Annals of Clinical Case Reports

Research Article
Published: 16 Dec, 2017

The Croatian Experience in Multidisciplinary Treating of Children with Muscular Dystrophy

Matijević Valentina[1,2], Kraljević Marija[1,2], Barbarić Bernarda[1,2] and Matijević Petra[3]*

[1]*Clinical Department of Rheumatology, Physical Medicine and Rehabilitation, Sestre milosrdnice University Hospital Centre in Zagreb, Croatia*

[2]*Josip Juraj Strossmayer University of Osijek – Faculty of Medicine in Osijek, Croatia*

[3]*University of Zagreb School of Dental Medicine*

Abstract

This review will briefly discuss the condition of muscular dystrophy in children and will explore the potential of multidisciplinary treating in managing the muscular dystrophy of children. Muscular dystrophy is the collective term used to represent genetic disorders characterized by progressive weakening of muscles and wasting of muscle tissue. Several forms of muscular dystrophy exist, that differ on the basis of clinical symptoms, disease severity and the way in which the disease is transmitted form one generation to the other. There are not many specialists and researchers in Croatia which are working in a multidisciplinary team with children who are suffering from muscular dystrophy. Life quality of people suffering from muscular dystrophy can be significantly improved by involving and supporting children and their parents in early intervention where they are supervised by the multidisciplinary team. Since that the aim of this research was not detecting the causes of muscular dystrophy and difference types of muscular dystrophy with their characteristics, in this report we have decided to write about the importance of multidisciplinary treatment children with muscular dystrophy and importance of early intervention. The multidisciplinary team presented in this report is considered by doctor specialist in physical medicine and rehabilitation, physiotherapist, speech therapist and psychologist. The lack of multidisciplinary treating probably happens due to poor understanding which early intervention can relate to the quality of children life in muscle disease. Therefore, the researchers have limited knowledge on which kind of multidisciplinary they should target the treatment muscle dystrophy.

Keywords: Muscular dystrophy; Therapeutic approach; Multidisciplinary team

OPEN ACCESS

Correspondence:
Matijević Valentina, Clinical Department of Rheumatology, Physical Medicine and Rehabilitation, Sestre Milosrdnice University Hospital Centre in Zagreb, Croatia;
E-mail: kraljevicmarija@yahoo.com

Received Date: *07 Oct 2017*
Accepted Date: *04 Dec 2017*
Published Date: *16 Dec 2017*

Citation:
Valentina M, Marija K, Bernarda B, Petra M. The Croatian Experience in Multidisciplinary Treating of Children with Muscular Dystrophy. Ann Clin Case Rep. 2017; 2: 1485.

ISSN: 2474-1655

Introduction

Muscular dystrophy is one group of genetic diseases in which various genes controlling muscle function are defective [1]. It's an autosomal recessively inherited disease which occurs at birth or within the first six months of life, with hypotonia, muscle weakness, contractures, and motor developmental delay. It has long been known that the clinical phenotype in this disease can vary substantially and until recently there was no explanation for this finding [2].

More than one form of muscular dystrophy existed and that these diseases affected people of either gender and of all ages [3]. The distribution of muscle weakness is different in various types of dystrophy.

With the development of diagnostics, it has become evident that muscular dystrophy is a heterogeneous entity composed of a large variety of neuromuscular disorders [4]. There are currently over 30 different types of muscular dystrophy have been recognized but generally, there are eight main types of muscular dystrophy called Duchenne muscular dystrophy, Becker muscular dystrophy, Emery-Dreifuss muscular dystrophy, limb girdle muscular dystrophy, facioscapulohumeral muscular dystrophy, distal muscular dystrophy, oculopharyngeal muscular dystrophy and congenital muscular dystrophy. The different muscular dystrophies are varied in who they affect and the symptoms of the specific type of muscular dystrophy [5]. In addition, because it is a hereditary disorder, it may recur again in some families.

The usual symptoms of muscular dystrophy besides the muscles weakness are a decrease in dexterity (quickness, readiness and grace in physical activities), increase in clumsiness, the stiffness of grip, cardiac complications and others difficulties. The cardinal symptoms are weak muscles

but weakness also occurs in the face, neck, hand and feet. New born infants with the muscular dystrophy generally have difficulties with sucking and breathing because of neonatal hypotonia, later orofacial weakness causes problems with indistinct articulation and speech [6].

Children who have speech difficulties (phonological, articulation and language) may be struggling in various aspects of everyday life such as social interaction in school and family life. This child may become isolated and withdrawn over time if these difficulties are ignored and would benefit greatly from the speech and language therapist in a multidisciplinary team. The larger number of speech and language pathologists should be available to work in the rehabilitation of these children from an early age [7].

Also, in the early stages child and their family can fall into depression and this depression can keep recurring throughout their lives due to the difficulty in coping with the disease [8]. Due to many limitations faced by muscular dystrophy patients, this disease can cause medical and social problems among the patients [8].

Even among the limited available researchers of muscular dystrophy disorder most of the researchers are focusing on the medical aspect of the disease [9]. The psychological and emotional effects of muscular dystrophy on the patients and their families are often ignored.

Many patients and their immediate families feel shocked and angry upon hearing the diagnosis so, therefore, such patients should be followed by a psychologist [10].

Professionally led early and multimodal stimulating treatment minimizes the short-term and long-term consequences of the child's neurophysiological development, and has multiple positive effects on their family and the environment, and thus allows the child to have equal or which participates more equally in society [11].

The goals of multimodal (re) habilitation of the child with muscular dystrophy are the most encouragement of all ability in accordance with psychophysical characteristics and life expectancy. Enable intellectual, physical, emotional and social development and ensure the conditions for successful integration into everyday life with the greatest possible potential degree of independence and successful socialization [11].

Materials and Methods

Every rehabilitation, including child habilitation, starts with the examination at the doctor's Office, then paediatrician, doctor specialist in physical medicine and rehabilitation and if necessarily neuropediatrician. The quality and positive results of therapy children with muscular dystrophy depends on mutual communication, continuity and regular therapy, early intervention, the motivation of child and parents and also teamwork from the primary paediatrician, child's doctor specialist in physical medicine and rehabilitation, physiotherapist, speech therapist and psychologist [12].

Methods for treatment of muscular dystrophy normally includes surgery, an orthopaedic device, physical therapy, speech and psychological therapy.

The child's doctor specialist in physical medicine and rehabilitation evaluates the spontaneous motors, active motions, qualitative analysis of general movement, the strength of the muscles, palpation and passive motility testing, reflexes testing by the Vojta principle. Doctor specialist in physical medicine and rehabilitation should have work experience in this area for at least five years or certification of completing the course of Bobath concept or Vojta principles [12].

The physiotherapist encourages the proper pattern about the movement of the child, but at the same time raises the child's neurophysiological development which is needed for the child to carry out the wanted or desired movement. The child physiotherapist used the Bobath concept which inhibits/controls the abnormal postural and locomotion pattern and simultaneous facilitation/activation of automatic postural reactions (equilibrium reactions and righting reaction). It all has an effect as there is an achievement with normal tonus and motorial functions which enables the child's variety of sensory experience in functional and purpose-directed activities [13].

Inactivation for the ontogenetic mechanisms of movement patterns Vojta therapy is applied. Vojta therapy is based on reflexive locomotion (rotation, crawling). In certain positions (supine, pertinent position and position on the hip), with the pressure from the therapeutic on specific parts (stimulating zones) cause muscle activation throughout the body. The musculature is always facilitated by the same recurring sequence, in the way it is used in an ideal motorcycle form [14].

Physical gymnastics with older children is divided according to the movement which is performed: active movement exercises, assistive active and passive exercises [11]. Electrotherapy is performed in various forms for those children where there is indicated (transcutaneous nerve stimulation, interferential current, magneto therapy, electro muscular stimulation, muscle training, muscular electrostimulation, functional electrostimulation), and thermotherapy surface modalities (paraffin, solux lamp, warm and cold compression, surface cooling ice). Because of relief, easiness or directional movement, in therapy, it is often to use orthoses, wrists, orthopedic shoes and dentures to replace the lost or entire extremities [15].

Speech and language therapist estimates the speech-language and communication development of the child as also improving communication skills of the children, which is mainly important for children with neuromotor disabilities, as they more often have a delay in their speech-language development. Based on standardized tests, the speech and language therapist evaluates understanding and speech in children with motor distortions. The most commonly used tests are the ones who investigate language comprehension, receptive vocabulary, language production and articulation. There are tests that examine the orofacial muscle weakness in children with difficulties with sucking, swallowing and chewing, as it is the case for children with muscular dystrophy. As a result of orofacial weakness, there are also difficulties in articulation and fluency in speech.

The psychologist evaluates the child's development of psycho motoric skills. According to the development gives advises to the parents on educational procedures and behaviour about the child's deviations in motor and neurophysiological development [11]. In addition, psychologist uses various types of tests to measure general intelligence and readiness for school.

Examination of physical medicine and rehabilitation specialist is 45 minutes long. Frequencies of control examination from age 0 to 16 months is every 6 (8) to 10 (12) weeks depending on functional status of the locomotion apparatus; from 16 months to 3 years every 3 to 4 months depending on functional status of the locomotion apparatus;

from age 3 to 6 every 4 to 6 months depending on the functional status locomotor device; after 6 years of age every 6 to 12 months depending on the functional status locomotor device. Paramedical examinations from the others in the multidisciplinary team: Speech and language therapist will examine every 3 to 6 months for 45 minutes, while the psychologist retests every 6 to 12 months for duration about 60 minutes [11].

Results and Discussion

In Croatia, the child's doctor specialist in physical medicine and rehabilitation is the leader of the multidisciplinary team as he/she is the first person in the chain to meet the family and the child. The doctor specialist in physical medicine and rehabilitation creates the appropriate spectrum of therapeutic interventions. It is important to recognize deviations in the motor and neurophysiological development of the child. The doctor specialist in physical medicine and rehabilitation will indicate by which kinesiotherapy stimulation procedure the child will be processed during treatment; also brings the decision of implementing the treatment in the ambulance and stationary and includes the speech and language therapists, psychologists and other specialists in the team [11].

Every rehabilitation of a child begins with the education of a parent or guardian. For continuous stimulation of neuromotor development, it is necessary to maintain and continue at home for at least two hours a day, although there is no consensus regarding it, through the motor and sensory activities appropriate to the age of the child [11,16].

After all, it cannot neglect that comprehensive non-motor stimulation is carried out throughout the course of feeding, carrying out hygiene, taking care of the child during the day, wearing, dressing and playing with the aim that the child learns the experience of a proper movement of each action which is stated above.

The goals of multimodal rehabilitation for the child is to maximally encourage of its abilities in accordance with psychophysical characteristics and life expectancy. It is necessary to enable intellectual, physical, emotional and social development as also providing conditions for successful integration into a lifetime with the highest degree of independence during it. Using the behavioural techniques as work to adopt those forms of behaviour which enables successful socialization [11].

Physiotherapist

Due to the wasting of muscle children with muscular dystrophies will struggle with many everyday activities. Physiotherapists can help with the management of the presenting neuromusculoskeletal problems. They can help slow the degression of a range of motion, muscle strength, daily function, work to improve gait pattern and posture [17].

Physiotherapy is a therapeutic science that makes use of manual skills to correct the deviations from normal health process. Muscular dystrophy is a life-disabling condition which makes children dependent upon a wheelchair in later stages of the disease, demands practice of the bodily functions and reconditioning of affected tissues. This can be primarily brought about by physiotherapy which has no side effects. An early referral from the hospital for physiotherapy is imperative to prevent unnecessary degradation of child's functioning. This disorder is disabling but consistent efforts from a team of a child and his/her physiotherapist, which is generally ignored, can help make this world a better place to live for the children having muscular dystrophy [18].

It is indispensable for the therapy to have children play in the hospital. Child's play is not only conditioned by an external stimulus but comes from a child's inner motivation. The game helps in coordination and develops proper muscle development, and in further helps children prepare for the future by playing those activities they have not equal to [19].

In a hospital environment in Croatian, children's play is often used as a form of psychotherapy. Thus, fears and anxieties can be more easily identified and hospital staff can alleviate them when it is necessary. Enabling parents to visit their children, implementing and introducing different activities with the help of professionals from different profiles facilitates a child's stay in the hospital during the therapy which at the end reduces the psychological consequences and positively influences on the mental and physical development of the child [20].

Speech therapist

Children with muscular dystrophy show orofacial muscle weakness. Preliminary evidence emerges suggests that speech, but not necessarily intelligibility is likely to be impaired when lingual weakness is severe [21].

Orofacial motor exercises are non-voice activities which involve sensory stimulation or initiation of the facial muscles, lips, jaw, tongue, soft palate, larynx and respiratory muscles which at the end affects the physiological of the oropharyngeal base in which their function is improving. Considering that people with muscular dystrophy have general muscle weakness, that weakness also affects the facial muscles responsible for chewing, swallowing and speech, so, therefore, the orofacial exercises are necessary for rehabilitation of people with muscular dystrophy [22].

The speech and language therapist is providing above mentioned exercises with children and advises parents what kind of exercises to do at home.[23] In a case study in one Clinic in Croatia, two girls with muscular dystrophy have shown great improvement after six months of intensive orofacial exercise. Progress in eliminating and reducing muscle weakness of orofacial area and significant improvement of orofacial muscle tone [23].

Feeding and swallowing problems are not uncommon in muscular dystrophies, which can lead to dehydration, malnutrition or aspiration pneumonia. In patients with chronic muscle disease, a prevalence of 35% of feeding problems and dysphagia was reported [2]. Facial muscle weakness can lead to craniofacial and dental malocclusion which aggravates problems with chewing [24]. Since that children with muscular dystrophy may have difficulties with chewing, if necessary, the speech therapist advises an expert of Dental Medicine who provides more information on oral practice to parents. Proper pronunciation of letters in most cases is conditioned with the developed oral practice. It is a prerequisite for proper articulation (pronunciation) of voice and represents the ability to voluntarily initiate the parts of the speech device (lips, cheeks, jaws, tongue, soft and hard palate). In this way, the speech organs are placed in the proper position which is necessary for correct pronunciation of voices. The maturation of oral praxis follows the physiological development of the child.

To improve their quality of life and nutritional state patients

with muscular dystrophy and also swallowing difficulties benefit from dietetic and swallowing recommendations by speech and language therapist, about safe swallowing techniques or advice about postural management and feeding aids. More liquid food is advised or alternating thick with thin consistencies, careful chewing and bolus preparation to a liquid consistency, effortful swallowing and double swallows, water after mealtime. The positioning of the head in sitting position can also be important to prevent residue [25].

Psychologist

Medical care incomplete without support for psychosocial well-being. During the medical care, it is welcomed to process counselling or other psychological help since it is not usually part of a normal medical procedure during treatment.

As the disease is acknowledged at the early stage, it is an often case and unpleasant situation where the patients hardly accept the reality that they are having an incurable illness at that moment. Patients can fall into a depression where it can develop signs of an aggressive behaviour. Problems with sleeping are in common and not foreign for patients with muscular dystrophy as psychological stress is building up [9].

Though, psychologist opinion and role is usually left unnoticed since that most of the pupil are not referred or advised to see the experts which are a specialist in the field of psychology and counselling patients during working with them.

Muscular dystrophy at the end is a rare and unique disorder for a person. Parents of children with muscular dystrophy sometimes are left feeling lonely and scared in facing all the upcoming challenges which are related and comes with the disease. Most of the time, they will centre their life to their children and by that are having rare opportunities for breaks for themselves. In some cases and some severe muscular dystrophy cases, parents face the deal with the loss of their child and have to cope with grief and anger. It is not rare that the parents deal with various feelings like guilt, shame and blame. Psychological assistance and intervention are also desired to help the parents as it's also important to the child.

Several psychotherapy techniques can help in various areas. One of them is parental respective management training which is recommended for externalising behaviours and the other technique is the individual therapy, recommended for patients with internalising behaviours. The psychological intervention also includes proactive approach in increasing the awareness of muscular dystrophy and the knowledge among school personnel, peer education about muscular dystrophy and promoting patient's independence and self-advocacy [26]. Education intervention is necessary to promote children independence and involvement in making decisions before entering school education. After that, it can be followed by the individualised educational programme. In addition, it is important to take measures to address deficits when they are identified [26].

Conclusion

There are some encouraging results in improving the quality of life patients with muscular dystrophy with early intervention and supported by the multidisciplinary team [27]. Early intervention in childhood consists multidisciplinary services which are provided to children with developmental risk factors in order to improve the health of the child, strengthen the development of ability, reduce the impact of difficulties and development lag, prevented the functional deterioration as much it is possible, improve adaptable parenting and overall family action [28].

This leads to a new set of challenges, including social and psychological impacts of longer-term survival which need to be addressed in parallel with the continued improvement in care and treatment. It also provides a learning experience for the development of new therapeutic approaches in other rare diseases.

Due to the lack of a comprehensive multidisciplinary treating program and module for the muscular dystrophy patients, there is a potential for research in finding proper approaches in managing the muscular dystrophy patients.

Given that the challenge of providing services in early intervention from specialists in various fields, this study shows the collabration of several experts involved in an indirect and immediate work with children with muscular dystrophy and their families. The desire is to find a common solution and to work on linking services in early intervention of children with developmental disadvantages.

Acknowledgments

The author thanked the speech therapist and psychologist for helping with the development of this work as well as support for everyday work with children.

References

1. Oxford University Press. Muscular Dystrophy (The Facts). New York; 2008.
2. Elsevier. Health Sciences Division. Muscle disorders in childhood. Philadelphia; 1995.
3. Amato AA, Griggs RC. Handbook of clinical neurology. In: Elsevier BV, editor. 3rd ed. Amsterdam, the Netherlands. 2011. 101: p. 1-9.
4. Boström K, Ahlström G. Living with a hereditary disease: person with muscular dystrophy and their next of kin. Am J Med Genet A. 2005;136:17-24.
5. Muscular dystrophy (perspectives on disease & disorders). In: Clay Farris Naff, editor. USA: Greenhaven Press; 2011.
6. Sjogreen L, Engvall M, Ekstrom AB, Lohmander A, Kiliaridis S, Tulinius M. Orofacial dysfunction in children and adolescence with myotonic dystrophy. Dev Med Child Neurol. 2007;49:18-22.
7. Blaži A, Kolarić B. Experiences and satisfaction of parents of children born with orofacial clefts with the support provided in Croatian systems of health and social care. Logopedija. 2015;5:18-24.
8. Che Ismail EH, Othman N. From Diagnosis to Treatment of Muscular Dystrophy: Psychology Meets Medicine. Int J Psychol Stu. 2016;8:85-91.
9. Michie S, Marteau TM. Predictive genetic testing in children: The need for psychological research. Br J Health Psychol. 1996;1:3-14.
10. Peay HL, Meiser B, Kinnett K, Tibben A. Psychosocial Needs and Facilitators of Mothers Caring for Children with Duchenne/Becker Muscular Dystrophy. J Genet Couns. 2017.
11. Matijević V, Marunica Karšaj J. (Re)habilitation guidelines for children with neuro developmental disorders. Physical and Rehabilitation Medicine. 2016;27:302-329.
12. Mikelić V, Košiček T, Crnković M, Radanović B. The participation of children with neurodevelopmental risk factors in the early rehabilitation program in relation to the level of parental education. Acta Clin Croat. 2011;50:457-461.
13. Vaughan-Graham J, Cott C. Defining a Bobath clinical framework - A modified e-Delphi study. Physiother Theory Pract. 2016;32:612-627.

14. Jung MW, Landenberger M, Jung T, Lindenthal T, Philippi H. Vojta therapy and neurodevelopmental treatment in children with infantile postural asymmetry: a randomised controlled trial. J Phys Ther Sci. 2017; 29:301-306.

15. De Groot IJM, Voet NBM, Van den Engel-Hoek MJL. Rehabilitation in Muscular Dystrophies: Changing Approach. In: InTechOpen, editor. The Netherlands: Radboud University Nijmegen Medical Centre; 2012. 12:235-246.

16. Matijaš T, Matijević V, Crnković M, Trifunović-Maček Z, Grazio S. Impulsivity and attention in children with mild motor disabilities. Paediat Croatica. 2011;55:239-242.

17. Tay SK, Lin JB. Current strategies in management of Duchenne Muscular Dystrophy: Allowing patients to live with hope. Ann Acad Med Singapore. 2012;41:44-46.

18. Gianola S, Pecoraro V, Lambiase S, Gatti R, Banfi G, Moja L. Efficacy of Muscle Exercise in Patients with Muscular Dystrophy: A Systematic Review Showing a Missed Opportunity to Improve Outcomes. PLoS One Online Journal.

19. Kolak Ž, Sečić A, Matijević V. Children's play in the humanization of children's stay in the hospital. Physical and Rehabilitation Medicine. 2013;25:42-49.

20. Divljaković K, Lang Morović M, Kraljević M, Matijević V, Maček Trifunović Z. Music therapy and rhythmic auditory stimulation with hospitalized children. Physical and Rehabilitation Medicine. 2014;26:1-11.

21. Van Ruiten H, Bushby K, Guglieri M. State-of-the-art advances in Duchenne muscular dystrophy. EMJ. 2017;2:90-99.

22. Arvedson J, Clark H, Lazarus C, Schooling T, Frymark T. The effectiveness of oral-motor exercises: An evidence-based systematic review. Dev Med Child Neurol. 2010;52:1000-1013.

23. Matijević V, Kraljević M. Therapeutic approach: orofacial-motor exercises in the rehabilitation of muscular dystrophy. In: Filodiritto, editor. 1st ed. Italy: Bologna; 2017. p. 17-22.

24. Kumin L. Intelligibility of speech in children with Down syndrome in natural settings: parents' perspective. Percept Mot Skills. 1994;78:307-313.

25. de Swart BJ, Padberg GW, van Engelen BG. Less is more: treatment of aggravating behaviour in myasthenia gravis patients with dysphagia. Eur J Neurol. 2012;9:688-689.

26. Bushby K, Finkel R, Birnkrant D J, Case LE, Clemens PR, Cripe L, et al. Diagnosis and management of Duchenne muscular dystrophy part 1: Diagnosis, and pharmacological and psychosocial management. Lancet Neurol. 2010;9:77-93.

27. Karjalainen K, Malmivaara A, Van Tulder M, Jauhiainen M, Hurri H, Koes B. Multidisciplinary rehabilitation for fibromyalgia and musculoskeletal pain in working age adults. Cochrane Database Syst Rev. 2000;CD001984.

28. Matijević V, Šečić A, Šafran I. Estimation of the difference in the effectiveness of neurodevelopmental stimulation programs in stationary and ambulatory conditions. Fiz Rehabil Med. 2016; 28:181-192.