Original Article

A Comparative Study of Mental Health Challenges in Parents of Children with Genetic versus Non-Genetic Conditions in Rwanda

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Cite as: Tuyishimire B, Niyonsenga J, Mutamuliza J, Kanazayire C, Ntete MJ, Mutesa L, et al. A Comparative Study of Mental Health Challenges in Parents of Children with Genetic versus Non-Genetic Conditions in Rwanda. Rwanda J Med Health Sci. 2024;7(2): 165-177. https://dx.doi.org/10.4314/rjmhs.v7i2.6

Abstract

Background
This study addresses a critical gap in research by examining the psychosocial impact on parents of children with genetic diseases compared to those with non-genetic conditions. While existing literature mainly focuses on medical aspects, it overlooks the emotional and mental health challenges faced by these parents.

Objectives
The main objective was to investigate and compare levels of depression, anxiety, perceived stress, parenting stress, self-esteem, and intimate partner violence between parents of children with genetic diseases and parents of children with non-genetic conditions.

Method
A cross-sectional comparative study was conducted with 100 caretakers of children with genetic diseases and 109 caretakers of patients with non-genetic diseases. Data were collected using standardized measures of self-esteem, intimate partner violence, perceived stress, and the Parenting Stress. Independent sample t-tests were performed to compare the means between the two groups.

Results
Parents of children with genetic diseases exhibited significantly higher levels of depression ($t(207) = 5.683$, $p < 0.001$), anxiety ($t(207)= 6.107$, $p<0.001$), perceived stress ($t(207)=11.680$, $p<0.001$), parenting stress ($t(207)= 12.893$, $p<0.001$), and intimate partner violence ($t(207)= 10.617$, $p < 0.001$) compared to parents of children with non-genetic conditions. Low self-esteem was also more prevalent in the case group ($t(207)= -14.565$, $p<0.001$).

Conclusion
These findings underscore the urgent need for comprehensive support systems to address the psychosocial challenges faced by parents and caregivers of children with genetic diseases. Recognition and targeted interventions for these issues can significantly enhance healthcare services, benefiting both patients and their parents.

Rwanda J Med Health Sci 2024;7(2):165-177

Keywords: Genetic diseases, Depression, Anxiety, Perceived stress, Parenting stress, Self-esteem, Intimate partner violence

Introduction

Genetic disorders, encompassing over 4,000 documented conditions, arise from anomalies within an individual's DNA, impacting a significant albeit relatively small global population.[1] Despite their rarity, these disorders bear profound implications for affected individuals and their families, giving rise to emotional and practical challenges that extend beyond the confines of the medical realm.[2] Children afflicted with genetic disorders often manifest distinct physical features, spanning from craniofacial dysmorphisms to intellectual disabilities. The diagnosis, frequently reliant on these features, imposes a considerable emotional burden on parents. Research findings suggest that 15.2% of parents with children affected by genetic abnormalities grapple with chronic adjustment difficulties, underscoring the unique psychological challenges faced by these families.[3]

This present research intends to explore the psychological impact on parents raising children with genetic disorders, comparing their experiences with those of parents of children with non-genetic diseases. Through an investigation into anxiety, depression, low self-esteem, parenting stress, perceived stress, and instances of intimate partner violence, this research seeks a profound understanding of the emotional and mental dynamics unique to families dealing with genetic health conditions. This study aims to bridge the existing gap in the literature by providing a comparative analysis of the psychosocial impact of genetic diseases on parents in Rwanda. The findings aspire to inform the development of targeted interventions and support systems, fostering not only the well-being of the patients but also addressing the overlooked emotional needs of their caregivers. The impetus for this study stems from the personal experiences gained in the Human Genetics department at Rwanda Military Hospital since 2020.

Daily encounters with patients exhibiting distressing indicators of genetic disorders prompted an exploration of the psychological challenges faced by parents in caring for these children. The introduction is enriched by narratives from parents of children diagnosed with genetic diseases, offering firsthand insights into the emotional and practical difficulties they face.[4] Concerns about the child's life, financial struggles, family turmoil, jealousy towards parents with non-disabled children, anxiety about the child's future, and worries about future pregnancies emerge as recurring themes.[4] Genetic diseases unleash a whirlwind of emotions for parents, ranging from shock and grief to fear and uncertainty. The initial diagnosis can be a profound shock, shattering the envisioned future for the child. Parents often find themselves on an emotional rollercoaster, oscillating between hope and despair as they navigate the unknown terrain of their child's.[5]

Depression, a pervasive mental disorder, emerges as a significant concern for families grappling with genetic diseases. The chronic nature of these conditions, such as Down syndrome, is associated with parental depression, impacting the developmental trajectory of affected children. [6] Depression, ranging from mild to severe forms, poses a substantial threat to the well-being of individuals and is recognized as a leading cause of disability globally.[7] On the other hand, Parents of children with genetic diseases, particularly intellectual disabilities like Down syndrome, report heightened stress levels. Mothers, in particular, exhibit both authoritative and less permissive parenting styles, with increased stress linked to verbal hostility. These stress levels signify the multifaceted challenges families encounter while caring for children with genetic disorders.[8]

In addition to that, Anxiety looms large in families dealing with genetic diseases, with concerns ranging from time limitations to the negative impact of family restrictions. Parents grapple with worries about providing adequate time for siblings, the financial burden of caring for a child with a disability, and the long-term impact on siblings when parents are no longer alive.[9]

This anxiety not only affects parents but also disadvantages siblings, creating a complex web of familial challenges.[10]

Moreover, the strain of caring for a child with a genetic disease can exert pressure on parental relationships. Partners may face communication challenges, differences in coping styles, and the need for shared decision-making. Recognizing and addressing these dynamics is essential for sustaining healthy relationships within families affected by genetic disorders.[8]

In Rwanda, the presence of a child with a disability in a family significantly impacts parents, imposing substantial emotional, financial, and social burdens.[11] Parents often experience high levels of stress and anxiety due to the cultural stigma associated with disabilities, which can lead to social isolation and psychological distress. The financial strain of medical expenses, specialized equipment, and educational needs further exacerbates their challenges, especially in a context where resources are limited. This constant pressure can lead to feelings of helplessness and burnout, as parents struggle to balance caregiving with other responsibilities. Additionally, the lack of accessible support services and community resources forces many parents to shoulder the burden alone, intensifying their emotional and physical exhaustion. [12]

The repercussions of genetic diseases extend beyond the afflicted individuals, permeating the lives of their caregivers. While existing research delves into the psychological challenges faced by parents of children with genetic diseases, a significant research gap persists in comparing the psychosocial aspects between this cohort and parents of children diagnosed with non-genetic conditions.[13] In the Rwandan context, where healthcare services predominantly focus on benefiting the patients, there exists a potential oversight of the psychological well-being of caregivers. This gap hinders the development of holistic support systems that cater to the comprehensive needs of families affected by genetic conditions. [14] Thus, we hypothesize that parents of children with genetic diseases experience elevated levels of depression, anxiety, perceived stress, and parenting stress compared to parents of non-genetic disease children. Additionally, parents of children with genetic diseases exhibit lower self-esteem levels in comparison to parents of non-genetic disease children. Moreover, parents of children with genetic diseases face a greater prevalence of intimate partner violence than parents of non-genetic disease children.

Methodology

Study design and Setting

This is a cross-sectional comparative study conducted to assess and compare the prevalence of psychological and social issues among families with members diagnosed with genetic diseases and families without such diagnoses. The research took place at Rwanda Military Hospital, located in Kigali city's Kicukiro district. Initially established in 1968 at Kanombe as a Military Referral Hospital, it exclusively served the military and their immediate families. However, since 1994, it has extended its services to the general population, offering both secondary and tertiary medical care. In the 2011-2016 strategic plan, the hospital evolved into a referral and teaching facility.

Study population and eligibility criteria

The study population comprised parents and caretakers of patients diagnosed with genetic diseases, and parents and caretakers of patients diagnosed with non-genetic diseases. To be eligible for participation in this study, individuals needed to be parents of children with confirmed genetic diseases or parents of children receiving treatment in the pediatrics department with diagnoses other than genetic diseases. Parents of children under six months of age were excluded from this study.

Sample size and sampling procedure

We employed a convenient sampling method, recruiting 100 caretakers of patients with genetic diseases and 109 caretakers of patients diagnosed with non-genetic diseases for interviews.This sampling method was non-probabilistic, with subjects selected based on specific criteria aligned with the research objectives.

Data collection instruments and procedure

To measure an individual's overall self-worth, the Rosenberg Self-Esteem Scale (RSE) comprises ten questions, utilizing a 4-point Likert scale from strongly agree to strongly disagree. Cumulative scores on this scale are categorized to indicate varying levels of self-esteem issues, providing a nuanced assessment that considers both positive and negative self-perceptions.

Moving on to the assessment of depressive symptoms and anxiety, the hscl-25 was used. It functions as a symptom inventory with 25 items divided into anxiety and depression components. Respondents rated each item on a four-point scale, and two distinct scores are computed: the total score, representing the average of all 25 items, and separate scores for depression and anxiety. Clinically significant symptoms are identified by an average score of 1.75 or higher for both depressive and anxiety scales.

The HITS tool, an acronym for Hurt, Insult, Threaten, and Scream, was employed as a straightforward screening method for assessing the risk of Intimate Partner Violence (IPV). With four questions and a point-based response system, this tool quantifies the frequency of abusive behaviors, providing a total score range of 4 to 20 points. A score surpassing 10 indicates the presence of intimate partner violence, offering a concise yet impactful assessment.

To gauge participants' subjective perception of stress, the Perceived Stress Scale (PSS) was used. It has 14 questions addressing various stressful situations. Scores on this scale, ranging from 0 to 56, are categorized into Low Stress, Moderate Stress, and High Stress, providing valuable insights into participants' overall stress perception and categorizing stress levels accordingly.

Finally, the Parenting Stress Index-Short Form (PSI-SF), a 36-item instrument, assesses the sources and extent of stress within the parent-child relationship. With three domains—Parental Distress, Parent-Child Dysfunctional Interaction, and Difficult Child—this self-report questionnaire provides a comprehensive understanding of parenting stress. In this study, only the subscales measuring parental distress and parent-child dysfunctional interaction were employed for infant-mother attachment testing, with the total PSI-SF score indicating overall parental stress levels based on percentile rankings.

The selection of the Rosenberg Self-Esteem Scale (RSE), Hopkins Symptom Checklist-25 (HSCL-25), HITS tool, Perceived Stress Scale (PSS), and Parenting Stress Index-Short Form (PSI-SF) is grounded in their demonstrated validity and reliability in psychological research. The RSE is a widely used measure for assessing self-esteem, providing a balanced view of positive and negative self-perceptions through its 10-item, 4-point Likert scale. This scale's established psychometric properties ensure accurate measurement of self-worth across diverse populations. Similarly, the HSCL-25 is a well-validated tool for detecting depressive and anxiety symptoms, offering a comprehensive symptom inventory that effectively captures the severity of these conditions with its two-component structure. The clinical relevance of the HSCL-25's threshold score of 1.75 enhances its utility in identifying significant mental health issues.

The HITS tool, with its focused four-item format, is an efficient screening measure for Intimate Partner Violence (IPV), providing a clear and actionable score to identify at-risk individuals. Its straightforward scoring system, where a score above 10 indicates IPV, is both practical and effective in clinical settings.

The PSS, with its 14-item scale, measures perceived stress, categorizing it into low, moderate, and high levels, offering valuable insights into participants' stress perceptions. Finally, the PSI-SF, particularly its subscales on Parental Distress and Parent-Child Dysfunctional Interaction, is crucial for assessing stress within the parent-child relationship. The use of well-documented, validated instruments ensures the reliability and validity of the data, supporting meaningful and accurate conclusions in the research study.

Detailed documentation of procedures, including data collection, entry, and analysis, was maintained, and any limitations or challenges encountered were transparently reported to provide context for the findings. These measures ensured the reliability and validity of the collected data, leading to more accurate and meaningful results.

Data Processing, Study Variables, and Analysis

Data processing for this cross-sectional study involved meticulous steps to ensure accuracy and reliability. Upon collection, all interview responses were entered into a robust data management system utilizing a double data entry method. Real-time validation checks were implemented to promptly identify and rectify any inconsistencies or errors. Subsequently, the dataset underwent thorough cleaning procedures to ensure completeness and accuracy before analysis. Throughout this process, stringent quality control measures were applied to maintain the integrity of the data.The study aimed to assess the psychosocial impact of genetic diseases on parents, with both dependent and independent variables playing a critical role. The dependent variables included self-esteem (measured by the Rosenberg Self-Esteem Scale),depressive and anxiety symptoms (measured by the HSCL-25), risk of intimate partner violence (measured by the HITS tool), perceived stress (measured by the Perceived Stress Scale), and parental distress and dysfunctional interaction (measured by the Parenting Stress Index-Short Form).

The primary independent variable was the type of disease in children, categorized as genetic or non-genetic.

To analyze these variables, descriptive statistics were first employed to summarize participant demographics and baseline measures, providing an overview of the sample's characteristics. This included calculating means, standard deviations, and frequencies for key demographic variables such as age, gender, and socioeconomic status. Following this, independent sample t-tests were conducted to compare the mean scores of the dependent variables between the two groups: parents of children with genetic diseases and parents of children with non-genetic diseases. These t-tests allowed for the determination of statistically significant differences in psychological outcomes between the groups, helping to identify the specific impacts of having a child with a genetic disease on parental well-being.

To confirm the reliability of these instruments in our study, we calculated Cronbach's alpha for each scale. The results demonstrated acceptable ranges, with the Rosenberg Self-Esteem Scale achieving a Cronbach's alpha of 0.80, the HSCL-25 yielding 0.78, the HITS tool showing 0.82, the Perceived Stress Scale scoring 0.85, and the Parenting Stress Index-Short Form producing 0.83. These values indicate a high level of internal consistency for each measure, affirming their reliability in assessing self-esteem, depressive and anxiety symptoms, risk of intimate partner violence, perceived stress, and parental stress.

Ethical consideration

The study adhered to ethical standards with approval obtained from the Institutional Review Board of the College of Medicine and Health Sciences at the University of Rwanda (No. 135/CMH IRB/2023). Prior permission was secured from the ethics committee of Rwanda Military Hospital before initiating data collection. Informed consent was obtained from all participants in accordance with ethical guidelines set forth

by the Institutional Review Board of the College of Medicine and Health Sciences at the University of Rwanda and the ethics committee of Rwanda Military Hospital. Prior to enrolment in the study, detailed information about the purpose, procedures, potential risks, and benefits was provided to each participant. Emphasis was placed on confidentiality measures to protect personal information. Participants were assured of their right to withdraw from the study at any time without penalty or impact on their ongoing medical care. Voluntary participation was underscored throughout the recruitment process, ensuring that individuals freely chose to participate based on their understanding of the study objectives and their ability to provide informed consent.

Results

Demographic characteristics of participants

Table 1. Demographic characteristics of participants

Demographics		n	%
Type of the disease	Genetic	100	48.1%
	Non-genetic	109	51.9%
Participant sex	Male	10	4.8%
	Female	199	95.2%
Education	Primary school	158	75.6%
	Ordinary level	2	1.0%
	Secondary school	33	15.8%
	University	16	7.7%
Socio-economic category	Category 1	3	1.4%
	Category 2	141	67.8%
	Category 3	60	28.8%
	Category 4	4	1.9%
Participant age	Below 30	16	4.9%
	30-40	102	44.6%
	41-51	79	33.3%
	Above 51	1	0.1%
Marital status	Legally married	195	96.1%
	Cohabitation	8	3.9%
	Divorced	0	0.0%

Table 1 indicates the demographic data of participants and their children. Of 209 participants, 100 (48.1%) were parents of children with genetic diseases while 51.9% were parents of children with non-genetic diseases. Almost participants (95.2%) were female. About socioeconomic status, 67.0 % and 28.8% of participants were respectively in the second and third socioeconomic categories. Academically, 75.6% of the participants had attended primary schools.

Clinical features among children of with non-genetic diseases

Figure 1 offers a comprehensive insight into the prevalence of different medical conditions among children with non-genetic diseases. It categorizes the diagnostic conditions into main groups, including respiratory conditions, skin conditions, gastrointestinal issues, and cardiovascular conditions. Within each group, several specific diseases or conditions are listed as examples.

The figure 1 emphasizes the percentages of these main condition groups. However, due to the multitude of conditions, only a few examples are presented under each group. Figure 1 indicates that respiratory conditions were the most common conditions, with 31 individuals (28.4%) having one or more of these conditions. Skin Conditions were the least common among the listed conditions, with only 7 individuals (6.4%) having these conditions. Neurological Conditions and Cardiovascular conditions both have 11 individuals each, making up 10.1% of the total for each category. Gastrointestinal Issues (abdominal pain, constipation, gastroenteritis, etc…) Are reported in 8 cases, accounting for 7.3% of the total. Other Conditions including anemia, meningitis and burn made up 37.3% of the total, with 41 individuals having these conditions.

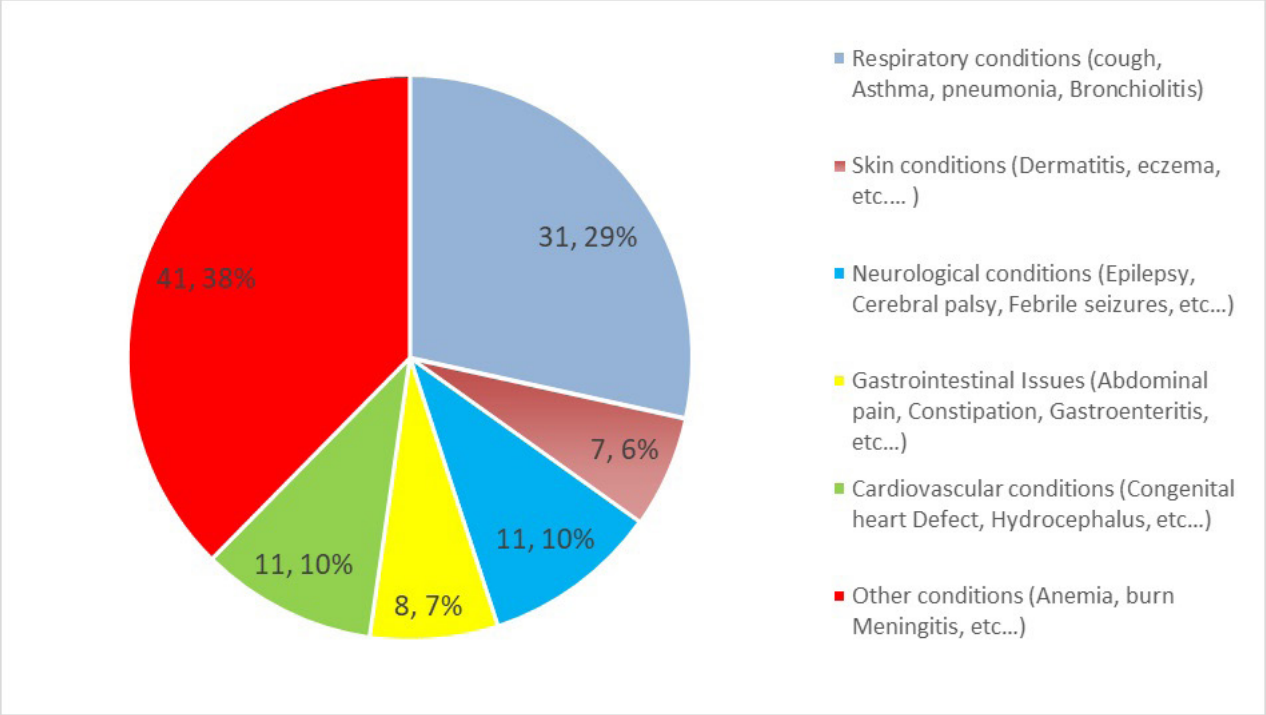

Figure 1. Clinical features among children with non-genetic diseases (n=109)

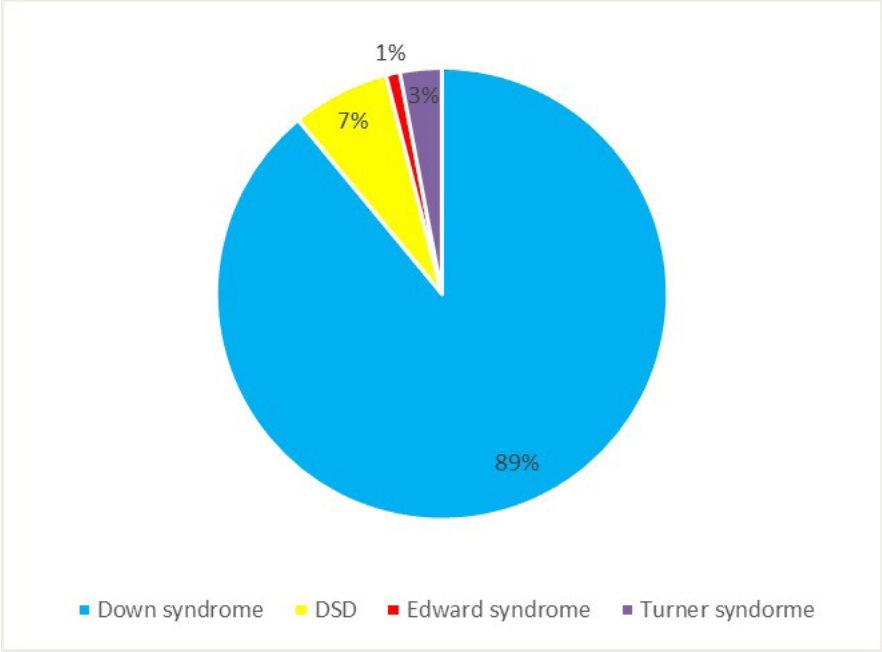

Figure 2. Clinical features among children of case group participants (n=100)

Figure 2 shows that nearly all the children in the were identified with Down syndrome (89%), while seven children were diagnosed with Disorders of Sex Differentiation (7%). A smaller number of children were found to have Edward syndrome (1%); and Turner syndrome (3%). These diagnoses were confirmed through karyotype test results. However, it is important to note that individuals with genetic disorders that could be identified through more advanced cytogenetic tests, rather than just karyotyping, were not included in this study.

Prevalence of mental disorders, low self-esteem and intimate partner violence between case and control groups

Table 2 presents a comparison of the prevalence rates of anxiety and depressive symptoms, perceived stress, low self-esteem, and intimate partner violence among participants in the case and control groups. This analysis considered the cutoff points for each scale. Clinical significance is indicated by an average score of 1.75 for both anxiety and depressive symptom scales. A score above 10 on the intimate partner violence scale indicates that an individual is experiencing abuse. For perceived stress, the score categories are as follows: Low Stress (scores 0 - 18), Moderate Stress (scores 19 - 37), and High Stress (scores 38 - 56). Regarding low self-esteem, cutoff scores less than 15 are considered severe, 15 to 20 is moderate, 21 to 25 indicates mild symptoms, and 26 to 30 is not clinically significant.

Table 2. Prevalence of mental disorders, low self-esteem and intimate partner violence among parents of children with genetic diseases and non-genetic diseases

Variable	Type of disease in children			
	Genetic		Non-genetic	
	n	%	n	%
Anxiety symptoms				
Symptoms not significant	45	45	89	81.7
Clinically significant symptoms	55	55	20	18.3
Depressive symptoms				
Symptoms not significant	55	55	80	73.4
Clinically significant symptoms	45	45	29	26.6
Self-esteem				
Low self esteem	89	89	17	16.8
Moderate esteem	7	7.2	10	9.9
Mild esteem	3	3.1	46	45.5
High esteem	1	1	28	27.7
Perceived stress				
Low stress	22	22.	87	79.8
Moderate stress	43	43.	22	20.2
High stress	35	35.	0	0.0
Intimate partner violence				
No abuse	29	29.0%	89	81.7%
Presence of abuse	71	71.0%	20	18.3%

Table 2 indicates scores of both parents of children with genetic diseases and parents of children with no-genetic diseases. The prevalence of anxiety symptoms, depressive symptoms, low esteem perceived stress and abuse were very high in parents of children with genetic diseases compared to parents of children with non-genetic diseases.

Prevalence of parenting stress among parents of children with genetic diseases and non-genetic diseases

Table 3 provides the prevalence of the Parenting Stress Index with respect to its three subscales: Parental Distress, Parent-Child Dysfunctional Interaction, and Difficult Child, each comprising 12 items. It outlines the number of participants experiencing significant parenting stress on each of these scales within both the case and control groups. To qualify as having significant stress, a score of 48 out of 60 on each subscale is necessary.

Table 3 indicates that parenting stress is more prevalent in case group compared to control group. 34.0% had parenting distress, 47.0%, had dysfunctional interaction with their children while 36.0% of them find their children as difficult to raise. Significant parenting stress was seen in less than 2% of control group participants in all the three scales.

Table 3. Prevalence of parenting stress index between case and control groups

Variable	Type of disease in children			
	Genetic		Non-genetic	
	n	%	n	%
Parenting distress				
No parenting stress	66	66	107	98.2
Significant parenting stress	34	34	2	1.8
Parent-child dysfunctional interaction				
No parenting stress	53	53	108	99.1
Significant parenting stress	47	47	1	0.9
Difficult child				
No parenting stress	64	64	107	98.2
Significant parenting stress	36	36	2	1.8

Independent sample t tests

Based on the results in the table 4 for the independent samples t-test, there is a statistically significant difference in the average test scores between case and control for all the variables. The mean score for anxiety symptoms is 18.82 for the case group 13.16 case and control group respectively (P= .0001). Results for depressive symptoms indicate that the mean score for case group is higher than the mean score in the control group 27.65 and 20.73 respectively with (P= .0001). Low self-esteem results indicate that the mean score for case group is higher than the mean score in the control group 27.65 and 20.73 respectively with (P= .0001) three scales.

Results of the three scales of parenting stress (parenting distress, difficult child, parent child dysfunctional interaction) indicate that the mean scores for case group are higher than the mean scores in control group respectively with the mean differences of 19.143, 19.555 and (P= .0001). The mean scores for the for the case group and control group on the intimate partner violence were respectively 11.91 and 6.98 with (P= .0001). Case group participants significantly scored higher than control group participants on the perceived stress with respective means; 31.41 and 16.91 and (P= .0001).

Table 4. Independent sample T tests

Variable	Type of disease	n	Mean (SD)	t	Mean difference	p	LCI	UCI
Anxiety symptoms	Genetic	100	18.82 (7.42)	6.107	5.664	.0001	3.855	7.473
	Non-genetic	109	13.16 (5.8)					
Depressive symptoms	Genetic	100	27.65 (10.607)	5.683	6.916	.0001	4.567	9.265
	Non-genetic	109	20.73 (6.227)					
Low self-esteem	Genetic	100	12.17 (4.557)	-14.565	9.9	.0001	-11.869	-9.039
	Non-genetic	109	22.62 (5.697)					
Parenting stress Index (PD))	Genetic	100	37.95 (12.280)	12.893	19.143	.0001		
	Non-genetic	109	18.81(3.985)				16.216	22.070
Parenting stress Index (DC)	Genetic	100	38.17(11.959)	12.893	19.555	.0001	16.861	22.249
Parenting stress Index (PCDI)	Non-genetic	100	43.76 (12.108)	22.400	28.650	.0001	26.128	31.172
	Genetic	109	15.11(5.398)					
Intimate partner violence	Genetic	100	11.91 (4.195)	10.617	4.928	.0001	4.013	5.843
	Non-genetic	109	6.98 (2.325)					
Perceived stress	Genetic	100	31.41 (12.280)	11.680	14.502	.0001	12.054	16.950
	Non-genetic	109	16.91 (3.985)	11.680				

Discussion

The objective of this study was to find if parents of children with genetic diseases suffer from psychological problems, and to compare the magnitude of those problems in comparison to parents of children with non-genetic diseases. As hypothesized, the prevalence of depressive symptoms, anxiety symptoms, low self-esteem, perceived stress, parenting stress and intimate partner violence were more prevalent among parents of children with genetic diseases (case group) than parents of children with non- genetic diseases.

The mean scores for all the variables were found to be significantly higher among parents of children with genetic diseases than parents of non- genetic diseases. As was revealed in other studies, families experience a range of emotions and stressors when raising children genetic diseases; mostly children with Down syndrome. psychological and emotional well-being of both parents and siblings in these families often raise due to challenges those diseases bring in a family leading to feelings of stress, anxiety, and depression.[13]

Similarly to this study, some researches revealed that a diagnosis of Down syndrome the child can be emotionally overwhelming. High stress levels were found in families with children with Down syndrome and other developmental disorders, shedding light on the emotional experiences of parents including depression.[10] Other researchers found that genetic disease are often associated with maladaptive behavior. This often lead to the emotional challenges among parents of, which can contribute in turn contributes to depression.[15] Parents may feel isolated from their social networks because of the demands of caregiving or because they perceive that others do not understand their unique challenges. Parents may worry about their child's long-term prospects, including educational opportunities, independence, and overall quality of life. These concerns can contribute to anxiety and depression. Insufficient access to support services, including respite care, therapy, and special education programs, can place additional stress on parents. Parents and their children with Down syndrome may encounter stigma and discrimination, which can be emotionally distressing and contribute to feelings of depression.

Anxiety is a common emotional response among parents of children with Down syndrome due to the unique challenges and uncertainties they may face. Here are some factors that can contribute to anxiety among these parents.[3] The initial diagnosis of Down syndrome can be emotionally overwhelming. Parents may experience anxiety as they grapple with uncertainties about their child's future, health, and development. Children with Down syndrome may have specific health issues, such as heart defects, respiratory problems, and a higher risk of certain medical conditions. These health concerns can lead to ongoing anxiety about the well-being of their child. Parents may worry about their child's educational opportunities and developmental progress.[16]

Findings of this research indicated more prevalent anxiety among parents of children with genetic diseases. In line with the findings of this study, different researchers have revealed the possible source of anxiety among those parents. Parents are concerned about their child's ability to reach milestones and achieve independence, in addition to costs associated with medical care, therapy, and special education services that place financial stress on families, providing the best possible care for their child can contribute to anxiety. Parents may worry about their child's long-term prospects, including independence and quality of life Planning for the future can be a source of anxiety. Parents may compare their child's progress to that of typically developing children or other children with Down syndrome, leading to anxiety if they perceive their child as falling behind. [17]

Low self-esteem as also been documented in other studies. Though those studies were not comparative in methodology, findings revealed that caring for a child with a genetic disease can have a significant impact on parents' self-esteem. Feelings of guilt, inadequacy, or self-blame may arise as parents grapple with the genetic nature of the condition and their role in their child's health contributing to low self-esteem. [18] Similar to our findings, some studies revealed that parents of children genetic diseases reported self-esteem. However, the researchers deeply investigated how stigma, and social support were associated with depressive symptomology. association between stigma and depressive symptomology and low self-esteem.[19]

Limitations of the study

This study utilized a cross-sectional approach with a quantitative methodology. A more comprehensive approach combining both quantitative and qualitative methods in longitudinal research would have enhanced the study's robustness. Additionally, although there was a reference framework connecting children to the genetic department for the case group and the pediatric department for the control group,

karyotype examinations were not conducted for all patients. The classification of participants in the control group was not determined through genetic tests. Consequently, there is uncertainty regarding whether patients in the control group had genetic diseases, as they were not subjected to testing.

Recommendations for future researches

For future research, it is recommended to use a longitudinal approach combining both quantitative and qualitative methodologies to better understand the psychological impact on parents of children with genetic versus non-genetic conditions. Systematic genetic testing for all participants is essential to ensure accurate classification of control and case groups. Implementing karyotype examinations will help clarify whether control group participants have genetic conditions, thereby enhancing the reliability of the findings. Additionally, incorporating in-depth qualitative interviews can provide valuable insights into the specific experiences and challenges faced by these parents, complementing the quantitative data.

Conclusion

The study revealed that parents of children with genetic diseases experience significantly higher levels of depression, anxiety, perceived stress, parenting stress, and intimate partner violence compared to parents of children with non-genetic conditions, with a greater prevalence of low self-esteem also observed. These findings highlight the urgent need for comprehensive support systems tailored to these parents' unique challenges. Healthcare providers should consider implementing targeted mental health interventions, stress management programs, and family support services. Genetic counseling is a recommended intervention, as it offers essential information, emotional support, and coping strategies. Additionally, creating resources to enhance self-esteem and address intimate partner violence can further improve the well-being of these families. By addressing these specific needs, healthcare providers can significantly enhance the quality of life for parents and their children with genetic diseases.

Add author's contribution statement

BT: Data collection, Data analysis, manuscript writing
NJ: Data analysis
JMN: Manuscript revision, correction and referencing
LM: Genetics part literature revision and overall supervisor
JM: Data analysis and manuscript revision
CK: Study design

Conflict of interest

Authors have no conflict of interest to reveal

References

1. Dator J. The Impact of Genetic Disease on the Family: Examining the Relationship Between Psychological Well-Being, Social Support, and Spirituality in Unaffected Carriers of Leber's Hereditary Optic Neuropathy. *Set Hall Univ Diss Theses* . 2014. https://scholarship.shu.edu/dissertations/2022
2. Cunningham F, Achuthan P, Akanni W, Allen J, Amode MR, Armean IM, et al. Ensembl 2019. *Nucleic Acids Research.* 2019;47:D745–51. https://doi.org/10.1093/nar/gky1113
3. Canavarro MC, Fonseca A. Parenting an infant with a congenital anomaly : An exploratory study on patterns of adjustment from diagnosis to six months post birth. *Journal of Child Health Care.* 2014. https://doi.org/10.1177/1367493512473856

4. Timpson NJ, Greenwood CMT, Soranzo N, Lawson DJ, Richards JB. Genetic architecture: The shape of the genetic contribution to human traits and disease. *Nature Reviews Genetics.* 2018;19:110–24. doi:10.1038/nrg.2017.101
5. Davis OSP, Butcher LM, Docherty SJ, Meaburn EL, Curtis CJC, Simpson MA, et al. A three-stage genome-wide association study of general cognitive ability: Hunting the small effects. *Behavior Genetics.* 2010;40:759–67. https://doi.org/10.1007/s10519-010-9350-4
6. D'Souza H, Lathan A, Karmiloff-Smith A, Mareschal D. Down syndrome and parental depression: A double hit on early expressive language development. *Research in Developmental Disabilities.* 2020;100:103613. https://doi.org/10.1016/j.ridd.2020.103613
7. Marcus M, Yasamy MT, van Ommeren M, Chisholm D. Depression, a global public health concern. *WHO Department of Mental Health and Substance Abuse.* 2012;1–8. http://www.who.int/mental_health/management/depression/who_paper_depression_wfmh_2012.pdf
8. Phillips BA, Conners F, Curtner-Smith ME. Parenting children with down syndrome: An analysis of parenting styles, parenting dimensions, and parental stress. *Research in Developmental Disabilities.* 2017;68:9–19. https://doi.org/10.1016/j.ridd.2017.06.010
9. Scherer N, Verhey I, Kuper H. Depression and anxiety in parents of children with intellectual and developmental disabilities: A systematic review and meta-analysis. *PLoS ONE.* 2019;14. https://doi.org/10.1371/journal.pone.0219888
10. Fidler DJ, Hodapp RM, Dykens EM. Stress in Families of Young Children with Down Syndrome, Williams Syndrome, and Smith-Magenis Syndrome. *Early Education and Development* .2000. https://doi.org/10.1207/s15566935eed1104_2
11. Talley L, Brintnell ES. Scoping the barriers to implementing policies for inclusive education in Rwanda: an occupational therapy opportunity. *International Journal of Inclusive Education.* 2016;20:364–82. http://dx.doi.org/10.1080/13603116.2015.1081634
12. Ministry of Health Rwanda. Violence Against Children and Youth with Disabilities in Institutional Settings in Rwanda. *Rwanda Minist Heal.* 2018.
13. Woodgate RL, Edwards M, Ripat JD, Borton B, Rempel G. Intense parenting: A qualitative study detailing the experiences of parenting children with complex care needs. *BMC Pediatrics.* 2015;15. https://doi.org/10.1186/s12887-015-0514-5
14. Ntizimira CR, Ngizwenayo S, Krakauer EL, Dunne ML, Esmaili E. Addressing End-of-Life Care in Cancer Patients through "Ubuntu": Lessons Learned from Rwanda in Global Health Perspective of Humanity. *Curr Obstet Gynecol Rep.* 2016;5:273–8. DOI 10.1007/s13669-016-0186-7
15. Evans DW, Gray FL. Compulsive-like behavior in individuals with down syndrome: Its relation to mental age level, adaptive and maladaptive behavior. *Child Development.* 2000;71:288–300. https://doi.org/10.1111/1467-8624.00144
16. Skotko BG, Levine SP, Goldstein R. Having a son or daughter with Down syndrome: Perspectives from mothers and fathers. *American Journal of Medical Genetics, Part A.* 2011;155:2335–47. https://doi.org/10.1002/ajmg.a.34293
17. Alrahili N. Burnout and anxiety among parents of children with neurodevelopmental disorders: a cross-sectional study in Saudi Arabia. *Middle East Current Psychiatry.* 2023;30. https://doi.org/10.1186/s43045-023-00331-3
18. Durmaz A, Cankaya T, Durmaz B, Vahabi A, Gunduz C, Cogulu O, et al. Interview with parents of children with Down syndrome: Their perceptions and feelings. *Indian Journal of Pediatrics.* 2011;78:698–702. https://doi.org/10.1007/s12098-010-0315-7
19. Cantwell J, Muldoon O, Gallagher S. The influence of self-esteem and social support on the relationship between stigma and depressive symptomology in parents caring for children with intellectual disabilities. *Journal of Intellectual Disability Research.* 2015;59:948–57. https://doi.org/10.1111/jir.12205