

EVALUATION OF A GIRL WITH 16p13.11 MICRODUPLICATION SYNDROME ACCORDING TO THE INTERNATIONAL CLASSIFICATION OF FUNCTIONING, DISABILITY AND HEALTH PERSPECTIVES

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Primljen/Received 11. 10. 2022.

Prihvaćen/Accepted 30.11. 2022.

Abstract

Objective: To present the functional status of a child with 16p13.11 microduplication syndrome by evaluating it under the International Classification of the World Health Organization's International Framework for Functioning, Disability and Health (ICF).

Case Description: An 11-year-old girl with 16p13.11 microduplication syndrome was assessed using the tools classified according to ICF for Children and Youth (ICF-CY) categories to evaluate body function, activity participation, and environmental factors. There was a wide range of problems, from body functions to activity participation and environmental factors. Besides these problems, there were social and cognitive disorders as well.

Conclusion: Physical and cognitive problems in body function and activity together constitute great barriers to participation in daily life.

Keywords: 16p13.11 microduplication syndrome, ICF, activity, participation, environmental factors

INTRODUCTION

16p13.11 microduplication syndrome is a rare chromosomal disorder associated with copy number variation on chromosome 16p13.11 locus (1). It is characterized by intellectual disability, behavioral disorders such as attention-deficit/hyperactivity, and an autism spectrum disorder. Other neurodevelopmental impairments include feeding disorder, gross motor

retardation, and seizure (1). The daily activities of children with this syndrome are generally adversely affected (2). Motor activities such as rolling, walking, and sitting may take longer to develop in these children. Children with this syndrome generally receive physical therapy (3). In addition, studies have shown that some children with this syndrome have speech delay, limited use of language, and repetitive speech (3). Fine motor skills, like grasping toys and holding a bottle have also been found to be affected (2). There are studies in the literature on children with 16p13.11 microduplication syndrome, but there is no evaluation made according to the ICF approach (4,5).

CASE REPORT

An 11-year-old girl presented to our Pediatric Neurology outpatient clinic with an intellectual disability and seizures. First parental concerns in terms of developmental milestones were noticed at the age of 8 months when she couldn't hold her head. She could control her head and sit with and without support at 8 months, 12 months, and 24 months, respectively. She was able to transfer objects from one hand to another at the age of 6 years when she also began to stand up from the floor with support. The family noted stereotypical hand movements for the last 3 years. Purposeful hand movements were not age-appropriate and complicated with task-oriented fine-motor activities. Further, she began to have seizures during sleep with sudden laughing and shouting attacks lasting up to 3 hours. During the last years, stereotypical movements, obsessive-compulsive behavior, hand clapping, and biting became prominent while the seizure frequency subsequently decreased. She was able to sit independently; however, she could not walk by herself. She had a spastic diplegic gait with support. She had no verbal output and did not respond to her name. In general, it was observed that the patient was highly dependent on the family in activities of daily living.

The patient presented to the genetic department when she was 6.5 years old. She had features reminiscent of Rett syndromes, such as hyperventilation episodes, postural abnormalities, stereotypical hand movements, teeth-grinding, sudden laughing and screaming attacks, task-related postural and intentional tremors, and insensitivity to pain.

After the patient's mother had been informed, the family agreed to involve her in the research, and her mother allowed us to evaluate the child.

Assessment

The evaluation was made via the Zoom program as a tele-assessment due to the current pandemic conditions. To evaluate this child, we decided on assessment tools, then categorized these tools according to the International Classification of Functioning, Disability, and Health-Children and Young major categories (Table 1).

Assessment Tools

Visual Analog Scale (VAS): The VAS was used to assess the child's pain level (6).

Viking Speech Scale (VSS): The VSS was used to evaluate the child's speech (7).

Gross Motor Function Measurement- 88 (GMFM-88): GMFM-88 was used to assess gross motor function (8).

Pediatric Evaluation of Disability Inventory (PEDI): PEDI was used to assess the child's activities and participation in daily living (8).

Eating Drinking Ability Classification System (EDACS): EDACS was used to assess the child's eating and drinking level (8).

European Child Environment Questionnaire (ECEQ): The ECEQ was used to assess factors in the child's environment (8).

RESULTS

Body Function

The child's pain level was 4 out of 10 according to **VAS**, and it did not affect her daily life. It was also observed that the patient had a severe impairment in intellectual function. However, in general, she is also interacted in the environment. Since the child had severe personal-social effects and autistic findings, her reaction to the environment was limited, and she did not return when called by her name.

Activity and Participation

According to the **VSS** performed to evaluate speech, her level was at level IV. At this level, the child does not have intelligible speech.

In the **GMFM-88** evaluation, only the sitting, standing, walking, running, and jumping sections were evaluated. Scores from these sections were determined and converted to percentages. According to the evaluation, the highest score was in the sitting section, while the lowest score

was in standing. However, it was found that the lowest percentage score was in the walking, running, and climbing sections (GMFM-88 and PEDI scores are shown in the figures below).

The total score for **PEDI** including self-care, mobility, and social function was 49, while the total score for caregiver-assisted was 7. According to the results of this scale, we see that the child was insufficient to do her daily activities on her own, and at the same time, she was mostly dependent on her mother to do these activities.

The patients eating and drinking skills were determined as level III according to **EDACS**. This means that the child eats and drinks with limitations in terms of safety, and some limitations in effectiveness might exist.

Environmental Factors

Environmental factors were evaluated as facilitators or barriers. According to **the ECEQ**, education and training services, accessibility of public buildings, income level of family, individual attitudes of immediate family members, auxiliary products, health systems, and policies were facilitators, whereas unfamiliar people, cognitive problems, and social attitudes were barriers.

DISCUSSION

This study aimed to evaluate the child with 16p13.11 microduplication syndrome according to the ICF approach. During this process, not only the patient's body function but also the social function of the person was taken into consideration. It is difficult to be objective and cooperate with the patient in the evaluations because autistic findings, intellectual disability, and personal-social areas are affected in this child. Different assessment tools were applied to the patient considering various factors.

Gross motor skills are an indispensable part of the activity, and motor impairments adversely affect participation in daily life. Considering the child's activity, she could sit and stand on her own but could not walk or run independently and could not go up and down stairs. It was also noticed that she could not communicate effectively and could not eat or drink alone. In studies conducted in children with this syndrome, it has been determined that there are developmental, cognitive, behavioral, and intellectual problems just like in our study (1-7).

Participation is a considerable element and emerges as a result of the interaction between body functions, structures, activities, and environmental factors. Participation in daily activities contributes to the development of children with and without disabilities and has a crucial

relationship with health and well-being. In our study, it was observed that the child was highly dependent on her mother for her activities in daily life. Being dependent on family or environment in daily life can be considered the most important problem in front of participation.

The ICF includes environmental factors such as recognition of the important role of the environment in people's functioning. These factors can be from physical factors to social factors. Interaction with environmental factors is an essential aspect of the scientific understanding of 'functioning and disability. In this study, ECEQ was used to evaluate the effectiveness of environmental factors. The social attitude was a barrier for the child, while the accessibility of the structures within the society was a facilitator. Whereas the child's parents were satisfied with the education given to the child at school, they thought the social assistance provided to them was not sufficient.

In conclusion, this child with 16p13.11 microduplication syndrome serves as an example to demonstrate the difficulties in a comprehensive evaluation of patients affected with various symptoms, including autistic features, intellectual disability, seizures, and stereotypical movements in combination under the heading of neurodevelopmental disorders.

Acknowledgment None.

Conflict of Interests: The authors declare no conflicts of interest related to this article.

Funding: None

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Sažetak

**EVALUACIJA DEVOJČICE SA SINDROMOM MIKRODUPLIKACIJE 16p13.11
PREMA MEĐUNARODNOJ KLASIFIKACIJI FUNKCIONISANJA,
NESPOSOBNOSTI I ZDRAVLJA**

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Cilj: Prikaz procene funkcionalnog statusa deteta sa 16p13.11 mikroduplicacionim sindromom prema međunarodnoj klasifikaciji Međunarodnog okvira za funkcionisanje, nesposobnosti i zdravlja (ICF) Svetske zdravstvene organizacije.

Prikaz slučaja: Jedanaestogodišnja devojčica sa sindromom mikroduplicacije 16p13.11 je pregledana korišćenjem instrumenata klasifikovanih prema kategorijama MKF za decu i omladinu (ICF-CI) za procenu funkcije tela, učešća u aktivnostima i faktora okoline. Postojao je širok dijapazon problema, od telesnih funkcija do učešća u aktivnostima i faktora okoline. Pored ovih problema, postojali su i socijalni i kognitivni poremećaji.

Zaključak: Fizički i kognitivni problemi u funkcionisanju i aktivnosti tela zajedno predstavljaju velike prepreke za učešće u svakodnevnom životu.

Ključne reči: 16p13.11 sindrom mikroduplicacije, ICF, aktivnost, učešće, faktori sredine

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How to cite this article: Fidan H, Kerem Gulen M, Haliloğlu G, Ütine GE, Kiper POS. Evaluation of a girl with 16p13.11 microduplication syndrome according to the International Classification of Functioning, Disability and Health Perspectives. Sanamed. Online First, December 2022. Doi: 10.5937/sanamed0-40595

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Table 1. Assessment tools categorized by ICF-CY major categories.

Body Function	Activity and Participation	Environmental Factors
VAS	VSS	ECEQ
Modified Hoehn & Yahr Scale	GMFM-88	
	PEDI	
	EDACS	

VAS, Visual Analog Scale; VSS, Viking Speech Scale; ECEQ, European Child Environment Questionnaire; GMFM-88; Gross Motor Function Measurement-88; PEDI, Pediatric Evaluation of Disability Inventory; EDACS, Eating Drinking Ability Classification System

Table 2. Codes, qualifiers, and assessment methods of body functions, activities, participation, and environmental factors

Body Functions (b)	Codes	Qualifiers	Assessment Methods
	b117.3	Intellectual functions	Observational
Activity and Participation (d)	Codes	Qualifiers	Assessment Methods
	d330.4	Speaking	VSS
	d4103.1	Sitting	GMFM-88
	d4104.2	Standing	GMFM-88
	d4158.1	Maintaining the sitting position	GMFM-88
	d4154.2	Maintaining the standing posture	GMFM-88
	d4301.3	Move objects by hand	PEDI
	d4400.3	Holding objects	PEDI
	d4401.3	Grasp	PEDI
	d4403.3	Drop an object	PEDI
	d4450.3	Pushing an object	PEDI
	d4452.3	Reach an object	PEDI
	d4500.2	Walking a short distance	GMFM-88
	d4552.4	Running	GMFM-88
	d4600.3	Getting around the house	PEDI
	d5100.3	Washing body parts	PEDI
	d5201.3	Dental care	PEDI
	d5300.3	Regulation of urination	PEDI

	d5400.3	Wearing clothes	PEDI
	d550.2	Eating	EDACS
	d560.2	Drinking	EDACS
	d7601.2	Child-parent relationship	PEDI
Environmental Factors (s)	Codes	Qualifiers	Assessment Method
	e1150+2	Auxiliary products and technology for personal use in daily life	ECEQ
	e1201+2	Auxiliary products and technology for personal use in mobility and transport inside and outside the home	ECEQ
	e1251+2	Auxiliary products and technology for communication	ECEQ
	e1301+1	Auxiliary products and technology for education	ECEQ
	e1500+2	Design, construction, and construction products, and technology for the entrance and exit of public buildings	ECEQ
	e1501+2	Design, building, and construction products and technology to	ECEQ

		increase the facilities inside public buildings	
	e310+3	Close family	ECEQ
	e340+1	Personal caregivers and personal helpers	ECEQ
	e460.2	Social attitude	ECEQ
	e5880+1	Health service	ECEQ
	e5801+2	Health systems	ECEQ
	e5802+2	Health policies	ECEQ
	e5850+2	Education and training services	ECEQ
	e5851+2	Education and training systems	ECEQ
	e5852+2	Education and training policies	ECEQ

VAS, Visual Analog Scale; VSS, Viking Speech Scale; GMFM-88, Gross Motor Function Measurement-88; PEDI, Pediatric Evaluation of Disability Inventory; EDACS, Eating Drinking Ability Classification System; ECEQ, European Child Environment Questionnaire