically explored the likely role of genetic causality on internalised stigma (i.e., “whether knowing the disease is genetic may affect decisions on getting married or having children”). However, despite this emphasis, our data suggest that many respondents across all FGDs held strong beliefs of other non-genetic disease explanations. These alternative explanations included: infectious disease, psychosocial, behavioural, and cultural causes. Infectious disease explanations for RHD have been previously described as the disease being caused by bacteria linked to having a sore throat (Strep A infection) (Faure et al., 2019). A few participants in our study held this view. For instance.

“I also think like the other participant, maybe you are in a village, and you are drinking water from the streams you don’t even know where it comes from. So there might be a germ or virus in that water that is how I think it can cause the disease. Maybe the water from the river would cause you to cough, have short breath, loss of appetite, it might be that germ forming all these things after that it becomes a heart disease.” (P.4: FGD 2).

In a study conducted with people who have schizophrenia (Matshabane et al., 2020), psychosocial causal beliefs were related to severe poverty, trauma, stress and past or present experiences of physical/emotional abuse. In our study many participants felt strongly about the impact of these factors on the onset of RHD. One said.

“Poverty is one of the difficult things that we go through and it can make you sick with stress.” (P.4: FGD 4).

While another participant said:

“I have been abused emotionally, and for me the heart disease was caused by this abuse, because I did not have this disease when I was growing up and in my family, we have no history of the heart disease but abuse from my husband caused this illness.” (P.1: FGD 2).

Behavioural explanations for understanding the cause of heart disease – in the context of genetics research – have largely been linked to behavioural actions such as eating unhealthy food, smoking and drinking (Condit et al., 2009). In our study, diet was suggested to be most important. For instance.

“It is because of the things that we eat daily, fatty foods, cooking oil consumption and so forth, they are the cause of heart disease because it’s not heredity.” (P.3: FGD 1).

“I think overthinking and consuming too much salt are the things that caused heart disease in my life.” (P.3: FGD 1).

In addition, in our study, participants also described what has been described in previous work (Matshabane et al., 2020; Staunton et al., 2018) as ‘cultural’ disease causal explanations. Cultural explanations, which can include witchcraft or influences of supernatural forces such as spirits, have been commonly described in health studies conducted with different African populations (Mshana et al., 2006, 2008; Swartz, 1998). This may not be surprising given that in an African paradigm of illness, spiritual and ancestral entities play an important role in understanding illness onset. These beliefs are often held alongside other attributions (Nwoye, 2015). In this study we also found that some people attribute RHD to cultural or supernatural causes.

For instance, one participant said:

“My mother, when I told her about it, she thought and believed that it was evil spirits that caused it. I told her that it is a common sickness, there is a lot of people who are affected by it. She was hurting so it was not easy for her to accept it.” (P.4: FGD 1).

Others implied that evil spirits or curses could also make the condition worse.

“And that is true, evil things will get to you through something that you have. If people want to bewitch you, they will use this heart disease, or use the illness that you suffer from.” (P.4: FGD 7).

While most participants focussed on a particular explanation for the onset of the disease, some participants held a cultural explanation alongside their other explanations for RHD.

“Me, it is said that there is my aunt who had it. I do not know how it connected with me. I would say it is heredity, but no, I also say it is evil spirits. [Laughter]. So I also think maybe it is that which caused the illness.” (P.3: FGD 6).

Importantly, during the FGDs participants described a range of causal beliefs, some of which were held simultaneously.

4. Stigma experiences

4.1. Stereotypes

Our study then moved on to understand more about the stigma experiences of our participants. The most commonly cited stereotypes about living with RHD reported in this study were 1) labelling of women with RHD as weak because they are perceived not to be able to bear a biological child or children to full term and 2) the labelling of men with RHD as weak and unable to obtain or maintain employment or to participate in socially and culturally important activities.

“For us women it can be difficult to date and to get married. We are always reluctant to get married. When you are married you are expected to bear children. While doctors say that it is difficult for us to carry children because of the disease. So, you always ask yourself what is the point of getting married if you cannot have children.” (P.3: FGD 5).

“For example, in the villages [in the Eastern Cape Province] in preparation of burial service, it is young men who perform the responsibility of digging the grave-hole. So, when it comes to him, people will say ‘oh that one, he is useless’, you see those negative comments. He cannot even make a suggestion and say, ‘Let us go and dig’, no, because people know that [character] is someone who cannot dig and cannot even carry a shovel, because he might just fall with it on his hands.” (P.5: FGD 6).

Furthermore, many participants felt that employers may not be comfortable hiring someone with RHD because of concerns that physical

limitations would compromise their productivity – which could explain the high rate of unemployment amongst our participants. This was considered particularly difficult for Xhosa men because in this culture, acceptable masculinity is associated with being able to financially provide for your family. The occupations referred to by our participants involved manual labour (i.e., construction or domestic work) since most people have low levels of education and are not professionally trained.

4.2. Prejudice

Participants reported negative attitudes relating to the above-mentioned stereotypes from members of the family, in-laws and the community. For instance, one participant said community members may perceive someone with RHD as inferior. See, "... they can make him [person with RHD] feel so small." (P.1 FGD 1) Another respondent said, "Yes, it can happen that people look down on him since he is sick. Some people can even go to an extent of keeping his plate and spoons separately from theirs as they think his condition might be contagious. Some people may even call him names." (P.1: FGD 4)

Another participant said.

"Yes, or if it comes to making decisions. Some people in his family may feel that they need to make decisions for him just because he has a heart condition and they think he cannot be told anything." (P.9 FGD 3).

4.3. Discrimination

Many women in this study voiced experiencing undesirable social moments including stigmatisation and emotional abuse by their partners and/or their in-laws or the community because of their difficulty to fall pregnant and carry a child to full term. These incidents placed strain on marriages and relationships, especially because in African cultures there is a perception that a good marriage is one in which children are born and womanhood is achieved primarily through motherhood. Therefore, single women in our study also expressed a deep longing to have children of their own.

"I can't have children because I have a heart disease, I was told by the doctor that I can't have children. In my community I'm not happy because people talk about me and I get to hear what they say about me." (P.6. FGD 1).

While it is true that pregnancy is a potentially lethal health challenge for females with RHD because of additional stress on the heart caused by greater blood volumes, there are measures which can be put in place to assist a woman with RHD to have a child of her own. Due to the real risks of pregnancy, however, generally female RHD participants reported being advised by medical professionals not to attempt pregnancy – especially in communities where there are minimal health resources to monitor their pregnancies (Chang et al., 2018). Internalising this possibility – of not bearing biological children – was deeply traumatic for many individuals in this study, resulting in some women weeping in the focus groups.

5. Genetic attribution and disease-stigma

Next in the study we were interested in exploring how participants thought that a partial genetic attribution of RHD – likely to come about as a consequence of the genomic focus in the genomics study that all FGD participants had taken part in – could possibly impact on internalised stigma. We did this by posing these questions, "How do you think [character's name] life will change after the doctor tells him that his disease is partly caused by genetics?", "How do you think his friends or family will relate to him when they know that the disease may be partly genetic?" and "How would you feel about becoming friends with [character] knowing that the disease may be partly genetic?" Responses of participants can be encapsulated in the following quote:

"I do not think it can change. Because, every person did not ask for what he/she has. I do not think they will change." (P.7. FGD 3).

By and large, we found that although stigma experiences were reported by some respondents in this study, these experiences were not at all recounted in relation to the causal belief of the disease being partly genetic. Importantly, because participants explicitly did not mention cause in their responses, our findings suggest that it is unlikely that a genetic attribution for disease impacts on the internalised stigma experiences of these participants. Moreover, the fact that many participants failed to mention the cause being genetic or non-genetic in their responses (despite it being emphasised in the guiding questions and vignettes), suggests that they did not consider causal explanations for RHD as an important contribution to disease-stigma.

6. Discussion

Our study found that participants held multiple disease causal explanations to explain their heart disease. Moreover, we found that even though all participants were exposed to a genetic explanation for their disease on more than one occasion, this exposure did not seem to push them towards a stronger genetic attribution for their disease. Instead, congruent with findings elsewhere, genetic explanations were added onto the multifaceted causal models that participants already held, which for some included genetics (Bates et al., 2003; Condit, 1999). In addition to genetic causal beliefs, other causal explanations articulated by our participants included infectious disease, psychosocial, behavioural, and cultural disease explanations. These alternative causal accounts were reported as more important explanatory models for their disease onset than the genetic account. Importantly, we found that historical and contemporary contextual realities of living in low-income poverty-stricken environments served as an overarching frame for how our participants understood the cause of their disease.

Second, we found that some Xhosa people with RHD do experience internalised stigma in the form of stereotypes, prejudice and discrimination. Mainly, such stigma experiences were gendered – for instance females were stigmatised for their perceived inability to bear children and men for their physical limitations which prohibited them from being employed or participating in socially and culturally valued activities. Both genders experienced labelling and name-calling related to connotations of being 'weak', 'different' or 'inferior' – similar to research about the stigma experiences of RHD patients conducted elsewhere in Africa (Chang et al., 2018; Petricca et al., 2009). These experiences were reported to negatively affect their social status. Importantly, we found that participants' accounts of disease-related stigma were intertwined with the structural stigmatising effects associated with historical discrimination experienced by people of colour in South Africa. This observation aligns with the 'intersectionality stigma' literature (Bowleg, 2012) which posits that when conducting research with groups which have multiple stigmatised identities (such as race, gender, and class) it is important not to consider forms of stigma as discrete (Crenshaw, 1989) but rather to consider their compounded effect (Rice et al., 2018). This is relevant to our work in that, for example, shared experiences of socio-economic inequality (i.e., difficulties finding employment which are intrinsically linked to most individuals having been schooled in an under-resourced education system) and psychological trauma (for instance in relation to high levels of violence and crime in their communities) were often articulated as important contributing factors to consider when investigating disease-stigma. This observation encourages researchers to think about the complexities of intersecting stigmas for different population groups in the context of genomics research.

Taken together, we found that although some Xhosa people with RHD did report internalised stigma experiences, these accounts were not related to conceptualising the cause of their disease as genetic. Despite the emphasis placed on genetic causal explanations in this study and in the genomics study that the participants had all enrolled in, the

participants' refusal to draw links between genetics and stigma suggests that being exposed to genetic information may not have an influence on internalised stigma for this population group. Instead, participants' existing causal knowledge, pre-existing structural stigma and contextual realities seem to play a crucial role in how they conceptualise their disease.

Overall, our research provides support for Condit's proposal that genomics information is strategically considered alongside other already held causal explanations (Condit, 2019). When conducting research with people from marginalised communities, there is often a concern that these individuals may be specifically vulnerable to being passive receivers of information and that genetic information is more likely to impact on their stigma – a concern that subsequently is translated into somewhat paternalistic or protective ethical attitudes. This study begins to challenge that perspective by showing that people from different cultural groups are capable of using genetic information in conjunction with their already held causal knowledge and that exposure to genetic explanations may not lead them to deterministic thinking. Moreover, evidence from this study and other African studies (Marsh et al., 2011) supports the analysis suggesting that pre-existing social, political, economic and cultural factors associated with specific populations should be seriously considered in the genomics research process, as these factors seem more likely to influence individuals' stigma accounts than the genomics study itself (de Vries et al., 2020).

This study has three important limitations. First, its qualitative nature means that it cannot report on causality and data cannot be used to make broad generalisations. Rather, the focus group method used in the study allowed us to gain a deeper understanding of the experiences and views of our participants on our research question. This assisted us to better understand why genetic attribution may not directly influence stigma. Second, our sample was relatively homogeneous in terms of socioeconomic and cultural characteristics, therefore future studies should investigate this phenomenon in populations which are socio-demographically different. Additionally, our sample is both small ($n = 46$) and skewed towards middle-aged females (84.78% with an average age of 43 years). Even though this sample is aligned to the recruitment pattern in the genomic study we drew our participants from, their accounts might not represent those of younger people or Xhosa males with RHD. Finally, the character in our vignette was male, whereas the majority of our participants were female. It is possible that this influenced the views and perspectives of our research participants.

Going forward, as more research is emerging to better understand genetic effects on a myriad of diseases present in the African continent, we encourage simultaneous investment in ethical studies carefully investigating in which situations and contexts genetic information may intersect with stigma for specific population groups, with specific attention to the effect of social, political, economic and cultural factors.

Funding

This work was part of the "Stigma in African Genomics research on schizophrenia and rheumatic heart disease" study funded by the National Institutes of Health [Grant Number: U01HG008226, PI: Jantina de Vries] and a member of the H3Africa Consortium (www.h3africa.org). The work also received funding from the National Research Foundation [Grant Number: 107664, PI: Olivia Matshabane] and the Harry Crossley Foundation [PI: Olivia Matshabane].

Declaration of competing interest

All authors state that there is no conflict of interest.

Appendix A. Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.socscimed.2021.113902>.

Contributors

All authors contributed to the conceptualisation of the work. OPM conducted the research. All authors commented on the findings and data analysis strategy. OPM wrote the first draft and all authors commented. Revisions were made by OPM and all the other authors agreed to the suitability of the submitted draft. JdV and MMC provided supervision of the work.

Disclosure statement

All authors are not aware of any affiliations, memberships, funding, or financial holdings that might be perceived as affecting the objectivity of the review.

References

- Angermeyer, M.C., Holzinger, A., Carta, M.G., Schomerus, G., 2011. Biogenetic explanations and public acceptance of mental illness: systematic review of population studies. *Br. J. Psychiatry* 199, 367–372.
- Barth, D.D., Zühlke, L.J., Joachim, A., Hoegger, T., Mayosi, B.M., Engel, M.E., 2015. Effect of distance to health facility on the maintenance of INR therapeutic ranges in rheumatic heart disease patients from Cape Town: no evidence for an association. *BMC Health Serv. Res.* 15, 219.
- Bates, B.R., Templeton, A., Achter, P.J., Harris, T.M., Condit, C.M., 2003. What does "a gene for heart disease" mean? A focus group study of public understandings of genetic risk factors. *Am. J. Med. Genet.* 119, 156–161.
- Bowleg, L., 2012. The problem with the phrase women and minorities: intersectionality—an important theoretical framework for public health. *Am. J. Publ. Health* 102, 1267–1273.
- Braun, V., Clarke, V., 2012. Thematic analysis. In: Cooper, H. (Ed.), *APA Handbook of Research Methods in Psychology*. American Psychological Association.
- Carapetis, J.R., McDonald, M., Wilson, N.J., 2005. Acute rheumatic fever. *Lancet* 366, 155–168.
- Chang, A.Y., Nabbaale, J., Nalubwama, H., Okello, E., Ssinabulya, I., Longenecker, C.T., et al., 2018. Motivations of women in Uganda living with rheumatic heart disease: a mixed methods study of experiences in stigma, childbearing, anticoagulation, and contraception. *PLoS One* 13.
- Condit, C.M., 1999. How the public understands genetics: non-deterministic and non-discriminatory interpretations of the "blueprint" metaphor. *Publ. Understand. Sci.* 8, 169–180.
- Condit, C.M., 2019. Laypeople are strategic essentialists, not genetic essentialists. *Hastings Cent. Rep.* 49, S27–S37.
- Condit, C.M., Gronnqvist, M., Landau, J., Shen, L.J., Wright, L., Harris, T.M., 2009. Believing in both genetic determinism and behavioral action: a materialist framework and implications. *Publ. Understand. Sci.* 18, 730–746.
- Condit, C.M., Templeton, A., Bates, B.R., Bevan, J.L., Harris, T.M., 2003. Attitudinal barriers to delivery of race-targeted pharmacogenomics among informed lay persons. *Genet. Med.* 5, 385–392.
- Corrigan, P.W., Watson, A.C., 2002. Understanding the impact of stigma on people with mental illness. *World Psychiatr.* 1, 16–20.
- Crenshaw, K., 1989. Demarginalizing the Intersection of Race and Sex: A Black Feminist Critique of Antidiscrimination Doctrine, Feminist Theory and Antiracist Politics. *139 u. Chi. Legal f.*
- de Vries, J., Jallow, M., Williams, T.N., Kwiatkowski, D., Parker, M., Fitzpatrick, R., 2012. Investigating the potential for ethnic group harm in collaborative genomics research in Africa: is ethnic stigmatisation likely? *Soc. Sci. Med.* 75, 1400–1407.
- de Vries, J., Landouré, G., Wonkam, A., 2020. Stigma in African genomics research: gendered blame, polygamy, ancestry and African epistemology impact on the risk of harm. *Soc. Sci. Med.* 113091.
- Duster, T., 2004. *Backdoor to Eugenics*. Routledge.
- Engel, M.E., Cohen, K., Gounden, R., Kengne, A.P., Barth, D.D., Whitelaw, A.C., et al., 2017. The Cape Town clinical decision rule for streptococcal pharyngitis in children. *Pediatr. Infect. Dis. J.* 36, 250.
- Engel, M.E., Stander, R., Vogel, J., Adeyemo, A.A., Mayosi, B.M., 2011. Genetic susceptibility to acute rheumatic fever: a systematic review and meta-analysis of twin studies. *PLoS One* 6.
- Faure, M.C., Matshabane, O.P., Marshall, P., Appelbaum, P.S., Stein, D.J., Engel, M.E., et al., 2019. Does genetics matter for disease-related stigma? The impact of genetic attribution on stigma associated with rheumatic heart disease in the Western Cape, South Africa. *Soc. Sci. Med.* 112619.
- French, K.A., Poppas, A., 2018. Rheumatic heart disease in pregnancy: global challenges and clear opportunities. *Circulation* 137, 817–819.
- Gray, D.E., 2002. 'Everybody just freezes. Everybody is just embarrassed': felt and enacted stigma among parents of children with high functioning autism. *Sociol. Health Illness* 24, 734–749.
- Heine, S.J., Dar-Nimrod, I., Cheung, B.Y., Proulx, T., 2017. Essentially biased: why people are fatalistic about genes. *Advances in Experimental Social Psychology*, 55. Academic Press, pp. 137–192.
- Holloway, I., 2005. *Qualitative Research in Health Care*. McGraw-Hill Education, UK.

- Hughes, R., Huby, M., 2002. The application of vignettes in social and nursing research. *J. Adv. Nurs.* 37, 382–386.
- Irlam, J., Mayosi, B.M., Engel, M., Gaziano, T.A., 2013. Primary prevention of acute rheumatic fever and rheumatic heart disease with penicillin in South African children with pharyngitis: a cost-effectiveness analysis. *Circulation: Cardiovascular Quality and Outcomes* 6, 343–351.
- Jayaratne, T.E., Gelman, S.A., Feldbaum, M., Sheldon, J.P., Petty, E.M., Kardia, S.L.R., 2009. The perennial debate: Nature, nurture, or choice? Black and White Americans' explanations for individual differences. *Rev. Gen. Psychol.* 13 (1), 24–33.
- Kitzinger, J., 2005. Focus group research: using group dynamics. *Qualitative research in health care* 56, 70.
- Kong, C., Dunn, M., Parker, M., 2017. Psychiatric genomics and mental health treatment: setting the ethical agenda. *Am. J. Bioeth.* 17, 3–12.
- Link, B.G., Phelan, J.C., 2014. Stigma power. *Soc. Sci. Med.* 103, 24–32.
- Livingston, J.D., Boyd, J.E., 2010. Correlates and consequences of internalized stigma for people living with mental illness: a systematic review and meta-analysis. *Soc. Sci. Med.* 71, 2150–2161.
- Marsh, V.M., Kamuya, D.M., Molyneux, S.S., 2011. All her children are born that way': gendered experiences of stigma in families affected by sickle cell disorder in rural Kenya. *Ethn. Health* 16, 343–359.
- Matshabane, O.P., Campbell, M.M., Faure, M.C., Marshall, P.A., Mayosi, B.M., Stein, D.J., de Vries, J., et al., 2020. Exploring how a genetic attribution to disease relates to stigma experiences of Xhosa patients with schizophrenia in South Africa. *Soc. Psychiatry Psychiatr. Epidemiol.* 55, 1679–1686.
- Mehta, S., Farina, A., 1997. Is being "sick" really better? Effect of the disease view of mental disorder on stigma. *J. Soc. Clin. Psychol.* 16, 405–419.
- Mocumbi, A.O., 2015. Rheumatic heart disease in Africa: is there a role for genetic studies? *Cardiovascular Journal of Africa* 26, S21.
- Mshana, G., Hampshire, K., Panter-Brick, C., Walker, R., 2008. Incidence Team. Urban-rural contrasts in explanatory models and treatment. *Science* 40, 35–52.
- Mshana, G., Plummer, M.L., Wamoyi, J., Shigongo, Z.S., Ross, D.A., Wight, D., 2006. 'She was bewitched and caught an illness similar to AIDS': AIDS and sexually transmitted infection causation beliefs in rural northern Tanzania. *Cult. Health Sex.* 8, 45–58.
- Muhamed, B., Parks, T., Sliwa, K., 2020. Genetics of rheumatic fever and rheumatic heart disease. *Nat. Rev. Cardiol.* 17, 145–154.
- Nteta, T.P., Mokgatle-Nthabu, M., Oguntibeju, O., 2010. Utilization of the primary health care services in the tshwane region of gauteng Province, South Africa. *PLoS One* 5, e13909.
- Nwoye, A., 2015. African psychology and the Africentric paradigm to clinical diagnosis and treatment. *S. Afr. J. Psychol.* 45, 305–317.
- Otto, H., Sæther, S.G., Banteyrga, L., Haugen, B.O., Skjærpe, T., 2011. High prevalence of subclinical rheumatic heart disease in pregnant women in a developing country: an echocardiographic study. *Echocardiography* 28, 1049–1053.
- Padmavati, S., 2001. Rheumatic heart disease: prevalence and preventive measures in the Indian subcontinent. *Heart* 86, 127.
- Parens, E., Appelbaum, P.S., 2019. On what we have learned and still need to learn about the psychosocial impacts of genetic testing. *Hastings Cent. Rep.* 49, S2–S9.
- Petricca, K., Mamo, Y., Haileamlak, A., Seid, E., Parry, E., 2009. Barriers to effective follow-up treatment for rheumatic heart disease in Jmma, Ethiopia: a grounded theory analysis of the patient experience. *Ethiop. J. Health Sci.* 19.
- Phelan, J.C., 2005. Geneticization of deviant behavior and consequences for stigma: the case of mental illness. *J. Health Soc. Behav.* 46, 307–322.
- Phelan, J.C., Cruz-Rojas, R., Reiff, M., 2002. Genes and stigma: the connection between perceived genetic etiology and attitudes and beliefs about mental illness. *Psychiatr. Rehabil. Skills* 6, 159–185.
- Ramsay, M., De Vries, J., Soodyall, H., Norris, S.A., Sankoh, O., 2014. Ethical issues in genomic research on the African continent: experiences and challenges to ethics review committees. *Hum. Genom.* 8 (15), 1–6.
- Rice, W.S., Logie, C.H., Napoles, T.M., Walcott, M., Batchelder, A.W., Kempf, M.-C., et al., 2018. Perceptions of intersectional stigma among diverse women living with HIV in the United States. *Soc. Sci. Med.* 208, 9–17.
- Rizvi, S., Khan, M., Kundi, A., Marsh, D., Samad, A., Pasha, O., 2004. Status of rheumatic heart disease in rural Pakistan. *Heart* 90, 394–399.
- Robertson, K.A., Mayosi, B.M., 2008. Rheumatic heart disease: social and economic dimensions. *S. Afr. Med. J.* 98, 780–781.
- Sanderson, S.C., Waller, J., Humphries, S.E., Wardle, J., 2011. Public awareness of genetic influence on chronic disease risk: are genetic and lifestyle causal beliefs compatible? *Public health genomics* 14, 290–297.
- Savulescu, J., Kerin, J., 1999. The "geneticisation" of disease stigma. *Lancet* 354, 16.
- Siccama, C.J., Penna, S., 2008. Enhancing validity of a qualitative dissertation research study by using NVivo. *Qual. Res. J.* 8, 91–103.
- Sliwa, K., White, A., Milan, P., Olga Mocumbi, A., Zilla, P., Wood, D., 2018. Momentum builds for a global response to rheumatic heart disease. *Eur. Heart J.* 39 (48), 4229–4232.
- Staunton, C., Tindana, P., Hendricks, M., Moodley, K., 2018. Rules of engagement: perspectives on stakeholder engagement for genomic biobanking research in South Africa. *BMC Med. Ethics* 19, 1–10.
- Swartz, L., 1998. *Culture and Mental Health: A Southern African View*. Oxford University Press Southern Africa.
- Tekola, F., Bull, S., Farsides, B., Newport, M.J., Adeyemo, A., Rotimi, C.N., et al., 2009. Impact of social stigma on the process of obtaining informed consent for genetic research on podoconiosis: a qualitative study. *BMC Med. Ethics* 10, 13.
- Thornicroft, G., Rose, D., Kassam, A., Sartorius, N., 2007. Stigma: ignorance, prejudice or discrimination? *Br. J. Psychiatry* 190, 192–193.
- Wade, C.H., 2019. What is the psychosocial impact of providing genetic and genomic health information to individuals? An overview of systematic reviews. *Hastings Cent. Rep.* 49, S88–S96.
- Watkins, D., Zuhlke, L., Engel, M., Daniels, R., Francis, V., Shaboodien, G., et al., 2016. Seven key actions to eradicate rheumatic heart disease in Africa: the Addis Ababa communiqué. *Cardiovascular Journal of Africa* 27, 184.
- Wong, L.P., 2008. Focus group discussion: a tool for health and medical research. *Singap. Med. J.* 49, 256–260.
- Zühlke, L., Perkins, S., Cembali, S., 2018. Rheumatic heart disease patient event: Cape Town hosts 4th Annual Listen to My Heart Rheumatic Heart Disease for patients at the South African Heart Association meeting in 2017. *Eur. Heart J.* 39 (19), 1669–1671.