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Positive Body Image and Quality of Life in Individuals with Marfan Syndrome

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AVISOS LEGAIS

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Estudo em colaboração

O presente estudo é parte integrante do projeto de investigação "A aceitação do corpo na doença: Estudo da imagem corporal positiva em diferentes condições clínicas".

Como colaboradora neste projeto de investigação, durante o presente ano letivo 2022/2023, participei na recolha e análise dos dados. O trabalho aqui apresentado baseia-se apenas numa parte dos dados recolhidos e constitui uma análise quantitativa das variáveis associadas à imagem corporal positiva em pessoas com síndrome de Marfan.

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Resumo

A imagem corporal positiva (ICP) refere-se a atitudes favoráveis de aceitação e admiração pelo corpo, englobando facetas que são inconsistentes com as típicas imagens idealizadas. Trata-se de um construto recente e multidimensional que é pouco estudado em junção com síndromes raros, como é o caso da Síndrome de Marfan. As aparentes alterações corporais características desta síndrome apresentam impacto não só a nível médico, como a nível das múltiplas áreas da vida do indivíduo, podendo influenciar a sua qualidade de vida.

O presente trabalho tem como objetivo explorar duas facetas da ICP (a apreciação corporal e a aceitação do corpo pelos outros) e a sua relação com os domínios da qualidade de vida (QdV) (saúde física, saúde mental, relações sociais e ambiente), em indivíduos diagnosticados com Síndrome de Marfan. A amostra é constituída por 13 participantes portadores desta doença genética e 13 participantes saudáveis. As análises estatísticas foram conduzidas para explorar diferenças na ICP e na QdV entre estes dois grupos, atendendo ao nível de escolaridade, idade e duração da doença.

Os resultados demonstraram não haver diferenças estatisticamente significativas entre o grupo clínico e o grupo não clínico nas facetas da ICP e nos quatro domínios da QdV. No grupo clínico, foram encontradas fortes correlações entre a ICP e a QdV. Adicionalmente, a ICP correlaciona-se negativamente com a escolaridade e positivamente com a duração da doença.

Considerando a escassez de literatura existente sobre o tema da ICP, em particular, em pessoas diagnosticadas com síndromes raras, como é o caso da Síndrome de Marfan, a continuidade do estudo deste tema torna-se pertinente.

Palavras-chave: Imagem corporal positiva; síndrome de Marfan; qualidade de vida

Abstract

Positive body image (PBI) refers to the favorable attitudes of acceptance and admiration towards one's body, embracing aspects that deviate from the typical idealized images. It is a multidimensional construct that has received little attention in rare syndromes like Marfan Syndrome. The noticeable bodily changes associated with this syndrome have medical implications and impact various aspects of an individual's life, potentially influencing their overall quality of life.

This study aims to investigate two aspects of PBI, namely body appreciation and body acceptance by others, and their correlation with different domains of quality of life (QoL), including physical health, mental health, social relationships, and environment, among individuals diagnosed with Marfan Syndrome. The sample comprises 13 participants with this genetic disease and 13 healthy participants. Based on education level, age, duration of the disease, statistical analyses were conducted to explore differences in PBI and QoL.

The results revealed no statistically significant differences between the clinical and non-clinical groups in PBI facets and the four QoL domains. However, strong correlations were observed between PBI and QoL in the clinical group. Additionally, PBI showed a negative correlation with education level and a positive correlation with the duration of the disease.

Given the limited literature on PBI, particularly in individuals diagnosed with rare syndromes like Marfan Syndrome, continuing research on this subject is crucial.

Keywords: Positive body image; Marfan syndrome; quality of life

Résumé

L'image corporelle positive (ICP) fait référence à des attitudes favorables d'acceptation et d'admiration pour le corps, englobant des facettes incompatibles avec les images idéalisées typiques. C'est un construit récent et multidimensionnel peu étudié en lien avec des syndromes rares, comme le syndrome de Marfan. Les changements corporels apparents caractéristiques de ce syndrome ont un impact non seulement sur le plan médical, mais aussi sur plusieurs domaines de la vie de l'individu et peuvent influencer sa qualité de vie.

Le présent travail vise à explorer les deux facettes de l'ICP (appréciation du corps et acceptation du corps par les autres) et sa relation avec les domaines de la qualité de vie (QdV) (santé physique, santé mentale, relations sociales et environnement), chez les individus diagnostiqués avec le syndrome de Marfan. L'échantillon est composé de 13 participants atteints de cette maladie génétique et de 13 participants sains. Des analyses statistiques ont été menées pour explorer les différences de PCI et de qualité de vie entre ces deux groupes, en tenant compte du niveau d'éducation, l'âge et la durée de maladie.

Les résultats n'ont montré aucune différence statistiquement significative entre le groupe clinique et le groupe non clinique dans les facettes de l'ICP et dans les quatre domaines de la qualité de vie. Dans le groupe clinique, de fortes corrélations ont été trouvées entre l'ICP et la qualité de vie. De plus, l'ICP est corrélé négativement avec l'éducation et positivement avec la durée de la maladie.

Compte tenu de la rareté de la littérature existante sur le sujet de l'ICP, en particulier chez les personnes diagnostiquées avec des syndromes rares, comme le syndrome de Marfan, la continuité de l'étude de ce sujet devient pertinent.

Mots-clés: Image corporelle positive; syndrome de Marfan; qualité de vie

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List of Acronyms and Abbreviations

PBI- Positive Body Image

NBI- Negative Body Image

QoL- Quality of Life

MFS- Marfan Syndrome

CG- Clinical Group

NCG- Non-Clinical Group

1. Introduction

1.1 Body Image

Body image can be conceptualized employing two dimensions: the evaluation of the body and the perception of body dynamics (Hansen et al., 2020). Body evaluation encompasses the critical appraisal of one's appearance and the sense of well-being in one's body. On the other hand, body dynamics refers to the perception of energy and vitality. This dimension describes the experience of power, fitness, and health-related to one's body (Hansen et al., 2020). Body image is, therefore, not a simple perceptual phenomenon. When we set out to measure an individual's perception of their own body, the judgments are highly influenced by cognitive, affective, attitudinal, and other variables (Slade, 1994), being a multidimensional concept. The study of this construct is of great importance since they are significantly associated with several others, namely with the individual's behavior, self-esteem, and interpersonal relationships (Hong & Hong, 2019).

Studying body image often leads us to the widely studied concept of negative body image. Negative body image (NBI) refers to the experience of negative thoughts and feelings toward one's body (Grogan, 2016). A NBI emerges due to several risk factors, such as low self-esteem, pressures imposed by the media, little social support from family or friends, and pressure from them, such as criticism about weight. Considering that the influence of media/social networks negatively affects body image during adolescence, age can also be seen as a risk factor. This construct is also related to anxiety disorders, frustration, eating disorders, and depression (Marques, 2020).

Body Image also comprises the concept of positive body image (PBI). While a negative body image constitutes a risk factor, a positive body image is a protective factor for the subject. A PBI is an extensive endearment and regard for the body, granting a high acceptance and admiration towards the body**,** encompassing facets inconsistent with typical idealized images. It encloses an appreciation for its singular beauty and the functions it performs. Therefore, there is an emphasis on the body's assets rather than having the focus directed toward imperfections. The incoming information is interpreted

in a body-protective manner through which most positive information is internalized, and most negative information is declined or reframed. Overall, PBI is often acknowledged as one feeling beautiful, comfortable, confident, and happy with their own body, which is often reflected as an outer radiance **(**Tylka & Wood-Barcalow, 2015).

Regarding the study of PBI, body appreciation has perhaps been the leading and studied concept at the level of research (Andrew, Tiggemann, & Clark, 2016; Tiggeman & McCourt, 2013). According to Tylka and Wood- Barcalow (2015), body appreciation consists of the appreciation of functionality, health, and characteristics of one's body and not so much the appreciation of the person's general appearance. General and physical welfare is also associated (Williamson & Karazsia, 2018).

Experimental studies have demonstrated a protective effect of body appreciation against media-induced body dissatisfaction in young women (Andrew et al., 2016). Andrew et al. (2015) noted that body appreciation is a protective element in circumstances where social patterns of beauty are present.

Tylka and Wood-Barcalow (2015) also reference bodily love and acceptance regarding the remaining facets of PBI. It concerns the display of love for the body and comfort felt within oneself, even if the person is not completely satisfied. It must be highlighted that this does not meet the concept of narcissism and vanity. A PBI also incorporates a broad idea of beauty, particularly employing a perception that several attributes can be beautiful, regardless of whether they can be modified. This facet of beauty is not exclusively meant to be found in the people fulfilling it, and it must be equally seen in themselves (Tylka & Wood- Barcalow, 2015).

Furthermore, these authors emphasize the component of self-care as an investment in one's appearance since it does not alter it to fit beauty standards negatively. The ability to filter information in a way that protects the person's body is also presented as a focal domain, which, in turn, is associated with accepting information that encourages positivity in the subject's body image and the refusal of information that may jeopardize this positivity. Menaces to body image include weight discomfort, advertisements encouraging dieting, and models who follow beauty ideals. PBI is also shaped by social identities, such as one's culture, gender, age, and weight, and the perception that others accept the person's body. In the Portuguese population, studies have revealed that men report higher levels of body appreciation compared to women in young people and adults

(Araújo, 2019; Lemoine, et al., 2018) and in middle-aged adults (Cabral & Vieira, 2019) but not in the older population, where no differences between men and women were identified (Meneses et al., 2019). The results of Alves's (2022) study revealed, in a non-clinical sample, gender differences in PBI and a positive relationship with age and a negative association with BMI. Older people and thinner people reported higher levels of body appreciation. Individuals who perceive that others accept their bodies as they are, regardless of any perceived defects, are more likely to accept and appreciate their bodies and exhibit higher levels of body appreciation (Andrew et al., 2016). Regarding the impact culture has on PBI, Jaeger et al. (2002) large-scale comparison of 1751 young women in 12 countries showed that women in typically Western societies (e.g., Sweden, Germany, Spain, France, Italy, and the UK) were more dissatisfied with their bodies than were women in non-Western cultures (e.g., Ghana, India, and Gabon).

According to Tylka and Wood-Barcalow (2015), PBI is a broad, stable, malleable concept that should be interpreted cautiously. It functions as a protective component that should secure physical and psychological well-being.

1.2 Marfan syndrome

Marfan syndrome is a rare, autosomal-dominant connective tissue disorder caused by mutations in the fibrillin one gene (Velvin et al, 2015). From a population-based perspective, the prevalence has been reported as 1 in 5–10,000 individuals (Sivasankari et al., 2017). Marfan syndrome has a very low prevalence with similar frequency in both genders and all countries and ethnicities. It is a severe, chronic, and life-threatening disease with multiorgan involvement without the availability of curative therapy. It is affiliated with chronic fatigue, pain, and psychological despair (Von Kodolitsch et al., 2015).

This disorder predominantly targets the cardiovascular system, eyes, and skeleton. The diagnosis is commonly considered in a young person with tall, thin body habitus, long limbs, arachnodactyly, pectus deformities, and sometimes scoliosis. The most highlighted and prominent skeletal feature is the long bones' out-of-proportioned overgrowth. The main ocular abnormality is lens dislocation; however, several other pathologies, such as cataracts or glaucoma, can be developed. The cardiovascular disorder most frequently presented is the dilatation of the ascending aorta at the aortic

sinuses level associated with aortic valve incompetence leading to aortic dissection. Cardiovascular complications are the leading cause of mortality for patients with Marfan syndrome (Sivasankari et al., 2017). Surgical intervention is highly recommended for affected individuals whose aortic roots are significantly dilated or dissected. Providing those managing cardiovascular symptoms prove successful, the life expectancy for individuals with Marfan syndrome increases significantly (Peters et al., 2001). Refraining from sports and limiting physical exertion is advised for many patients to reduce the risk of aortic dilatation and lens dislocation. Consequently, inactivity and a sedentary lifestyle are expected (Velvin et al., 2016).

For many affected individuals, the diagnosis of Marfan syndrome is usually made during childhood or early adulthood. Thus, earlier diagnoses allow affected individuals more opportunities to engage in health-enhancing health behaviors, such as cardio-vascular medication regimens and prophylactic surgery, before substantial condition-related morbidity occurs (Peters et al., 2002).

Three international nosologies have been proposed for the diagnosis of Marfan syndrome: the Berlin nosology in 1988; the Ghent nosology in 1996 (Ghent-1); and the revised Ghent nosology in 2010 (Ghent-2) (Von Kodolitsch et al., 2015). Presently, clinicians use the 2010 revised Ghent nosology with greater frequency, including optional genetic sequencing of the FBN1 gene to diagnose patients (Groth et al., 2015), improving patient management.

1.3 Body Image in Marfan Syndrome

Marfan syndrome is a major disorder through its skeletal abnormalities, namely an apparent long, slender body shape with an above-average body height (Hansen et al., 2020). Due to their many physical limitations, patients with Marfan syndrome do not often engage in physical fitness. This can be a risk factor, considering that exercise and physical fitness significantly affect body image (Hansen et al., 2020). Verified risk factors also include the degree of impairment and the experience of surgery resulting in a negative body image in patients with congenital heart disease since this is a frequent occurrence (Hansen et al., 2020). Research regarding the body in Marfan syndrome is presently limited. However, the few studies available prove essential to understanding

this subject. The studies of Hansen et al. (2020) and Warnink-Kavelaars et al. (2019) elaborate specifically on body image in Marfan Syndrome.

In the study of Hansen et al. (2020), the dimensions of body image disturbances in male (N = 9) and female (N = 33) Marfan patients, where the mean age was 45.1 years, were assessed and compared with a representative normative sample. It was found that people with Marfan syndrome presented a significantly impaired body image compared to the normative sample. Marfan syndrome patients claimed to not feel well in their bodies and evaluated it much more negatively. They also perceived themselves as much less powerful and energetic than healthy individuals of comparable age.

In addition, the study by Hansen et al. (2020) explored whether physical, psychological, and sociodemographic variables could predict patients' satisfaction with body image. The researchers identified psychological variables as the main predictors of differences in body image in individuals with Marfan syndrome. Disease-related variables nor variables indicating objective physical fitness supported a more influential impact on body image than the psychological state of health. Patients with Marfan syndrome may feel more vulnerable, given higher levels of anxiety, which may lead them to evaluate their bodies negatively (Hansen et al., 2020). Moreover, the above-average height predominantly perceived in Marfan syndrome patients was related to a negative body assessment. Considering the atypical appearance of individuals with this syndrome, social interactions may generate feelings of insecurity, which are then attributed to body image, mainly if social anxiety is present (Hansen et al., 2020).

The physical body undergoes many changes as people age, and losing a youthful body may be challenging to accept altogether. Mindful, adapting to a new physical reality of one's body may be increasingly challenging in the presence of a congenital condition such as Marfan syndrome (Hansen et al., 2020).

Psychological variables such as depression and anxiety determine the way individuals with Marfan Syndrome experience their body bodies rather than measures of fitness or cardiac impairment. Hansen et al. (2020) found individuals with MFS tend to evaluate their bodies much more negatively and feel less powerful and energetic than individuals from the general population. This finding is feasible given that being noticeably different, and expecting or perceiving negative reactions from others, will most likely impact psychological functioning, potentially resulting in a compromised

body image. Treatment and rehabilitation concepts should be aware that body image is mainly predicted by depression and anxiety for it to be appropriately addressed and managed. Therefore, psychological interventions targeted at depression and anxiety should also be considered rather than exclusively focusing on medical treatment and aspects of physical rehabilitation (Hansen et al., 2020).

A study by Warnink-Kavelaars et al. (2019) on adolescents' perspectives (N = 19) on physical functioning and other constituents reports that adolescents with Marfan syndrome feel different from their peers due to their condition's appearance and disability. Bullying creates negative feelings about themselves and their peer status; some reported a negative self-image/esteem. As a result, these adolescents avoided various social activities such as going to the swimming pool, beach, parties, or sports.

However, most of them outlined that the key to feeling accepted and less different from their peers would be to accept their appearance.

A few studies support the evidence found thus far. Rumsey and Harcourt (2004) report that individuals with visible physical differences often report difficulties regarding social interactions and possess negative self-esteem or body image. These hardships in interpersonal relationships are evident through the difficulty in making new friends, meeting new people, or even establishing long-term relationships. To explain this, Rumsey (2002) references other people's negative attitudes, such as staring, audible comments, and direct questions about their physical condition. Thereby, when individuals experience negative social situations, they begin to internalize them and anticipate similar experiences in the future, resulting in a poor interaction style, shyness, aggression, or excessive social avoidance (Rumsey & Harcourt, 2004).

1.4 Body Image and Quality of Life in Marfan Syndrome

Quality of life can be described as a person's sense of well-being stemming from satisfaction or dissatisfaction with areas of life that are significant to an individual (Peters et al., 2002). The concept of QoL has become progressively acknowledged as an essential patient-reported result measure in assessing care and treatment (Hunter & Swan, 2016).

Studies regarding PBI and Quality of Life (QoL) in Marfan Syndrome are scarce. Nonetheless, some research was done on the quality of Life in MFS.

Moon et al. (2016) found that a lower QoL was associated, along with other variables, to lower body image. Body image issues were associated with depression, anxiety, and QoL. These issues comprise significant height, long and thin fingers, scoliosis, and the need for thick eyeglasses in individuals with the syndrome (Moon et al., 2016).

Fusar-Poli et al. (2008) found that subjects affected with Marfan syndrome reported lower QoL in the mental domain but not in the physical domain, compared with the general population. It was stipulated that MFS can impose a burden on daily life, particularly on school attendance, work opportunities, and social behavior, as well as on the development of an introverted personality, possibly generating defensive psychological traits, such as denial and isolation (Fusar-Poli et al., 2008). Contrariwise, Fusar-Poli, et al. (2008) mentioned that having MFS can nourish self-awareness, awareness of the existence, and eventually personal identity, thus increasing QoL regardless of the disorder's severity.

Additionally, Andonian et al. (2021) compared adults with different congenital heart defects with adults with Marfan syndrome and concluded that subjects affected by the syndrome reported an overall reduced QoL. This study highlighted emotional distress as a harmful factor for the patient's health, leading to increased cardiovascular morbidity and premature mortality. A few patients with Marfan syndrome were at high risk for unfavorable psychosocial outcomes, distinctly in the psychological and physical domains.

Velvin et al. (2019) found that being male, being older, having a lower educational level and lower income, not working, having low private insurance, and having little social support were variables significantly associated with decreased QoL in patients with MFS. Symptoms such as pain, fatigue, psychosocial distress, and learning disability can substantially impact the QoL (Velvin et al., 2019). Nevertheless, high QoL can be experienced despite numerous issues, for instance, having an absence of awareness of one's illness, successful social embedding, and mild symptom manifestations of the syndrome during childhood. Pediatric patients with Marfan syndrome greatly benefit from being taught to engage in positive coping skills and having informed parents about

the disorder (Velvin et al., 2019). As mentioned by Goldfinger et al. (2017), some predictors of better QoL include having a college education, full-time employment, a higher household income, health insurance, a reduced number of prior surgeries, a careful consumption of alcohol, absence of depression, and less severe manifestations of Marfan syndrome.

1.5 The present study

Future research needs to be wary of body image in treating Marfan syndrome. Increased attention should be directed towards enhancing subjective well-being to potentially improve the QoL and long-term health outcomes of people with MFS.

The present study looks to polish some of the previous research and (1) assess the PBI and the QoL in people with MFS, compared with a Non-Clinical Group, as well as (2) determine to what extent PBI and sociodemographic factors influence the QoL in people with MFS. We will use these two constructs (PBI and QoL) in people with Marfan syndrome to better understand the dimensions previously analyzed and find more precise answers to this puzzling subject. Given our methodological design and our review, we can propose the following hypothesis:

Hypothesis 1: The PBI facets (body appreciation and body acceptance by others) and the QoL domains (physical and psychological health, social relationships, and environment) are expected to display significantly higher values in the Non-Clinical Group than in the Clinical Group (Hansen et al., 2020).

Hypothesis 2: In the clinical sample, positive and significant associations are expected between the facets of PBI and Quality of Life domains (da Ponte Cordeiro, 2021). Also, positive associations are expected between age, education level (Goldfinger et al., 2017; Velvin et al., 2019), and duration of the disease with QdV. Similarly, are expected positive associations between these three variables and PBI (Alves, 2022; Tiggermann, 2015).

2. Method

2.1 Design

The present study follows a quantitative methodology (self-report measures) with an observational, analytical, and cross-sectional research design, with a Clinical Group (CG) and a Non-Clinical Group (NCG).

2.2 Participants

The inclusion criteria of participants in the clinical group encompass evidence of a medical diagnosis of Marfan syndrome, normal (or rectified to normal) visual acuity, and being 18 years of age or older. Exclusion criteria covered the existence of some cognitive deficit; pregnancy or a 6-month post-pregnancy period; significant ophthalmologic pathology; gender dysphoria; self-harming behaviors; severe psychiatric comorbidity, as well as the exclusion of all participants who were in critical condition or palliative care.

The inclusion criteria for the control or non-clinical group were chronological age equal to or greater than 18 years, normal weight (a body mass index between 18.5 and 25 kg/m2), and a regular menstrual period (in premenopausal women). All participants with a current or history of severe medical or psychiatric illness were restricted from participating.

The clinical group (CG) consists of 13 participants, ten men and three women, with ages between 18 and 70 (M = 37.46; DP = 17.13) and an education level that ranges from 2 up to 19 years (M = 12.69; DP = 4.07). Regarding marital status, seven are single, and six are married or in a non-marital partnership.

The non-clinical (NCG) group is made up of 13 participants, ten men and three women, with ages between 18 and 71 (M = 37.62; DP = 17.36) and an education level ranging from 12 up to 17 (M = 12.85; DP = 1.73). Concerning marital status, six are single, and seven are married or in a non-marital partnership.

It should be noted that a prior matching was carried out between the participants of the clinical group and the non-clinical group according to gender and age to ensure homogeneity between groups concerning the present sociodemographic variables.

2.3 Materials

Sociodemographic questionnaire. To collect sociodemographic and clinical data from the participants, a questionnaire was designed to gather information on gender,

age, marital status, education level, professional status, anthropometric data, diagnosis, duration of the disease, and treatments.

Body Appreciation Scale-2 (BAS-2). The Body Appreciation Scale-2 was developed by Avalos, Tylka, and Wood-Barcalow (2005) and was later validated for the Portuguese population (Lemoine et al., 2018) to assess body acceptance, favorable opinions, and respect for the body. It is a self-response questionnaire comprising 10 items that must be answered using a five-point Likert Scale (1=never, 2=rarely, 3=sometimes, 4=often, 5=always). It has good psychometric qualities and a good internal consistency in the validation study for the Portuguese adult population (Cronbach's alpha of .94 for women; .91 for men) (Lemoine et al., 2018). In this study, the Cronbach's alpha for the Marfan syndrome sample was .95 and for the NCG was .94, revealing good psychometric properties. Its administration is simple and easy to score while applying to various fields, such as assessing body appreciation within the research area, clinical practice, or an educational environment.

Body Acceptance by Others Scale (BAOS). The Body Acceptance by Others Scale was developed by Avalos and Tylka (2006; Portuguese version by Barbosa et al., 2018), and it is a unifactorial instrument that intends to understand the degree to which the individual perceives the acceptance of his body by people external to him (family, friends, boy/girlfriends, and media). It has ten items, which should be answered according to a Likert Scale of 5 points: from 1 (never) to 5 (always) (Avalos & Tylka, 2006). Avalos and Tylka (2006) revealed the presence of a good internal consistency (α =.91). The internal consistency values of the Portuguese version of this scale have shown satisfactory levels, both in samples of adolescents (α = .93) and adults (α = .94; Barbosa et al., 2018). In this study, the internal consistency indexes were .95 in the CG and .96 in the NCG group.

Quality of Life. Quality of life was assessed using an instrument developed by the WHOQOL group (World Health Organization Quality of Life [WHOQOL], 1998), specifically by the WHOQOL-BREF, an abbreviated version of the WHOQOL-100. The WHOQOL-BREF provides a valid and reliable alternative to the WHOQOL-100, being more useful in studies that require a brief assessment of QoL and for health professionals to assess the effectiveness of the treatment since it is easy to apply, and the filling is less time-consuming. In this study, the Portuguese version of the WHOQOL-BREF by Vaz

Serra et al. (2006) was used. The scale is made up of 26 items, organized into four domains related to QoL: physical and psychological health, social relationships, and environment. It also includes a facet of QoL and general health. The Cronbach's alpha of this instrument varies between .64 (social relationships QoL) and .87 (physical QoL), and the domains together explain 58.8% of the variance (Serra et al., 2006). In this study, Cronbach's alpha varies between .74 (social relationships) and .90 (environment) in the CG and between .87 (physical and psychological health) and .96 (environment) in the NCG.

2.4 Procedure

The sample collection was based on a non-probabilistic sampling process by snowball sampling or convenience.

After considering the inclusion and exclusion criteria defined, their doctor invited participants in the clinical group to participate in the study. All participants were informed about (a) the purpose of the study; (b) the absence of expected risks in the participation; (c) the procedures that comprise the study (e.g., filling in self-report measures), and (d) the time required for participation (approximately 20 minutes). The information provided orally by the researcher was likewise written on an Information Sheet and delivered to the participant. It was reinforced that their participation would be voluntary and confidential, and before data collection began, the Informed Consent was also completed.

The in-person collection of the clinical sample took place in a central hospital in Greater Porto. Due to difficulties accessing the internet in these locations, the questionnaires were administered on paper. Simultaneously, data was collected virtually through contacts in the Portuguese Association of Marfan Syndrome and some social connections.

The sample from the non-clinical group was gathered online through the UP LimeSurvey platform. Participants were contacted directly via email or social networks to present the study and display interest in collecting a sample. People who were prone to partake filled out the questionnaire through the link shared by the researcher. In addition, after reading the information concerning the study and its aim, all participants had to give their informed consent, without which completing the questionnaire was impossible.

The protocol consists of 14 pages, arranged counterbalanced, and its execution took, on average, 15 minutes.

This project complies with the ethical and data protection procedures required. It considers the ethical principles set out in the Declaration of Helsinki, which considers voluntary participation, informed consent and its clarification, confidentiality, and access to data.

All instruments used had the authorization of the original authors or the authors of the national versions.

The study was approved by the Ethics Committee of the Hospital Center (approval number 301/18), by the Faculty of Psychology and Education Sciences of the University of Porto (approval number 2028/12-6b), and the University of Porto Data Protection Impact Assessment (AIP-UP, approval number P-4/2020).

In terms of statistical procedures, after administering the test battery, the data was coded and subsequently statistically processed using the statistical analysis program IBM SPSS Statistics (Statistical Package for the Social Sciences), version 28.

The database was cleaned, and any violations of statistical assumptions that could distort the results were evaluated. In total, no participants were eliminated in either of the groups, even though there were missing values.

A descriptive analysis was conducted to characterize the sample and the study variables. The assumption of normal distribution in the dependent variables was checked but not confirmed, necessitating non-parametric statistics. Specifically, the Mann-Whitney U test was employed to compare the Clinical Group with the Non-Clinical Group regarding the facets of PBI and the four QoL domains. To determine the effect size, the r measure was used, following Cohen's (1988) guidelines: .1 indicating a small effect, .3 a medium effect, and .5 a large effect. Subsequently, Spearman's correlation coefficient was utilized to study the relationship between the PBI facets and QoL domains. Furthermore, an attempt was made to explore the relationship between the four QoL domains and three sociodemographic and clinical variables (age, education level, and duration of the disease).

3. Results

3.1 Comparative analysis between the Clinical Group and the Non-Clinical Group regarding PBI and QoL

As shown in Table 1, there were no statistically significant differences found between the CG and NCG regarding the two facets of the PBI: body appreciation, *Z* (U)

= -0.257, p = .797, r = .05 and body acceptance by others, Z (U) = -1.292, p = .196, r = .25.

Similarly, no statistically significant differences were observed between the CG and NCG concerning the four domains of the QoL: physical health, Z (U) = -0.164, p = .870, r = .03, psychological health, Z (U) = -0.082, p = .934, r = .02, social relationships, Z (U) = -1.182, p = .237, r = .24 and environment, Z (U) = -0.164, p = .870, r = .03. [1]

Table 1

Mann-Whitney U test for differences in PBI and QoL between the CG and NCG

		Clinical Group (N = 13)			Non-Clinical Group (N = 13)			Mann-Whitney U Test		
		MR	SR	Mdn	MR	SR	Mdn	Z	*p*	*r*
PBI Facets	BAS-2	13.12	170.50	4.10	13.88	180.50	4.10	-0.257	.797	.05
	BAOS	11.58	150.50	4.00	15.42	200.50	4.60	-1.292	.196	.25
Quality of Life domains	Physical	12.75	153.00	80.35	13.23	172.00	75.00	-0.164	.870	.03
	Psychological	12.88	154.50	72.91	13.12	170.50	75.00	-0.082	.934	.02
	Relations	11.21	134.50	54.17	14.65	190.50	75.00	-1.182	.237	.24
	Environment	12.75	153.00	73.44	13.23	172.00	75.00	-0.164	.870	.03

**. Significant differences at the .01 level
*. Significant differences at the .05 level
MR – Mean Rank; SR – Sum Rank; Mdn - Median

3.2 Correlations between PBI, QoL, Age, Education level, and Duration of the disease in the Marfan participants

Table 2 presents the correlations between the variables mentioned above. Upon closer examination, the correlations between the PBI facets and the QoL domains exhibit positive and moderate to strong associations, aligning with expectations.

[1] Given the fact that statistically significant differences were found between the two groups regarding the Body Mass Index (BMI) [Z (U) = -2.69, p = <.01, r = .53], ANCOVAs were carried out between the CG and the NCG concerning the PBI and QoL using the BMI as a covariate. However, no significant differences were found either PBI(body appreciation, F (1, 23) = 0.279, p = .603, η^2 = .012; body acceptance by others, F (1, 23) = 2.789, p = .108, η^2 = .108 or QoL domains (physical health, F (1, 22) = 0.232, p = .635, η^2 = .01; psychological health, F (1, 22) = 0.021, p = .887, η^2 = .001, social relationships, F (1, 22) = 0.977, p = .334, η^2 = .043, and environment, F (1, 22) = 0.016, p = .899, η^2 = .001.

Regarding body appreciation, moderate to strong associations were found with all QoL domains: Psychological Health, *rs* = .827, n = 12, *p* < .01, Physical Health, *rs* = .752, n = 12, *p* < .01, and Social Relationships, *rs* = .662, n = 12, *p* = .019). Additionally, a moderate association was observed between the Environment and Body Appreciation, *rs* = .550, n = 12, *p* = .064), although it did not reach statistical significance due to the small sample size. These results suggest that higher body appreciation and functionality levels are linked to a greater perception of one's QoL, and vice versa.

Similarly, positive and moderate to strong associations were found between body acceptance by others and the four domains of the QoL: Physical Health, *rs* = .866, n = 12, *p* < .01), Psychological Health, *rs* = .856, n = 12, *p* < .01), Social Relationships, *rs* = .707, n = 12, *p* = .01), Environment, *rs* = .683, n = 12, *p* = .014. The better one perceives body acceptance by others, the better one's quality of life perception, and vice versa.

Only two significant correlations were observed regarding the associations between the sociodemographic and clinical variables (age, education level, and syndrome duration) and PBI facets. Education level displayed a moderate and negative association with body appreciation, *rs* = -.598, n = 13, *p* = .031, indicating that higher education levels are associated with lower levels of body appreciation. On the other hand, the duration of the disease (in months) exhibited a moderate and positive association with the body acceptance by others, *rs* = .604, n = 13, *p* = .029, suggesting that the longer the duration of the disease, the higher the perception of body acceptance by others.

Despite not being statistically significant (perhaps due to the sample size), there were one negative and two positive moderate correlations. A negative correlation was found between the education level and body acceptance by others, *rs* = -.439, n = 13, *p* = .134, implying that the higher the education level, the lower the perception of body acceptance by others. Two moderate and positive correlations were found between the duration of the disease and two QoL domains: Physical Health, *rs* = .541, n = 12, *p* = .069) and Social Relationships, *rs* = .532, n = 12, *p* = .075) suggesting that longer disease durations are associated with better physical health and social relationships.

Table 2

Pearson Correlations between PBI, QoL, and Sociodemographic Variables

		1	2	3	4	5	6	7	8	9
PBI	1. BAS-2	—								
	2. BAOS	.866**	—							
Quality of Life	3. Physical	.752**	.866**	—						
	4. Psychological	.827**	.856**	.712**	—					
	5. Relations	.662*	.707*	.630*	.648*	—				
	6. Environment	.550	.683*	.750**	.590*	.739**	—			
Sociodemographic Variables	7. Age	.240	.225	-.183	.163	.436	.093	—		
	8. Education	-.598*	-.439	-.154	-.140	-.040	.152	-.120	—	
	9. Duration	.388	.604*	.541	.375	.532	.393	.240	-.104	—

**. Correlation is significant at the .01 level
*. Correlation is significant at the .05 level

4. Discussion

The primary objective of this study was to assess PBI and QoL in individuals with Marfan Syndrome (MFS) compared to a Non-Clinical Group (NCG) as a control group. The study aimed to examine the influence of PBI and sociodemographic factors on QoL in individuals with MFS. By exploring these constructs in the context of MFS, the study sought to contribute to a better understanding of the related domains and facets.

The study found no significant differences in PBI between the Clinical Group (CG) and the NCG. The effect sizes observed were small. These results differ from those reported by Hansen et al. (2020), where Marfan patients were compared to a normative sample and demonstrated significantly impaired body image. The authors concluded that individuals with Marfan syndrome have negative perceptions of their bodies and consider themselves less powerful and energetic than healthy individuals of similar age. However, it is important to note that the present study had a smaller sample size compared to Hansen et al. (2020), and different scales were used to evaluate body image.

Nonetheless, Rumsey and Harcourt (2011) suggest that individuals with dysmorphic conditions, such as those in the clinical group of this study, may develop a

positive body image by learning to embrace their differences as unique and valuable characteristics. Tiggemann (2015) also suggests that individuals with visible differences or disfigurements can adopt a positive body image by perceiving their visual difference as only a small part of their lives. Furthermore, Feragen (2012) argues that most individuals with visible physical differences can cope adaptively and even transform the challenge of living with differences into a positive experience. In a study conducted by Marques (2020) involving a sample with various genetic conditions, no significant differences were found in the facets of PBI between the CG and the NCG. These results were attributed to the individuals' resilience in overcoming the issues related to their condition. It is suggested that this resilience may contribute to developing more positive attitudes towards their bodies among individuals in the clinical group, fostering acceptance and appreciation of their unique physical characteristics.

Additionally, individuals without visible physical differences or disfigurements tend to express positive body image as being comfortable with and appreciating their bodies, including perceived “imperfections” (Tiggemann, 2015).

Similarly, this study found no significant differences in QoL between individuals with MFS and those without the syndrome. The effect sizes were also small, indicating that both groups have similar quality of life. These results contrast with a systematic review conducted by Velvin et al. (2019), which concluded that living with a lifelong, potentially disabling disease like Marfan syndrome may lead to increased challenges in daily life, decreased QoL, and psychological distress. However, Peters et al. (2002) reported results suggesting that individuals with MFS generally have a satisfactory quality of life. Furthermore, De Bie et al. (2004) found that most Marfan patients cope well with their condition, despite the syndrome's impact on their lives.

In exploring the Body Mass Index (BMI) variable, which differed significantly between the groups, the study aimed to determine if it could act as a covariate influencing PBI and QoL. However, once again, no significant associations were found. This aligns with the findings of Rand-Hendriksen et al. (2010), who observed no relationship between QoL and BMI in individuals with Marfan syndrome. Furthermore, Swami et al. (2017) identified a negative association between BAS-2 and BMI, indicating that higher BMI scores were linked to lower body appreciation.

The correlations observed between the studied variables, including PBI, QoL, age, education level, and duration of the disease, provide insights into the relationships between these factors. Strong correlations were found between PBI and QoL, including physical QoL with BAS, physical QoL with BAOS, psychological health QoL with BAS, and psychological health QoL with BAOS. Moderate correlations were identified between social relationships QoL and BAS, social relationships QoL and BAOS, and environment QoL and BAOS. These findings are consistent with the importance of unconditional acceptance from others, as Tiggemann (2015) mentioned. Perceiving acceptance and valued qualities that are not dependent on appearance from meaningful individuals is crucial for positive body image. The family environment also significantly shapes body image, particularly in children with visible differences. Children assimilate acceptance or discomfort from their parents, and a family environment where the condition has minimal impact and body image, and self-esteem are not dependent on the disfigurement is most beneficial (Rumsey, 2012). For adults, cognitive beliefs are a crucial predictor of psychological well-being. A lack of concern about negative evaluations from others and a perception of social acceptance are essential for positive adjustment (Rumsey, 2012).

Individuals who can adjust positively to their condition tend to be comfortable in their skin and believe that their physical appearance can provide them with highly regarded characteristics, such as communication skills, inner strength, and genuine friendships not based on external factors (Feragen, 2012).

The severity of a visible difference does not necessarily predict the intensity of distress or negative body image, but psychological difficulties have been reported in individuals with noticeable differences (Rumsey & Harcourt, 2011; Tiggemann, 2015). The psychological factors surrounding visible differences play a significant role in their impact on body image.

The qualitative study by Wood-Barcalow et al. (2010) further supports the present study's findings. Women with specific illnesses affecting their appearance reported that managing their disease led to gratitude for the optimal functioning of their bodies, shifting the focus away from appearance-related concerns and fostering positive body image.

Overall, the studies mentioned support the strong and moderate correlations found in the present study between PBI, QoL, and the studied variables. They emphasize the importance of acceptance, valued qualities beyond appearance, family and peer

environment, cognitive beliefs, and psychological factors in shaping body image and its relationship with quality of life.

The positive correlations found between the domains and facets of PBI and QoL in the present study align with the idea that higher quality of life is associated with a more positive body image. Notably, the reduced sample of the present study will not represent all people with this syndrome. Nevertheless, this finding is consistent with the study by Moon et al. (2016), which found that lower QoL was associated with a lower body image. It suggests that individuals who experience a higher quality of life tend to have a more positive perception of their bodies. Additionally, the study of da Ponte Cordeiro (2021) supports these findings by verifying the existence of positive and significant associations between the PBI facts and the QoL domains in individuals with chronic kidney failure.

While this study did not find a direct relationship between the environment QoL and body appreciation, the study by Rhoten et al. (2013) in patients with head and neck cancer highlighted the moderating effect of environmental factors on body image. Environmental factors such as socioeconomic status, work status, and insurance status can influence the impact of dysfunction and disfigurement on body image. A positive and welcoming environment can contribute to a more positive body image.

Regarding the negative correlation between education level and body appreciation (BAS) and body acceptance by others (BAOS), the findings of the present study contradict those of Swami et al. (2008), where higher educational qualifications were associated with improved body appreciation. It is important to note that the sample in Swami et al.'s study did not mention any underlying health issues, while the present study focused on individuals with Marfan syndrome. However, the study by Gilbert-Diamond et al. (2009) supports the results of the present study, suggesting that higher education levels may lead to a more idealized body image, which can ultimately result in dissatisfaction with one's body. Body dissatisfaction and body concerns appear to be correlated to poorer academic achievement and functioning in college students (Yanover & Thompson, 2008).

The correlation between the duration of the disease and body acceptance by others in the present study indicates that individuals who have had Marfan syndrome for a longer period tend to experience greater acceptance of their bodies by others. This finding aligns with the study by Augustus-Horvath and Tylka (2011), which suggested that body appreciation becomes more reliant on the acceptance of others in women. A similar effect

may occur in a male sample, which could explain the present study's findings. Still, the statement that body appreciation increases with age is supported by the results of Alves (2022). This study indicates that individuals tend to have higher levels of body appreciation as they grow older. As people age, they often better accept their bodies and prioritize self-acceptance and self-care over societal beauty standards or external judgments.

The associations found between physical health and social relationships with the duration of the disease in the present study suggest that as individuals with Marfan syndrome live with the condition for a longer time, they may develop better strategies for taking care of their physical health and become more socially engaged. This could be attributed to the increased experience and knowledge gained over time in managing the challenges associated with the syndrome. The findings indicate that longer disease duration may contribute to improved self-care and a more outgoing attitude toward social interactions.

Furthermore, the study by Goldfinger et al. (2017) supports the association between education level and quality of life (QoL) in individuals with Marfan syndrome, indicating that lower education is linked to lower QoL. This suggests that individuals with higher educational qualifications may have access to more resources and knowledge that positively impact their overall well-being and quality of life.

The systematic review conducted by Velvin et al. (2019) on the hereditary thoracic aortic aneurysm and dissection found that increased age was associated with decreased quality of life in individuals with this diagnosis. Although the reviewed study focused on a different condition, it highlights the potential impact of demographic factors, such as age, on the quality of life of individuals with a hereditary disease. This finding may have implications for individuals with Marfan syndrome as well.

The associations observed in the present study and supported by previous research suggest that factors such as disease duration, education level, and age can influence the quality of life and social experiences of individuals with Marfan syndrome. These findings provide valuable insights into the complex interplay between demographic factors and the well-being of individuals with the condition.

5. Conclusion

The present study focused on exploring the relationship between positive body image (PBI) and quality of life (QoL) in individuals diagnosed with Marfan syndrome (MFS). This clinical population is particularly interesting in the field of psychology due to the significant impact of the disease on their lives. The study suggests that QoL and PBI are intertwined, meaning that the presence of one is closely related to the presence of the other. The findings also indicate that individuals with Marfan syndrome may learn to accept their bodies over time, despite the various challenges they face due to their condition.

It is important to note that further research is needed to expand our understanding of body image in people with Marfan syndrome and its implications for their quality of life. Future studies could benefit from employing qualitative methodologies, providing deeper insights into the participants' subjective experiences and the meanings they attribute to living with the disease. Qualitative research allows for a more comprehensive exploration of individuals' perspectives, emotions, and coping strategies, shedding light on the complex nature of their experiences.

By conducting qualitative studies in the future, researchers can gain a more nuanced understanding of how body image and quality of life intersect in individuals with Marfan syndrome. This would contribute to developing targeted interventions and support services that address this population's specific needs and challenges.

These findings suggest that PBI and QoL in individuals with MFS are complex and can be influenced by various factors, including individual experiences, perceptions of differences, and adaptation over time. Further research with larger sample sizes and comprehensive assessments may provide a deeper understanding of these constructs in the context of Marfan Syndrome.

Indeed, the present study's findings align with previous research that has explored the multifaceted interplay between various factors in the context of Marfan syndrome. The complex relationships between quality of life, environmental factors, education level, duration of the disease, and body image have been recognized in the literature. Awareness of these relationships is crucial for understanding the experiences and needs of individuals with Marfan syndrome.

By building upon the existing body of knowledge, the present study contributes to understanding the intricate connections between these variables. The findings shed light on how the quality of life and body image are intertwined, the potential moderating role of environmental factors, the influence of education level on body appreciation and acceptance by others, and the impact of disease duration on body acceptance.

By highlighting these relationships, the study provides valuable insights that can inform future research, clinical practice, and support services for individuals with Marfan syndrome. These findings contribute to a more comprehensive understanding of the factors that shape the experiences and well-being of individuals living with the condition.

The present study, in line with Moon et al. (2016), contributes to the limited research exploring the relationship between positive body image (PBI) and quality of life (QoL) in individuals with Marfan syndrome (MFS). By expanding our understanding of this relationship, the study emphasizes the importance of providing psychological support, specifically targeting self-esteem and body image concerns to preserve the overall quality of life for individuals with MFS.

The findings also highlight the role of education level in shaping body appreciation and acceptance by others. The notion that higher education may lead to a more idealized body image, which in turn can result in dissatisfaction with one's own body, is supported by the research of Gilbert-Diamond et al. (2009). The authors suggest that higher education often correlates with higher socioeconomic status, and individuals from these backgrounds may be more susceptible to distorted perceptions of body image. Thus, interventions aimed at challenging and restructuring harmful conceptions of body image may be particularly beneficial for these communities.

These insights have important implications for healthcare professionals working with individuals with MFS. By recognizing the significance of body image struggles and their impact on overall well-being, healthcare professionals can provide targeted support and guidance to help patients navigate these challenges. This may involve referring individuals to appropriate support structures, implementing interventions that promote positive body image, addressing underlying psychological factors related to self-esteem and body acceptance, and acknowledging the influence of education level and socioeconomic factors.

Overall, the present study contributes to the existing literature on Marfan syndrome by underscoring the need for psychological support and interventions to address body image concerns and preserve the quality of life.

Acknowledging limitations in the present study is essential for comprehensively considering its findings and implications. The first limitation mentioned is the predominantly male sample, which may restrict the generalizability of the results and hinder a thorough exploration of potential gender differences in positive body image and quality of life in individuals with Marfan syndrome. Future research should strive to include more gender-balanced samples to provide a more comprehensive understanding of the topic and potential gender-related nuances.

The second limitation mentioned is the small sample size of the clinical population, which may limit the statistical power of the analyses and the ability to draw robust conclusions. With a larger sample size, conducting more reliable statistical analyses and drawing more generalizable conclusions about the population of individuals with Marfan syndrome would be possible. Therefore, future studies should aim to include a larger sample to enhance the validity and generalizability of the findings.

Acknowledging these limitations shows that further research is needed to build upon the current study's findings. Conducting studies with more extensive and diverse samples, including both males and females, would allow for a more comprehensive understanding of positive body image and quality of life in individuals with Marfan syndrome. This expanded knowledge base will contribute to developing more effective interventions and support strategies tailored to the specific needs of individuals with Marfan syndrome, ultimately enhancing their overall well-being and quality of life.

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