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Development and disordres in the process of communication of people with the chromosome 13q deletion syndrome

Abstract: Chromosome 13q deletion syndrome is a kind of chromosome aberration which belongs to rarely occurring genetic abnormalities. Chromosome 13q deletion results in phenotypic disorders such as malfunctions in the central nervous system, anatomical changes of the body, as well as disorders in large and small motor skills. One of the possible consequences of the problems in motor skills development are speech development disorders, particularly concerning speech production. Other problems may relate to breathing, phonation and articulation. Additionally, on the one hand, some persistent reflexes in the orofacial area can be observed, while, on the other hand, some reflexes are absent. In the case of chromosome 13q deletion, the logopedic therapy contains also swallowing, as well as exercises which aim at improving the muscle tension in the area of face, articulators, neck, shoulders, chest and upper limbs. Disorders in speech production may take form of dysarthria, for example. Whereas, learning to communicate may require to introduce alternative communication – non-verbal communication within language production.

Keywords: speech development, speech disorders, dysarthria, chromosome 13q deletion syndrome

Rozwój i zaburzenia w procesie komunikacji osób z zespołem delecji chromosomu 13q

Streszczenie: Zespół delecji chromosomu 13q jest rodzajem aberracji chromosomowej, która należy do rzadko występujących nieprawidłowości genetycznych. Delecja chromosomu 13q skutkuje zaburzeniami fenotypowymi, takimi jak nieprawidłowości w funkcjonowaniu ośrodkowego układu nerwowego, zmiany anatomiczne, a także zaburzenia w zakresie motoryki dużej i małej. Jedną z możliwych konsekwencji problemów w rozwoju zdolności motorycznych są zaburzenia rozwoju mowy, zwłaszcza w zakresie produkcji mowy. Problemy mogą dotyczyć także oddychania, fonacji i artykulacji. Ponadto z jednej strony można zaobserwować pewne przetrwałe odruchy w okolicy orofacjalnej, a z drugiej strony niektóre odruchy są nieobecne. W przypadku delecji chromosomu 13q terapia logopedyczna obejmuje ćwiczenia połykania, ćwiczenia mające na celu poprawę napięcia mięśniowego w obrębie twarzy, artykulatorów, szyi, barków, klatki piersiowej i kończyn górnych. Zaburzenia w produkcji mowy mogą przybierać postać np. dyzartrii. Nauka komunikacji może wymagać wprowadzenia komunikacji alternatywnej – niewerbalnej w ramach produkcji językowej.

Słowa kluczowe: rozwój mowy, zaburzenia mowy, dyzartria, zespół delecji chromosomu 13q

Chromosome deletion belongs to genetic abnormalities and is defined as the loss of a part of chromosome along with its genetic material. It is a result of the malfunctions in the structure of chromosome (Moore, Persaud & Torchia, 2013, p. 280). This situation may occur as early as at the stage of gamete production or in the process of genetic code recombinations after the combination of stem cells. The lost part of the chromosome may have different size (Tobias, Connor & Ferguson-Smith, 2011, p. 100). Deletion may affect genes of different significance to development in life. Disorders occurring in specific examples of people depend on how much genetic material is lost and on its meaning in the development.

Common consequences of chromosome 13q deletion are malfunctions in the central nervous system as well as anatomical changes (13). The most frequently mentioned anatomical disorders that have been observed and described so far in the literature about deletion of the long arm of chromosome 13 are: microcephaly, triangular-shaped head, changes in the area of face and facial bones, microphthalmia, hypertelorism, blepharophimosis, epicanthus, defects of iris, cataract, big and low-set ears, widened nasal bridge, short neck, club feet, hip joint dysplasia, thumb hypoplasia (Pecyna, 2000, p. 133), as well as hearing loss reaching to 90–95db, asymetric set of shoulder blades (Binkuńska, 2003, pp. 123–126) and retinoblastoma (Matsunaga, 1980, pp. 53–58; Baud et al., 1999, pp. 478–82).

The disorders occurring in specific people with deletion depend on the extent of deletion process, that is to say, on how many genes have been lost and on their meaning. Therefore, the symptoms that have their basis in the discussed genetic process may also have different manifestations that are not listed above (Kasyan & Benirschke, 2005, pp. 658–665).

Chromosome 13q deletion belongs to rarely occurring disorders, therefore the literature concerning it is scarce and concentrates mainly on the genetic, phenotypic and neurological aspects (Allderdice et al., 1969, pp. 499–512; Amor, Voullaire, Bentley, Savarirayan & Choo, 2005, pp. 151–157). Among materials about 13q deletion, we may find articles comparing the changes in differential disorders that appear within chromosome 13. Kasyan & Benirschke, for instance, posted the article in 2005: *Genetic halpoinsufficiency as phenotypic determinant of a deletion 13q syndrome* in Padiatric and Developmental Pathology (Kasyan & Benirschke, 2005, pp. 658–665). The authors analyse mainly neurological disorders in two infants with chromosome 13q disorder. Lucia Ballarati et al. in the article: 13q Deletion and central nervous system anomalies: Further insights from karyotype-phenotype analyses of 14 patients, in 2007, presented an analysis of development and possible disorders in a considerably big number of people with 13q deletion (Ballarati et al., 2007). The literature on the subject, though, lacks elaborations presenting the therapy of speech development and of motor development – small and large motor skills. The author of this work has undertaken the subject of speech therapy of a person with 13q deletion by analysing therapeutic methods

which allow to develop the functions of the speech apparatus (Binkuńska, 2003, pp. 123–126). The author has analysed the quality of communication (Binkuńska, 2010, pp. 123–128) and its disorders, mainly in the form of dysarthria (Binkuńska, 2013, pp. 63–71).

Purpose of the study

The subject of this work is concerned with language communication development and disorders in the functioning of speech organs in people with the chromosome 13q deletion syndrome. The aim of the work is to present the difficulties in specific areas of communication – active and passive speech, as well as to point to possible malfunctions of the speech organs. The following questions are examined: how does language communication develop in people with the chromosome 13q deletion syndrome? What kind of speech organ malfunctions can be observed in people with the chromosome 13q deletion? What kind of speech disorders may occur in people with the chromosome 13q deletion? Which aspects of the language develop well enough for the speech communication to be possible? Which aspects of the language develop with such difficulties that they considerably or completely prevent communication? The attempt to answer the above questions requires not only the analysis of the literature concerning the discussed genetic disorder but, most of all, it requires longer period of observation of the development of a person with the above mentioned disorder.

Methods of proceeding in speech therapy

The case study presented in this work is based on twelve years of speech therapy with a person with chromosome 13q deletion. The therapeutic work started when the person was 6 years old and finished when she was 18.

The anatomical changes that occurred in this case were: changes in the facial bones: triangular-shaped head, microphthalmia (small eyes), hypertelorism (wide-set eyes), blepharophimosis (reduced lid size), epicanthus (skin fold in the inner corner of eyes), defect in vision, low-set ears, widened nasal bridge, short neck. In the area of anatomy there occurred: thumb hypoplasia, Sprengel's deformity – asymetric set of shoulder blades, feet changes – club feet, hip joint dislocation (Binkuńska, 2013, p. 65).

Malfunctions in the work of muscles were also observed. There was a pathological distribution of muscle tension between the shoulder and hip girdle on the right side of the body. The weakened muscle tension affected also the muscles of larynx and the center of the body. However, in upper and lower limbs the muscle tension increased. In the area of arms the work of forearm rotation was hardly noticeable.

The muscle functioning disturbance was observed during making purposeful movements. When the muscles were at rest, their tension was lowered, whereas during attempts to make purposeful requested movements the tension increased in the upper and lower limbs. In the central area of the body it remained unchanged. As it was mentioned before, sudden altering of muscle tension affected mainly movements done at request, less frequently spontaneous movements.

Apart from upper and lower limbs, the increased muscle tension was observed in the area of orbicularis oris muscle while doing purposeful movements. However, in this case, this situation was connected with positioning body, lips and jaw in a way that they leaned forward.

As a consequence of asymmetric set of shoulder blades, shoulder girdle and hip girdle, there were disturbances in the muscle tension in the area from face and facial bones, through shoulders to intercostal spaces and the muscle structures of abdomen and diaphragm.

There was a slower pace of motor skills development. This was the result of the disturbance in sensorimotor processing and in muscle tension distribution. It took a longer period of time to master the ability of rotating around herself, groveling, crawling and sitting on her own. Walking was not mastered in the analysed case.

Therefore, the developmental deficits affected both small and large motor skills. In the area of small motor skills poor eye-hand coordination and lack of precision in the performed movements were observed.

Apart from the aforementioned changes in the area of child's speech apparatus, anatomy and physiology, there were also reflexes in the orofacial area such as the reflexes of searching, biting, pushing the tongue outside and rooting reflex. The vomiting reflex was weakened and moved inward the oral cavity. The reflexes of chewing and swallowing were absent. Typically, the child kept her mouth open and was slavering (excessive salivation).

The disorders were also concerned with breathing. When breathing was not accompanied by phonation, both breathing in and out were performed through mouth, even though the nasal cavity was unobstructed. The breath was shallow with domination of one side of the chest. When the child was in recumbence while breathing, abdomen part was dominant and ribs did not move. In the early stage, the child was not able to consciously breathe in and direct the air flow while breathing out. When these abilities were mastered, while consciously breathing in and directing the breath, the phases of breathing in and out were shallow,

performed by the upper part of the chest. They were short and fast. Therefore, despite activating the upper part of the chest, the breath was not performed with the whole breathing capacity. Rather, depending on the child's position, with the lower part of the chest, when the child was in recumbence, or with the upper part, when it was sitting. The absence of deeper breath may have resulted from the anatomical changes in torso, including asymmetric set of shoulder blades and disturbances in the muscle work in these parts.

Disorders in the ability to consciously breathe in and breathe out, as well as damages in the central nervous system lead to the lack of respiratory and phonation coordination, thus creating the basis for difficulties in speech sound production.

Additional factor causing disturbance in performing full and deep breath was positioning shoulders foreward while sitting, which entailed constricting intercostal muscles.

Relaxed breathing through mouth was accompanied by the weakening in the area of orbicularis oris muscle and of cheek muscles (Rządzka, 2011, p. 24; Mitrinowicz-Modrzejewska, 1963). Weakening of face muscles tension was an unfavorable element in keeping oral cavity closed.

Tongue malfunctions were connected with a weakened muscle tension, with aforementioned reflex of pushing the tongue outside and with positioning the speech organs, such as jaw and lips, foreward. This kind of positioning the jaw and lips prevented tongue from being placed upright. It did not settle on the jaw, rather it extended outside the mouth. Initially, the muscle tension of the tongue was weakened and the tongue was positioned asymmetrically. Gradually though, when the person mastered the ability of making purposeful movements of the tongue, the muscle tension increased during the tongue's activity. The aforementioned reflex of pushing the tongue outside appeared as well. Little mobility was observed also in the area of the soft palate. There were difficulties in making purposeful movements there.

When the speech sound production is concerned, the child produced individual, spontaneous sounds, which were not always logically connected with the context and the situation in which they occurred. The sounds produced were of exclamation kind. Speech disorders were related to dysarthria. The ability to produce speech sounds was limited. While producing sounds there was no coordination of breathing, voicing and articulation. Disorders in the speech production were also the result of the malfunctions in the motor skills of speech organs (Tłokiński, 2005, p. 911). Both, the lack of coordination in breathing, voicing and articulation, as well as the malfunctions in the motor skills of speech organs are the result of genetically based impairments in the functioning of the central and peripheral nervous system (Jastrzębowska, 2003, pp. 120–142). Another characteristic connected with dysarthria that occurred in the child's case was the lack

of swallowing and chewing abilities. Despite not developing active speech, there was passive speech comprehension observed. The fact that the child tried to react adequately to the context of statements, proves that particular levels of language analysis – syntax, vocabulary, morphology and phonology – were developed. She tried to respond to commands by gestures, showed anxiety when grammatical mistakes were made, and she also recognised linguistic jokes.

Speech therapy started from the attempts to position the child upright. The aim was to improve the asymmetric setting of the hip and shoulder girdle as much as possible. This would allow better breathing, especially deepening the breath and teaching the child to consciously direct the air flow while breathing out. Working against the forces of gravity was to provide proprioceptive stimuli that would affect the improvement of feeling the body.

The initial stage of the speech therapy included the work on eliminating the reflexes of searching, biting and rooting, as well as the reflex of pushing the tongue outside. The aim of the therapy at that time was to trigger the reflex of swallowing, chewing and the emetic reflex.

The sitting position of the child made it possible to start teaching her how to consciously take the air in and direct the air flow while breathing out. Positioning the child appropriately with the tendency to place child's head, chest and body rather symmetrically, contributed to the improvement of phonation. It also contributed to creating better opportunities to work on sound articulation. The therapeutic work on conscious breathing also required placing the child in the upright position.

Therefore, activities concerned with breathing functions included work on consciously directing the flow of air while breathing out, on breathing deeply, on prolonging the exhaust phase and on changing the direction of breathing to breathing through nose.

The objective of the initial stages of the therapy was to improve the muscle tension in the area of the organs responsible for verbal communication – the areas of face and facial bones, speech articulators, neck and shoulders, as well as chest and abdomen. During the therapy various massages were done, with a special emphasis on face and oral cavity massage. The speech organs were exercised in the direct and indirect way. The aim was to improve muscle tension and the motor skills of speech articulators. That included working on erect position of the tongue, putting the tip of the tongue behind the lower teeth and raising the middle part of the tongue to the hard palate. The exercises of raising up and lowering down the soft palate and the massage of these areas were additional factors stimulating the swallowing reflex. The exercises mentioned above included also the muscle around the mouth and the groups of cheek muscles.

The next phase of the speech therapy concentrated on phonation activities. The impediments in this area resulted from the lack of coordination between

breathing, phonation and articulators, as well as from the abnormal glottal stop which occurred during attempts to produce sounds. In that situation the sounds were produced with difficulty, the phonation was usually preceded by articulation movements and the voice was hoarse.

The speech therapy was concerned with the attempts to shape verbal communication within the active speech. As the results of sound production and combining sounds into word structures were unsatisfactory, nonverbal sounds started to be introduced. The ability to use passive speech were developed.

Results of speech therapy

The speech therapy has resulted in the improvement of the breathing functions. The child managed to learn to breathe through nose and direct the air consciously. The exhaust phase got prolonged and the breathing became deeper.

The muscle tension of the articulators and the motor skills of the tongue have also been improved. Considerable change for the better has been observed in the work of the tongue, lips, as well as the soft palate. The child has learnt to swallow, to raise the tongue to the hard palate and to maneuver it inside and outside the oral cavity. The ability to push the lips forward has also been mastered. The improvement in the muscle tension was essential in learning to keep the mouth closed. Keeping the mouth closed, in turn, has made previously occurring salivation less troublesome.

The child has mastered the articulation of vowel sounds and few consonants. These sounds allowed for producing several words such as, for example, "mama" ('mum'), "nie" (no), "ja" (I), "lala" ('doll'). The lexical system, though, remained poor and the vocabulary did not allow to create sentences. Therefore, non-verbal signals started to be introduced. As the consequence of developing speech comprehension, the child with 13q deletion syndrome has started to react adequately to instructions and speech in general.

However, phonation has not improved. The malfunctions in phonation stem from dysarthria in the discussed case. There were the following characteristic features of phonation: the lack of coordination between breathing, phonation and articulators, as well as glottal stop. The voice has remained to be hoarse.

Conclusion

The results achieved during the speech therapy were varied in different spheres, in the functioning of the speech organs – breathing, mouth and facie reflexes – as well as in communication development – active speech and passive speech. The disproportion between understanding speech, which was well enough to develop communication, and the speech production was discernible. The disturbances that occurred during speech production were of dysarthria nature. There has been a considerable improvement in the area of breathing functions, in eliminating the mouth and face reflexes and in eliciting the reflexes which did not occur previously.

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