International Journal of
Health Systems and Medical Science

ISSN: 2833-7433
Volume 1 | No 6 | Dec-2022

Clinical and Audiological Aspects of Hearing Impairment in Children with Down Syndrome

Z. Kh. Karimova [1]

Abstract: This article presents data from a review of the literature on the etiology, epidemiology of Down syndrome, the prevalence of hearing loss, as well as the impact of hearing impairment on the development of cognitive, psychomotor, and communication skills. It should be remembered that in young children and newborns, hearing impairment is predominantly of a neurosensory nature and most often occurs in the first year of life. Children with Down syndrome experience some hearing loss, which is more common in the early years of life.

Keywords: Down syndrome, hearing impairment, sensorineuraland conductive hearing loss, audiological diagnostics.

Introduction. Down syndrome (DS) (ICD 10: Q90.0; OMIM: 190685) is one of the most common chromosomal aberrations, with a prevalence of 1:1000 newborns (15). Down syndrome is the most common genetic abnormality. It is caused by an inadvertent mutation in the 21st chromosome pair, in which another chromosome appears. Such children lag behind their peers with hearing loss and speech formation much later [1,6,7,9,10]. The syndrome was first described in 1866 by John Langdon Down in children born to different mothers but with similar external features as "mental retardation in patients with high susceptibility to infection and low life expectancy."[12] According to statistics from the World Health Organization, one in every 700-800 babies worldwide is born with a diagnosis of Down syndrome. The rate is the same in different countries, climatic zones and social strata. Genetic failure occurs regardless of the parents' lifestyle, health, habits and education.

A study conducted by scientists at the University of Mysore, India, has identified four factors that influence a child's chances of having Down's syndrome. These are the age of the mother, the age of the father, close marriages, and, oddly enough, the age of the maternal grandmother. The last of the four factors proved to be the most significant. The older the grandmother was when she gave birth to her daughter, the more likely she was to give birth to a grandchild or granddaughter with Down syndrome. This probability increases by 30 per cent for every year the grandmother-to-be "missed out".

For the study, published online in the British Medical Journal, experts analysed data from a national register which contained information on 26,000 identified cases of Down syndrome in England and Wales, diagnosed in both the prenatal and postnatal stages of development. According to the Downside Up charity, around 2,500 such children are born each year in Russia. 85% of families abandon a child with Down syndrome in the maternity home, including on the advice of medical staff. In comparison, no cases of abandonment of these children have been recorded in Scandinavia. In America, 250 families are on the waiting list to adopt them. As for statistics on our republic, official figures have not been found. According to unofficial statistics, around 2,000 children with Down syndrome are born in Uzbekistan every year. In the International Classification of Diseases, 10th revision, Down syndrome is represented by class Q90: Q90.0 Trisomy 21, meiotic non-discrimination; Q90.1 Trisomy 21, mosaicism (mitotic non-discrimination); Q90.2 Trisomy 21, translocation; Q90.9 Unspecified Down syndrome; - Trisomy 21 BDU. The karyotype of people with Down syndrome is as follows 47,XX,+21 (if a girl is sick) and 47,XU,+21 (if a boy is sick). Let us

talk a little more about the classification of Down syndrome. There are three genetic variations (or forms) of Down syndrome:

1. trisomy 21. More than 90% of cases of Down syndrome are caused by trisomy 21. Children with trisomy 21 have three chromosomes in the 21st pair instead of the normal two. In this case, all of the child's cells have this defect. This disorder is caused by an abnormality of cell division during the development of the egg or sperm. In most cases, it is due to a chromosome mismatch during oocyte maturation (about 2/3 of cases).
2. Mosaicism. In this rare form (about 2-3% of cases) of Down syndrome, only some cells have an extra chromosome in the 21st pair. This mosaic of normal and abnormal cells is caused by a defect in cell division already after fertilization.
3. Robertson's translocations. Down syndrome can also occur when part of the chromosome 21 pair is shifted to another chromosome (translocation), which occurs before or during conception. Children with this condition have two chromosomes in the 21st pair, but they have extra material from chromosome 21 that is attached to the other chromosome. This form of Down syndrome is rare (about 4% of cases).

Down syndrome is one of the most common forms of chromosomal abnormalities today. It is characterised by intellectual impairment as well as specific changes in appearance. It is noteworthy that the number of boys and girls born with Down syndrome is approximately equal, and the frequency is 1 child per 600 to 800 babies born (Baranova Yu.A. et al., 2020)[4]. Nervous system, speech and mental disorders are the main cause of disability and social disadaptation in children and most originate in the perinatal period (Aleksandrovich Y.S., et al., 2010). [2,14]. In addition to small abnormalities, children with DM have muscular hypotonia, developmental delays and congenital abnormalities of internal organs, including the heart and gastrointestinal tract, as well as a number of syndrome-associated diseases such as hypothyroidism, celiac disease, etc.[20]. Delayed physical and mental development is evident as children with this syndrome grow. Their height is usually low and they are at an increased risk of obesity.The average IQ is around 50. Behaviour is suggestive of attention deficit hyperactivity disorder, which is common in childhood, and the frequency of autistic behaviour increases (particularly in children with profound intellectual disability).Most children with DM have some degree of cognitive impairment, ranging from severe (IQ = 20 - 35) to mild (IQ = 50 - 75)[22,34].

There is an increased risk of depression in children. Severe delay in motor and speech development is also evident at an early age [35]. Mental developmental problems in children with diabetes are related to the OLIG1 and OLIG2 genes (oligodendrocytes), which are located on chromosome 21. This region is also thought to contribute to cognitive defects in DM. They are responsible for the formation of so-called GABAergic neurons, the main 'brakes' of the nervous system, with the first DNA site suppressing their formation and the second accelerating this process [29, 31, 31]. DM patients also do not have normal lymphocyte counts and produce fewer antibodies, which increases the risk of infection.

Muscle hypotension affects the muscles of the bronchial tree, making it difficult to expel mucus. An accumulation of mucus can lead to upper respiratory tract infections and a subsequent increase in the incidence of otitis media [5]. A high incidence of ear disease in children with diabetes has been reported by many authors. For example, muscle hypotonia is a factor of this fact, which may contribute to an increased incidence of ear infections, and is associated with anatomical defects such as Eustachian tube dysfunction, prolonged presence of mesenchymal tissue in the tympanic cavity, and stenosis of the external auditory canal and hypoplasia of the mastoid process. [13].

Histopathological studies have shown that in cases of secretory otitis media, diffusion of bacterial toxins can occur and cause abnormal inner ear structures and lead to neurosensory hearing loss. [17,36,38].

Congenital abnormalities of the inner ear are uncommon, although people with DM have anatomically smaller cochlea sizes than typically developing children[23]. Furthermore, beginning in

the second decade of life, people with diabetes show a reduced hearing threshold with progressive presbyacusis[21,28,34].

Although many children with Down syndrome suffer from hearing loss, little research has been done on its effects on speech and language development.

Delayed speech development in Down syndrome due to the presence of anatomical features, reduced muscle tone, and intellectual impairment presents a particular challenge. Research in this area has shown that 40-80% of these children have a hearing impairment. For most professionals working with them, the link between speech development and hearing is obvious. However, very few studies are available that rigorously investigate the impact of hearing impairment on speech development. And the results are not always complete or reliable because they exclude children who have the most significant hearing loss. For example, it has been found that staff from Downside Up have made many visits to educational institutions in the UK and have seen how much attention teachers and parents pay to monitoring children's hearing. Many more children with Down syndrome in England use hearing aids or have had a cochlear implant. In Russia for instance these children are rarely seen.

All children have to attend an audiology centre. For example, in Portsmouth, which is near London, the audiological centre is attached to the public hospital and linked to an organization that provides educational support for children with Down syndrome. This means that the family of a newborn baby arrives for a consultation with doctors and teachers literally from the comfort of the building to undergo a full audiological examination, and doctors and teachers can meet quickly to discuss each individual child's situation and develop methods of support. Working together, doctors and teachers can also examine the problem holistically and systematically, keeping track of the child's hearing in relation to speech development and ensuring that the necessary help is provided in good time. Specialists of Downside Up recommend all children with Down syndrome to undergo hearing screening and audiogram once a year for the timely detection and correction of hearing loss [3, 8,9, 10, 16, 18].

Children need basic sensory and perceptual skills to develop speech. Sensory skills include hearing, seeing, tasting, smelling and touching. Perceptual skills, or perceptual skills, relate to the ability to make sense of information from sensory systems. Developmental disorders - impairments in the formation of developmental functions - figure prominently in the structure of these impairments. Congenital and acquired disorders of the analysers - vision and hearing - play a special role among the causes of developmental disorders. According to the World Health Organisation (WHO) more than 5% of the world's population (360 million people) has a disabling hearing loss, of which 32 million are children (WHO, 2006). According to the European Downs Syndrome Association (EDSA), 80% of people of varying ages with Down syndrome have some form of hearing loss, however there are also cases of sensorineural hearing loss.Epidemiological studies show the prevalence of moderate to profound hearing loss in children, including sensorineural hearing loss and conductive hearing loss is as high as 6:1000, with 10% of children having profound hearing loss.

Children with Down syndrome are at higher risk of hearing loss compared to their typically developing peers.The literature suggests that 1.4 per 1000 newborns and 5 per 1000 children aged 3 to 7 years have hearing loss (Centers for Disease Control and Prevention, National Center for Birth Defects and Malformations, Division of Birth Defects and Malformations 2013).

Many children with Down syndrome experience some hearing loss especially in the early years of life. Up to 20% of children may have sensorineural hearing loss caused by ear and auditory nerve defects. They have narrow nasal passages, small mouths and tongue deviation. The specific horizontal location of the Eustachian tube causes frequent middle ear infections, and this can lead to hearing loss. Hearing loss occurs in 38-78% of children with Down syndrome. These children find it difficult to look, listen, and focus on anything at the same time, and as a consequence they are unable to process signals from more than one stimulus at any one time.[5,17,18,21]. Several studies have shown an assessment of the peripheral auditory system in people with diabetes. Because hearing loss can impair speech development, a complete audiological evaluation of these individuals is crucial for differential diagnosis and further treatment and rehabilitation tactics.

Among the few works on the subject, some are worth mentioning. For example, colleagues from Cincinnati conducted a comprehensive study of hearing function in 135 children with various forms of DM between the ages of 1 and 15 years[17]. According to the authors, the most informative type of objective audiometry available was broadband acoustic impedanceometry at low frequencies, which provided a complete picture of the degree of hearing loss and the severity of auditory tube patency in the study group. Such data could not be obtained with tonal threshold audiometry alone.

Another American scientist from Jackson University Laboratory [16] has performed a very interesting fundamental study in which the pathogenetic mechanisms of middle exudative otitis media were analysed in mice with the Ts65Dn genotype showing many phenotypic characteristics in common with human Down syndrome. In a group of mice, otitis media was found to be characterised by middle-ear effusion and hearing loss. Thus, in the 53 Ts65Dn mice tested, 81.1% had high ABR thresholds for at least one of the stimulus frequencies (click, 8kHz, 16kHz and 32kHz) and in at least one ear.

The aim of a study by Portuguese[11, 14]scientists was to characterize the peripheral auditory system of people with Down syndrome (DS) using conventional and high-frequency audiometry between the ages of 7 and 15 years. A comprehensive audiological examination revealed signs of a mild conductive hearing loss in one or both ears. Mean hearing thresholds for conventional audiometry were below 20 dB SP and between 20 and 40 dB SP for high-frequency audiometry. Pearson's correlation coefficient showed a moderate positive correlation between 9-14 kHz thresholds and age. Also, middle ear abnormalities and conductive hearing loss were found in most children. Moreover, high-frequency audiometry suggested the onset of cochlear dysfunction, which may be related to frequent episodes of otitis media and/or early degeneration of the cochlea.

Thus, there have been many studies aimed at studying the hearing in persons with Down syndrome, using both screening and objective methods of examination, but despite the numerous scientific works, the correlation mechanisms and correlation between hearing loss and the degree of speech delay in children with Down syndrome have not been fully investigated. It is known that one of the functions of the auditory analyzer is the activation of the limbic system, which in turn plays an important role in the formation of the child's neuropsychological and mental status. The type and degree of hearing loss and the likelihood of one type becoming dominant depending on the form of ASD remain unknown. Ways of correction and methods of medical and therapeutic rehabilitation taking into account the underlying pathology are not disclosed.

A detailed review of the literature on this problem did not reveal any works devoted to the effectiveness (auditory training and speech work in children) of the correction of hearing loss in children with Down's syndrome in practice. Based on the analysis of the literature review, we identified the relevant goals and objectives of the planned work, namely, to study the features of the ENT, in more detail the functional state of the hearing in children with Down syndrome, the impact of its violations on the psychological and speech development, the definition of corrective and rehabilitative tactics, as well as preventive measures.

Thus, Andijan Medical Institute, Department of Otorhinolaryngology in cooperation with adjacent specialists (child neurologists, geneticists, neurophysiologists, psychoneurologists and pediatricians) initiated research work on children with Down syndrome and issues of auditory and speech development, which we plan to highlight in our future publications.

Literature:

1. Madjidova Ya.N., Salikhova S.M.: Possible pathogenetic mechanisms of progression and the occurrence of intellectual and cognitive impairment in patients with Down syndrome, International Journal of Psychosocial and Rehabilitation, Vol.24, special issue 1, 2020. 789-793.
2. Баранова Ю.А. с соавт.,20205 Н. А. Бенис, Т. В. Самсонова // Материалы XVI Конгресса педиатров России «Актуальные проблемы педиатрии». — М., 2012.-С. 80.

3. Жиянова П. Л., Поле Е. В., Нечаева Т. Н. Формирование основных двигательных навыков у детей с синдромом Дауна. Благотворительный фонд "Даунсайд Ап", 2010. - 68 с. - ISBN 978-5-904828-02-8, - УДК 6.16.7-053.2, ББК 57.31.
4. Гориянова А.В., Семенова Н.А. Оценка темпов психомоторного развития детей с синдромом Дауна.// Вестник Российского Государственного Медицинского Университета. Специальный выпуск. - 2013.-№1, с.311
5. Одинокова Г.Ю. Выявление и преодоление неблагополучия в развитии общения матери и ребенка раннего возраста с синдромом Дауна: автореф. дисс. … канд. психол. наук : 13.00.03 / Одинокова Галина Юрьевна. – Москва, 2015. – 26 с.
6. Самиева Г. У. Дисбиотические расстройства верхних дыхательных путей у детей с острым стенозирующим ларинготрахеитом //медицинские новости. – 2015. – №. 7 (250). – С. 70-71.
7. Самиева Г. У., Абдирашидова Г. А., Собирова Ш. Б. Прогностическое значение спектра цитокинов и их изменения при первичных и рецидивирующих ларинготрахеитах у детей //инновационные исследования: проблемы внедрения результатов и направления развития. – 2017. – С. 103.
8. Самиева Г. У., Карабаев Х. Э. Влияние эндогенной интоксикации на клиническое течение различных форм острого стенозирующего ларинготрахеита у детей //Вестник оториноларингологии. – 2016. – Т. 81. – №. 1. – С. 37-39.
9. Самиева Г. У. Состояние респираторного тракта у детей с острым стенозирующим ларинготрахеитом //Вісник наукових досліджень. – 2015. – №. 1.
10. САМИЕВА Г. У. и др. HEARING IMPAIRMENT IN PATIENTS WITH ADHESIVE OTITIS MEDIA //ЖУРНАЛ БИОМЕДИЦИНЫ И ПРАКТИКИ. – 2022. – Т. 7. – №. 1.
11. Barbara Carrico1, Alessandra Giannella Samelli1, Carla Gentile Matas1, Fernanda Cristina Leite Magliaro1, Renata Mota Mamede Carvallo1, Suelly Cecília Olivan Limongi1, Ivone Ferreira Neves-Lobo1Peripheral hearing evaluation in children with Down syndrome valiação auditiva periférica em crianças com síndrome de Down
12. Bunt CW et al: Role of the family physician in the care of children with Down syndrome. Am Fam Physician. 90(12):851-8, 2014
13. Bain, S. K., & Allin, J. D. (2005). Book review: Stanford-Binet intelligence scales, fifth edition. Journal of Psychoeducational Assessment, 23, 87-95
14. Crawford D, Dearmun A. Down's syndrome /2016 Nov 8;28(9):17. PMID: 27821010 DOI: 10.7748/ncyp.28.9.17.s19.
15. Cauwenbege P, Watelet JB, Dhooge I. Uncommon and unusual complications of otitis media with effusion. Int J Pediatr Otorhinolaryngol. 1999;49 Suppl 1:S119-25. http://dx.doi.org/10.1016/S0165-5876(99)00214-1
16. Fengchan Han*, Heping Yu*, Jiangping Zhang*, Cong Tian*, Cecilia Schmidt , Casey Nava*, Muriel T. Davisson and Qing Y. Zheng* Otitis media in a mouse model for Down syndrome *Department of Otolaryngology-HNS, Case Western Reserve University, Cleveland, OH, USA and The Jackson Laboratory, Bar Harbor, ME, USA
17. Hunter, L. L., Keefe, D. H., Feeney, M. P., Brown, D. K., Meinzen-Derr, J., Elsayed, A. M., Amann, J. M., Manickam, V., Fitzpatrick, D., & Shott, S. R. (2017). Wideband acoustic immittance in children with Down syndrome: prediction of middle-ear dysfunction, conductive hearing loss and patent PE tubes. International journal of audiology, 56(9), 622–634. https://doi.org/10.1080/14992027.2017.131455
18. Hauser P. C. et al., 2006; Woll B., 2008; Van Naarden B. K. et al., 2015 Hook E. G. Epidemiology of Down syndrome.In: Pueschel, S. M.; Rynders, J. E. :Down Syndrome.

Advances in Biomedicine and the Behavioral Sciences. Cambridge:Ware Press (pub.)- 1982,- P. 11.

19. Hassmann E, Skotnicka B, Midro AT, Musiatowicz M. Distortion products otoacoustic emissions in diagnosis of hearing loss in Down syndrome. Int J Pediatr Otorhinolaryngol. 1998;45(3):199-206. http://dx.doi.org/10.1016/S0165-5876(98)00106-2
20. Hankinson TC et al: Craniovertebral junction abnormalities in Down syndrome. Neurosurgery. 66(3 Suppl):32-8, 2010
21. Harada T, Sando I. Temporal bone histopathologic findings in Down´s syndrome. Arch Otolaryngol. 1981;107(2):96-103
22. Hook E. G. Epidemiology of Down syndrome.In: Pueschel, S. M.; Rynders, J. E. : Down Syndrome. Advances in Biomedicine and the Behavioral Sciences. Cambridge: Ware Press (pub.)- 1982,- P
23. Irving C., Basu A., Richmond S. et al. Twenty-year trends in prevalence and survival of Down syndrome// Eur J Hum Genet.- 2008.-16- p.1336-1340.
24. Jensen K.M. et al: Primary care for adults with Down syndrome:adherence to preventive healthcare recommendations. J. Intellect Disabi / Res. 57 (5): 409-21, 2013
25. Klosowska A et al: Down syndrome, increased risk of dementia and lipid disturbances. Dev Period Med.21 (1):69-73,2017
26. Marcell MM. Relationships between hearing and auditory cognition in Down's syndrome youth. Downs Syndr Res Pract. 1995;3(3):75-91. http://dx.doi.org/10.3104/reports.54
27. McDowell KM et al: Pulmonary complications of Down syndrome during childhood. J Pediatr. 158(2):319-25, 2011
28. Mustacchi Z. Síndrome de Down. In: Mustacchi Z, Peres S. Genética baseada em evidências: sídromes e heranças. São Paulo: CID; 2000. p. 817-94.
29. Raitano Lee N, Pennington BF, Keenan JM: Verbal short-term memory deficits in Down syndrome: phonological, semantic, or both? J.NeurodevDisord. 2010; 2(1):9–25. 10
30. Rex A. P., Preus M. A diagnostic index for Down syndrome//J. Pediatr.-1982.- 100- p. 903-906.
31. Sullivan WF et al: Primary care of adults with intellectual and developmental disabilities: 2018 Canadian consensus guidelines. Can Fam Physician. 64(4):254-79, 2018
32. Shah NB: Trisomy 21. In : Florin TA et al, eds: Netters Pediatrics. Saunders; 2016:740-4
33. Tuomanen EI. Pathogenesis of pneumococcal inflammation: otitis media. Vaccine. 2000;19 Suppl 1:38-40. http://dx.doi.org/10.1016/S0264-410X(00)00276-0
34. Tomé DC, Sanchez TG, Bento RF. Síndrome de Down e otorrinolaringologista: características gerais e aspectos otológicos (Parte I). Arq Int Otorrinolaringol. 1999;3(3):93-8.