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The Self-Concept, Family Functioning, Psychological and Emotional Symptoms, in Children and Adolescents with Neurofibromatosis Type 1

Nörofibromatozis Tip 1 Tanılı Çocuk ve Ergenlerde Benlik Kavramı, Aile İşlevleri, Ruhsal ve Emosyonel Semptomlar

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ABSTRACT

Aim: This study aimed to examine the self-concept and related psychological factors in children and adolescents with neurofibromatosis type 1 (NF1).
Material and Methods: A total of 71 participants (35 patients and 36 controls) were included in this study. As measurement tools the Piers-Harris children's self-concept scale (PHSCS), family assessment device (FAD), Turgay DSM-IV-based screening and rating scale for disruptive behavior disorders (T-DSM-IV-S), children's depression inventory (CDI), Beck depression inventory (BDI), state-trait anxiety inventory (STAI), children's sleep habits questionnaire (CSHQ), and autism spectrum quotient-adolescent (AQ-adolescent) were used.
Results: NF1 patients had lower intellectual functioning/academic achievement (p=0.002) and popularity/social appreciation (p<0.001) subscale scores in addition to total self-esteem (p=0.002) scores compared to the control group. Inattentiveness (p=0.024), state anxiety (p=0.028), and trait anxiety (p=0.012) symptoms were more common in these patients, and autistic features were more conspicuous in adolescents (p<0.001). Mother's depression (p=0.045), and state and trait anxiety (p=0.016, and p=0.006, respectively) levels were higher in the NF1 group. Depression, state anxiety, and hyperactivity were found predictors of self-concept in children and adolescents with NF1 (p<0.001, p=0.036, and p=0.016, respectively).
Conclusion: Turkish children and adolescents with NF1 face substantial challenges related to their self-concept and psychological health, particularly in academic and social domains, compared to their peers. It seems important to be aware of poor self-concept and related psychological factors in NF patients early in life and to develop intervention programs to address this issue.
Keywords: Self-concept; anxiety; neurofibromatosis 1.

ÖZ

Amaç: Bu çalışma, nörofibromatozis tip 1 (NF1) tanısı olan çocuk ve ergenlerde benlik kavramını ve ilişkili psikolojik faktörleri incelemeyi amaçlamaktadır.
Gereç ve Yöntemler: Bu çalışmaya toplam 71 katılımcı (35 hasta ve 36 kontrol) dahil edildi. Ölçme aracı olarak Piers-Harris çocuklar için benlik kavramı ölçeği (PHBKÖ), aile değerlendirme ölçeği (ADÖ), Turgay yıkıcı davranım bozuklukları için DSM-IV'e dayalı tarama ve değerlendirme ölçeği (YDB- TDÖ), çocuklar için depresyon ölçeği (ÇDÖ), Beck depresyon envanteri (BDE), durumluk-sürekli kaygı envanteri (DSKE), çocuk uyku alışkanlıkları anketi (ÇUAA) ve otizm spektrum anketi-ergen formu (OSA-ergen) uygulandı.
Bulgular: NF1 hastaları, kontrol grubuna kıyasla daha düşük toplam benlik kavramı puanlarının (p=0,002) yanı sıra daha düşük zihinsel işlevsellik/akademik başarı (p=0,002) ve popülerlik/sosyal beğeni (p<0,001) alt ölçek puanlarına sahipti. Dikkat eksikliği (p=0,024), durumluk anksiyete (p=0,028) ve sürekli anksiyete (p=0,012) semptomları bu hastalarda daha sıktı ve ergenlerde otistik özellikler (p<0,001) daha belirgindi. Annelerin depresyon (p=0,045) ve durumluk ve sürekli kaygı (sırasıyla p=0,016 ve p=0,006) düzeyleri NF1 grubunda daha yüksekti. NF1 tanılı çocuk ve ergenlerde depresyon, durumluk anksiyete ve hiperaktivite, benlik kavramının yordayıcıları olduğunu gösterdi (sırasıyla, p<0,001, p=0,036 ve p=0,016).
Sonuç: NF1 tanısı olan Türk çocuk ve ergenler, akranlarına kıyasla, özellikle akademik ve sosyal alanlarda olmak üzere benlik kavramları ve psikolojik sağlıkları ile ilgili önemli zorluklarla karşılaşmaktadır. NF hastalarındaki zayıf benlik kavramının ve ilişkili psikolojik faktörlerin yaşamın erken dönemlerinde farkında olmak ve buna yönelik müdahale programları geliştirmek önemli görünmektedir.
Anahtar kelimeler: Benlik kavramı; anksiyete; nörofibromatozis 1.

INTRODUCTION

Neurofibromatosis (NF) is a progressive, multisystemic, genetic disorder that primarily affects the nervous system and skin, increasing the risk of tumors and affecting the skeletal, endocrine, reproductive, and cardiovascular systems. Additionally, elevates the risk of developing mental disorders (1,2). These include NF type 1 (NF1, known as Von Recklinghausen disease), NF type 2 (NF2), and schwannomatosis (SWN) (3). NF1 and NF2 are autosomal dominant disorders caused by mutations in genes located on chromosomes 17 and 22, respectively. They can be inherited or caused by de novo mutations in germ cells, and each mechanism is responsible for approximately half of the cases. The pooled prevalence of NF1 and NF2 was 1:3164 (95% CI, 1:2132-1:4712) and 1:50.000 (95% CI, 1:32.829-1:65.019), respectively (4). The exact prevalence of mosaic cases is not known but is estimated at 1:36.000 to 1:40.000 (5). NF1 is a RASopathy caused by mutations in the neurofibromin gene and characterized by café-au-lait patches, Lisch nodules, and neurofibromas (5). Neuropsychologically, NF1 is characterized by problems in visual-spatial, visual-perceptive, and executive functions along with maintaining and shifting attention. These problems may increase with age and may be present in mosaic cases (6). Up to 80.0% of children with NF1 may have psychiatric disorders while 25.0-36.8% may be diagnosed with autism spectrum disorder (ASD). Otherwise, 40.0% of NF1 individuals have attention deficit hyperactivity disorder (ADHD) (6-10). In the preschool period, up to 68.0% of these children may display delays in speech/language development and motor coordination problems. Preclinical studies suggest that learning and memory in NF1 may be disrupted by p21RAS activity and long-term potentiation, whereas lesions in the basal ganglia, thalamus, brain stem, and cerebellum may contribute to the development of ADHD (5-10). A genetic disorder with far-reaching consequences, such as NF1, may also affect self-concept in addition to causing psychopathology (11).

The set of beliefs (also known as self-schemas) about oneself is known as one's self-concept. It can be also termed as, self-identity, self-perspective, self-construction, and self-structure (12,13). It differs from self-awareness, self-knowledge, self-esteem, and the social self, as it interacts with them to form the self as a gestalt. Self-schemas may pertain to the past, the current, and the future, as well as various domains of daily functioning (e.g., academic, social, etc.). They affect behavior and are composed of cognitive/descriptive components rather than evaluations or opinions (12,13). Although it develops and is elaborated upon throughout one's lifespan, it is most malleable in childhood and adolescence (12-14). Age, gender, developmental, social, and educational processes, as well as chronic disorders affect its development (11-15). Studies conducted on self-concept and related constructs among patients with NF1 at various ages suggest that children may focus on the effects of the disorder on physical abilities and pain (11,16), while adolescents report broader effects on self-concept (17,18), and that the clinical severity of the disorder may not correlate with changes in self-concept (11).

Studies conducted on patients with NF1 at various ages suggest that they may have reduced self-esteem compared to their peers (19). Having peers with NF1, attending dedicated support groups, receiving care at specialized clinics, and genetic counseling were found to predict higher self-esteem (19). Almost five to seven million people in Turkey are thought to be affected by rare genetic or metabolic disorders, including NF (20). Limited knowledge of these disorders, limited numbers of specialist physicians and centers, elevated costs of treatment, and high rates of consanguineous marriages further contribute to this problem (20). Studies on Turkish children with NF are limited to single centers (21,22) and focus on the neurocognitive abilities of affected children and their correlates (23-25). To the best of our knowledge, no research has evaluated self-concept, family functioning, or reported emotional/psychological symptoms among Turkish children with NF. Therefore, this study aimed to assess self-concept and related characteristics in Turkish children and adolescents with NF1 and compare them with healthy controls.

MATERIAL AND METHODS

Study Center, Sampling, and, Ethics

This is a cross-sectional, case-control study of children and adolescents diagnosed with NF1 and is in the process of treatment and follow-up in the Pediatric Oncology Department of Mersin University conducted between May 15, 2021, and October 15, 2021. The following were the inclusion criteria for the patient group: 1) diagnosis of NF1 according to the National Institutes of Health (NIH) criteria (26); 2) age between 8 and 18 years (including 8 and 18 years old); 3) absence of hearing loss and visual impairment; 4) absence of tumors, hepatic, and renal disorders; 5) normal ambulation skills; 6) fluency in Turkish; 7) ability to complete scales at a level that can be analyzed; and 8) acceptance of participation in this study and approval of the informed consent form for both the parents and children. The following were the inclusion criteria for the control group: 1) age between 8 and 18 years (including 8 and 18 years); 2) absence of hearing loss and visual impairment; 3) absence of any chronic illness, medication use, major trauma, surgical history, and physical disability; 4) fluency in Turkish and ability to complete scales at a level that can be analyzed; 5) absence of psychopathology in the diagnostic and statistical manual of mental disorders, 5th edition (DSM-5)-based psychiatric interviews (27); and 6) acceptance of participation in this study and approval of the informed consent form by both the parents and themselves. Sixty patients with NF were followed up in the Department of Pediatric Oncology, and 35 met the inclusion and exclusion criteria. Therefore, 71 participants (35 patients and 36 controls) were included in this study. The study was conducted in accordance with the principles of the Declaration of Helsinki, following the approval from the ethics committee of Mersin University Faculty of Medicine (08.05.2019, 186).

Data Collection Tools

Piers-Harris Children's Self-Concept Scale (PHSCS)

This scale was developed by Piers (28) to evaluate the self-concept and adapted to Turkish by Öner (29). It consists of 80 items with six subscales and is standardized for those aged 9-20. The internal consistency of the PHSCS was determined as 0.87 for the children sample,

and 0.86 for the adolescent sample. The scale score is between 0 and 80 points, with high scores indicating positive self-concept. PHSCS ≤39 is classified as reflecting "poor self-esteem", and PHSCS ≥40 is "average, and normal self-concept." In this study, the cut-off score was set at 40 to distinguish between poor and average/normal self-concept (29). In this study, the children completed the PHSCS.

Family Assessment Device (FAD)

This scale measures family functioning on seven distinct subscales by means of this 4-point Likert-type self-report scale: problem-solving, communication, roles, emotional responsiveness, attention span, behavior control, and general functions. It comprised 60 items. Mean scores greater than 2 are recognized as indicators of a trend toward poorer family functioning (30). The reliability and validity of the Turkish version have already been determined by Bulut (31). In the study, Cronbach's alpha value was found as 0.91 for general functioning, and ranging from 0.38 to 0.86 for the subscales.

Turgay DSM-IV-Based Screening and Rating Scale for Disruptive Behavior Disorders (T-DSM-IV-S)

This 4-point, Likert-type scale evaluates symptoms of attention deficit, hyperactivity/impulsivity, oppositional defiant, and conduct disorder according to parents'/teachers' reports. It consisted of 41 items. Its validity and reliability were established by Ercan et al. (32), and Cronbach's alpha coefficients of the subscales were found to be 0.88 for attention deficit, 0.95 for hyperactivity/impulsivity, 0.89 for oppositional defiant, and 0.85 for conduct disorder. High scores indicate increased symptom levels. The children's mothers completed the scale.

Children's Depression Inventory (CDI)

This self-report scale consists of 27 items evaluating symptoms of depression and is standardized between the ages of 6 and 17 years (33). The cut-off score was 19, and the maximum score was 54. Öy (34) conducted a validity and reliability study of the CDI's Turkish version, and Cronbach's alpha value was determined as 0.77. Children completed the CDI in this study.

Beck Depression Inventory (BDI)

This self-report scale consists of 21 items evaluating symptoms of depression, each of which is scored between 0 and 3 (35). Hisli (36) conducted a validity and reliability study of the BDI's Turkish version, and the internal consistency coefficient of the scale was found as 0.74. The mothers completed the BDI for themselves.

State-Trait Anxiety Inventory (STAI)

This is a 40-item, 4-point Likert-type scale, 20 items tapping state, and 20 items evaluating trait anxiety. A total score can range from 20 to 80 with each item receiving a value between 1 and 4 (37). Öner and Le Compte (38) conducted a reliability and validity study on the Turkish version. Cronbach's alpha value was found as 0.93 for STAI-State and 0.85 for STAI-Trait in the study. The mothers and children completed the STAI.

Children's Sleep Habits Questionnaire (CSHQ)

This scale evaluates the sleep habits and associated problems in children by 3-point Likert-type 33 items (39). Fiş et al. (40) performed a validity and reliability study of the CSHQ's Turkish version, and Cronbach's alpha coefficient was determined as 0.78. The parents completed the CSHQ for their children.

Autism Spectrum Quotient-Adolescent (AQ-Adolescent)

This is a parental report-based screening tool for measuring the degree of subthreshold autistic characteristics of adolescents with normal intelligence (41). Çetinoğlu and Aras (42) conducted a validity and reliability study in Turkey, and Cronbach's alpha value was found as 0.83. The cut-off score for the AQ-adolescent scale was 24 points. Parents completed AQ-adolescent for their children in this study.

Statistical Analysis

The Shapiro-Wilk test was used to test the assumptions of normality. The normally distributed data were compared between groups using an independent samples t-test, while the non-normally distributed data were compared using a Mann-Whitney U test. The categorical data was analyzed using either Fisher's exact test or Pearson's chi-square, depending on the expected value. Pearson's correlation analysis was used in order to examine the correlations between the continuous variables and the PHSCS score. The effects of variables that were significant in correlation analyses as predictors of PHSCS were analyzed using multiple regression analysis (backward method). Statistical analyses were performed using the IBM SPSS v.22.0 software (IBM Inc., Armonk, NY, USA), and the level of significance was considered 0.05.

RESULTS

The NF1 and control groups had mean ages of 12.9±3.4 and 13.1±3.0 years, respectively. The majority of both groups (65.7%, n=23 in NF1 and 58.8%, n=21 in control) were male. There were no significant differences in age and gender between the groups (p=0.766, and p=0.522, respectively). The frequency of having one or more psychiatric disorders in the NF1 group (82.8%, n=29) compared to the control group (2.8%, n=2) was significantly higher (p<0.001). The NF1 group had significantly lower PHSCS total (p=0.002), intellectual and school status (p=0.002), anxiety (p=0.044), and popularity/social appreciation (p<0.001) scores compared to the control group (Table 1).

According to the results of the parent T-DSM-IV-S scale, only the attention deficit subscale score was higher in the NF1 group (p=0.024). CDI scores showed no statistically significant difference (p=0.074), however, both the STAI-state (p=0.028) and STAI-trait (p=0.012) scores were significantly higher in the NF1 group. This group had

Table 1. Evaluation of subscale and total scores of the PHSCS between NF1 and the control groups

	NF1 (n=35)	Control (n=36)	p
Intellectual and school status	4.00±1.88	5.19±1.19	**0.002**
Happiness	7.03±2.24	6.64±2.31	0.473
Anxiety	5.00±2.44	6.03±1.73	**0.044**
Physical appearance and attributes	4.57±1.56	5.11±1.09	0.095
Behavioral adjustment	9.91±2.48	10.75±1.46	0.090
Popularity/Social appreciation	5.14±1.68	7.31±0.86	**<0.001**
PHSCS-Total	35.66±8.31	41.03±5.57	**0.002**

NF1: neurofibromatosis type 1, PHSCS: Piers-Harris children's self-concept scale

higher AQ-adolescent scores (p<0.001), and no significant difference in total sleep scores (p=0.461, Table 2).

In the FAD scale, only the FAD involvement score was significantly different between the groups (p=0.034). Mothers' BDI (p=0.045), STAI-trait (p=0.016), and STAI-state anxiety scores (p=0.016) were higher in the NF1 group (Table 3).

FAD affective involvement (r=-0.379, p=0.032), FAD behavior control (r=-0.443, p=0.011), T-DSM-IV-S attention deficit (r=-0.474, p=0.004), T-DSM-IV-S hyperactivity (r=-0.422, p=0.012), CDI (r=-0.599, p<0.001), STAI-trait (r=-0.605, p=0.001), and STAI-state (r=-0.547, p=0.001) scores were significantly correlated with the PHSCS total score. No correlation was found between the T-DSM-IV-S oppositional defiant (r=-0.223, p=0.199), T-DSM-IV-S conduct disorder (r=-0.115, p=0.511), mother's BDI depression (r=-0.177, p=0.331), mother's STAI-trait anxiety (r=-0.012 p=0.947), mother's STAI-state anxiety (r=-0.012 p=0,420), CSHQ (r=-0.088 p=0,622), AQ-adolescent (r=-0.264 p=0.184), and PHSCS total score. In the NF1 group, there was no correlation between T-DSM-IV-S attention deficit score (r=0.102, p=0.613), T-DSM-IV-S hyperactivity (r=0.034, p=0.865) score, and AQ-adolescent score. In the multiple regression analysis, the PHSCS total score among children with NF1 was negatively predicted by T-DSM-IV-S hyperactivity, CDI, and STAI-state scores (F=16.936; p<0.001; Table 4).

Lastly, the PHSCS total scores were divided into two groups, PHSCS ≤39 (n=22) and PHSCS ≥40 (n=13), and the study variables were compared between the groups. There were no statistically significant sex and age differences between the groups (p=0.736, and p=0.411, respectively). The CDI (p=0.017), STAI-state (p=0.006), STAI-trait (p=0.002) anxiety scores, and FAD affective involvement (p=0.033) scores were significantly higher in the group with PHSCS ≤39 (Tables 5, and Table 6).

DISCUSSION

This single-center study used a case-control cross-sectional design to evaluate self-concept and associated factors in Turkish children and adolescents with NF1 and compared them with healthy controls. The rate of psychopathology was found to be significantly elevated among the children with NF1, they had lower self-esteem (especially in terms of intellectual/academic status and popularity/social appreciation), and their state and trait anxiety were significantly elevated compared to controls. Their mothers rated them as having significantly elevated symptoms of inattention and ASD compared to the controls. Self-reported depressive and state/trait anxiety scores were also significantly elevated among the mothers of children with NF1. Families of children with NF1 have been found to show significantly different levels of parental involvement. The self-esteem of children with NF1 correlated negatively with parental involvement, behavior control, self-reported depression, state and trait anxiety, and parent-reported inattention/hyperactivity. In regression analyses, self-reported depression, state anxiety, and parent-reported hyperactivity emerged as significant predictors of self-esteem among children with NF1. Bivariate analyses showed that lower self-esteem among children with NF1 was also associated with lower affective involvement.

Up to five-fourths of children with NF1 may have psychopathology, while less than half may be diagnosed with ADHD (6-10). Supporting these results, the frequency of psychopathology among our sample of children with NF1 was reported to be 82.8%, which was significantly higher than that in controls. Mothers also rated

Table 2. Evaluation of T-DSM-IV-S, CDI, STAI, CSHQ, and AQ-Adolescent scores between NF1 and the control groups

	NF1 (n=35)	Control (n=36)	p
T-DSM-IV-S, attention deficit	13.60±8.08	9.49±6.82	**0.024**
T-DSM-IV-S, hyperactivity	10.71±8.65	8.80±5.76	0.280
T-DSM-IV-S, oppositional defiant	8.71±7.39	7.89±6.66	0.624
T-DSM-IV-S, conduct disorder	2.43±5.51	1.86±2.93	0.590
CDI	11.74±6.81	8.94±6.18	0.074
STAI-state	36.34±10.44	31.33±8.26	**0.028**
STAI-trait	41.09±9.23	35.56±8.84	**0.012**
CSHQ	55.88±11.76	53.92±10.41	0.461
AQ-adolescent	21.74±5.03	16.11±5.69	**<0.001**

T-DSM-IV-S: Turgay DSM-IV-based screening and rating scale for disruptive behavior disorders, CDI: children's depression inventory, STAI: state-trait anxiety inventory, CSHQ: children's sleep habits questionnaire, AQ-adolescent: autism spectrum quotient-adolescent, NF1: neurofibromatosis type 1

Table 3. Evaluation of FAD, mother's BDI, and mother's STAI scales between NF1 and control groups

	NF1 (n=35)	Control (n=36)	p
FAD, general functioning	1.90±0.70	1.74±0.48	0.278
FAD, problem solving	1.82±0.61	1.80±0.48	0.909
FAD, communication	1.92±0.59	1.78±0.62	0.371
FAD, roles	2.13±0.58	2.20±0.44	0.547
FAD, affective involvement	2.52±0.50	2.29±0.36	**0.034**
FAD, behavior control	2.23±0.54	2.15±0.47	0.527
FAD, affective responsiveness	2.19±0.86	1.96±0.79	0.254
Mother's BDI	16.81±10.26	12.25±8.09	**0.045**
Mother's STAI-state	42.86±10.74	36.28±11.60	**0.016**
Mother's STAI-trait	48.76±10.93	41.75±9.80	**0.006**

FAD: family assessment device, BDI: Beck depression inventory, STAI: state-trait anxiety inventory, NF1: neurofibromatosis type 1

Table 4. Predictors of PHSCS total score

	Beta	95% CI	p
(Constant)		47.587 - 61.684	**<0.001**
T-DSM-IV-S, hyperactivity	-0.291	-0.553 - -0.061	**0.016**
CDI	-0.530	-1.010 - -0.344	**<0.001**
STAI-state	-0.281	-0.437 - -0.016	**0.036**

PHSCS: Piers-Harris children's self-concept scale, T-DSM-IV-S: Turgay DSM-IV-based screening and rating scale for disruptive behavior disorders, CDI: children's depression inventory, STAI: state-trait anxiety inventory, CI: confidence interval

their children as having significantly elevated symptoms of inattention and ASD compared to the controls. Anxiety, depression, and learning disorders may also be elevated in children with NF1 (43). Partially supporting these results, children with NF1 in our study reported significantly elevated state and trait anxiety, while their depressive symptom levels were similar to those of controls. This discrepancy may be due to our dependence on self-report instruments, which may be subject to reporting and recall bias, as well as shared method variance. We did not evaluate ADHD, ASD, or learning disorders using semi-structured clinical interviews (44). Future studies on psychopathology among children with NF1 should use structured clinical interviews and parent and clinician ratings of depression, anxiety, and various psychopathologies. They may also evaluate the correlations between these psychopathologies and the severity of the NF1.

Studies conducted on patients with NF1 at various ages suggest that they may have reduced self-esteem compared to their peers (19). Barton et al. (11) reported that children with NF1 may have lower self-esteem, especially in terms of physical and sports-related skills, while their academic self-esteem may be elevated. In our study, children with NF1 reported lower self-esteem, especially in terms of intellectual and academic status and popularity. Lower self-esteem in the academic/intellectual domain may be due to the presence of learning disorders (45). Children with NF1 have also been reported to be less popular and more prone to peer bullying (46). Supporting these views, children with NF1 in our study rated themselves as less popular among their peers. Lower self-esteem in our sample was predicted by elevated self-reported depression and state anxiety, and higher parent-reported hyperactivity. There are also signals that lower family functioning may contribute to reduced self-esteem. Although the study's cross-sectional nature prevents hypotheses on causality, it may be posited that lower family support may have led to a negative self-concept and internalizing symptoms, while hyperactivity/impulsivity may lead to problems with peers, further compounding the problem (46). Future studies on self-esteem and related factors among children with NF1 should use longitudinal designs with multiple evaluation waves to test this hypothesis.

We need to consider evaluating the results in light of these limitations. First, the results are valid for the study center and may not be generalizable to children with NF1 followed up at other centers or those without access to specialized care. Second, we did not evaluate the level of social support available to children and their victimization rates in peer bullying. Third, we did not evaluate ADHD, ASD, or learning disorders in children with NF1 using psychometric instruments. Fourth, we did not evaluate the effects of NF1 symptom severity or visibility on self-esteem or psychopathology. Fifth, dependence on multiple self- and parent-report instruments may have biased our results. Sixth, we did not evaluate the effects of genetic counseling on the symptoms of children and mothers. Lastly, we did not evaluate mothers with structured clinical interviews for psychopathology. Regardless of these limitations, our results suggest that Turkish children with NF1 have lower self-esteem and higher symptoms of psychopathology compared to peers and that interventions to address these problems may be beneficial.

Table 5. Evaluation of emotional symptoms and family functioning according to the PHSCS total score in the mothers of patients with NF1

	PHSCS ≤39 (n=22)	PHSCS ≥40 (n=13)	p
FAD, general functioning	1.93±0.80	1.86±0.52	0.798
FAD, problem solving	1.70±0.64	2.01±0.54	0.166
FAD, communication	1.89±0.66	1.95±0.47	0.786
FAD, roles	2.04±0.62	2.27±0.50	0.298
FAD, affective involvement	2.64±0.58	2.31±0.25	**0.033**
FAD, behavior control	2.32±0.51	2.09±0.57	0.257
FAD, affective responsiveness	2.21±1.00	2.15±0.57	0.827
Mother's BDI	17.57±11.89	15.36±6.31	0.572
Mother's STAI-state	41.73±11.53	44.77±9.37	0.426
Mother's STAI-trait	47.82±12.38	50.50±7.80	0.503

PHSCS: Piers-Harris children's self-concept scale, NF1: neurofibromatosis type 1, FAD: family assessment device, BDI: Beck depression inventory, STAI: state-trait anxiety inventory

Table 6. Evaluation of the emotional and behavioral symptoms, sleep habits, and autistic features according to the PHSCS total score in the patients with NF1

	PHSCS ≤39 (n=22)	PHSCS ≥40 (n=13)	p
T-DSM-IV-S, attention deficit	15.05±7.91	11.15±8.08	0.172
T-DSM-IV-S, hyperactivity	11.73±8.95	9.00±8.16	0.375
CDI	13.82±6.34	8.23±6.31	**0.017**
STAI-state	39.95±10.05	30.23±8.22	**0.006**
STAI-trait	44.64±8.64	35.08±6.98	**0.002**
CSHQ	54.45±11.96	58.50±11.41	0.345
AQ-adolescent	22.24±3.72	20.90±6.87	0.581

PHSCS: Piers-Harris children's self-concept scale, NF1: neurofibromatosis type 1, T-DSM-IV-S: Turgay DSM-IV-based screening and rating scale for disruptive behavior disorders, CDI: children's depression inventory, STAI: state-trait anxiety inventory, CSHQ: children's sleep habits questionnaire, AQ-adolescent: autism spectrum quotient-adolescent

CONCLUSION

The study highlights that Turkish children and adolescents with NF1 face substantial challenges related to their self-concept and psychological health compared to their peers. Elevated rates of anxiety, ADHD symptoms, and reduced self-esteem, particularly in academic and social domains, underscore the complex impact of NF1 on daily life and emotional well-being. These findings emphasize the importance of comprehensive care strategies that address not only medical needs but also psychosocial support for affected individuals and their families. Future research should explore longitudinal effects and interventions to mitigate these challenges effectively.

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