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ORIGINAL ARTICLE

Validity of the Gross Motor Function Measurement in a sample of Turkish Children with Neurofibromatosis Type 1

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Purpose: Muscle weakness and delays in motor development are more common problems in children with neurofibromatosis type 1. Gross Motor Function Measurement-88 is widely used tool to evaluate motor functions in children with developmental disabilities. We aimed to investigate validity of the Gross Motor Function Measurement-88 in a sample of Turkish children with neurofibromatosis type 1.
Methods: Aged between 5 to 17 years 40 children (20 male/20 female) with neurofibromatosis type 1 participated in this study. To asses validity of Gross Motor Function Measurement-88, Manual Muscle Test was done seven muscle groups in upper and lower limbs bilaterally by a physical therapist.
Results: The mean age was 9.7±3.81 years. A positive moderate to strong correlation was found between Manual Muscle Test and Gross Motor Function Measurement-88 subdomains (r=0.317-0.668; p <0.05).
Conclusion: Gross Motor Function Measurement-88 is a valid measurement for evaluating gross motor functions in children with neurofibromatosis type 1. Identifying motor developmental delays in children with neurofibromatosis type 1 will be a guide for establishing early intervention programs and determining symptom-specific rehabilitation goals. We recommend the use of Gross Motor Function Measurement-88 in children with neurofibromatosis type 1 for evaluating gross motor function.
Keywords: Neurofibromatosis, Validity, Physical therapy.

Nörofibramatosis Tip 1 tanılı Türk çocukları örnekleminde Kaba Motor Fonksiyon Ölçütü'nün geçerliği

Amaç: Kas güçsüzlüğü ve motor gelişimdeki gecikmeler nörofibromatozis tip 1 olan çocuklarda sık görülen problemlerdir. Kaba Motor Fonksiyon Ölçümü-88, gelişim geriliği olan çocuklarda motor fonksiyonları değerlendirmek için yaygın olarak kullanılan bir araçtır. Bu çalışmanın amacı tip 1 nörofibromatozisli Türk çocuklarından oluşan bir örneklemde Kaba Motor Fonksiyon Ölçümü-88'in geçerliğini araştırmaktır.
Yöntem: Bu çalışmaya nörofibromatozis tip 1 olan 5 ile 17 yaş arası 40 çocuk (20 erkek/20 kız) katıldı. Kaba Motor Fonksiyon Ölçümü-88'in geçerliliğini değerlendirmek için Manuel Kas Testi, fizyoterapist tarafından üst ve alt ekstremitelerde yedi kas grubuna bilateral olarak yapıldı.
Bulgular: Yaş ortalaması 9,7±3,81 yıldı. Manuel Kas Testi ve Kaba Motor Fonksiyon Ölçümü-88 alt boyutları arasında pozitif yönde orta ile güçlü korelasyon bulundu (r=0.317-0.668; p <0.05).
Sonuç: Kaba Motor Fonksiyon Ölçümü-88, nörofibromatozis tip 1 tanılı çocuklarda kaba motor fonksiyonların değerlendirilmesi için geçerli bir ölçümdür. Nörofibromatozis Tip 1'li çocuklarda motor gelişimsel gecikmelerin belirlenmesi, erken müdahale programlarının oluşturulması ve semptoma özgü rehabilitasyon hedeflerinin belirlenmesi için bir rehber olacaktır. Kaba motor fonksiyonu değerlendirmek için Nörofibromatozis tip 1 tanılı çocuklarda Kaba Motor Fonksiyon Ölçümü-88'in kullanılmasını öneriyoruz.
Anahtar kelimeler: Nörofibromatozis, Geçerlilik, Fizyoterapi.

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Neurofibromatosis type 1 (NF1) is a rare autosomal dominant disorder that affects multiple organ systems. The average prevalence of NF1 is 1/3000-3500. Clinical features include neurofibromas or nerve sheath tumors, skin pigmentary abnormalities, low-grade gliomas and skeletal dysplasia, as well as the involvement of numerous other organ systems are of the NF1.[1]

The condition may gradually progress over lifetime, although the specific manifestations, rate of progression and severity of complications vary widely. At present, no cure is available, and clinical management is typically limited to surveillance and symptomatic treatment, usually surgical or symptomatic treatments for particular complications such as physiotherapy, speech therapy, or medication for pain or attention deficit.[2,3]

Common motor problems reported in children with NF1 are delay in motor development, muscle weakness, and tone abnormalities.[1,2] Movement Assessment Battery for Children version 1 and 2 (M-ABC-1 and 2)[2] (3 years to 16 years 11 months) and Bayley Scales of Infant Development-III (BSID-III)[4] (0 to 42 months) were used to assess mental and motor skill development in children with NF1. But these assessment tools are not validated in children with NF1 and age ranges are not comprehensive for motor assessment. The Gross Motor Function Measurement (GMFM) (5 months to 16 years of age) is a detailed test measuring motor functions and an optimal one for assessing functional mobility in children with neurological conditions.[5] It was first created for children with cerebral palsy (CP).[5] The GMFM evaluates motor function and activity in children with CP[6] and the only one assessment tool has Turkish version.[7] Until now GMFM's validity and reliability was shown in children with different types of disabilities such as Down syndrome (DS)[8], spinal muscular atrophy (SMA)[9], cerebral visual impairment (CVI)[10], Fukuyama-type congenital muscular dystrophy (FCMD).[11] It was found only valid in traumatic brain injury (TBI) and its reliability has not been investigated.[12] However, to our knowledge, no validation study has been reported on GMFM in children with NF1. Therefore, we aimed to investigate the validity of the GMFM in a sample of Turkish children with NF1.

METHODS

Study design

This study was designed as a psychometric evaluation study.

Setting and participants

The study was conducted between April 2018 and April 2020. Ethical approval was obtained from Non-Interventional Clinical Research Ethics Committee (GO 17/935-10), date: 02.01.2018 according to the Declaration of Helsinki.

Children between 5-17 years of age diagnosed with NF1 according to National Institutes of Health (NIH) criteria.[13] Who had no additional neurological or metabolic disease, any condition that would prevent testing such as severe vision, hearing, or communication problems, and who agreed to participate in the study were included. Written consent was obtained from the families and children. All children were evaluated by a pediatric neurologist before the physiotherapy assessments.

Assessments

Sociodemographic characteristics (age, sex) of the children with NF1 were recorded.

Gross Motor Function Measurement (GMFM): GMFM is a widely used scale developed by Palisano et al[4] to evaluate and follow-up the evolution of the gross motor functions of children with CP. GMFM is a valid and reliable scale.[5] It's validity and reliability have been determined in several neuromotor diseases such as DS. There are two versions of the GMFM. The GMFM-88 is the original 88-item measure and GMFM-66 is a 66-item subset of the original 88 items. Items span the spectrum of gross motor activities in five dimensions: lying supine-prone (GMFM-A), sitting (GMFM-B), crawling (GMFM-C), standing (GMFM-D), walking, running and jumping (GMFM-E). GMFM is scored as four-point likert between "0" (cannot initiate) and "3" (completed) and calculated as percentage. The score of each section can be used alone or the total score can be calculated. The scale ranging from 0 to 100.[4] In this study the Turkish version of GMFM-88[7] was applied according to the previously translated manual by a 13-years experienced pediatric physiotherapist who had completed the one-day GMFM course. The

GMFM-88 was chosen because more detailed evaluation of motor function was required.

The Manual Muscle Test (MMT): It is a valid and reliable test used both in typically developing and disabled children worldwide. MMT's validity was shown in children with disabilities as Duchenne Muscular Dystrophy[14], Spina Bifida[15] and CP in previous studies.[16] Muscle Test has also been used as a criteria in SMA validation of GMFM.[8] Additionally, manual muscle testing is an assessment method was used for children with NF1 before.[17] The measurement is made when the extremities and body complete the movement against gravity and resistance. There are different scoring systems in MMT. In this study, the "0" (no contraction) to "5" (completes the movement against gravity and takes maximum resistance) scale was applied according to the Medical Research Council.[16] Shoulder flexors, elbow flexors and extensor muscles in upper extremities and hip extensor, hip abductor, knee extensor muscles were selected for lower extremity. All muscles were evaluated in standardized test positions.

Statistical analysis

In the power analysis (80% power and 95% confidence interval), the number of individuals to be included in the study was determined as at least 40 individuals.

Software package program SPSS 23 (SPSS Statistics for Windows, Version 23.0. IBM Corporation Armonk, NY, USA) was used for demographic data, and concurrent validity analysis.

Kolmogorov Smirnov test was performed to examine whether numerical variables were normally distributed. As a descriptive statistic in numerical variables, depending on nonparametric distribution median (minimum-maximum) values were used.

Concurrent validity was analyzed with Spearman's rho correlation coefficient between GMFM-88 subdomain scores and MMT (r=0.10–0.29 as weak, r=0.30–0.49 as moderate, and r≥.50 as strong correlation).[18]

RESULTS

The study is completed with 40 children (20 male, 20 female) with NF1. The mean age of the children was 9.7±3.81 years. Descriptive of the GMFM and MMT is given in Table 1.

A positive moderate to strong correlation is found between MMT and GMFM-88 subdomains (r=0.317-0.668; p<0.05). There was a positive moderate to strong correlation between global GMFM score and shoulder flexors (left: r=0.668, p<0.001; right: r=0.604, p<0.001), elbow flexors (left: r=0.461, p=.004; right: r=0.550, p<0.001), elbow extensors (left: r=0.507, p=0.001, right: r=0.575, p<0.001), hip extensors (left: r=0.587, p<0.001, right: r=0.607, p<0.001), hip abductors (left: r=0.641, p<0.001, right: r=0.650, p<0.001), knee extensors (left: r=0.336, p=0.037, right: r=0.336, p<0.037). The correlation between MMT and GMFM-88 subdomains is given in the Table 2.

Table 1. Descriptive of the Gross Motor Function Measurement-88 (GMFM-88) and Manuel Muscle Test (MMT) in children with neurofibromatosis type 1 (NF1).

	Right	Left
	Median (min-max)	Median (min-max)
Muscles		
Shoulder flexor	4 (3-5)	4 (3-5)
Elbow flexor	5 (3-5)	5 (3-5)
Elbow extensor	5 (3-5)	5 (3-5)
Hip flexor	4 (2-5)	4 (2-5)
Hip extensor	3 (2-5)	3 (2-5)
Hip abductor	4 (2-5)	4 (2-5)
Knee extensor	5 (3-5)	5 (3-5)
	Median (min-max)	
GMFM-88		
GMFM-A	96 (92-100)	
GMFM-B	95 (90-100)	
GMFM-C	92.5 (85-100)	
GMFM-D	84.5 (69-100)	
GMFM-E	80,5 (61-100)	
GMFM-T	91.5 (83-100)	

GMFM: A) Lying and rolling, B) Sitting, C) Crawling and kneeling, D) Standing, E) Walking, running and jumping.

DISCUSSION

Gross Motor Function Measurement was used in children with different neuromuscular diseases to evaluate motor functions and to monitor motor development. It has not

Table 2. Correlations between Gross Motor Function Measurement-88 (GMFM-88) subdomains and Manuel Muscle Test.

		GMFM-A	GMFM-B	GMFM-C	GMFM-D	GMFM-E	GMFM-T
Right							
Shoulder flexor	rho	0.485	0.511	0.512	0.591	0.648	0.668
	p	0.002*	0.001*	0.001*	<0.001	<0.001	<0.001
Elbow flexor	rho	0.443	0.525	0.508	0.329	0.458	0.461
	p	0.005*	0.001*	0.001*	0.044*	0.004*	0.004*
Elbow extensor	rho	0.402	0.508	0.495	0.454	0.511	0.507
	p	0.014*	0.001*	0.002*	0.005*	0.001*	0.001*
Hip flexor	rho	0.232	0.226	0.240	0.321	0.262	0.243
	p	0.155	0.166	0.141	0.046*	0.107	0.135
Hip extensor	rho	0.499	0.417	0.406	0.579	0.547	0.587
	p	0.001*	0.008*	0.010*	<0.001	<0.001	<0.001
Hip abductor	rho	0.463	0.443	0.437	0.655	0.596	0.641
	p	0.003*	0.005*	0.005*	<0.001	<0.001	<0.001
Knee extensor	rho	0.194	0.317	0.328	0.261	0.359	0.336
	p	0.236	0.050*	0.042*	0.109	0.025*	0.037*
Left							
Shoulder flexor	rho	0.439	0.459	0.460	0.499	0.577	0.604
	p	0.005*	0.003*	0.003*	0.001*	<0.001	<0.001
Elbow flexor	rho	0.514	0.594	0.579	0.424	0.548	0.550
	p	0.001*	<0.001	<0.001	0.006*	<0.001	<0.001
Elbow extensor	rho	0.508	0.594	0.582	0.530	0.573	0.575
	p	0.001*	<0.001	<0.001	<0.001	<0.001	<0.001
Hip flexor	rho	0.232	0.226	0.240	0.321	0.305	0.291
	p	0.155	0.166	0.141	0.046*	0.059	0.072
Hip extensor	rho	0.498	0.415	0.404	0.570	0.563	0.607
	p	0.001*	0.009*	0.011*	<0.001	<0.001	<0.001
Hip abductor	rho	0.442	0.414	0.408	0.618	0.617	0.650
	p	0.005*	0.009*	0.010*	<0.001	<0.001	<0.001
Knee extensor	rho	0.194	0.317	0.328	0.261	0.359	0.336
	p	0.236	0.050*	0.042*	0.109	0.025*	0.037*

*p<0.05. rho: Spearman's correltion coefficient. A) Lying and rolling, B) Sitting, C) Crawling and kneeling, D) Standing, E) Walking, running and jumping.

previously used in children with NF1, to our knowledge. In our preliminary study, we aimed to investigate the validity of the GMFM in a sample of Turkish children with NF1. According to our results GMFM has an acceptable concurrent validity in children with NF1.

We used MMT to evaluate validity of the GMFM. There was moderate to strong correlation (r=0.317-0.668) between GMFM-88 dimensions and MMT. The biggest correlation was between hip abductor muscles and GMFM-D subdomain (r=0.655). Also, it is found that proximal muscle groups in both upper and lower limbs (especially in lower limbs) were weaker than other muscle groups according to MMT in children with NF1. Cornett et al.[19] investigated muscle weakness with 15 muscle groups in children with NF1 and stated reduced muscle force in all muscle groups ranging from 3% (tip pinch) to 43% (hip abductors) in children with NF1. Nelson et al.[9] investigated the validity of the GMFM-88 with muscle test in children with SMA. They included 40 children with SMA aged between 5-18 years in their study. Quantitative muscle testing was applied to bilateral grip, knee extension, knee flexion and elbow flexion

muscles. GMFM-88 was used as total score and percentage values were not calculated. The study showed the GMFM-88 was valid and sensitive in measuring motor function in children with SMA. Sato et al.[11] included 41 children with genetically diagnosed FCMD aged ranged 0.6–24.4 years in the validity and reliability study of GMFM-88. They stated GMFM-88 as a useful, valid and reliable measure as assessed by Spearman's correlation coefficient between GMFM-88 and Ueda classification score. Our study is similar to previous studies as sample size, age distribution and evaluation method. Russel et al.[8] studied the validity of GMFM-88 in 123 preschool children with DS assessed twice over a 6-month period, comparing with the motor scale of the Bayley Scales of Infant Development- second edition (BSID-II). They observed the GMFM-88 was relatively more sensitive to changes in gross motor function than the motor scale of the BSID-II, demonstrated better evidence of reliability and validity in children with DS. Linder-Lucht et al.[11] investigated the validity of GMFM-88 in 73 children and adolescents with TBI in a multicenter trial. They compared the parental scores, video assessments and physiotherapists' GMFM-88 scores for validity and examined test-retest reliability. They found the GMFM-88 is a reliable and valid measurement to evaluate gross motor function in children and adolescents with TBI. Salavati et al.[9] modified the GMFM-88 for children with both spastic CP and CVI: they showed GMFM-88 adapted for CVI is a useful and reliable instrument for pediatric physical therapists who work with CP.

In this study we aimed to investigate validity of the GMFM in children with NF1 and according to our results GMFM showed acceptable validity in children with NF1.

Limitations

This study has some limitations. First, all participants were between 5-17 years and ambulatory yet, it is recommended that in future studies younger age, different types of severity children with NF1 and different types of children with NF should be included. The second, MMT is a valid measurement for assessing muscle force but it is not detecting minimal differences. Further researches may be use hand held dynamometer to show minimal changes in muscle force. Third we only investigated GMFM-88 validity so in other studies responsiveness and reliability of the GMFM-88 may be study in children with NF1.

Conclusion

The GMFM-88 is a valid measurement for evaluating gross motor functions in children with NF1. Identifying motor developmental delays in children with NF1 will be a guide for establishing early intervention programs and determining symptom-specific rehabilitation goals. We recommend the use of GMFM-88 in children with NF1 for evaluating gross motor function.

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REFERENCES

1. Gutmann DH, Ferner RE, Listernick RH, et al. Neurofibromatosis type 1. Nat Rev Dis Primers. 2017;3:1-17.
2. Rietman AB, Oostenbrink R, Bongers S, et al. Motor problems in children with neurofibromatosis type 1. J Neurodev Disord. 2017;9:1-10.
3. Miller DT, Freedenberg D, Schorry E, et al. Health supervision for children with neurofibromatosis type 1. Pediatric. 2019;143.
4. Lorenzo J, Barton B, Acosta MT, et al. Mental, motor, and language development of toddlers with neurofibromatosis type 1. J Pediatr, 2011;158:660-665.
5. Palisano R, Rosenbaum P, Walter S, et al.

Development and reliability of a system to classify gross motor function in children with cerebral palsy. Dev Med Child Neurol. 1997;39:214-223.
6. Lee BH. Relationship between gross motor function and the function, activity and participation components of the International Classification of Functioning in children with spastic cerebral palsy. J Phys Ther Sci. 2017;29,1732-1736.
7. Kerem-Günel M. Kaba Motor Fonksiyon Ölçütü (KMFÖ-66 & KMFM-88) Kullanıcı Kılavuzu. Kaba Motor Fonksiyon Ölçütü (KMFÖ-66 & KMFM-88) Kullanıcı Kılavuzu, ed. M. Kerem-Günel. 2019, Ankara: Hipokrat.
8. Russell D, Palisano R, Walter S, et al. Evaluating motor function in children with Down syndrome: validity of the GMFM. Dev Med Child Neurol. 1998;40:693-701.
9. Nelson L, Owens H, Hynan LS, et al. The gross motor function measure™ is a valid and sensitive outcome measure for spinal muscular atrophy. Neuromuscul Disord. 2006;16:374-380.
10. Salavati M, Krijnen WP, Rameckers EAA, et al. Reliability of the modified gross motor function measure-88 (GMFM-88) for children with both spastic cerebral palsy and cerebral visual impairment: a preliminary study. Res Dev Disabil. 2015;45:32-48.
11. Sato T, Adachi M, Nakamura K, et al. The gross motor function measure is valid for Fukuyama congenital muscular dystrophy. Neuromuscul Disord. 2017;27:45-49.
12. Linder-Lucht M, Othmer V, Walther M, et al. Validation of the Gross Motor Function Measure for use in children and adolescents with traumatic brain injuries. Pediatrics. 2007;120:880-886.
13. DeBella K, Szudek J, Friedman JM. Use of the national institutes of health criteria for diagnosis of neurofibromatosis 1 in children. Pediatrics. 2000;105:608-614.
14. Florence JM, Pandya S, King WM, et al. Intrarater reliability of manual muscle test (Medical Research Council scale) grades in Duchenne's muscular dystrophy. Phys Ther. 1992;72:115-122.
15. Mahony K, Hunt A, Daley D, et al. Inter-tester reliability and precision of manual muscle testing and hand-held dynamometry in lower limb muscles of children with spina bifida. Phys Occup Ther Pediatr. 2009;29:44-59.
16. Manikowska F, Chen BPJ, Jóźwiak M, et al. Validation of Manual Muscle Testing (MMT) in children and adolescents with cerebral palsy. NeuroRehabilitation. 2018;42:1-7.
17. Helmers KM, Irwin KE. Physical therapy as conservative management for cervical pain and headaches in an adolescent with neurofibromatosis type 1: a case study. J Neurol Phys Ther. 2009;33:212-223.
18. Akoglu H. User's guide to correlation coefficients. Turk J Emerg Med. 2018;18:91-93.
19. Cornett KM, North KN, Rose KJ et al. Muscle weakness in children with neurofibromatosis type 1. Dev Med Child Neurol. 2015;57:733-736.