© Borgis New Med 2021; 25(3): 71-73 DOI: https://doi.org/10.25121/NewMed.2021.25.3.71

Maja Skwarska[1], Bartosz Rigall[2], *Lidia Zawadzka-Głos[2]

Case study of a patient with Cornelia de Lange syndrome – Department of Pediatric Otolaryngology, Medical University of Warsaw

Studium przypadku pacjentki z zespołem Cornelii de Lange – Klinika Otolaryngologii Dziecięcej Warszawskiego Uniwersytetu Medycznego

[1]Student Audiological and Phoniatric Scientific Group in Medical University of Warsaw, Poland
[2]Department of Pediatric Otolaryngology, Medical University of Warsaw, Poland
Head of Department: Associate Professor Lidia Zawadzka-Głos, MD, PhD

Keywords

de Lange syndrome, genetic disorder, hear loss, ABR

Summary

Cornelia de Lange syndrome (CdLS) is a genetic disorder characterized by facial dysmorphia, growth failure, otolaryngological and cardiovascular problems. Due to its multiorgan occurrence, it is crucial for a multidisciplinary team to cooperate and provide regular check-ups for these patients.
3-month old girl with CdLS was admitted to Pediatric Otolaryngology Clinic to diagnose hearing loss. The child prenatal and neonatal life was complicated with IUGR, heart defect, respiratory insufficiency, infection and feeding problems. In our clinic hearing examination has been conducted and bilateral hearing loss was diagnosed. Further tests are being conducted.
Most of the children with CdLS experience otolaryngological problems which result in hear loss. Objective hearing examination is crucial in this group of patients due to mental retardation and early onset of symptoms. Some of these patients have severe speech development problems. That is why it is essential for specialists to examine these patients regularly and provide early medical interventions, which can result in better outcomes for them.

Słowa kluczowe

zespół de Lange, zaburzenie genetyczne, utrata słuchu, ABR

Streszczenie

Zespół Cornelii de Lange jest zaburzeniem genetycznym charakteryzującym się dysmorfią twarzy, zaburzeniem wzrastania, problemami otolaryngologicznymi oraz kardiologicznymi. Z racji tego, że może on powodować zaburzenia w wielu różnych organach, kluczowe jest, aby nad pacjentami z tym zespołem sprawował opiekę wielospecjalistyczny zespół prowadzący regularne konsultacje.
Trzymiesięczna dziewczynka z zespołem Cornelii de Lange została przyjęta na Oddział Otolaryngologii Dziecięcej w celu diagnostyki zaburzeń słuchu. Okres prenatalny oraz noworodkowy był u pacjentki powikłany IUGR, występowaniem wrodzonej wady serca, niewydolnością oddechową, infekcją oraz problemami z przyjmowaniem pokarmu. W Klinice zostały przeprowadzone badania słuchu i stwierdzono obustronny ubytek słuchu. Dalsza diagnostyka jest prowadzona.
Większość dzieci z zespołem Cornelii de Lange posiada różnorakie zaburzenia otolaryngologiczne, które mogą skutkować ubytkiem słuchu. Obiektywne badania słuchu są kluczowe w tej grupie pacjentów, ze względu na występowanie upośledzenia umysłowego oraz wczesnego rozwoju objawów. Niektórzy z tych pacjentów doświadczają poważnych problemów z rozwojem mowy. Z tego powodu jest to niezwykle ważne, aby specjaliści prowadzili w tej grupie pacjentów regularne kontrole oraz stosowali wczesne interwencje medyczne, które mogą prowadzić u nich do lepszych wyników.

Introduction

Cornelia de Lange syndrome was first described by the Dutch pediatrician Cornelia de Lange (CdLS) in 1933. It can have many clinically varying presentations (1). It is estimated that 1 in 30 000 to 1 in 10 000 children are affected (2). It is a genetic disorder, in which, most commonly, a de novo mutation occurs. In a classical form it is easily recognized on the base of clinical features, although genetic testing is often used to confirm the diagnosis (1). The most common phenotypical features include: synophrys and/or thick eyebrows; short nose, concave nasal ridge and/or upturned nasal tip; long and/or smooth philtrum; thin upper lip vermilion and/or downturned corners of mouth; hand oligodactyly and/or adactyly; congenital diaphragmatic hernia (3). Growth failure is also very usual, which can sometimes be observed prenatally. Besides that an intellectual disability is present, as well as behavioral issues, neurologic, gastrointestinal, ophthalmologic, otolaryngologic, genitourinary, cardiovascular and immunologic problems (4). It is therefore crucial, that a multidisciplinary team cares for the patient and provides regular check-ups.

The aim of this article is to present a case report of a 3-months old patient with Cornelia de Lange syndrome and hearing loss and to underline the importance of an early diagnosis, multidisciplinary approach and regular check-ups in patients with this syndrome.

Case report

3-month old girl (adjusted age – 1 month) with Cornelia de Lange syndrome was admitted to Pediatric Otolaryngology Department in order to further diagnose hearing loss. The patient has face dysmorphia, genetically confirmed CdLS, is fed by a feeding tube and is under multidisciplinary care. Previous hearing examinations were nondiagnostic.

The girl was born in 32nd week of gestation (3rd pregnancy, 3rd birth), weighing 1400 g, with a score of 5/6/6/6 in Apgar scale. The baby was delivered by a caesarean section, during the pregnancy IUGR, polyhydramnios, fetus dysmorphia and heart defect were reported. During neonatal period the patient experienced respiratory insufficiency (surfactant therapy), urinary tract infection, feeding problems, anemia of prematurity (treated with blood transfusions), neurologic problems. The patient also had temporary hypogammaglobulinemia and lymphopenia. Some laryngological problems were also reported: cleft palate, Pierre Robin sequence, disrupted sucking reflex and abnormal hearing test results. The child was also treated with gentamicin during its stay at neonatal clinic.

During examination the child was in good state, cleft palate was observed, besides that without abnormalities. The child is receiving propranolol (2 x 1 mg) for its congenital heart defect (DORV/VSD).

To assess whether the patient has hearing loss, otoacoustic emission (OAE) and auditory brainstem response (ABR) were performed. In OAE the results were to control (further tests are needed). Consequently, further diagnostics should be carried out and the functioning of the middle and inner ear should be monitored. In ABR in right ear: 500 Hz – 50 dB, 1000 Hz – 40 dB, click – 30 dB. In ABR in left ear: 500 Hz – 60 dB, 1000 Hz – 50 dB, click – 50 dB. Bilateral hearing loss was diagnosed. The patient is going to be diagnosed further.

Discussion

CdLS is often characterized by many otolaryngological symptoms. These include: craniofacial abnormalities (cleft/high-arched palate), laryngomalacia (due to reflux laryngitis) chronic middle ear effusions (due to eustachian tube dysfunction), anatomic abnormalities in cochlea and finally hearing loss (5, 6). In this syndrome occur many abnormalities in external, middle and inner ear, which can be observed in CT scans of the temporal bone. They are more common in patients with a classical form of the syndrome and their severity corelates with the advancement of hearing loss (7).

Hearing loss is very common in children with CdLS, reaching more than 80% of cases. Unfortunately, due to mental retardation and very early onset of symptoms, hearing examination can be hindered, which is why it is essential to use objective tests in order to properly evaluate those patients (7, 8). In our case OAE and ABR were used.

Both sensorineural and conductive hearing loss can be present in patients with CdLS (9). Numerous studies confirm that secretory otitis media (SOM) is an important cause of hear loss in children with this condition and that it occurs frequently in this population (7, 9, 10). Moreover, many patients with this syndrome experience significant problems with development of speech, even absence of speech in some cases, which can partially be caused by hearing impairment (11). That is why it is very important to assess these patients regularly from an early age and, if necessary, employ medical interventions (10, 12). What is more it could be beneficial for patients with CdLS and hear loss to be fitted with hearing aids, receive auditory training or be qualified for cochlear implantation. These procedures can result in better quality of life and audiological outcomes, even if speech abilities remain below average, and ABR examination can allow early assessment of hearing impairment and assessment in children with severe mental retardation (8, 13). Moreover early therapy can result in better outcomes, both in terms of tolerance and better auditory and speech development (8).

Our patient is being taken care of by a multidisciplinary team from a very early age and has already been examined to assess whether hear loss is present. Hopefully this can result in better outcomes in terms of her communicational skills and overall quality of life, for both the patient and her family. It is important to remember that regular check-ups with specialist are essential for adequate care and early intervention in patients with CdLS.

Conflict of interest
Konflikt interesów

None
Brak konfliktu interesów

Correspondence
Adres do korespondencji

*Lidia Zawadzka-Głos
Klinika Otolaryngologii Dziecięcej
Warszawski Uniwersytet Medyczny
ul. Żwirki i Wigury 63A, 02-691 Warszawa
tel.: +48 (22) 317-97-21
laryngologia.dsk@uckwum.pl

submitted/nadesłano:
13.07.2021
accepted/zaakceptowano do druku:
5.08.2021

References/Piśmiennictwo

1. Cascella M, Muzio M: Cornelia de Lange syndrome [Updated 2022 Feb 5]. StatPearls Publ [Internet]. 2022 Jan. Available from: https://www.ncbi.nlm.nih.gov/books/NBK554584/.
2. Selicorni A, Mariani M, Lettieri A, Massa V: Cornelia de Lange syndrome: From a Disease to a Broader Spectrum. Genes 2021; 12(7): 1075.
3. Kline AD, Moss JF, Selicorni A et al.: Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nat Rev Genet 2018; 19(10): 649-666.
4. Deardorff M, Noon S, Krantz I: Cornelia de Lange Syndrome [Updated 2020 Oct 15]. GeneReviews® [Internet]. 2005 Sep 16; Available from: https://www.ncbi.nlm.nih.gov/books/NBK1104/.
5. Eliason MJ, Melzer JM, Gallagher TQ: Cornelia de Lange Syndrome: What Every Otolaryngologist should Know. Ear Nose Throat J 2017; 96(8): E6-9.
6. Hamilton J, Clement WA, Kubba H: Otolaryngological presentations of Cornelia de Lange syndrome. Int J Pediatr Otorhinolaryngol 2014; 78(9): 1548-1550.
7. Kim J, Kim EY, Lee JS et al.: Temporal Bone CT Findings in Cornelia de Lange Syndrome. Am J Neuroradiol 2008; 29(3): 569.
8. Sakai Y, Watanabe T, Kaga K: Auditory brainstem responses and usefulness of hearing aids in hearing impaired children with Cornelia de Lange syndrome. Int J Pediatr Otorhinolaryngol 2002; 66(1): 63-69.
9. Marchisio P, Selicorni A, Bianchini S et al.: Audiological findings, genotype and clinical severity score in Cornelia de Lange syndrome. Int J Pediatr Otorhinolaryngol 2014; 78(7): 1045-1048.
10. Marchisio P, Selicorni A, Pignataro L et al.: Otitis media with effusion and hearing loss in children with Cornelia de Lange syndrome. Am J Med Genet A 2008; 146A(4): 426-432.
11. Grados MA, Alvi MH, Srivastava S: Behavioral and psychiatric manifestations in Cornelia de Lange syndrome. Curr Opin Psychiatry 2017; 30(2): 92-96.
12. Mikołajewska E: Interdisciplinary therapy in Cornelia de Lange syndrome – review of the literature. Adv Clin Exp Med 2013; 22(4): 571-577.
13. Psillas G, Triaridis S, Chatzigiannakidou V, Constantinidis J: Cornelia De Lange Syndrome and Cochlear Implantation. Iran J Otorhinolaryngol 2018; 30(101): 369-373.