


# Major Ear Aplasia and Cochleovestibular Dysplasia: Rare Congenital Malformation about a Case

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## Abstract

**Introduction:** The congenital malformations of the ear are rare and often isolated, may be unilateral or bilateral, and can be associated with another syndromic malformation. Such malformations so not necessarily impact aesthetics and social relations. **Case Presentation:** The authors report the case of Samuel M, male born at 38 WA, who is the first child of healthy parents from the same socio-cultural area. His birth weight was 2800 g and he did not have any risk factors for deafness or concept of obstetrical trauma. He presented with congenital malformation manifesting as bilateral ear aplasia with unilateral facial paralysis. Computed tomography revealed abnormalities of the inner ear; functional explorations, such as PEA and OEA, showed findings in favor of bilateral cophosis. ASSR (Auditory Steady-State Responses) was not performed. The announcement of the serious diagnosis of deafness requires multidisciplinary care in order to plan a therapeutic program to limit the impact on the development of language, schooling, and consequently, the socio-professional future of children. **Conclusion:** This clinical case underlines the fact that interest of the clinical interview before possible multiple surgery does not always guarantee the satisfaction of the desire for repair in this type of patient.

## Keywords

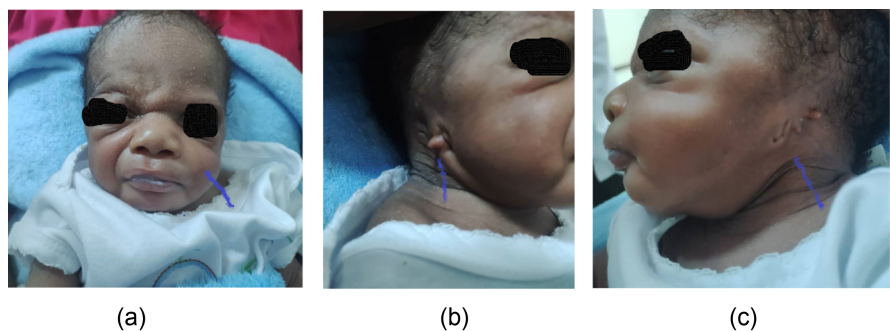
Ear Malformations, Congenital Malformations, Major Aplasia, Congenital Facial Paralysis, Congenital Deaf-Mutism

## 1. Introduction

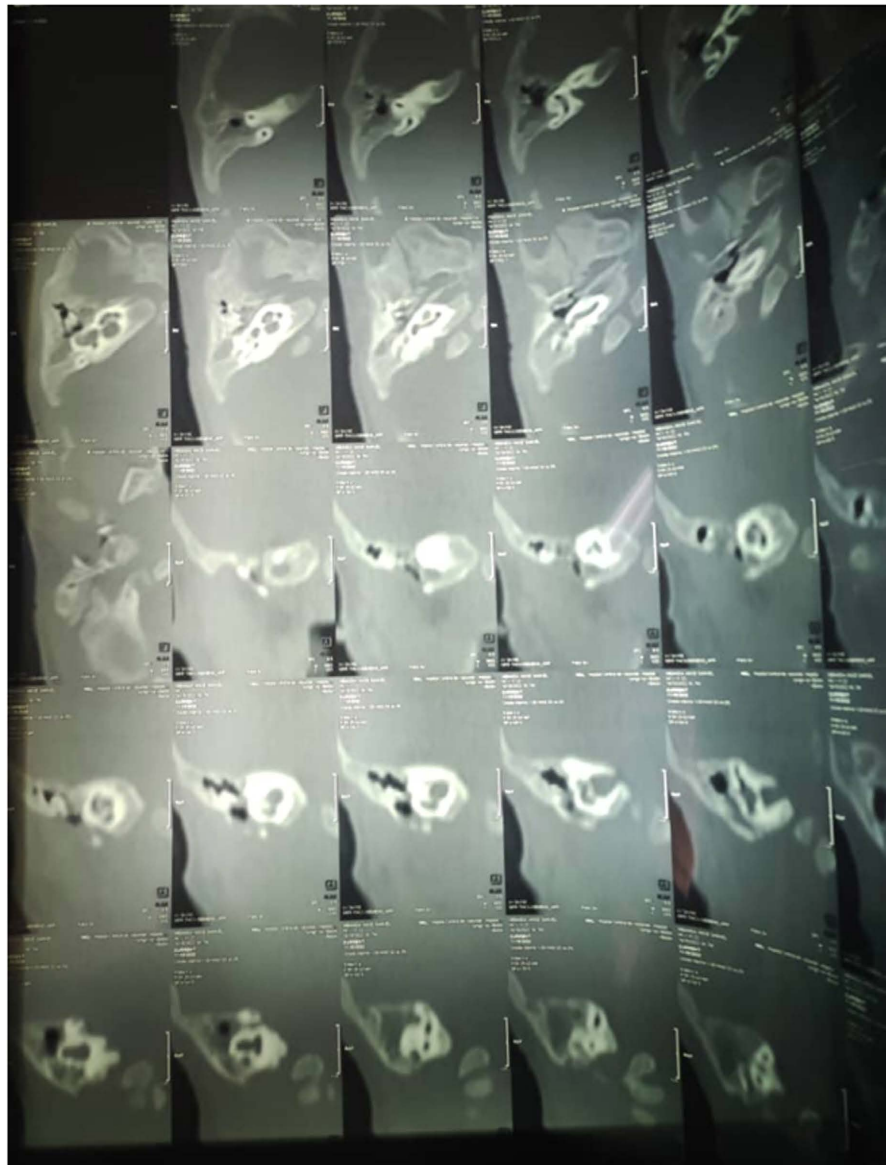
Congenital defects, also called birth defects, are physical defects that are already present before birth. “Congenital” means “present at birth”. Bilateral major ear aplasia is a rare complaint in ENT (Oto-Rhino-Laryngology) consultation. When this aplasia is associated with facial paresis and bilateral cochleovestibular dysplasia, this discovery is often made during the evaluation of a congenital deafness in the child. The World Health Organization has stated in its global report on hearing that by 2050 nearly 2.5 billion people will be living with hearing loss [1]. As a result, inaction will be costly [2] in terms of exclusion, education, communication, and employment. The number of children born with congenital anomalies is very low and to the best of our knowledge, no cases have been described in Cameroon. We report through this case study, the correlation between the morphological aspect and functional impairment in an environment where there is no systematic neonatal screening of organized deafness and where cochlear implantation (CI) is just starting to gain recognition.

## 2. Case Presentation

The patient is a 3-day-old boy born at 38 WA, brought to consultation for an ear anomaly discovered by his mother. The mother had a normal pregnancy, labor, and delivery. He is the first child of hearing parents (father, 45 years; mother, 35 years) from the same socio-cultural area. His birth weight was 2800 g and his temperature 36.5°C. He has no risk factors for deafness or notion of obstetrical trauma. A clinical examination showed a congenital malformation manifesting as bilateral ear aplasia grade III and IV with unilateral facial paralysis (facial wrinkle removal, Charles Bell sign) (Figures 1(a)-(c)). He had no additional congenital morphological malformations. Computed tomography (CT) scan revealed bilateral cochlea-like inner ear abnormalities with 1.5 turns (Figure 2). Functional explorations revealed an absent V wave on the AEP (Auditory Evoked Potentials); the OAE (Acoustic Otoemissions) was inconclusive (Figure 3). Cardiac and abdominopelvic ultrasound were normal. The ASSR (Auditory Steady-State Responses) has not been performed. After consulting the related specialties, namely pediatric surgeon, maxillofacial surgeon, ophthalmologist,



**Figure 1.** Image of Microtia with unidentifiable reliefs grade III of Meurman (a), of Anotia grade IV of Meurman (b) and left congenital facial paresis (c).



**Figure 2.** CT Scan image showing cochlea with 1.5-turn bilateral coils.

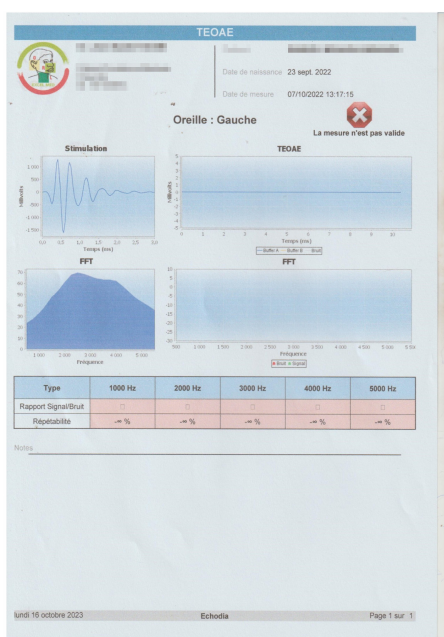
psychologist, we concluded that there is a deep bilateral deafness and major bilateral aplasia of the ear associated with bilateral cochleovestibular dysplasia. We introduced vaccination according to the EPI (Expanded Vaccination Program) and referred the child to the implantology team.

### 3. Discussion

The ear is divided into three parts: the outer ear, the middle ear, the inner ear. Bilateral congenital ear aplasia is a rare malformation with an incidence of 0.83 - 17.4/10000 births, and is present and visible at birth [3]. It is more common in boys [4], who accounts for 15% - 20% of cases [3] [4]. Embryologically, this obvious ear aplasia results from an early involvement of the fusion process of the 06 “collicles of His” occurring between the 4th and 12th week of intrauterine life



(a) (b)



(c)

**Figure 3.** Bilateral image of OAE (a), (b) and AEP (c) showing respectively an inconclusive test and an absence of wave V.

[5] [6]. The cause as described in the literature is either syndromic (familial) or sporadic (multifactorial), as in this case [3]. These atrial formation abnormalities have varied clinical presentations and are either isolated or form a part of a malformative syndrome (otomandibular syndrome, Goldenhar syndrome, Franceschetti syndrome, etc.). Apart from the Altmann classification [7], several classifications [3] have been proposed for adequate management, which include the ones by Tanzer (1978) and Weerda (1988), but the classification given by Meur-

man is the most quoted in Europe, which is contrast to America [4]. It is representative of the four main types of damage: horned ear (grade I), microtia with verticalized chondro-cutaneous bead but main identifiable reliefs (grade II), microtia with non-identifiable reliefs (grade III), and anotia (grade IV). **Figure 1** allowed us to classify our clinical case and thus consider the probable repair hypotheses for this defect. However, in 80% to 90% of cases there is no damage to the inner ear. The clinical case presented with profound bilateral deafness with major bilateral aplasia of the ear associated with bilateral cochleovestibular dysplasia. A few cases associating major ear aplasia and cochleovestibular dysplasia have been described, but the prevalence remains unknown. It is a congenital anomaly resulting in the formation of an incomplete cochlea composed of a turn and a half of spire. Such malformation constitute 26% of malformations of the inner ear and 13% of all malformations of the ear; only 20% of these malformations present with radiological anomalies [8]. Several classifications of these dysplasias have been proposed in order to further study this congenital anomaly. Diagnostic methods include clinical examination, hearing tests, sometimes skull imaging examinations, genetic tests. The initial assessment consists of a complete ENT examination, opinions of maxillofacial surgery, pediatric surgery, psychology, ophthalmology, and when possible, a genetic investigation and examination to detect any associated lesions. Auditory functional explorations of screening and the subsequent confirmation, including subjective and objective tests, such as acoustic otoemission, early AEP, and even ASSR (auditory brainstem response or auditory steady-state response test), must be carried out as soon as possible and according to a precise chronology and age [9] [10]. CT is an essential part of the operability of malformations. Its purpose is to identify the facial/cochleovestibular nerve. In this case, the anomaly turned out to be facial paresis; other structures, such as the otic capsule, and the pneumatization vessels were also to noted by the radiologist. Although extremely expensive in our context, combined functional explorations and imaging techniques allow a very good evaluation of the cases presented to us. Imaging (CT scan, MRI) according to age, context, accessibility is a systematic part of the diagnostic assessment. Two situations therefore arise: useful and non-urgent imaging, urgently essential imaging. CT may be useful in special situations, malformations and trauma. The two examinations are necessary before cochlear implantation. Magnetic resonance imaging can be used to identify the nerves for CI. It is notable that in case of bilateral major aplasia, the hearing loss varies from 60 to 70 dB associated with cochleovestibular dysplasia a loss of 30 to 40 dB on acute frequencies that sometimes extends to severe frequencies, or is confined to a cophose, thus prohibiting any attempt at classical functional rehabilitation [11]. Deafness is a symptom defined by a loss of hearing, regardless of its importance, whatever its etiology. Auditory functional explorations dedicated to congenital anomalies are non-invasive methods: early auditory evoked potentials (AEP), known as trunk cerebral, Otoacoustic emissions (OAE). they have limits, indeed the presence of otoemissions does not make it possible to eliminate deafness by auditory

neuropathy, nor to affirm that the child will not have later deafness, the auditory evoke potentials do not allow a frequency-by-frequency study of the responses, the depth of the deafness can interfere with the interpretation of the curves for the analysis of the latencies. Deafness is therefore of the deep neurosensory type. Treatment may be either reconstructive surgery to create a normal-looking outer ear and to create an external auditory canal, or consist of hearing aids. Most often, in cases of major aplasia, it is impossible to use an air conduction device; a bone conduction device is then used which can be external, applied to the mastoid or directly through an implant. An alternative option envisaged to surgery consists of placing an external prosthesis in silicone: pavilion epithesis. The advantages seem multiple: non-invasive technique, good aesthetic result, prosthesis removable, painless installation. The main drawback in the use of an epithesis lies in the choice of the fixing system (glued or implants). The treatment will therefore consist of the stimulation of the auditory pathways as early as possible, *i.e.*, from 4 months using bone conduction equipment (Baha®, PontoTM) on a headband [10], followed by cochlear implants intended to treat sensorineural hearing loss severe to profound bilateral. They are rehabilitation devices hearing intended to allow the restoration or the development of the oral communication. An improvement in hearing is observed on the abilities hearing, language skills and verbal communication. Progression on these criteria is major in the months following implantation and continues for the long term. Polyhandicap is no longer a contraindication to this treatment according to recent literature. Primary prevention does not exist and support for children and families through psychological support and the creation of a care network is the paramount at the school level [10]. All this requires an adapted technical platform, something that is not found in sub-Saharan Africa, although the WHO objectives are aimed at sustainable development (SDGs) in health. The people of Africa and the rest of the world have hope for these Millennium Development Goals.

#### **4. Conclusion**

The morphogenesis of the ear responds to complex mechanisms that are still poorly understood. Parents/caregivers of patient with outer and inner ear abnormality also experience difficulties that may be the subject of further study. Indeed raising a child with craniofacial abnormalities is challenging and stressful due to the multiple medical appointments, functional problems, teasing, psychosocial problems.

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#### **Data Availability**

The data that support the findings of this study are available from the corres-

pending author upon request.

## Consent of Parental Rights

Written informed consent was obtained from the patient's mother for publication of this case report and all accompanying images.

## Conflicts of Interest

The authors declare that they have no conflicts of interest.

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