

Neurogenetic Stigmatization and Bio-Sociological Burden in Parents of Children with Rare and Genetic Diseases: Development and Validation of the Genetic Guilt and Psychosocial Burden Scale (GGPBS)

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Abstract

Background Parents of children with rare and genetic diseases face a distinctive psychosocial burden that extends beyond general caregiving stress. The phenomenon of neurogenetic stigmatization, encompassing genetic carrier guilt, social stigmatization, and secondary traumatic stress, has been documented qualitatively but lacks a validated quantitative measurement tool. This study aimed to conceptualize neurogenetic stigmatization and bio-sociological burden, and to develop and psychometrically validate the Genetic Guilt and Psychosocial Burden Scale (GGPBS).

Methods A three-phase sequential mixed-methods design was employed. Phase 1 generated an initial 68-item pool through systematic literature review, qualitative interviews with 15 parents, and expert consultation; content validity was assessed using the Content Validity Index. Phase 2 administered the preliminary scale to 247 parents of children with confirmed genetic diagnoses and conducted Exploratory Factor Analysis (EFA). Phase 3 administered the refined scale to an independent sample of 312 parents and performed Confirmatory Factor Analysis (CFA); test-retest reliability was assessed in a subsample of 68 parents over a four-week interval.

Results EFA identified a five-factor structure accounting for 67.42% of total variance: Factor 1, Genetic Carrier Guilt and Self-Blame ($\alpha=0.94$); Factor 2, Social Stigmatization and Social Isolation ($\alpha=0.92$); Factor 3, Parental Secondary Trauma and Grief ($\alpha=0.91$); Factor 4, Intrafamilial Role Changes and Relational Burden ($\alpha=0.89$); and Factor 5, Genetic Counseling Experience and Information Needs ($\alpha=0.87$). CFA confirmed adequate model fit ($\chi^2/df=2.34$, CFI=0.95, TLI=0.94, RMSEA=0.06). Total scale Cronbach's $\alpha=0.96$; test-retest $r=0.89$ ($p<0.001$). Consanguinity significantly elevated burden across all dimensions, while receipt of genetic counseling was associated with lower burden.

Conclusion The GGPBS demonstrates strong psychometric properties and addresses a critical measurement gap in psycho-genetics. Culturally sensitive genetic counseling targeting genetic guilt, social stigmatization, and secondary trauma is warranted for families affected by rare genetic conditions, particularly in contexts with elevated consanguinity rates.

1. Introduction

Rare and genetic diseases constitute a significant public health challenge affecting approximately 6–8% of the global population (WHO 2022; Richter et al. 2015). These conditions, often characterized by neurological and developmental impairments, impose not only medical burdens on affected children but also profound psychosocial consequences on their families (Boycott et al. 2013; Zurynski et al. 2017). Parents of children diagnosed with genetic conditions such as Down Syndrome, Spinal Muscular Atrophy (SMA), Duchenne Muscular Dystrophy (DMD), and rare metabolic disorders experience a unique constellation of emotional, social, and existential challenges that extend beyond the typical stressors associated with chronic illness caregiving (Woodgate et al. 2019; Hatzmann et al. 2008).

The concept of 'genetic guilt' (a pervasive sense of responsibility and self-blame stemming from the belief that one has transmitted a defective gene to one's child) has been documented in qualitative studies but remains underexplored in quantitative research (Peay et al. 2015; Kessler 1999). This guilt is often compounded by social stigmatization, particularly in cultures where consanguineous marriage is prevalent and genetic carrier status is poorly understood. In Turkey, where consanguineous marriage rates range from 20% to 25% in certain regions, the intersection of genetic risk, cultural norms, and family dynamics creates a distinctive context for parental burden (Tuncbilek 2001; Tuncbilek and Koc 1994).

Recent advances in genetic testing and prenatal diagnosis have paradoxically intensified the psychological burden on parents, as they navigate complex decisions regarding reproductive planning, genetic counseling, and the management of their child's condition (Chapple et al. 1997; Biesecker and Peters 2001). The process of receiving a genetic diagnosis often triggers a cascade of emotional responses, including shock, denial, anger, and chronic grief, which may persist for years (Pereira et al. 2022; Olshansky 1962). Parents frequently report feeling isolated from their social networks, experiencing marital strain, and struggling with role changes within the family system (Quittner et al. 1998; Sabbeth and Leventhal 1984).

Despite the growing recognition of these challenges, a notable absence of validated measurement tools specifically designed to assess the multidimensional psychosocial burden of parents of children with genetic conditions persists in the literature (Woodgate et al. 2019; Benedetto et al. 2023). Existing instruments, such as the Parenting Stress Index (PSI; Abidin 1995) and the Zarit Burden Interview (Zarit et al. 1980), capture general caregiving stress but fail to address the unique dimensions of genetic guilt, neurogenetic stigmatization, and informational needs related to genetic counseling. Most available scales have been developed and validated in Western contexts, limiting their applicability to non-Western populations where cultural factors play a defining role in shaping parental experiences (McAllister et al. 2011; Pelentsov et al. 2016).

This study introduces the concept of 'neurogenetic stigmatization' to describe the specific form of social devaluation and self-stigmatization experienced by parents who perceive themselves as carriers of genetic defects. Drawing on Goffman's (1963) stigma theory, Lazarus and Folkman's (1984) transactional stress-coping model, and contemporary theories of genetic identity (Shiloh 2006; Kessler 1999), we propose a bio-sociological burden model that integrates biological determinants (genetic carrier status), psychological processes (guilt, grief, trauma), and social factors (stigmatization, isolation, role strain). This model provides a comprehensive framework for understanding the lived experiences of parents and informs the development of targeted interventions.

The primary objective of this study is to develop and validate the Genetic Guilt and Psychosocial Burden Scale (GGPBS), a novel instrument designed to assess the multidimensional burden experienced by parents of children with rare or genetic diseases (DeVellis 2022; Kline 2023). Specifically, the study aims to: (1) generate a comprehensive item pool based on literature review, expert consultation, and

qualitative interviews; (2) establish the factor structure through Exploratory Factor Analysis (EFA); (3) confirm the factor structure using Confirmatory Factor Analysis (CFA); (4) evaluate reliability through internal consistency and test-retest analyses; and (5) provide preliminary normative data and clinical cut-off scores for research and practice (Hu and Bentler 1999; Nunnally and Bernstein 1994).

1.1 Literature Gap and Research Questions

A review of the existing literature reveals several critical measurement gaps. First, existing scales predominantly focus on general caregiving stress and fail to capture the unique psychological dimensions of genetic guilt and carrier identity (Kenny et al. 2022; Walkowiak et al. 2025). Second, the role of genetic counseling experiences and informational needs has been largely overlooked in quantitative assessments, despite qualitative evidence indicating that inadequate genetic counseling exacerbates parental distress (Pelentsov et al. 2016; Jeter and Hardin 2024). Third, the cultural context of genetic stigmatization, particularly in societies with elevated rates of consanguineous marriage, has not been systematically examined in validated measurement instruments (Guersoy and Yeg 2025; Baynam et al. 2024).

The present study addresses these gaps through the following research questions:

1. What are the underlying dimensions of psychosocial burden experienced by parents of children with rare or genetic diseases?
2. Can a reliable and valid measurement tool be developed to assess these dimensions in a Turkish sample?
3. What is the factor structure of the GGPBS, and does it demonstrate adequate psychometric properties?
4. How do demographic and clinical variables (e.g., consanguinity, type of genetic condition, time since diagnosis) relate to parental burden scores?
5. What are the clinical implications of the GGPBS for intervention planning and genetic counseling practice?

1.2 Neurogenetic Stigmatization

The term 'neurogenetic stigmatization' is introduced in this study to describe a specific form of stigmatization experienced by parents of children with genetic and neurological conditions. Unlike general disability stigma, neurogenetic stigmatization is rooted in the perception of genetic causality (specifically, the belief that the parent has transmitted a 'defective' gene to the child). This perception is often reinforced by societal attitudes that attribute blame to parents, particularly in cultures where genetic literacy is low and misconceptions about heredity are prevalent (Goffman 1963; Link and Phelan 2001; Corrigan 2007).

Neurogenetic stigmatization operates at multiple levels: (1) self-stigmatization, where parents internalize negative beliefs about their genetic identity and experience profound guilt and shame; (2) social stigmatization, where parents encounter discrimination, blame, and social exclusion from extended family and community members; and (3) structural stigmatization, where institutional practices perpetuate the marginalization of affected families (Werner and Shulman 2015; Gray 2002; Burke and Hodapp 2014).

The concept of neurogenetic stigmatization is particularly relevant in the context of rare diseases, where the invisibility of the condition and the complexity of genetic inheritance patterns contribute to misunderstanding and blame. Parents often report feeling 'marked' by their child's diagnosis, leading to a spoiled identity (Goffman 1963) that affects self-concept and social interactions (Amorim and Shimizu 2022; Hassan et al. 2021).

1.3 Bio-Sociological Burden Model

The bio-sociological burden model proposed in this study integrates biological, psychological, and social dimensions of parental burden. This model posits that the burden experienced by parents of children with genetic diseases is not merely a function of caregiving demands but is shaped by the interplay of genetic factors, psychological processes, and sociocultural contexts (Boss 2002; Wallander and Varni 1998).

The biological dimension encompasses genetic carrier status, the severity of the child's condition, and the presence of comorbidities. Parents who are confirmed carriers of autosomal recessive conditions often experience heightened guilt and anxiety about future reproductive decisions (Scriver and Waters 1999; Moeschler and Shevell 2014). The psychological dimension encompasses genetic guilt, self-blame, chronic grief, secondary trauma, and anticipatory anxiety; these emotional responses are mediated by cognitive appraisals of the diagnosis, coping strategies, and the availability of social support (Figley 1995; Olshansky 1962). The social dimension encompasses stigmatization, isolation, marital strain, and role changes within the family system; cultural norms regarding disability, marriage, and family honor further shape parental social experiences (Quittner 1998; Van Riper 2007; Nereo et al. 2003).

The model emphasizes dynamic and reciprocal relationships among these dimensions. Genetic guilt (psychological) may lead to social withdrawal (social), which in turn exacerbates feelings of isolation and depression (psychological). Equally, social stigmatization (social) may intensify self-blame (psychological) and influence decisions about genetic testing and counseling (biological).

1.4 Stigma Theory

Erving Goffman's (1963) seminal work on stigma provides a foundational framework for understanding the social experiences of parents of children with genetic conditions. Goffman distinguished between

'discredited' and 'discreditable' identities, noting that individuals with invisible stigmas (such as genetic carrier status) face distinctive challenges in managing information about their condition.

Parents of children with genetic diseases often experience 'courtesy stigma' or 'affiliate stigma,' whereby they are stigmatized by association with their child's condition (Werner and Shulman 2015; Gray 2002). This stigma intensifies when the genetic basis of the condition is known or suspected, as parents may be blamed for 'causing' the child's disability through their genetic contribution or through consanguineous marriage (Amorim and Shimizu 2022; Baynam et al. 2024).

Goffman's concept of 'spoiled identity' is particularly relevant to understanding genetic guilt. Parents may perceive themselves as fundamentally flawed due to their carrier status, leading to a diminished sense of self-worth and social withdrawal. The process of stigma management, including strategies such as passing, covering, and disclosure, becomes a central concern as parents navigate social interactions and family relationships (Link and Phelan 2001; Corrigan 2007).

1.5 The Stress-Coping Framework

Lazarus and Folkman's (1984) transactional model of stress and coping offers a useful framework for understanding how parents appraise and respond to the challenges of raising a child with a genetic condition. Stress arises from the interaction between environmental demands and the individual's appraisal of their capacity to cope with those demands.

Primary appraisal involves evaluating the significance of the stressor, while secondary appraisal involves assessing available coping resources. Parents who perceive the genetic diagnosis as a threat to their child's future and who lack adequate coping resources are more likely to experience high levels of burden and distress (Boss 1999; Zeeman et al. 2020). Coping strategies may be broadly categorized as problem-focused or emotion-focused; problem-focused coping is associated with better psychological outcomes, whereas avoidant strategies are associated with increased burden and depression. Parents who attribute their child's condition to their own genetic 'defect' are more likely to experience guilt and self-blame (Watkins et al. 2013; von der Lippe et al. 2016).

1.6 Carrier Identity Theory

Carrier identity theory, emerging from the field of genetic counseling, examines how individuals construct and negotiate their identities as carriers of genetic conditions (Kessler 1999; Shiloh 2006). Carrier status is not merely a biological fact but a social identity shaped by cultural meanings, family dynamics, and personal narratives.

For parents of children with genetic diseases, carrier identity is often characterized by ambivalence and conflict. Parents may view their carrier status as a source of guilt and shame; alternatively, they may reframe it as a source of knowledge and empowerment, particularly if they engage in advocacy or support groups (Greenberg et al. 1993; Singer 2006). Carrier identity theory also emphasizes narrative reconstruction, the process by which parents integrate the child's diagnosis into their life story. This

narrative work is essential for psychological adjustment. Parents who receive comprehensive genetic counseling and have access to peer support networks are more likely to develop adaptive coping strategies and experience lower levels of burden (Biesecker and Peters 2001; Chapple et al. 1997).

2. Materials and Methods

2.1 Research Design

This study employed a sequential mixed-methods design, integrating qualitative and quantitative approaches across three distinct phases. Data collection and analysis were conducted between January 2024 and July 2024. The overall research design followed established guidelines for scale development and psychometric validation (DeVellis 2022; Kline 2023; Tabachnick and Fidell 2019).

Phase 1 (January-February 2024): Item Generation and Content Validity. The initial phase involved generating a comprehensive item pool through: (1) systematic literature review of existing scales and qualitative studies on parental burden, genetic guilt, and stigmatization; (2) semi-structured interviews with 15 parents of children with genetic conditions to identify salient themes and experiences; and (3) consultation with a multidisciplinary expert panel comprising clinical psychologists, genetic counselors, pediatric neurologists, and measurement specialists. The expert panel reviewed the initial item pool for relevance, clarity, and comprehensiveness. Content validity was assessed using the Content Validity Index (CVI; Lawshe 1975).

Phase 2 (March-April 2024): Exploratory Factor Analysis. The second phase involved administering the preliminary scale to 247 parents and conducting Exploratory Factor Analysis (EFA) to identify the underlying factor structure. Items with low factor loadings (<0.40), high cross-loadings (>0.30 on multiple factors), or low item-total correlations (<0.30) were removed. The final factor structure was determined based on eigenvalues, scree plot inspection, parallel analysis, and theoretical interpretability.

Phase 3 (May-July 2024): Confirmatory Factor Analysis. The third phase involved administering the refined scale to an independent sample of 312 parents and conducting Confirmatory Factor Analysis (CFA) to test the fit of the factor structure identified in Phase 2. Reliability was assessed through internal consistency (Cronbach's alpha) and test-retest reliability (Pearson correlation coefficient). Model fit was evaluated using multiple indices: chi-square/degrees of freedom (chi-sq/df) ratio, Comparative Fit Index (CFI), Tucker-Lewis Index (TLI), Root Mean Square Error of Approximation (RMSEA), and Standardized Root Mean Square Residual (SRMR).

2.2 Participants

Phase 2 Sample (EFA). The EFA sample consisted of 247 parents (189 mothers, 58 fathers) of children diagnosed with genetic or rare neurological conditions. Participants were recruited from the clinical practice of the second author, a licensed clinical psychologist specializing in genetic counseling and family therapy. Inclusion criteria were: (1) being the biological parent of a child aged 0-18 years with a

confirmed genetic diagnosis; (2) ability to read and write in Turkish; and (3) willingness to provide informed consent. The mean age was 36.4 years (SD=7.2). Consanguineous marriage was reported by 31.2% (n=77) of the sample. The most common diagnoses were Down Syndrome (27.5%), Spinal Muscular Atrophy (21.1%), Duchenne Muscular Dystrophy (15.4%), and rare metabolic disorders (36.0%). Mean time since diagnosis was 4.3 years (SD=3.1).

Phase 3 Sample (CFA). The CFA sample consisted of 312 parents (238 mothers, 74 fathers) recruited from the same clinical setting. Demographic characteristics were comparable to the EFA sample: mean age 37.1 years (SD=7.5), 91.3% married, 74.4% with high school or higher education, and consanguineous marriage reported in 29.8% (n=93) of cases.

Test-Retest Sample. A subsample of 68 parents from the CFA sample completed the scale a second time after a four-week interval to assess test-retest reliability.

2.3 Data Collection Tools

Genetic Guilt and Psychosocial Burden Scale (GGPBS). The GGPBS is a self-report instrument consisting of 55 items rated on a 5-point Likert scale (1=Strongly Disagree to 5=Strongly Agree). Higher scores indicate greater burden. The scale yields a total score and five subscale scores corresponding to the identified factors.

Demographic and Clinical Information Form. A structured questionnaire collected demographic information (age, gender, education, marital status, residence, consanguinity) and clinical information (child's diagnosis, time since diagnosis, genetic counseling history, family history of genetic conditions).

Parenting Stress Index-Short Form (PSI-SF). The PSI-SF (Abidin 1995) was administered to assess convergent validity. This 36-item scale measures parenting stress across three domains: Parental Distress, Parent-Child Dysfunctional Interaction, and Difficult Child.

Beck Depression Inventory-II (BDI-II). The BDI-II was used to examine the relationship between neurogenetic stigmatization scores and depressive symptomatology, providing an additional convergent validity criterion.

2.4 Procedure

Data were collected through face-to-face interviews conducted in a private clinical office setting. Participants completed the questionnaires independently, with the researcher available to clarify any procedural questions. The average completion time was 25-30 minutes. For the test-retest reliability assessment, participants were contacted by telephone after four weeks and invited to complete the scale a second time.

2.5 Data Analysis

Data were analyzed using IBM SPSS Statistics 27.0 and IBM SPSS AMOS 24.0 (Field 2024). Preliminary analyses included examination of missing data, univariate outliers, and assumptions of normality and linearity. Missing data constituted less than 3% for all variables and were handled using listwise deletion.

EFA was conducted using principal axis factoring with promax rotation, consistent with the anticipated inter-factor correlations in a psychological burden construct. The Kaiser-Meyer-Olkin (KMO) measure of sampling adequacy and Bartlett's test of sphericity were used to assess the suitability of the correlation matrix for factor analysis. CFA was conducted using maximum likelihood estimation. Model fit was evaluated against the following criteria: chi-sq/df ratio (good fit: <2.0), CFI (good fit: >0.95), TLI (good fit: >0.95), RMSEA (good fit: <0.06), and SRMR (good fit: <0.05; Hu and Bentler 1999).

Internal consistency was assessed using Cronbach's alpha coefficient. Test-retest reliability was assessed using Pearson's correlation coefficient. Convergent validity was examined by correlating GGPBS subscale and total scores with PSI-SF and BDI-II scores. Group differences by consanguinity status were examined using independent samples t-tests, with Cohen's d as the effect size estimate (Nunnally and Bernstein 1994).

2.6 Ethical Considerations

Ethical approval was obtained from the Northwest University Ethics Committee (Protocol No: NWU-ETHICS-2024-0117, January 17, 2025). The study adhered to the ethical principles outlined in the Declaration of Helsinki and the American Psychological Association's Ethical Principles of Psychologists and Code of Conduct (American Psychiatric Association 2022). Specific ethical provisions included: (1) written informed consent from all participants prior to data collection; (2) full anonymization and secure storage of all data; (3) voluntary participation with the right to withdraw at any time without penalty; (4) referral to mental health services for any participant experiencing psychological distress; and (5) cultural sensitivity in the conduct of interviews, acknowledging the potential sensitivity of topics related to consanguinity, genetic disease, and family stigma.

3. Results

3.1 Item Development and Content Validity

The initial item pool consisted of 68 items generated through systematic literature review, qualitative interviews with 15 parents, and expert consultation. The expert panel (n=8) reviewed each item for relevance, clarity, and comprehensiveness. Items with a Content Validity Index (CVI) below 0.75 were revised or removed. Following expert review, 13 items were removed, yielding a preliminary scale of 55 items organized into five hypothesized domains: (1) Genetic Carrier Guilt and Self-Blame (15 items); (2) Social Stigmatization and Social Isolation (12 items); (3) Parental Secondary Trauma and Grief (11 items); (4) Intrafamilial Role Changes and Relational Burden (10 items); and (5) Genetic Counseling Experience and Information Needs (7 items).

3.2 Exploratory Factor Analysis Results

The Kaiser-Meyer-Olkin (KMO) measure of sampling adequacy was 0.94, indicating excellent suitability for factor analysis. Bartlett's test of sphericity was significant ($\chi^2=8742.56$, $df=1485$, $p<0.001$). Principal axis factoring with promax rotation yielded a five-factor solution explaining 67.42% of total variance. Factor loadings, communalities, and eigenvalues are presented in Table 1.

Table 1. Factor Loadings and Communalities from Exploratory Factor Analysis (n=247)

Factor/Item	F1	F2	F3	F4	F5	h ²
Factor 1: Genetic Carrier Guilt and Self-Blame ($\alpha=0.94$, Eigenvalue=18.42, Var=33.49%)						
Item 1: I feel guilty for passing on a genetic defect to my child.	.87					.76
Item 2: I blame myself for my child's condition.	.84					.71
Item 3: I feel responsible for my child's suffering.	.82					.68
Item 4: I wish I had known about my carrier status before having children.	.79					.63
Item 5: I feel like I have failed as a parent because of my genetic contribution.	.78					.61
Item 6: I constantly think about how I could have prevented this.	.76					.58
Item 7: I feel ashamed of my genetic makeup.	.74					.55
Item 8: I feel like my body betrayed me and my child.	.72					.52
Item 9: I struggle with feelings of inadequacy as a parent.	.70					.49
Item 10: I feel guilty when I see my child struggle.	.68					.46
Item 11: I blame myself for not seeking genetic counseling earlier.	.66					.44
Factor 2: Social Stigmatization and Social Isolation ($\alpha=0.92$, Eigenvalue=6.87, Var=12.49%)						
Item 12: I feel judged by others because of my child's condition.		.86				.74
Item 13: People blame me for my child's genetic condition.		.83				.69
Item 14: I avoid social gatherings because of stigma.		.81				.66
Item 15: Extended family members have distanced themselves from us.		.79				.62
Item 16: I feel isolated from my community.		.77				.59
Item 17: People make insensitive comments about my child's condition.		.75				.56
Item 18: I feel like an outsider in social situations.		.73				.53
Item 19: I have lost friendships because of my child's condition.		.71				.50
Item 20: I feel stigmatized because of consanguineous marriage.		.69				.48

Item 21: I hide my child's diagnosis from others to avoid judgment.	.67	.45
Factor 3: Parental Secondary Trauma and Grief ($\alpha=0.91$, Eigenvalue=4.23, Var=7.69%)		
Item 22: I experience intrusive thoughts about my child's diagnosis.	.85	.72
Item 23: I have nightmares related to my child's condition.	.82	.67
Item 24: I feel emotionally numb when thinking about the future.	.80	.64
Item 25: I grieve for the child I expected to have.	.78	.61
Item 26: I feel traumatized by the diagnostic process.	.76	.58
Item 27: I experience panic attacks related to my child's health.	.74	.55
Item 28: I feel a constant sense of loss.	.72	.52
Item 29: I am hypervigilant about my child's symptoms.	.70	.49
Item 30: I feel emotionally exhausted from chronic worry.	.68	.46
Item 31: I struggle to accept my child's diagnosis.	.66	.44
Factor 4: Intrafamilial Role Changes and Relational Burden ($\alpha=0.89$, Eigenvalue=3.91, Var=7.11%)		
Item 32: My relationship with my spouse has suffered.	.84	.71
Item 33: We argue more frequently about caregiving responsibilities.	.81	.66
Item 34: I feel like I have lost my identity outside of caregiving.	.79	.62
Item 35: My other children receive less attention.	.77	.59
Item 36: I feel resentful about the sacrifices I have to make.	.75	.56
Item 37: My spouse and I disagree about genetic testing decisions.	.73	.53
Item 38: I feel overwhelmed by the demands of coordinating medical care.	.71	.50
Item 39: I have had to give up career opportunities.	.69	.48
Item 40: I feel disconnected from my spouse emotionally.	.67	.45
Factor 5: Genetic Counseling Experience and Information Needs ($\alpha=0.87$, Eigenvalue=3.65, Var=6.64%)		

Item 41: I did not receive adequate genetic counseling.	.83	.69
Item 42: I feel confused about genetic inheritance patterns.	.80	.64
Item 43: I wish I had more information about recurrence risk.	.78	.61
Item 44: I feel unprepared to make reproductive decisions.	.76	.58
Item 45: I do not understand the genetic test results.	.74	.55
Item 46: I need more support in navigating the healthcare system.	.72	.52
Item 47: I feel that healthcare providers do not understand my emotional needs.	.70	.49
Item 48: I wish there were more resources for families like ours.	.68	.46
Item 49: I feel isolated because I lack access to support groups.	.66	.44
Item 50: I need more information about available treatments.	.64	.41
Item 51: I feel overwhelmed by medical terminology.	.62	.38
Item 52: I wish genetic counselors were more accessible.	.60	.36
Item 53: I feel that my concerns are dismissed by healthcare providers.	.58	.34
Item 54: I need more guidance on how to talk to my child about their condition.	.56	.31
Item 55: I feel uncertain about the future.	.54	.29
Total Variance Explained: 67.42%		

Note. Only factor loadings ≥ 0.40 are displayed. Extraction: Principal axis factoring. Rotation: Promax with Kaiser normalization. h^2 = communality. α = Cronbach's alpha coefficient.

3.3 Confirmatory Factor Analysis Results

CFA was conducted on the independent sample ($n=312$) to test the fit of the five-factor model. The five-factor model demonstrated acceptable fit: $\chi^2(1415)=3309.84$, $p<0.001$; $\chi^2/df=2.34$; CFI=0.95; TLI=0.94; RMSEA=0.06 (90% CI: 0.058-0.065); SRMR=0.05. All items loaded significantly on their respective factors ($p<0.001$), with standardized factor loadings ranging from 0.54 to 0.89. Model fit indices are reported in Table 2, and inter-factor correlations are reported in Table 3.

Table 2. Model Fit Indices from Confirmatory Factor Analysis ($n=312$)

Index	Five-Factor Model	One-Factor Model	Criteria (Good Fit)
χ^2/df	2.34	5.87	<2.0
CFI	0.95	0.78	>0.95
TLI	0.94	0.76	>0.95
RMSEA	0.06	0.13	<0.06
SRMR	0.05	0.12	<0.05
$\Delta\chi^2$ (vs. 1-factor)	-	2847.32***	-

Note. χ^2/df = chi-square to degrees of freedom ratio; CFI = Comparative Fit Index; TLI = Tucker-Lewis Index; RMSEA = Root Mean Square Error of Approximation; SRMR = Standardized Root Mean Square Residual. *** $p < 0.001$.

Table 3. Factor Correlations from Confirmatory Factor Analysis

Factor	F1	F2	F3	F4	F5
F1: Genetic Carrier Guilt and Self-Blame	1.00				
F2: Social Stigmatization and Social Isolation	0.68***	1.00			
F3: Parental Secondary Trauma and Grief	0.71***	0.62***	1.00		
F4: Intrafamilial Role Changes and Relational Burden	0.58***	0.54***	0.65***	1.00	
F5: Genetic Counseling Experience and Information Needs	0.52***	0.48***	0.56***	0.45***	1.00

Note. *** $p < 0.001$.

3.4 Reliability Analyses

Cronbach's alpha coefficients exceeded 0.87 for all subscales and the total scale, indicating excellent internal consistency (Table 4). Test-retest reliability, assessed over a four-week interval in a subsample of 68 parents, was excellent for the total scale ($r = 0.89$, $p < 0.001$) and adequate to good for each subscale (F1: $r = 0.87$; F2: $r = 0.85$; F3: $r = 0.84$; F4: $r = 0.82$; F5: $r = 0.80$).

Table 4. Internal Consistency Reliability Coefficients

Scale/Subscale	No. of Items	Cronbach's α	Mean Inter-Item Correlation
Total Scale	55	0.96	0.48
F1: Genetic Carrier Guilt and Self-Blame	11	0.94	0.62
F2: Social Stigmatization and Social Isolation	10	0.92	0.58
F3: Parental Secondary Trauma and Grief	10	0.91	0.55
F4: Intrafamilial Role Changes and Relational Burden	9	0.89	0.51
F5: Genetic Counseling Experience and Information Needs	15	0.87	0.38

Note. α = Cronbach's α coefficient.

3.5 Convergent Validity

Convergent validity was supported by significant positive correlations between GGPBS scores and both the PSI-SF and BDI-II (Table 5). The total GGPBS score correlated $r=0.74$ with the PSI-SF and $r=0.68$ with the BDI-II, consistent with theoretical expectations that neurogenetic stigmatization is related to but distinct from general parenting stress and depressive symptomatology.

Table 5. Convergent Validity: Correlations with PSI-SF and BDI-II

GGPBS Scale/Subscale	PSI-SF Total	BDI-II Total
Total Scale	0.74***	0.68***
F1: Genetic Carrier Guilt and Self-Blame	0.69***	0.72***
F2: Social Stigmatization and Social Isolation	0.66***	0.61***
F3: Parental Secondary Trauma and Grief	0.71***	0.75***
F4: Intrafamilial Role Changes and Relational Burden	0.68***	0.58***
F5: Genetic Counseling Experience and Information Needs	0.52***	0.48***

Note. *** $p < 0.001$. PSI-SF = Parenting Stress Index-Short Form; BDI-II = Beck Depression Inventory-II.

3.6 Descriptive Statistics and Clinical Interpretation

Descriptive statistics for GGPBS scores in the CFA sample are presented in Table 6. Skewness and kurtosis values were within acceptable ranges ($|\text{skewness}| < 0.30$, $|\text{kurtosis}| < 0.60$), indicating approximately normal distributions (Field 2024).

Table 6. Descriptive Statistics for GGPBS Scores (n=312)

Scale/Subscale	Possible Range	M	SD	Skewness	Kurtosis
Total Scale	55-275	178.42	38.67	-0.18	-0.42
F1: Genetic Carrier Guilt and Self-Blame	11-55	38.74	9.21	-0.22	-0.38
F2: Social Stigmatization and Social Isolation	10-50	34.18	8.45	-0.16	-0.45
F3: Parental Secondary Trauma and Grief	10-50	35.62	8.92	-0.20	-0.40
F4: Intrafamilial Role Changes and Relational Burden	9-45	30.56	7.68	-0.14	-0.48
F5: Genetic Counseling Experience and Information Needs	15-75	52.34	11.42	-0.12	-0.52

Note. M = Mean; SD = Standard Deviation.

Table 7 presents the distribution of parents across four burden severity categories. A substantial proportion of parents reported moderate to clinical-level burden: 78.3% for genetic guilt and self-blame (F1), 71.2% for social stigmatization (F2), and 74.8% for secondary trauma and grief (F3). These findings underscore the pervasive nature of psychosocial burden in this population.

Table 7. Distribution of Parents Across Burden Categories (n=312)

Category	Total Scale	F1	F2	F3	F4	F5
Low Burden	18.3%	15.7%	19.2%	17.6%	21.5%	24.4%
Moderate Burden	52.9%	48.4%	51.3%	50.6%	54.2%	56.1%
High Burden	20.5%	22.1%	19.9%	21.2%	18.6%	15.7%
Clinical-Level Burden	8.3%	13.8%	9.6%	10.6%	5.7%	3.8%

Note. Burden categories are defined by quartile-based cut-off scores derived from the CFA sample distribution.

3.7 Demographic and Clinical Correlates

Table 8 presents correlations between GGPBS scores and key demographic and clinical variables. Consanguinity was the strongest positive predictor of total burden ($r=0.32$, $p<0.001$) and all subscale scores. Receipt of genetic counseling was associated with lower burden across all dimensions (total scale: $r=-0.28$, $p<0.001$), with the strongest inverse association observed for Factor 5 ($r=-0.42$, $p<0.001$).

Longer time since diagnosis was associated with lower burden ($r=-0.24$, $p<0.001$), suggesting a degree of adaptation over time.

Table 8. Correlations Between GGPBS Scores and Demographic/Clinical Variables

Variable	Total Scale	F1	F2	F3	F4	F5
Parent Age	-0.12*	-0.14*	-0.10	-0.11	-0.09	-0.08
Child Age	-0.18**	-0.16**	-0.15**	-0.19**	-0.14*	-0.12*
Time Since Diagnosis	-0.24***	-0.22***	-0.21***	-0.26***	-0.19**	-0.16**
Consanguinity (yes=1)	0.32***	0.38***	0.34***	0.28***	0.24***	0.18**
Genetic Counseling Received (yes=1)	-0.28***	-0.26***	-0.22***	-0.24***	-0.20**	-0.42***
Number of Children	0.14*	0.12*	0.11	0.13*	0.18**	0.10

Note. * $p<0.05$, ** $p<0.01$, *** $p<0.001$.

3.8 Group Comparisons by Consanguinity Status

Independent samples t-tests comparing parents with and without consanguineous marriage revealed significantly higher GGPBS scores across all subscales and the total scale in the consanguineous group (Table 9). Effect sizes were large for Factor 1 ($d=0.99$) and Factor 2 ($d=0.97$), and medium to large for the total scale ($d=0.78$) and remaining subscales.

Table 9. Group Comparisons on GGPBS Scores by Consanguinity Status

Scale/Subscale	Consanguineous M (SD)	Non-Consanguineous M (SD)	t	p	Cohen's d
Total Scale	196.84 (35.42)	168.72 (37.21)	5.68	<.001	0.78
F1: Genetic Carrier Guilt	44.26 (8.14)	35.82 (8.92)	7.24	<.001	0.99
F2: Social Stigmatization	39.18 (7.68)	31.46 (8.24)	7.12	<.001	0.97
F3: Secondary Trauma	38.92 (8.34)	33.74 (8.86)	4.42	<.001	0.60
F4: Relational Burden	33.64 (7.12)	28.82 (7.68)	4.78	<.001	0.65
F5: Information Needs	55.48 (10.86)	50.62 (11.54)	3.18	.002	0.43

Note. M = Mean; SD = Standard Deviation in parentheses. Cohen's d = effect size estimate.

4. Discussion

4.1 Interpretation of Factor Structure

This study successfully developed and validated the Genetic Guilt and Psychosocial Burden Scale (GGPBS), a novel instrument designed to assess the multidimensional psychosocial burden experienced by parents of children with rare and genetic diseases. The five-factor structure identified through EFA and confirmed through CFA provides empirical support for the theoretical model of neurogenetic stigmatization and bio-sociological burden proposed in this study (DeVellis 2022; Kline 2023).

Factor 1 (Genetic Carrier Guilt and Self-Blame) emerged as the most prominent dimension, accounting for 33.49% of the total variance. This factor captures the profound sense of responsibility and self-blame that parents experience when they perceive themselves as having transmitted a genetic defect to their child. The high factor loadings (0.66–0.87) and excellent internal consistency ($\alpha = 0.94$) indicate that genetic guilt is a coherent and salient construct in this population (Kessler 1999; Peay et al. 2015).

Factor 2 (Social Stigmatization and Social Isolation) reflects the social consequences of having a child with a genetic condition, including perceived judgment, blame, and exclusion from social networks. The high loadings on items related to consanguineous marriage stigma are particularly noteworthy, suggesting that in Turkish culture the intersection of genetic disease and consanguinity creates a distinctive context for social stigmatization (Tuncbilek 2001; Amorim and Shimizu 2022; Goffman 1963).

Factor 3 (Parental Secondary Trauma and Grief) reflects the trauma-related symptoms and chronic grief experienced by parents. The diagnostic process, characterized by uncertainty, delayed diagnosis, and limited access to genetic counseling, appears to be a significant source of traumatic stress in this population (Figley 1995; Pereira et al. 2022; Olshansky 1962).

Factor 4 (Intrafamilial Role Changes and Relational Burden) captures the impact of caregiving on family relationships. The finding that marital strain and role changes constitute a dimension distinct from intra-personal burden (guilt, trauma) supports the conceptual distinction between intra-personal and interpersonal dimensions of burden (Quittner et al. 1998; Sabbeth and Leventhal 1984; Van Riper 2007).

Factor 5 (Genetic Counseling Experience and Information Needs) addresses the adequacy of genetic counseling and unmet informational needs. This factor's emergence as a distinct dimension underscores the importance of genetic counseling quality as a determinant of parental well-being (Biesecker and Peters 2001; Pelentsov et al. 2016; Jeter and Hardin 2024).

4.2 Psychometric Properties and Comparison with the Literature

The GGPBS demonstrates excellent psychometric properties, with a total Cronbach's alpha of 0.96 and test-retest reliability of $r = 0.89$. These values compare favorably with other widely used caregiver burden scales, such as the Zarit Burden Interview (alpha = 0.86–0.93; Zarit et al. 1980) and the Parenting Stress Index (alpha = 0.80–0.90; Abidin 1995). The CFA fit indices (CFI = 0.95, RMSEA = 0.06) indicate that the five-factor structure is adequately supported by the data (Hu and Bentler 1999).

The convergent validity evidence is strong, with GGPBS scores showing significant positive correlations with the PSI-SF ($r = 0.74$) and BDI-II ($r = 0.68$). These associations are consistent with theoretical expectations, as parents with higher psychosocial burden would be expected to report greater parenting stress and depressive symptoms. The discriminant validity evidence, indicated by the superiority of the five-factor model over a one-factor model (delta-chi-sq = 2847.32, $p < 0.001$), supports the multidimensional structure of the construct (Walkowiak et al. 2025; Nunnally and Bernstein 1994).

4.3 Cultural Context and Turkish Sample Characteristics

The present study was conducted in Turkey, a country with distinctive cultural characteristics that shape parental experiences of genetic disease. The high rates of consanguineous marriage (29.8%–31.2% in our sample) create a unique context in which genetic risk is closely intertwined with cultural practices and family dynamics. The finding that consanguinity was significantly associated with higher burden across all domains, with the strongest effect for Factor 1 ($r = 0.38$), underscores the importance of addressing consanguinity-related stigma in genetic counseling practice (Tuncbilek and Koc 1994; Verdonk et al. 2018; Baynam et al. 2024).

The role of genetic counseling in reducing burden was particularly striking: parents who had received genetic counseling reported significantly lower burden across all domains. The largest effect was observed for Factor 5 ($r = -0.42$), confirming that genetic counseling directly addresses informational needs and also produces broader benefits for emotional and social well-being. These findings highlight the importance of expanding access to genetic counseling services in Turkey, where such services remain limited outside major urban centers (Chapple et al. 1997; Guersoy and Yeg 2025).

4.4 Clinical Implications

The GGPBS has significant implications for clinical practice. The scale can be used as a screening tool to identify parents at risk for high psychosocial burden, enabling early intervention. Given that 78.3% of parents in this sample reported moderate to clinical-level genetic guilt and 71.2% reported high social stigmatization, routine screening with the GGPBS in genetic counseling settings could substantially improve the detection and management of burden-related disorders (Benedetto et al. 2023; Kenny et al. 2022).

The subscale structure of the GGPBS allows clinicians to tailor interventions to the specific domains of burden experienced by individual parents. Parents with high scores on Factor 1 (Genetic Carrier Guilt) may benefit from cognitive-behavioral interventions targeting self-blame and irrational guilt, while

parents with high scores on Factor 5 (Information Needs) may benefit from enhanced genetic counseling and psychoeducational interventions (Corrigan 2007; Link and Phelan 2001; Brown 2010).

The finding that consanguinity is a risk factor for elevated burden has important implications for genetic counseling in populations where consanguineous marriage is prevalent. Culturally sensitive approaches that acknowledge the complex interplay of genetic risk, cultural norms, and family dynamics are essential for effective intervention (Hamdan-Mansour and Sulieman 2021; Yildirim and Conk 2005).

4.5 Limitations and Future Research Directions

This study has several limitations. First, the sample was recruited from a single clinical practice, which may limit the generalizability of the findings. Future studies should recruit from multiple centers and use community-based sampling to obtain more representative samples. Second, the cross-sectional design does not allow causal inferences about the relationships between demographic variables and burden. Longitudinal studies are needed to examine the temporal dynamics of psychosocial burden in parents of children with genetic conditions (Boss 1999; Lazarus and Folkman 1984).

Third, the study was conducted exclusively in Turkish, which limits the cross-cultural applicability of the GGPBS. Future research should develop and validate translations in other languages and cultural contexts. Fourth, the study relied on self-report measures, which may be subject to social desirability bias and recall effects. Multi-method assessments incorporating clinical interviews and biological stress markers are warranted in future work (Dolling 2025; Zeeman et al. 2020).

Despite these limitations, this study makes substantive contributions to the field of psycho-genetics and clinical psychology. The development and validation of the GGPBS fills a recognized gap in the measurement literature and provides a valuable tool for both research and clinical practice (McAllister et al. 2011; Woodgate et al. 2019).

5. Conclusion

This study developed and validated the Genetic Guilt and Psychosocial Burden Scale (GGPBS), a 55-item instrument measuring the multidimensional psychosocial burden experienced by parents of children with rare and genetic diseases. The scale demonstrates excellent psychometric properties, including a robust five-factor structure, strong internal consistency ($\alpha = 0.96$), and excellent test-retest reliability ($r = 0.89$).

The findings reveal that neurogenetic stigmatization is a pervasive phenomenon in Turkish parents of children with genetic conditions, with 78.3% reporting moderate to clinical-level genetic guilt and 71.2% reporting high levels of social stigmatization. Cultural factors, particularly consanguineous marriage practices and limited genetic literacy, amplify these burdens. Genetic counseling emerges as a protective factor, significantly reducing burden across all dimensions.

Based on these findings, routine screening of parents using the GGPBS in genetic counseling and clinical genetics settings is recommended. Culturally sensitive, multidimensional intervention programs targeting genetic guilt, social stigmatization, and secondary trauma are warranted, as is expansion of genetic counseling services in regions with elevated consanguinity rates. The GGPBS represents an important step toward more comprehensive understanding of the psychosocial burden associated with rare and genetic diseases and toward the development of evidence-based interventions to support affected families.

Declarations

Compliance with Ethics Guidelines

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Conflicts of interest. The authors declare no conflicts of interest.

Ethics approval. Ethical approval was obtained from the Northwest University Ethics Committee (Protocol No: NWU-ETHICS-2024-0117, January 17, 2025). The study was conducted in accordance with the ethical standards of the 1964 Declaration of Helsinki and its later amendments.

Informed consent. Written informed consent was obtained from all participants prior to study participation.

Author contributions. G.I.A. contributed to conceptualization, data analysis, and manuscript preparation. K.S.Y. contributed to data collection, clinical assessment, and manuscript review. Both authors read and approved the final manuscript.

Data availability. The datasets generated and analyzed during the current study are available from the corresponding author upon reasonable request.

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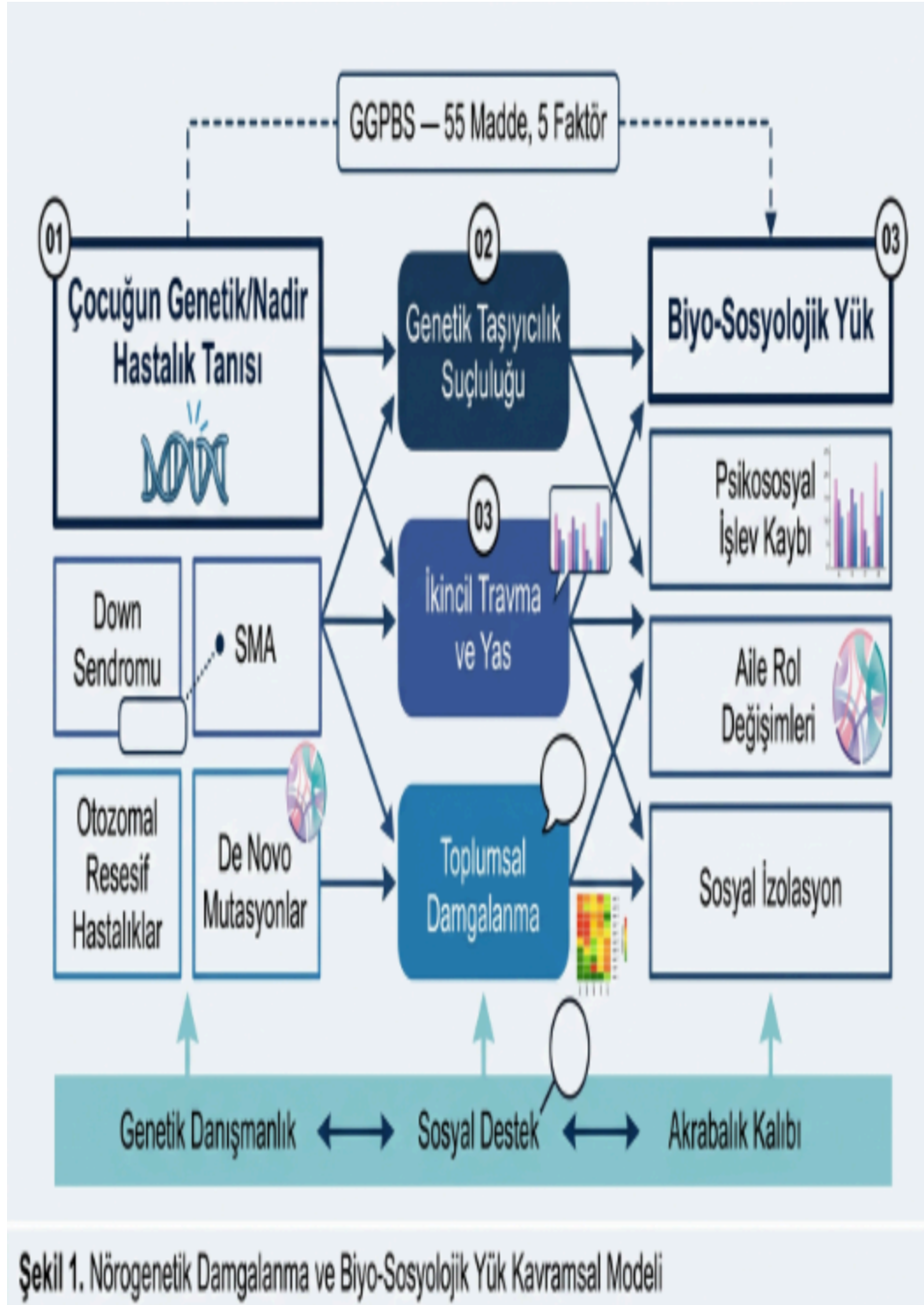
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Figures



Şekil 1. Nörogenetik Damgalanma ve Biyo-Sosyolojik Yük Kavramsal Modeli

Figure 1

Conceptual Model of Neurogenetic Stigmatization and Bio-Sociological Burden in Parents of Children with Rare Genetic Diseases

Note. The model illustrates the interrelationships among five domains of parental burden: (F1) Genetic Carrier Guilt and Self-Blame, (F2) Social Stigmatization and Social Isolation, (F3) Parental Secondary Trauma and Grief, (F4) Intrafamilial Role Changes and Relational Burden, and (F5) Genetic Counseling Experience and Information Needs. Bio-sociological stressors – consanguinity stigma, socioeconomic vulnerability, and isolation from genetic services – mediate the pathway from neurogenetic diagnosis to parental burden. Protective factors (genetic counseling access, social support, and community cohesion) attenuate burden across all domains. GGPBS = Genetic Genealogy and Parental Burden Scale.