[原著]Two cases of Dysostosis Mandibulo-Facialis

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Two cases of Dysostosis Mandibulo-Facialis

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Congenital atelia (= incomplete development) of maxilla and mandibula have been named Treacher-Collins' syndrome, Berry's syndrome, Franceschetti's syndrome, Dysostosis mandibulo-facialis, the first branchial arch syndrome, the first and second branchial arch syndrome, etc[1]. We experienced two cases of ateloprosopia accompanied by hearing loss, the details of which are reported below together with studies of the literatures mainly referring to development of ears.

CASE I: 5 year-old female

Chief complaints: Bilateral hearing loss and lalopathy

Past history: No anomalous development was seen at birth and after birth except facial region.

History of present illness: Unabled to speak until about 3 years old, probably due to hearing loss. Afterwards she began to talk, only simple matters, though she seemed to have good memory. No other specifities in general.

Family history: Parents, a sibling, all alive well. No consanguineous marriage. Hereditary diseases and hearing loss are not seen.

Clinical findings: Physically small but nutrition favorable. Neck, thoracoabdominal region and extremities were normal. Facial expression is specific, bilaterally palpebral fissures turned down outwards, being so-called antimongoloid (Fig. 1). Poor development of mandibula was seen. Both auricles are nearly normal positioned symmetrically, but morphologically anomalous and somewhat larger in general showing no auricular fold like auricular concha, scaphoid fossa, etc. (Fig. 2 and 3). Otoclesis was observed on both ears, only showing dents of 3 mm at right and 5 mm at left respectively. No other abnormalities with oral cavity, denture and pharynx.

Laboratory examinations: General blood test, urinal test, etc., indicated normal. Syphilitic reaction was negative, no abnormal finding with electro-encephalogram.

Audiometry: Bone conduction normal on both sides, air conduction bilaterally 50 – 60 dB on the average, showing bilateral conductive hearing loss.

Surgical findings:

Exploratory tympanotomy at left: External audiory meatus existed with cerumen. Something like tympanic membrane existed, and upon removing same entered into small cavity corresponding to tympanic cavity. Auditory ossicles not existed at all, oval window as well as round window not clear, tubal closing either.

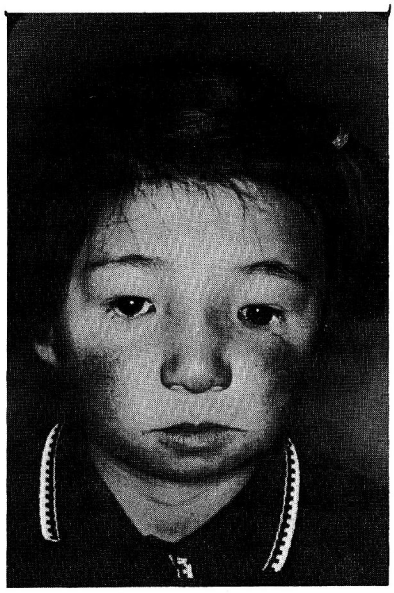

Fig. 1. The face of Case 1.

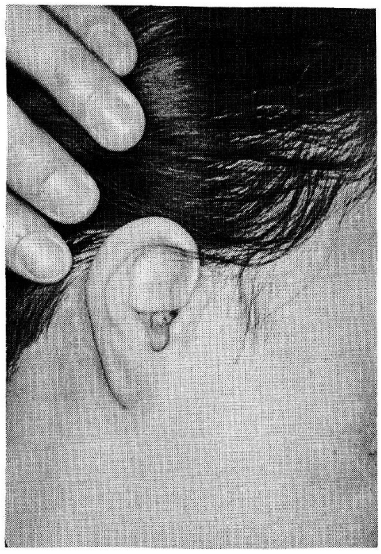

Fig. 2. The right ear of Case 1.

Exploratory tympanotomy at right: Entered from back of the ear. External auditory meatus obscure, only recognized osseous elevation at corresponding part. Zygomatic process looked like only two elevations having no contact with malar bone. Those corresponding to tympanic cavity and auditory ossicles not clear at all.

Post-operative hearing: Nearly the same level as before. Upon formation of external auditory meatus, hearing aid was mounted.

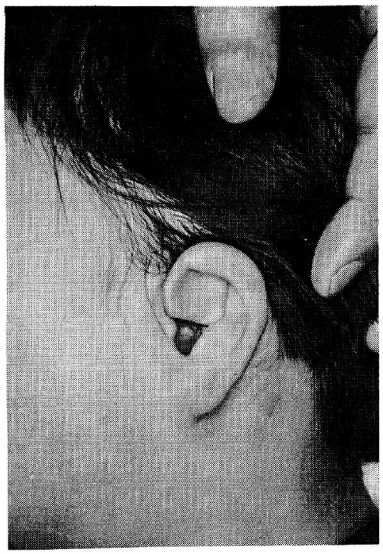

Fig. 3. The left ear of Case 1.

CASE II: 14 year-old male

Chief complaint: Hearing loss of the right ear

Past history: Nothing special abnormal with growth was seen at birth and after birth.

History of present illness: Up to 4 – 5 years old the boy was healthy with no abnormality noticed. About 5 years old, his hearing loss was noted after tonsillectomy and adenotomy.

Family history: Neither consanguineous marriage nor hearing loss observed.

Clinical findings: Physically built well and nutrition is favorable. Thoraco-abdominal region and extremities are normal. Facial expression: Bilateral palpebral fissures turned down outwards (antimongoloid). Maxilla developed comparatively well. Both auricles were normal in shape and position, with external auditory meatus. The tympanic membranes were present but strongly cloudy. There was hearing loss about 30 – 40 dB with left air conduction and about 50 dB with right,

bilateral bone conduction were within normal limit showing conductive hearing loss. X-ray examination on ears revealed incomplete development of mastoid antrum, mastoidal cells and tympanic cavity at left.

DISCUSSION

Referring to development of face[2)3)], the skull develops first at an early embryonal stage and face develops apparently in the fetal stage. During the 3rd and 4th weeks of gestation primordium of ears and eyes, and in the 7th week of gestation middle and external nasal processes are fused with maxillary process originated from the 1st branchial arch to make the upper layer, thus nasal cavity is separated from the oral cavity. Eyes move from exterior to interior and gradually fixed at the normal positions. Auricles are formed around hyomandibular cleft and positioned low, and face structure is nearly completed by the 8th week, i.e., development of the face starts from about the 4th week and almost completed in the 8th week.

Referring to formal genesis of ears[2)3)], genesis of inner ear differs from either middle or external ears. Inner ear is ectodermal, and differentiates from about the 3rd week of gestation. Membraneous labyrinth of cochlea and vestibule is completed in about the 8th week. Meanwhile, bony labyrinth is formed from mesodermal tissue around the otocyst. As to middle ear, starting from the 5th week mesodermal tissue proliferates between the first branchial cyst and the 1st branchial cleft, and the blind of the 1st branchial cyst expands to make tympanic cavity, while the interior becomes tuba auditiva. Mesodermal tissue around the tympanic cavity becomes mucous tissue, in which auditory ossicles are made. When primordium of auditory ossicle is cartilaginified, still the cartilage is in a continuous state. After some time auditory ossicles are said to lose their bonds and articles are developed therein. Among auditory ossicles bodies of malleus and incus are made from Meckel's cartilage of the 1st branchial arch in about the 6th week and completed by the 6th month. Crus longum of incus and stapes develop from Reichert's cartilage of the 2nd branchial arch at the end of the 4th week, which is completed in the 6th month. Tympanic membranes are made from mesoblastic band between external auditory meatus and tympanic cavity yielded on branchial cleft becoming thinner. Starting from about the 2nd month the lower part of the 1st branchial cleft sinks to be the primary external auditory meatus, and phorocyte on the bottom of which proliferates arriving at tympanic membrane, thus in the 7th month on this epithelial plate a cavity is made, which becomes the secondary external auditory meatus getting through the above primary external auditory meatus. At the end of the 1st month on the margin of the 1st and 2nd branchial arches surrounding the 1st brancial cleft, 3 each, totalling 6 processes are made, which are differentiated and fused to auricles.

The first report available on Dysostosis mandibulo-facialis is said to be the one studied by Berry on two cases of mother and child. After Collins made a report designating Treacher-Collins syndrome, the name has so far been widely know and used. Furthermore, Franceschetti and Zwahlen called this disease as Dysostosis madibulo-facialis and reported in detail about manifestations[4)]. According to their report, the major symptoms are as follows:

1) Turning down of palpebral fissures outwards (anti-mongoloid) accompanied by coloboma at lower part of the upper eyelid on looking downward.
2) Insufficient development of malar bone and madibula.
3) Macrostomia, hypsistaphylia, odontoparallaxis and malocclusion.

4) Deformation of external-, middle- and internal ears.
5) Blind-end fistulation between angle of the mouth and external ear.
6) Lingulate hair lines on cheeks.
7) Occasionally facial fissure, skeletal malformation and so on.

There is no established theory concerning the origin of the disease[5]. As stated about development of face before, a face at this time (embryonal stage, 2 month) is characteristic that compared with mandibula the upper half of the face is abnormally large. That is, eyes are wide open, mouth is extremely wide and auricles are positioned very low. When the growth is inhibited at this stage (embryonal stage, 2 month), micrognathia, macrostomia, abnormal positioning of auricles, and abnormalities on eyes as shown in Dysostosis mandibulo-facialis occur. An explanation is made in view of heredity[6] as one of the reasons, since heredity of this disease is seen in many cases as single onset, but when several cases are seen in one family tree, either of them appears in successive generations, and there is no case of consanguineous marriage, thus it is said to be probably an autosomal dominant heredity in view of the family tree.

In addition to the characteristic face, Case 1. identifies the presence of bilateral otocleisis and by exploratory incision on both ears defect of audityory ossicles has been identified. Referring to auditory ossicles morphological abnormalities, defects, etc., are reported[7]. The most popular one is malformation related to incus followed by malformation of stapes. The total defect of auditory ossicles as reported in this paper was also known in the reports of Suzuki and Sakurai[2)7)].

Referring to Case 2. anti-mongoloid eyes, ateliosis of mandibula, etc., suggest a form of Dysostosis mandibulo-facialis. No marked change is observed on external ears, external auditory meatus and tympanic membranes, however, hearing indicates a typical conductive hearing loss, which would be due to fusion and defect of auditory ossicles. It is earnestly hoped that efforts on detailed examinations by tympanotomy as well as improvement of hearing should further be extended.

CONCLUSION

Two cases of Dysostosis mandibulo-facialis with bilateral palpebral fissures turned down outwards as well as mandibular ateliosis were studied and reported together with some referential considerations.

In Case 1 otocleisis and total defect of auditory ossicles were observed, while in Case 2 malformation of auditory ossicles was considered from the image of hearing although no tynpanotomy was performed yet.

Development of face, mainly related to ears, were studied bibliographically.

The summary of this paper was reported at the 170th Regular Meeting of the Nihon University Medical Association and the 7th Regular Meeting of Okinawa Branch, the Oto-Rhino-Laryngological Society of Japan.

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