SOCIETY. INTEGRATION. EDUCATION
Proceedings of the International Scientific Conference. Volume III, May 27th - 28th, 2016. 160-165

PROBLEMATIC OF MÖBIUS SYNDROME - CASE STUDY

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Abstract. *The article focuses on the problematic of Möbius syndrome, a specific description of a child with this syndrome. This is a very rare syndrome. At the time of diagnosing this girl's syndrome, doctors in the Czech Republic had almost no experience and had no seen any other child with Möbius syndrome. So she got the intensive care specific for this syndrome just before her third birthday. The aim of our study was to obtain data in motor development, cognitive skills and abilities and especially to obtain information about speech development. In this article, we try to capture the main therapeutic approaches that led to the development of communication skills and promote the development of all language levels.*
Keywords: *case study, Möbius syndrome, speech development, therapeutic approaches.*

Introduction

Moebius syndrome is a rare congenital neurological disease, which was first described in 1888 by Paul Julius Möbius. The most obvious symptom is a facial paresis, which arises as a result of damage to the cores VII. cranial nerve. Most often is paresis bilateral, but they may occur asymmetric variation. People with this syndrome are unable facial expression and are often referred to as a person with a mask or deadpan. Palsy is on the face lead to a decrease of muscle tone mimic muscles, triggering the drop side of his mouth and eyelids. For Möbius syndrome is also typical disability of VI. cranial nerve and that is the reason why they aren't capable of lateral movement of the eye (Abott, 1998; Zuker, Manktelow, 2014).

At the Möbius syndrome are damaged other cranial nerves, the combination is usually individual. Most often are affected these nerves: V., IX., X. and XI. These nerves innervate the muscles of the jaw, tongue, larynx and gullet and are involved in the production of speech. Because the people with Möbius syndrome have facial paresis, they can't smile, frown, raise eyebrows, blink or close their eyes. Other symptoms include strabismus, dental problems, high palate, cleft palate, hypersalivation, deformities of the ears, hearing difficulties (conductive or mixed disorder), vision disorders (photophobia, corneal erosion). There is a lag of motor development due to hypotony. Cognitive skills are in the norms range, at about 10 % of people with Möbius syndrome the mental disorder occurs (Rosenfeld-Johnson, 1999; Redett, Hopkins, 2006; Cole, Spalding, 2009; Čápová, 2010; Ilenčíková, 2013).

DOI: http://dx.doi.org/10.17770/sie2016vol3.1441

Based on the facts described above it is necessary during the therapy the interdisciplinary cooperation. The Möbius syndrome is usually diagnosed by a neurologist and an ophthalmologist abroad. The team also usuallyincludes a speech therapist, physiotherapist, paediatrician, psychologist, neurosurgeon, plastic surgeon and the other experts based on the individual needs (Tabachová, 2014).

The aim of this article is to describe the development of girl with Möbius syndrome from birth to her five years of age - how did the mother's pregnancy to birth and then proceeded motor and speech development of a child. To obtain the data the medical history questionnaires, reports from individual professional and recording sheets from the examination of speech were utilized.

Case report

The described person is a girl who was born in 2009. She was born like a third child to parents, who were 35 and 32 years old. The pregnancy was physiological. At 24th week of pregnancypes equinovaruscongenitus using ultrasound (in the normal control) was diagnosed. Also absence of the umbilical artery was discovered. Therefore Doctors recommended parents terminate pregnancy. Parents seek other experts who told them that there is no reason to terminate the pregnancy. During third trimmest of pregnancy, the mother had elevated blood pressure, so she must taking medications. Month before the birth, the hypertrophy of the fetus was diagnosed and mother had preeclampsia, which was grounds for immediate termination of pregnancy by caesarean section in the 35th week of gestation. After birth the girl was hypotonic and slightly immature hypotrophy. Apgar scores were 3/7/8. The girl weighed 1900 grams and measured 40 cm. Immediately after birth the child had irregular hyperventilation, bradypnoea and bradycardia. The girl had to be connected to the artificial lung ventilation and there was also an indirect heart massage. The child's crying was week and whiny. Already after birth the face stigmatization was evident. Gradually spontaneously condition occurred and hypotonia and occasional tapping of the bulbs prevailed.

In the fourth day of life the child's pneumonia was diagnosed. In the fifth and sixth day lack of interest in food prevailed and reflexes weren'tnearlyperceptible. For persistent difficulties in sucking the girl was not breastfed. The first Vojta's reflex locomotion method was launched, leading to better reflexes and momentum. The girl was fed through a nasogastric tube with breast milk. A week after birth, she was transferred to the neonatology in Prague. Doctors began to think about Prader-Willy syndrome there. Based on the examination this hypothesis was refuted, doctors didn't determine another syndrome. After three weeks she was transported again into a hospital in the

hometown. Here she underwent hearing tests - otoacoustic emissions presented on the right, left absent; also eye examination - without proof of pathology. After nearly eight weeks the girl was released to home care.

To treat orthopaedic disability conservative Ponseti treatment was recommended. Every fifth day she had to go to hospital for an exchange redresser plaster bandages. These bandages covered the entire leg from the fingertips to the end of the groin. In third month percutaneous tenotomy of the Achilles tendon was realized. After this treatment the feet were fixed again in a cast for a period of three weeks. After removing the plaster Denis Browne splint was applied. Girl wore this plaster 23 hours per day for a period of three months. After this period she wore it only during night sleeping until her forth birthday.

In third month there was a slight momentum improvement. There were still problems with feeding. The girl was fed by syringe, bottle feeding was not possible. The girl could not coordinate the movements of sucking, swallowing and breathing, plus suction force was very small. The girl grew by weight tables, so the paediatrician had no reason to deal with feeding. In this period she began to turn his head toward the sound, began to hum and babble and watch toys.

Because of the persistent problems the family turned to the Early Care Centre. The Early Care Centre worker helped to find to family the expert examinations for girl - visual, psychological and audiology. Audiology examination revealed a severe lesion on the left and retro-cochlear or pre-cochlear lesion on the right. Eye examination revealed nystagmus and astigmatism. As the girl could not blink her parents regularly instilled artificial tears eye to prevent drying of the corneas.

Problems with feeding was still not resolved, in her sixth month doctors wanted to proceed with the insert of PEG. Parents rejected this solution. Mother found a contact to a speech therapist with whom she consulted the situation. The position during feeding was changed, the feeding was more frequently and in smaller portions, parents began trained sucking with the child. Also speech therapist acquainted the family with Bobath concept. Vojta’s method was changed by Bobath concept and girl’s development began to move forward again.

The girl could rewind from back to stomach in ninth month (but only on the right side). She was able to lean on the forearm in the prone position. In this period, she began use soft drink cup and spoon for feeding. At the end of ninth month, she underwent scheduled strabismus surgery. After the surgery, she had to wear eyeglasses. In order to develop binocular vision, she had to exercise regularly at home.

When she was approximately one year old, the girl began to sit, eyes fixed the communication partner as well astoys. If she wanted to watch the moving object shehad to rotate the whole head. She began to play with more intonation,

duplicate syllables. She was not using words yet. Oral motoric was delayed, voice was in unnatural high position. After the first year there were frequent upper respiratory tract infections with a high fever and inflammation of the middle ear.

When she was two years old endoscopic adenoidectomy and bilateral tympanostomy were performed soit solved the otitis. At this age her mouth was filled by milk teeth. She had an overbite, high palate and soft palate dysfunction. Elevation, retraction and lateral movements of the tongue absented.

In three years of age, she began to walk around furniture, in a speech vowels prevailed, she used around 40 words actively. In this period she showed a good musical ear – she could imitate melody, intonation and rhythm. Only her mother understood her speech. Understanding of another person's corresponded to the child's age. She still wore diapers. The girl was sent to the hospital department of paediatric neurology, where she underwent a detailed examination and subsequently Möbius syndrome was diagnosed. Doctors had no experience with this syndrome. This syndrome was diagnosed by accident when one doctor read article about the syndrome, and thanks to the information gathered from this article doctors began to think whether it is really Möbius syndrome. Doctors were unable to disclose further information or to answer parents' questions. Parents contacted foreign organisations, which provided them very important information. Girl has bilateral palsy of facial muscles. Function of III., IV., VII., IX., X. and XII. cranial nerve is damaged. Other symptoms include language abnormality, high palate, submucosal cleft palate, short mandible, clubfoot turning inwards, breathing difficulties. In newborns and infants age she had difficulty during sucking and she was fed through the NG tube. Girl is photophobic, short-sighted, hypersensitive to noise and she has hearing loss. Sleep disorders are associated too.

When she was four she started attending the kindergarten where she came together with her mother, who made assistance to her daughter every day from 8 to 12 pm. The girl was under speech therapy and psychological examination, and she had individual education plan. Adaptation in kindergarten proceeded very well. During kindergarten attendance gross motor skills significantly improved, fine motor skills and consequently articulation were improved too. Walking is safer, girl manages to walk upstairs with support railings, she doesn't go downward even with support. At the age of five she learned how to ride the bike with side wheels. The level of gross motor skills is delayed relative to peers. She performs almost all of the self-service operations. She has problems with fastening buttons and tying shoelaces. There is still bedwetting during sleep and she have to wear diapers. She has problems with spatial orientation and walking on uneven terrain.

In terms of speech dysarthria is evident, articulation is more understandable but all speech sounds are not still induced. There is still a movement disorder of tongue, lips, cheeks, soft palate and hypernazality. The girl has a bad tongue position, does not protraction of the lips, jaw angle is small. Morphological-syntactic and lexical-semantic language skills corresponding to average age. The pragmatic language skillisdisturbed duediscontinuous and partlynon-created visual contact and stigmatization in the face - the girl cannot pull up the eyebrows upwards, inflate the cheeks or pucker nose.

The first meeting with a speech pathologist led to food intake improve by changing feeding techniques and tools. Speech therapist began with orofacial stimulation based on the Bobath concept, which led to the improvement of oral reflexes and oral motor skills. Parents were trained how to use orofacial stimulation and massage techniques of soft palate to improve velopharyngeal mechanism. Girl regularly carried out breathing exercises and phonation. With improving muscle tone and momentum of the orofacial region she was introduced with active therapy exercises aimed at developing motor coordination. Speech therapist started with inferring speech sounds. Because great incomprehensibility of speech persisted and girl wanted to speak a lot, they started use elements of augmentative communication and natural gestures. The girl can communicate with the people around her. Her speech is more understandable. The therapy is aimed at the overall development of gross motor skills (there is cooperation with the physiotherapist), fine motor skills and graphomotory (cooperation with occupational therapist) and the sensory perception development. The goal of therapy is the practical language use and communication with other people.

Discussion

The aim of this article was a partial view on the issue of Möbius syndrome at particular child. Overhand we described the development of a child with Möbius syndrome from birth to age five. We see that the development of the girl was delayed in all areas, but we also see a positive potential of development. Girl reaches the particular periods later than its peers but the development does not stagnate. Early physiotherapy, speech therapy and intensive work of parents can affect the development of person with Möbius syndrome and improve his life. It is important to know detailed specifications of symptoms for each child because symptoms are variable and individual. We proceed in the preparation of an individual plan of therapy on the basis of the detailed information. It leads tomore effective therapy for individual client. It is necessary to approach to each client individually and collaborate with other professionals who contribute to the

improvement of the client's condition. With interdisciplinary cooperation we can achieve better results.

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