Hojan-Jezierska et al., J Clin Case Rep 2018, 8:1
DOI: 10.4172/2165-7920.10001067

Journal of Clinical Case Reports

Case Report Open Access

Diagnosis of the Auditory System of a CCD Case Treated Surgically and Orthodontically

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Abstract

Cleidocranial dysplasia (CCD) represents a rare genetic disease characterized by disturbances in osseous structures, face dysmorphia, hypoplasia or aplasia of clavicles and numerous supranumerary teeth causing permanent teeth impaction. The study aimed at presenting a case of 12-year-old girl with CCD at the phase of active orthodontic treatment, involving successive extraction of multiple supranumerary teeth and repositioning of additional teeth using elastic traction. The patient was also subjected to a complex auditory examination which detected a hearing loss. Patients with CCD from their youngest years should be covered by a broad diagnosis and therapy, involving a comprehensive collaboration of multiple specialities.

Keywords: Cleidocranial dysplasia (CCD); Hearing loss; Impacted teeth; Supranumerary teeth

Introduction

Cleidocranial dysplasia (CCD) involves a rare genetic disease, manifesting an autosomal –dominant inheritance. The responsible mutation is located on the 6th chromosome, in the *RUNX2* (*CBFA1*) gene, responsible for development of osseous structures [1].

Cases and Methods

The most frequently noted signs of cleidocranial dysplasia include low stature, in males on the average 156.6 cm to 168.8 cm, in women 144.6 cm to 148.5 cm, facial dysmorphia, uni- or bilateral complete or partial aplasia of clavicles [2]. The face seems small since maxilla and zygomatic bone are hypoplastic, while the nose is broad, with indented base. The skull is large, with broad sutures and very large, occasionally incompletely fused fontanelles [2,3]. Moreover, other osseous defects may be manifested, such as clawed hip joints, deviation of vertebral column, scapular hypoplasia, abnormal number of ribs and other [2,4].

Oral cavity manifests high a high palate and small maxilla. Teeth manifest an abnormal anatomic structure, they are supranumerary and frequently impacted, so that they have to be exposed and surgically extracted [2,5,6]. Permanent teeth have to be repositioned in the arch using orthodontic forces [7-9].

In cleidocranial dysplasia cleft palates are also noted, which may result in conductive hypoacusis due to dysfunction of auditive tube [4]. Moreover, structural and functional alterations in temporal bone may promote abnormal function of external, middle and internal ear, resulting in hypoacusis [9].

This study aimed at presenting a case of 12-year-old girl with CCS syndrome in an active phase of orthodontic treatment, in whom a complete examination of auditory system was conducted.

A 12-year-old ZW girl, born following third pregnancy complicated with hydramnion in the 38th week by cesarean section, with detection of a very large fontanelle, broad cranial sutures and a slight facial dysmorphia. In the first year of her life, hypertrophy of pharyngeal tonsil and serous otitis media were diagnosed, the latter diagnosis repeated within the following year. The entire clinical pattern caused that she was referred to a genetic outpatient clinic, where the cleidocranial dysplasia was diagnosed. In general anamnesis neither developmental errors nor genetic diseases were noted in parents or two brothers of the girl. The persisting chronic otitis provided cause for ventillation draining of the middle ear. In the left ear a tube was applied, in the right ear a tympanotomy was used. In the subsequent year a conductive bilateral hearing loss was noted. The first orthodontic consultation took place at the age of 10 years. The extraoral examination en face documented evident frontal eminences, a broad nose with indented base, eyeballs positioned with traits of hypertelorism and a slight internal squint. The profile showed an increased naso-labial angle, hypoplastic maxilla and normal structure of mandible with a marked mento-labial sulcus (Figure 1). No disturbances could be detected in temporo-mandibular joints, the path of mandible abduction and adduction was normal. Intraoral examination detected a highly arched, gothic palate. The dental age was clearly delayed, presence of persisting immobile, deciduous teeth and a delayed eruption of permanent teeth were documented. Dental diagram was as follows: 55, 54, 53, 52, 51, 61, 62, 63, 64, 65, 26, 36, 75, 74, 73, 32, 31, 41, 42, 83, 84, 85, 46. General clinical examination documented low stature and abnormal structure of the chest with symmetrical steepness of shoulders and brachydactylia. The patient as able to bring her

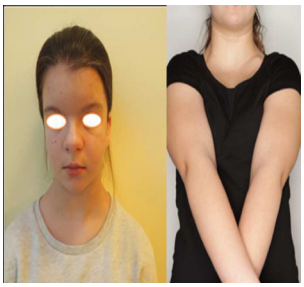

Figure 1: Extraoral documentation of ZW.

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Received December 18, 2017; **Accepted** January 16, 2018; **Published** January 22, 2018

Citation: Hojan-Jezierska D, Kuras M, Turska-Malińska R, Urbaniak M, Matthews-Brzozowska T (2018) Diagnosis of the Auditory System of a CCD Case Treated Surgically and Orthodontically. J Clin Case Rep 8: 1067. doi: 10.4172/2165-7920.10001067

shoulders close to body's median line, which confirmed the hypothesis of clavicular hypoplasia (Figure 1).

Conical tomography CBCT was performer in order to appraise supranumerous teeth and to evaluate their positions in maxilla and the mandible. Eleven supranumerous teeth were disclosed: 6 in the mandible and 5 in the maxilla. Moreover, CBCT examination demonstrated hypoplasia of maxillary sinuses. Both pantomogram and lateral teleradiogram confirmed the typical parallelism of the frontal and posterior edges of mandibular rami, linked to hypoplasia of masseter muscles (Figures 2 and 3).

The accumulated diagnostic material allowed that the team collectively worked out the plan of orthodontic-surgical treatment. The girl was qualified to surgery in the local anaesthesia. The treatment was started with extraction of partially resorbed deciduous teeth: the canine and first molar tooth on the left mandibular side and of supranumerary teeth in the region. Subsequently, the deciduous canine on the right side and the supranumerary tooth were extracted. After 2 weeks the child was supplied with removable appliance (two Schwarz plates with a screw). At the subsequent stage, deciduous upper incisors and supranumerary teeth were extracted, situated on the palatal side from the unerupted teeth in the region. A trapezoid cut was made at the apex of alveolar process, supranumerary teeth were extracted and the wound was sutured tight. Even if the plates were worn for 8 months, the unerupted central incisors failed to self-standingly erupt and, therefore, it was decided to uncover the unerupted teeth 11 and 21. The surgical treatment involved elevation of the muosal/periosteal flap from the trapezoid cut, exposure of crowns of central maxillary incisors by removal of the bone covering the unerupted teeth and application of orthodontic adhesive buttons, to which hooks were bound, bent from the ligature wire. The close technique of uncovering teeth was used, involving suturing of the gingival flap in such a way that only the wire hook communicated with oral cavity while the dental crowns remained covered with mucosa. The wound was supplied with sutures, removed after 7 days.

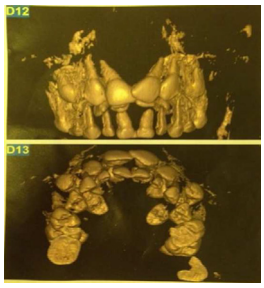

Figure 2: CBCT in ZW patient with CCD-the original condition.

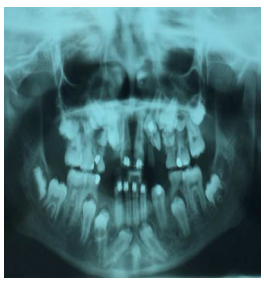

Figure 3: Pantomogram in ZW patient with CCD: condition in the course of orthodontic therapy.

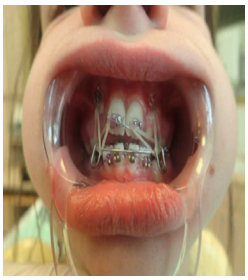

Figure 4: Intraoral photograph: the manner of active trans placement of permanent teeth in frontal maxilla to the arch.

Directly after the procedurę orthodontic treatment could be continued. The patient began to self-standingly apply the elastic tractions, attaching them from wire hooks to the labial arch in the upper Schwarz plate. After a month margins of teeth were visible in the oral cavity. After subsequent three months a similar surgical-orthodontic procedure was onducted for uncovering unerupted premolars in the mandible in order to transplace them to the arch. Half-a-year after the procedure of uncovering the medial incisors permanent upper canines were uncovered and they began to be transplaced to the lower plate as well as lower premolars using elastic tractions. Currently, the patient carries a fixed appliance on lower teeth, covering incisors and premolars (Figure 4).

Soon procedures are planned of removing supranumerous teeth in the premolar area of maxilla, uncovering the additional teeth in the premolar region of maxilla, uncovering lateral upper incisors situated on palatine side and continuation of replacement procedures on unerupted teeth using a fixed lower appliance. The subsequent stage will involve transplacement of lower canines to the arch. During the treatment another round of radiological diagnosis was conducted. A very good collaboration of the patient in carrying the elastic traction should be stressed.

Due to information on deterioration of hypoacusis, the patient was referred to the Chair of Biophysics Department of Hearing Healthcare, Poznan University of Medical Sciences in Poznań, Poland to conduct audiological studies checking efficacy of hearing organ. Before conducting all the tests, the patient was subjected to otoscopic examination for establishing patency of external auditory meatus and for evaluation of tympanic membrane.

A full set of tests was conducted, both subjective and objective one, including examination of air conduction thresholds for the range of frequencies from 125 to 16000 Hz, bone conduction thresholds for the range of frequencies from 250 to 4000 Hz. The examination took advantage of diagnostic audiometer, Madsen Itera II, Otometrics, high frequency earphones Sennheiser HD300 and bone conductance earphones. Thresholds of air and bone conduction was tested using the ascending 2/3 method. Determination of osseous conductance for the right and the left ears required masking. Moreover, classical tympanometry was conducted using clinical tympanometer, Madsen Zodiac 901 (Otometrics) for the test frequency of 226 Hz. This examination allows to estimate compliance of tympanic membrane, capacity of the outer auditory tract as well as to check and evaluate work of auditive tube. DPOAE otoemission was also examined, using the equipment of Madsen Capella 2, (Otometrics), which allows to evaluate relatively accurately passive and active processes taking place in the labyrinth. In addition, evoked auditory potentials induced in brain stem (ABR) were measured using ICS Chartr EP 200 equipment (Otometrics). For the study insert earphones were used. Using Titan

tympanometer (Interacoustic) a broad band tympanometry was performed, which permits to define slight abnormalities in tympanic cavity, as it involves frequency range of 226-8000 Hz and a broad band stimulus (a click). Due to the so broad range of frequencies, the measurement is more accurate than that in clinical audiometry and independent of outer disturbancs, such as a noise or movements of tested individual. In cases of tested children this property is very important [10]. Using this equipment the absorbance was measured which defines value of acustic energy absorbed by middle ear structures and resonance frequency of middle ear. This allows to check the frequency at which the mass and rigidity of auditory system exert the same effect on on admittance value of middle ear, allowing to evaluate anormalities in the chain of auditory ossicles [10,11]. The broad band tympanometer was used to examine reflexes from stapedius muscle or involuntary defensive reflex of hearing organ, protecting basement membrane of labyrinthus of from pronounced and acute sounds.

Results of Hearing Examination

Following otoscopic examination the patient was found to manifest no contraindications to conduct audiological studies.

Tonal audiometry disclosed a slight conductive hearing loss in the right ear, since the cochlear reserve ranged between 30 dB and 15 dB. The mean hearing loss calculated for four frequencies of 500 Hz, 1000 Hz, 2000 Hz and 4000 Hz amounted to 25 dB HL.

Hearing threshold of the left ear was normal in the entire range of frequencies although it contained a small conductive component since cochlear reserve was present. The tests proved the worsening of hearing in high frequencies (ranging from 8000 to 16000 Hz), observed for both, right and left ear (Figure 5).

The tympanometric curve for the left ear manifested type As, it was slightly flattened with normal pressure. Such a curve pointed to a reduced compliance of tympanic membrane. In the right ear the tympanometric curve manifested type A, with low amplitude or it was normal (Figure 6).

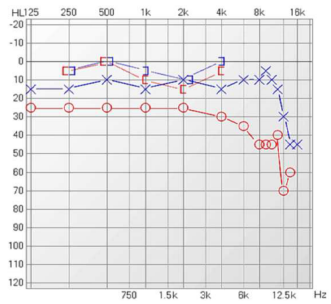

Figure 5: Hearing threshold for air and osseous conductance in both ears.

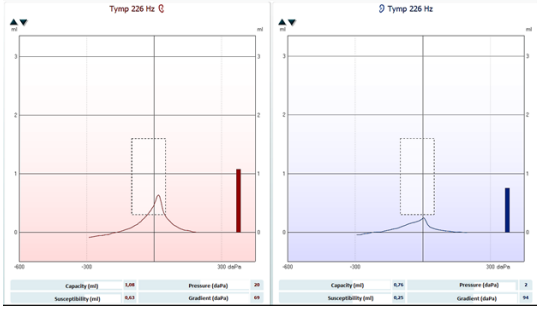

Figure 6: Results of impedance audiometry for the two ears.

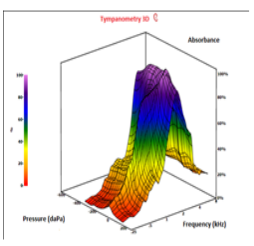

Figure7: Results of broad band tympanometry for the two ears.

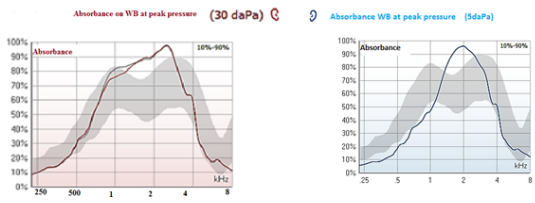

Figure 8: Spectra of absorbance for the two ears.

In the right ear no acoustic reflexes from stapedius muscle were recorded. The measurement was conducted only to 95 dB HL. It is possible that the reflex would manifest itself at a higher intensity, above 100 dB HL. In the left ear such reflexes are present for frequencies of 500 and 1000 Hz at the level of 95 dB HL and for the frequency of 2000 Hz at the level of 90 dB HL. This indicates manifestation of a protective reflex against sounds of an excessively high intensity. It aims at protecting the Cortis's organ against damage due to acoustic waves of an excessive energy.

Results of broad band tympanometry confirmed those obtained in classical tympanometry (Figure 7).

Absorbance, or the ability of absorbing sounds in the right ear was normal. In the left ear only the frequency range of 1500 to 3000 Hz remain normal while the other frequencies manifested lowered absorbance (Figure 8): Resonance frequency of 775 Hz for the right ear and of 891 Hz for the left ear). WB absorbance at the peak pressure (30 daPa) WB absorbance at the peak pressure (5 daPa).

Otoemission in the right ear was not recorded for the entire range of frequencies while in the left ear it was recorded only for the frequency range of 1000 to 4000 Hz, these results are consistant with results obtained in the audiometric examination. For the frequency range of 4000 to 8000 Hz otoemission of DPOAE was not recorded because in this range of frequencies hearing levels in tonal audiometry remained at the level of 40 to 70 dB HL (Figure 9).

Examination of auditory potentials evoked in the brainstem confirmed the result of tonal audiometry, latency of V wave in the left ear was slightly elongated, pointing to a slight conductive component. In the left ear V wave manifested a normal course up to the level of 40 nHL, for the lower intensities it could not be unequivocally defined. This reflected the fact that the patient could not rest peacefully during examination. Latency of V wave in the right ear reached even 9 ms and, therefore, the entire record of potentials was shifted to the right, typically for the conductive hearing loss. Moreover, the course of the function of latency vs intensity manifested shape typical for conductive hearing loss (Figure 10).

Discussion

The cleidocranial syndrome (CCD) represents a rare genetic disease, resulting from mutation of the *RUNX2* (*CBFA1*) gene [1].

DP Ear right

Diag. 1-8 kHz, 4 PPO 2016-11-09 10:05

F2	GM	L1/L2	DP1	NF	SNR	Result
996	904	64/55	8	-1	9	Accepted
1191	1078	64/54	4	3	1	Rejected
1416	1283	65/55	2	-4	7	Accepted
1679	1520	65/55	-6	-6	1	Rejected
2001	1812	65/54	-19	-8	-11	Rejected
2382	2157	65/54	-14	-16	2	Rejected
2832	2560	65/55	-9	-12	3	Rejected
3359	3041	65/54	-8	-12	4	Rejected
4003	3624	65/55	-15	-20	4	Rejected
4755	4304	64/55	-12	-21	9	Rejected
5654	5121	65/54	-10	-20	10	Rejected
6728	6092	65/55	-20	-18	-2	Rejected
7998	7239	66/61	-3	-9	6	Accepted

DP Ear Left

Diag. 1-8 kHz, 4 PPO 2016-11-09 10:07

F2	GM	L1/L2	DP1	NF	SNR	Result
996	904	64/54	-1	0	-1	Rejected
1191	1078	64/55	8	1	7	Accepted
1416	1283	65/55	2	2	0	Rejected
1679	1520	65/55	14	7	7	Accepted
2001	1812	65/55	5	-5	10	Accepted
2382	2157	65/55	4	-12	16	Accepted
2832	2560	65/55	4	-18	22	Accepted
3359	3041	66/55	0	-16	16	Accepted
4003	3624	66/55	0	-16	17	Accepted
4755	4304	64/55	-8	-15	7	Rejected
5654	5121	64/54	-12	-18	7	Rejected
6728	6092	66/55	-5	-11	6	Accepted
7998	7239	65/55	-15	-13	-2	Rejected

Figure 9: Results of otoemission for the two ears.

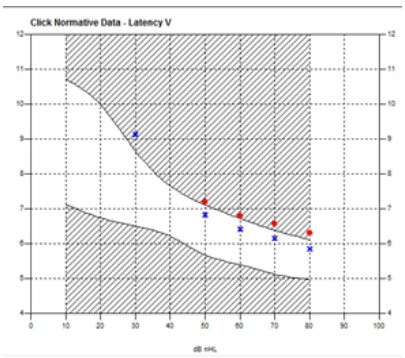

Figure 10: The latency vs intensity function for the two ears.

The phenotype of the syndrome includes hypoplasia of clavicles, a specific dental/occlusive pattern and facial dysmorphia. Literature of the topic stresses also manifestation of a conductive or sensorineural hearing loss.

In the described above case of a patient the spectrum of signs included also many supranumerary teeth. The surgical-orthodontic treatment of children and teenagers with CCD involves extraction of persisting deciduous teeth and stimulation of permanent teeth eruption after removal of blocking them additional teeth [12-14]. Out of the described in he literature procedures of management, such as Toronto, Belfast-Hamburg, Bronx or Jerusalem approach, the method resembling the Jerusalem approach was chosendue to the age of the patient and stage character of the procedures. The treatment was started with mobilization of upper central incisors, in an attempt to improve patient's outlook. Moreover, stimulation of dental eruption in the region allowed to reduce tendency to pseudoprognatism [14]. The optimum moment for starting the surgical-orthodontic treatment involves the time when the persisted incisors reach 2/3 of their expected length of roots [15]. The early treatment allows to avoid formation of primordial cysts [16] and dilaceration of roots, preventing an effective orthodontic treatment [17]. Due to dense trabeculation of bones teeth-surrounding bones, defective structure of cementum [18] and abnormal structure of PDL cells [7,18], the awaiting approach is improper. Therefore, in the presented case a gradual surgical-othodontic treatment was implemented but after completion of skeletal growth skeletal discrepance will have to be evaluated on the basis of a new cephalogram and the possible need for its treatment by orthodontic camouflage or orthognatic surgery will have to be considered.

It remains important to regularly control hearing of CCD patients to detect hearing disorders, which may develop due to structural or functional lesionsin temporal bone, dysfunction of auditory tube, induced by improper shape of the palate, narrowed auditory canals or abnormalities linked to formation of the auditory ossicles chain [10,19]. The discussed patient is regularly subjected to audiological diagnosis but till now no steps were taken to treat or prosthetically supply hearing of the patient. Following the set of audiological studies we could detect a slight conductive hearing impairment, in line with the literature data [1,10,11]. The literature indicates that patients with CCD are frequently suffered from infection of ears, which transform into otitis media [20]. Our patient in her early childhood frequently suffered from otitis media. Patients with CCD syndrome should be routinely examined by otolaryngologists an should be subjected to a complete audiological examination. In the presented above case examination of the hearing system will be repeated after completion of orthodontic treatment.

Conclusion

From their youngest years, patients with CCD should be covered by a broad diagnosis and therapy exercised by collaborating units specializing in stomatology, audiology, radiology, surgery and orthodontics. Upon such a partner collaboration on may expect good effects of rehabilitation.

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