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Original Article

Assessment of functional capacity of teens who are Mucopolysaccharidosis type II carriers

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Abstract: Introduction: Mucopolysaccharidosis II (MPS II) is an X-linked genetic disease which almost exclusively affects males. The disease presents a multisystem form, caused by the progressive accumulation of glycosaminoglycans (GAGs) in tissues and organs. It causes developmental delays resulting in limitations on the performance of day-to-day activities. Objective: The aim of this study was to identify occupational performance areas in which people with MPS II have greater impairment and analyze their family's socioeconomic profile. Method: The research was done through interviews with an individual responsible for three adolescent brothers, (A1-19 years, A2-16 years and A3-15 years) who are MPS II carriers from Alagoas, Brazil. Data collection was performed by a trained professional using the Functional Independence Measure (FIM) as a questionnaire to identify the functional losses and the ABEP for socioeconomic profile analysis. Results: The results showed that the family is included in the C1 socioeconomic class. Among the siblings, A1 presents total dependence on the investigated skills; A2 presents the most disabling functional losses requiring moderate assistance and A3 has much of his physical and cognitive skills preserved, but has modified dependence. Conclusion: Taking into consideration the difficulties identified in this study, we highlighted the importance of early diagnosis and rehabilitation therapies combined with enzyme therapy, which can delay disease progression.

Keywords: *Mucopolysaccharidosis II, Reabilitation, Occupational Therapy.*

Avaliação da capacidade funcional de adolescentes portadores de Mucopolissacaridose do tipo II

Resumo: Introdução: A Mucopolissacaridose II (MPS II) é uma doença genética, ligada ao cromossomo X e que afeta quase que exclusivamente o sexo masculino. Apresenta-se de forma multissistêmica, ocasionada pelo acúmulo progressivo de glicosaminoglicanos (GAG) em tecidos e órgãos, o que causa atraso no desenvolvimento neuropsicomotor, ocasionando limitações na realização de atividades de vida diária. Objetivo: O objetivo deste estudo foi a identificação das áreas de desempenho nas quais os portadores de MPS II apresentam maior comprometimento, além da análise do perfil socioeconômico da respectiva família. Método: A pesquisa se deu através de entrevista com a responsável por três irmãos adolescentes, A1-19 anos, A2-16 anos e A3-15anos, portadores de MPS II, no Estado de Alagoas. A coleta foi efetuada por profissional capacitado, utilizando a Medida de Independência Funcional (MIF) como questionário para identificação de perdas funcionais, e ABEP, para análise do perfil socioeconômico. Resultados: Os resultados apontaram que, em termos socioeconômicos, a família está inserida na classe C1. Em relação ao desempenho, verificou-se que, entre os irmãos, A1 apresenta dependência total nas habilidades investigadas; A2 apresenta perdas funcionais mais incapacitantes, necessitando de auxílio moderado, e A3 possui

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boa parte das habilidades físicas e cognitivas preservada, porém possui dependência modificada. Conclusão: Diante das dificuldades identificadas neste estudo, foi analisada a importância do diagnóstico precoce e das terapias de reabilitação aliadas à terapia enzimática, para que haja um retardo na progressão da doença.

Palavras-chave: *Mucopolissacaridose II, Reabilitação, Terapia Ocupacional.*

1 Introduction

According to Camelier (2011), metabolism, inborn errors (MIE) are genetically determined diseases, caused by a specific enzyme defect, which leads to blocking a particular metabolic route.

Currently, the most widely used classification divides the mucopolysaccharidoses in seven subtypes, based on clinical findings and laboratory tests (NEUFELD; MUENZER, 2001). Even showing chronic and progressive typical characteristics, clinical manifestations of each subtype vary according to the enzyme that is missing in the carrier of the disease, which determines the type of Mucopolysaccharidosis (ROCHA et al., 2012).

Charles Hunter was the first reporter of Mucopolysaccharidosis type II (MPS II) in 1917, describing specific features of this type of MPS (JONES, 1998).

> Mucopolysaccharidoses (MPS) are a group of genetic disorders caused by a deficiency of Lysosomal enzymes responsible for a specific step in the degradation of glycosaminoglycans (GAG). It presents X-linked inheritance, being caused by deficient activity of the enzyme iduronate sulfatase, with consequent increase of the urinary concentration of GAG dermatan sulfate and heparan sulfate (PINTO et al., 2006, p. 274).

MPS II is the most common type in Brazil and most of the people involved is male, although there are reports of occurrence in females. It is rare and characterized by an abnormal translocation or a non-random X chromosome inactivation (TUSCH et al., 2004 apud PEREIRA et al., 2011). Boy and Schwart (2011) claim that MPS II has an approximate incidence of 0.31 to 0.71 per 100000 live births.

It is worth mentioning the genetic and progressive characteristic of MPS II, which causes risk of recurrence in the same family and even in the same offspring, which makes essential orientation and genetic tracking for families that already have a member diagnosed (ROCHA et al., 2012).

According to Boy and Schwart (2011), mucopolysaccharidoses II is characterized by precocious manifestations, which consist of delayed language development, school performance, behavior disorder and mental retardation. The skeletal changes consist of skeletal and vertebral malformations, and joint stiffness. Respiratory impairment and hepatosplenomegaly, umbilical and inguinal hernias which are also frequent. Changes in skin and heart diseases are also frequent manifestations.

It is important to realize the early diagnosis of Mucopolysaccharidosis by enzyme analysis and molecular studies, in order to allow an adequate genetic monitoring and support to the family in early stages of the disease (PEREIRA et al., 2011).

The differential diagnosis should be made with the other mucopolysaccharidoses and other Lysosomal diseases; for this, it is necessary to carry out additional tests and enzyme specific dosages (PINTO, 2005).

There is no curative treatment for MPS. The therapeutic options available include interventions at the level of clinical phenotype, such as surgeries to correct hernias and adenoids, ventilatory support, etc. (BOY; SCHWART, 2011). The treatment of MPS II is symptomatic and supportive, but there have been reports of the use of steroids and bone marrow transplant; However, today, the most common is the enzyme replacement, by which the child receives the enzymes that are deficient in their body (PEREIRA et al., 2011).

According to Giugliani et al. (2010), the accumulation of GAG occurs in various organs and tissues in patients with Mucopolysaccharidosis II, resulting in a series of signs and symptoms, which are part of a multysistemic clinical scenario, which compromises bones and joints, respiratory system, cardiovascular system and many other organs and tissues, including, in some cases, cognitive functions. According to Guarany (2011), these clinical manifestations cause several limitations in these patients' daily living activities (DLA). The development of chronic diseases during life can result in a continuous loss of organic and biological functions, which can lead to functional incapacity.

For Neri (2001), functional independence is the capacity to perform activities by own means, that is, with no help or other people interference. It is related to mobility and functional capacity, for which the individual does not require help to

carry out the daily life activities, i.e. independence supposes satisfactory motor and cognitive conditions to perform these tasks.

Complementing this thought, the World Health Organization (ORGANIZAÇÃO..., 2003) states that the functionality encompasses all functions of the body, the performance, activities and social participation of the individual.

For Mello and Mancini (2007), occupational therapy aims of action is, facing the patient, occupational dysfunction, which is brought in the daily life of the individual as a difficulty for the realization of any activity that is routine, regardless of whether the cause for such difficulty is physical, cognitive, social or other. The authors still complete that "[...] the ability or inability to perform ADL (activities of daily living) can be used as a practical measure to measure disability" (MELLO; MANCINI, 2007, p. 50).

Analyzing the clinical manifestations present in MPS II and realizing that these limit the functional independence of these individuals, so that their quality of life is reduced, the level of functional independence of these carriers was analyzed through the MIF so that, it is possible to identify what areas of performance are affected more quickly.

2 Method

This is a cross-sectional, descriptive study, attended by three teenagers, brothers, males with Mucopolysaccharidosis II, being one 19 years old, the other 16 years old and the last with 15 years of age, in genetic treatment by CER III (Specialized in Rehabilitation Center), located in Maceió-AL. It should be noted that these guys were the only MPS II carriers registered in the institution.

2.1 Functional Independence Measure

The application of the questionnaire was carried out in a reserved room of a philanthropic institution, in which the teenagers carry out perform phonoaudiology, physiotherapy and occupational therapy sessions, in the city of Maceió-AL, being the interview conducted by a professional adequately trained to use the instrument. At the time, only the principal investigator stayed on the scene, a research assistant and the mother of the person searched.

The data-gathering instrument used was the functional independence measure (FIM), which, according to Riberto et al. (2012), the questionnaire aims to evaluate 18 levels of functions and their scores, which are subdivided into personal care (food, morning hygiene, bathing, dressing- upper top, dressing- bottom top, using the toilet); sphincters controls (urine control, bowel control); mobility (transferring bed-chair, transferring to the toilet, transferring shower); locomotion (horizontal locomotion, stairs); communication (comprehension, expression), and social cognition (social interaction, problem solving, memory).

The answers are objective and classified as 1 to 7, varying according to the level of help that is need in carrying out that activity, being: 7-complete independence; 6-modified independence; 5-supervision; 4-minimum help (75% of the activity is held >); 3- moderate help (50% of the activity is held >); 2-maximum help (25% of the activity is held >) and 1-total help.

2.2 Socio-economic classification

To define the social situation of the subjects involved in the survey, we used the criterion of Economic Classification of Brazil, 2013 version (ASSOCIAÇÃO..., 2013), that categorizes individuals, as the score obtained in the economic classes A1, A2, B1, B2, C1, C2, D and E, which consists of a sum established from the possession of consumable items (tv, radio, bathroom, car, refrigerator and others) and the level of education of the head of the family, so that the greater the number of points obtained, the closer to family is to the socioeconomic class A and, in the other direction, the lowest score corresponds to the class E. this classification was used for, among other things, relating the socio-economic situation and the late diagnosis, taking into account the access to health services and general information about the disease.

Data collection counted with a qualified researcher for application of the questionnaire. As part of the search procedure, the respondent was informed about the objectives of the study and asked to express their agreement, with the signature of informed consent (TFCC) as Bioethical standards of research in Human Beings of the Ministry of Health of Brazil. This study was reviewed and approved by a Research Ethics Committee of the Universidade Estadual de Ciências da Saúde de Alagoas-UNCISAL, with the record Protocol ZIP CODE No. 33297014.3.0000.5011.

3 Results

For this study, three individuals were assessed, brothers, patients with Mucopolysaccharidosis II, male, black and identified by codes: A1-19 years;

A2-16 years and A3-15 years. They live in Teotônio Vilela a municipality of Alagoas, which is approximately 101 km from Maceió, where the therapies (enzymatic therapy, monitoring with geneticist and other medical specialties, occupational therapy, physiotherapy and speech therapy) are carried out, once a week.

All of them were referred to a geneticist only when A1 was already seven years old, due to similar symptomatology of the brothers; on occasion, A2 was four years old and A3 three years old.

According to economic classification proposed by the Brazilian Association of Research (ASSOCIAÇÃO…, 2013), the family of the teens studied is inserted in the C1 class, comprising an average income of R $1,685.00, as the instrument used.

They receive government social benefit, the benefit of Continued Provision of the Organic Law of Social Assistance (BPC/LOAS), granted by the Instituto Nacional de Seguridade Social – INSS to those that present some crippling deficiency or have more than 65 years, proving they do not have means of providing their own maintenance and their family. This benefit has direct cooperation with the family maintenance.

Table 1 describes the performance areas evaluated through the MIF, obtained through interview with the mother of the teenagers, by which, through the results, it was observed the progressive condition of the disease before the dependency in activities of daily living (ADL) and instrumental activities of daily living (IADL).

Considering the order of birth, the eldest son, A1 (19 years), presents total dependency on the skills investigated, spending most of the day in his bed. The teenager, identified as A2 (16 years) presents more disabling functional losses, requiring moderate help, and the teenager A3 (15 years) has still many of the physical and cognitive skills preserved, being the only one that goes to school.

4 Discussion

The present research aimed to assess the level of functional independence of patients with Mucopolysaccharidosis II through a cross-sectional study, to analyze the occupational performance of subjects involved in the study and contribute to the understanding of the specific therapeutic needs of individuals with MPS II.

As for the result obtained on the socioeconomic status of the family and the information about the place where they reside, associated with poor schooling, it is observed that such reality directly affects access to information about the disease, which may have had as a result the delay in diagnosis. It should be taken into consideration that, according to the mother of the subject, she looked for a health service to the younger brothers only because of the similar the symptoms we to the older brother's. Before this discussion, we take into account the statement of Coelho and Rezende (2007), who report that, the earlier it is detected the development delay, the faster intervention will be and, consecutively, the smaller the impact of problems in the life of this child.

According to information provided by the mother, the three teenagers are the fourth, fifth and sixth children of six offspring, being the first three female. She also reports that all are born of natural childbirth and in term, with appropriate weight for gestational age and, until two years of age, had the typical neuropsychomotor development. Then, they began to present difficulties in flexioning the fingers, with deficits in motor coordination, joint stiffness, short stature and distended abdomen. She also reported that, as the symptoms of the disease appeared, there was a loss of previously acquired skills.

According to these findings, the case reports on MPS II show that the course of the disease can get through typical development and, after a certain age, there's a regression in the activities that have been developed

Table 1. Characterization of functional capacity of adolescents evaluated by MIF.

Cathgory (reference score)	A1*	A2**	A3***
		Score obtained	
Self-care (6-42)	6	16	21
Control of sphincters (2-14)	2	8	14
Transferrings (3-21)	3	18	21
Locomotion (2-14)	2	6	8
Social communication (2-14)	2	14	14
Social cognition (3-21)	4	5	11
MIF total	19 (complete dependence)	67 (modified dependence)	89 (modified dependence)

A1* - adolescent 1, 19 years; A2** - adolescent 2, 16 years; A3*** - adolescent 3, 15 years.

independently, or presents backwardness since the beginning of their development (PEREIRA et al., 2011; ROCHA et al., 2012).

Vieira et al. (2008) examined, in their research, carried out between May 2005 and November 2006, in the medical genetics service of the Hospital das Clínicas de Porto Alegre and six other medical genetics services in Brazil (five in the Southeast and one in the northeast), all the history from birth to the biochemical diagnostic of the Mucopolysaccharidoses. Of these, the MPS II is the group that presented the greatest delay in development before diagnosis and whose symptoms appeared early. MPS II was still regarded as the MPS that presented greater delay in diagnosis, because the average age for the onset of symptoms is 24 months, while the average for the diagnosis was 95 months. This study still brings that most cases of Mucopolysaccharidoses, analyzed by them, were referred to doctors geneticists and these professionals made the diagnosis of the disease, through specific tests.

It was observed, in this analysis, the brothers present progressive aid need in carrying out their activities of daily living and instrumental activities of daily living, and the older totally depends on help to perform his ADL. These results strengthen what was quoted by Guarany (2011), which states that the Mucopolysaccharidosis is a chronic and progressive disease, causing their carriers stay increasingly dependent on the realization of their activities of daily life, although its symptoms can be delayed with enzyme therapy and multidisciplinary treatment in rehabilitation.

The subject identified as A1 is the one who needs greater help to carry out his activities, obtaining minimum score in all categories, except in the memory item, social cognition, in which he scored 2, which shows he needs maximum help; while A2, 16 years old, has a total of 67 points, showing high score in social communication and lowest in social cognition, and A3, 15 years old, has 89 points, expressing greater need for help in self-care actions. The two younger brothers fit into the modified dependency scores, which shows a lack of assistance of up to 25% in the task.

The youngest guy, A3, requires more help to fulfill activities with self-care, which raises the hypothesis that this is the area of occupational performance that features initial functional loss. In support, Camargos et al. (2012) reported that, in their study, it was identified the relationship between physical function domain and self-care and mobility. The research conducted by Morini (2007) brings that, in patients with Mucopolysaccharidosis II, the deficit is greatest in upper limbs than in inferior limbs; we can associate this fact with the difficulty they present to take care of themselves, as, for that purpose, the upper limbs are needed. Still complementing with ideas of Morini (2007), he states that treatment should be focused on upper limbs, since they represent the main complaint of the subjects of their search.

Among the factors identified in this study, which hinder the implementation of the activities of daily life, is included the motor impairment and reduced muscle strength. Corroborating this idea, Guarany (2011) reports that another factor that affects the independence of these patients are the clinical manifestations of MPS II and short range of motion, in addition to the joint stiffness and contractures, also caused by the accumulation of GAG in these regions, hampering mobility, locomotion and the completion of tasks. Accordingly, Morini (2007) performs a study on Inborn Errors of Metabolism from the outpatient clinic of the Hospital das Clínicas da Universidade Estadual de Campinas, between February 2005 and May 2006, with eight individuals with Mucopolysaccharidosis II, male, between 4 and 46 years, in which all of them showed strength loss in at least one muscle group, which affected them in the execution of their daily life activities. It is known that the motor experience is directly related to cognitive learning, since discoveries become familiar through attempts, i.e. If the motor component is damaged, consequently there will be cognitive changes.

The intellectual damage was also identified as another factor that interferes with the independence of these teenagers, directly related to the understanding of what must be done.

The MPS Network Brazil (REDE…, 2011) states that the Mucopolysaccharidosis type II features accumulation of GAG in the brain, which brings as a consequence the loss of acquired skills and the delay in development; There are also other problems such as hydrocephalus, which can hinder learning, in addition to inadequate oxygen levels and sleep deprivation (caused by sleep apnea), that can affect brain function.

According to Guarany (2011), the identification of the reduction of the independence on their daily activities and restrictions on social participation enables patients with MPS can be encouraged and receive monitoring appropriate to their needs.

The subjects of this research had or have occupational roles common to children and adolescents, however, with the advancement of the MPS, symptoms receded and they lost the roles that were part of their lives. Barrett and Kielhofner's (2002) reported that occupational behavior involves mundane activities developed by human being and that, when this restrictes by some factor, the feeling of competence may be threatened. Completing this thought, Beer (2002) states that the occupational performance can be affected by active pathologies, which is evident in this study.

Occupational therapy helps people in carrying out activities that are important to them, despite their weaknesses and inefficiencies, expanding their autonomy and reinserting them in the community/family (SOARES, 2007; NEISTADT; CREPEAU, 2002).

The occupational therapist can rehabilitate the patient, through everyday activities, by evaluating the context in which the patient is inserted. Their beliefs and convictions, the environment in which he lives and the family dynamics are just some of the items analyzed by occupational therapists for planning the most appropriate treatment plan for each individual. It is also important not to overestimate or underestimate the abilities of the individual and work to increase self-esteem is key in the treatment.

According to Rogers and Holm (2002), the most widely used ingredient to analyze the level of incapacity is the degree of independence shown in performing a task. Holm, Rogers and James (2002) state that the treatment proposed by the occupational therapy is the acquisition of greater independence in the execution of their daily life activities. Pedretti and Early (2005) mention that because of the achievement of independence is the central axis of the occupational therapy process, the method of intervention shall be the achievements in the areas of performance of the patient, in order to return to their social roles.

Until the present moment, there is no cure for the Mucopolysaccharidoses and, for treatment, should be initially considered the identification, understanding and managing, analyzing each case, the needs that stand out in each patient and combine this conduct occupational therapy to the enzymatic treatment to, essentially, increase quality of life through the delay of disease progression (CAMELIER, 2011; REDE..., 2011; GIUGLIANI et al., 2010; GUARANY, 2011). Before this statement, it is possible to say that the treatment methods adopted by patients studied follow the line adopted by other researchers.

5 Conclusion

Although few studies report the motor and cognitive difficulties of patients with Mucopolysaccharidosis II, it was observed, in this research, the areas of occupational performance of these individuals suffer progressive loss, corroborating with the findings in the literature. Such losses lead these carriers to the functional dependency still in adolescence, considering that according to the results obtained in this research, self-care, social cognition and locomotion are the first areas to present performance losses, since, according to the assessment of the youngest patient, these areas are already very damaged, and his older brothers, lost the ability to perform tasks independently.

Thus, the importance of early diagnosis, aimed at establishing an immediate therapeutic intervention, with the goal of slowing functional losses, enabling the individuals affected by Mucopolysaccharidosis II to keep as much time performing their activities of daily life with the least possible help, increasing the quality of life.

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Author's Contributions

Ingrid Alves Barros Silva Amaral was responsible for the collection of data and drafting of the text. Reinaldo Luna de Omena Filho was responsible for the design and revision of the text. João Ancelmo dos Reis Neto was responsible for translating and organizing the sources. Monique Carla da Silva Reis was responsible for data collection, analysis and conception of the text. All authors approved the final version of the text.