Auris Nasus Larynx xxx (2016) xxx–xxx

Contents lists available at ScienceDirect

Auris Nasus Larynx

journal homepage: www.elsevier.com/locate/anl

Tympanoplasty for chondrodysplasia punctata: Case report

Makoto Hosoya, Sho Kanzaki *, Satoko Wakabayashi, Kaoru Ogawa

Department of Otorhinolaryngology-Head and Neck Surgery, Keio University School of Medicine, 35 Shinanomachi, Shinjuku-ku, Tokyo 160-8582, Japan

ARTICLE INFO

Article history:
Received 2 May 2016
Accepted 2 September 2016
Available online xxx

Keywords:
Chondrodysplasia punctata
Hearing loss
Tympanoplasty

ABSTRACT

Chondrodysplasia punctata (CP) is a systemic disorder of chondrogenesis. The most prominent features of patients with CP are abnormal faces characterized by a flat nose and short stature. CP patients show various types and levels of hearing loss. This disease is rare, and no successful tympanoplasties with hearing recovery have been reported. Here, we report on a CP case, in which hearing recovery was successfully treated with tympanoplasty.

1. Introduction

Chondrodysplasia punctata (CP) is a systemic disorder in which chondrogenesis is impaired [1]. The most characteristic features of CP patients are their distinct faces (large forehead, flat nose due to facial hypoplasia) and short stature. CP derives its name from the radiographic appearance of bones and cartilage, which show abnormal cartilaginous stippling resulting from aberrant deposition of calcium during endochondral bone formation. This disease comprises a clinically heterogeneous group of bone and cartilage dysplasias. Etiologies of this disease are diverse, with some subtypes caused by metabolic deficiencies (e.g., vitamin K and cholesterol deficiency), maternal systemic lupus erythrematosus (SLE), or maternal alcoholism [1]. Disease severity also varies widely. As mentioned above, the most common symptoms are distinct facial morphology (e.g., depressed nasal bridge secondary to maxillofacial hypoplasia) and a short stature with short limbs. Many CP patients are developmentally delayed and exhibit mental retardation. Life expectancy in most cases is short [1].

In otological exams, some CP patients are found to have hearing loss [2,3]. Although all types of hearing loss (sensorineural, conductive, or mixed) can be present, types and severity vary among individuals with CP and depend on disease subtype.

2. Patient

We present here a 17-year-old male diagnosed with CP in childhood based on X-ray findings and phenotypic presentation (i.e., short stature, characteristic face with hypoplasia of facial bone and abnormalities of fingers (Fig. 1)). He showed no obvious mental retardation. From childhood onward, he was hard of hearing and had left unilateral mild conductive hearing loss in an auditory test (Fig. 2, left). Stapedius reflex was absent in left. CT scan revealed low absorption areas in the incus and stapes (Fig. 3). These findings suggested conductive hearing loss caused by ineffective sound conduction due to insufficient calcification of the left incus and stapes, resulting in reduced rigidity.

3. Surgical approach

We performed left-ear tympanoplasty to assess the actual condition of the left incus and stapes and to restore hearing

* Corresponding author.
E-mail address: skan@a7.keio.jp (S. Kanzaki).

http://dx.doi.org/10.1016/j.anl.2016.09.001
0385-8146/

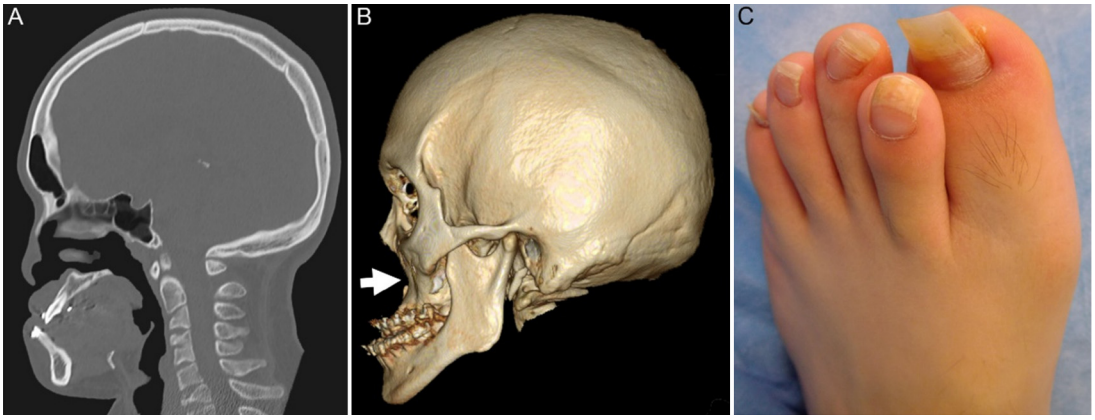

Fig. 1. Hypoplasia of facial bone (A: head CT, B: 3D image) and (C) abnormalities of fingers.

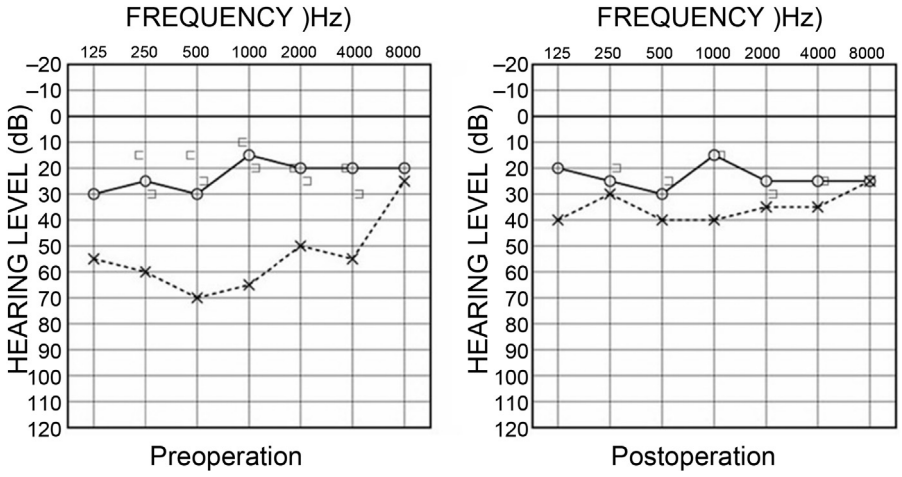

Fig. 2. Preoperative audiogram showing mild conductive hearing loss of the left ear. Postoperative audiogram showing hearing improvement.

conduction. During the operation, we found hypoplasia of the incus and head of the stapes with being replaced by pale color cord-like structure (Fig. 4). Direct visual inspection of the stapes footplate revealed it was normal and flexible. To restore sound conduction, we performed type IVc tympanoplasty, attaching the columella to the footplate of the stapes. After tympanoplasty, hearing recovered (preoperative hearing threshold: 61.7 dB, postoperative hearing threshold: 38.3 dB) and the air-bone gap was reduced about 25 dB (Fig. 2, right). Improved hearing was maintained during postoperative observation period (over 3 years).

4. Discussion

CP is a very rare and heterogeneous disease, having various etiologies, types, and levels of symptom severity (Table 1). These sometimes include mental retardation and short lifespan. The life expectancy of many CP patients is short, but some live longer than expected. Long-lived patients sometimes suffer from hard of hearing. In the current case, precise etiology is unknown, however based on the accompanying systemic complications (hypoplasia of facial bones without developmental delay) maternal SLE type was

Table 1
Etiologies of CP.

Chondrodysplasia punctata	Inborn errors of metabolism	Abnormalities of peroxisomal function
		Abnormalities of cholesterol synthesis
		Other metabolic disorders
	Disruption of vitamin K metabolism	
	Chromosomal abnormalities	
	Unknown etiology	Maternal factors (SLE, fetal alcohol), etc.

Modified from Ref. [1].

Please cite this article in press as: Hosoya M, et al. Tympanoplasty for chondrodysplasia punctata: Case report. Auris Nasus Larynx (2016), http://dx.doi.org/10.1016/j.anl.2016.09.001