The Egyptian Journal of Radiology and Nuclear Medicine (2012) **43**, 535–542

Egyptian Society of Radiology and Nuclear Medicine

The Egyptian Journal of Radiology and Nuclear Medicine

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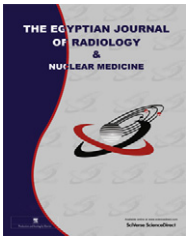

ORIGINAL ARTICLE

Revisit to congenital anomalies of the inner ear: The spectrum of aplastic/dysplastic labyrinthine malformations (ADLM). A new concept for classification☆

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Received 29 April 2012; accepted 17 July 2012
Available online 24 August 2012

KEYWORDS

Aplastic/dysplastic labyrinthine malformations;
Congenital anomalies;
Inner ear;
Sensorineural hearing loss;
Cochlear implant

Abstract *Aim:* The aim of this work is to establish a new radiological classification system for the congenital anomalies of the inner ear depends on congenital gross morphological changes that occur in the bony labyrinth as seen with today's multi-slice CT imaging technology.
Material and methods: This study was conducted on 26 congenitally deaf ear showing gross morphological changes of the bony labyrinth as seen with today multi-slice CT machines. The concept of ADLM radiological classification system depend on that both aplastic and dysplastic congenital anomalies of the bony labyrinth are linked. One or more component(s) of the labyrinth may be aplastic and other(s) may be dysplastic.
Results: The cochlea was the most common component of bony labyrinth prone to aplasia appeared in 7 ears (26.9%) followed by semicircular canals (19.23%). The vestibule was the most common component of the bony labyrinth resistant to aplasia and on the other hand it was the most common labyrinthine component that was prone to dysplasia.
Conclusion: ADLM is a numerical system that gives a total idea to the referring physician about the status of the bony labyrinth in a short simplified way. It could entail these anomalies with high degree of compliance.

1. Introduction

Mondini in 1791 described flat cochlea having 1.5 turns instead of the normal 2.5–2.75 turns. He also described a large vestibule. However, many anomalies of the inner ear continue to be documented and have a wide range of pathologies that have had variable classifications over years. The traditional nomenclature used to describe these congenital anomalies of the labyrinth involves a confusing array of eponyms that stem from the first reports usually by the 18th and 19th century authors

☆ Poster presentation in scientific exhibit of 43rd annual meeting of the American Society of Head and Neck radiology (ASHNR), New Orleans, USA, 7–11 October, 2009.

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Peer review under responsibility of Egyptian Society of Radiology and Nuclear Medicine.

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like Mondini, Bing-Siebenmann, Schiebe and Alexander dysplasia as well as Michel deformity (Complete labyrinthine aplasia). The original classification of these anomalies was by discrete patterns of histopathological change described by Valvassori et al. (1) in 1969.

In 1978, Valvassori and Clemis (2) drew attention to the large vestibular aqueduct malformation and its accompanying hearing loss. The width of the vestibular aqueduct lumen within the temporal bone cortex close to the endolymph sac and the posterior fossa dura is normally less than 1.5 mm (3–5).

Based on embryogenesis, another attempt to make this topic logical, and more clinically relevant, was by Jackler et al. (6) in an article published in Laryngoscope in 1987. A descriptive classification system taking into consideration the traditional eponyms was established. The congenital anomalies of the inner ear were classified into congenital malformation limited to the membranous labyrinth, anomalies of the osseous and membranous labyrinth and eighth nerve anomalies. Several forms of aplasia and dysplasia as well as the common cavity anomalies were described (6). This classification system seems to depend on histopathological basis and defined clear limits between different entities, more than actually seen in the imaging practice.

Phelps (1990) (7) described the pseudo-Mondini malformation, which resembles the Mondini malformation. The developmental arrest occurs during an early stage of cochlear partitioning. On CT, as in the Mondini malformation, the cochlea is short and the apical turn is absent, but in the pseudo-Mondini malformation, the basal turn is enlarged. Clinically, the hearing loss is total. However Mondini malformation in now an obsolete term with multiple confusing definitions (8).

Cystic cochleovestibular anomaly (CCVA) and large endolymphatic sac anomaly (LESA) or its synonym, the large vestibular aqueduct syndrome (LVAS) are now used to describe the dysplasia involving different portions of the labyrinth (8). Sennaroglu and Saatci (9) proposed the term of incomplete partition (IP) in 2002. IP type I was equal to CCVA and IP type II defined as cochlea with 1.5 turns (coalesce of middle and apical turns) accompanied by dilated vestibule and enlarged vestibular aqueduct.

Nowadays, there are anomalies that have been and continue to be documented with thin-section, high resolution multi-slice CT that do not fit neatly into single entity (10). On the other side, many reported congenital anomalies showed features belong to more than one entity (11–16).

The introduction of cochlear implantation surgery enhanced the radiological interest in that topic. This new developing era is now offering a promising hope in the management of pediatric sensorineural hearing loss (SNHL), regaining one of the human basic senses and significantly help in rehabilitation of handicapped children (17–19).

Cochlear implantation (CI) is an effective rehabilitation method for profoundly hearing impaired patients who do not benefit from hearing aids. It is a multi-component electronic device that provides auditory information by direct stimulation of auditory fibers in the cochlea (17–19). Establishment of a classification system that depends primarily on congenital gross morphological changes that occur in the bony labyrinth as seen with today's imaging technology, will be of great help for surgeons before cochlear implantation.

The role of radiologist in the preoperative assessment is to identify absolute and relative contraindications of surgery as well as to elicit the pre-implantation findings that may complicate surgery (8).

The aim of this work is to establish a new radiological classification system for the congenital anomalies of the inner ear depends on congenital gross morphological changes that occur in the bony labyrinth as seen with today's multi-slice CT imaging technology.

2. Material and methods

This work included 18 patients, two thirds (twelve patients) were females and the last third were males. Eight patients (44%) had bilateral involvement. The total studied ears were 26. The patient's age ranged between 1 to 16 years. The mean age was 6.22 years. All of these children were suffering from congenital deafness and refereed for radiological assessment prior to cochlear implantation surgery. All of them were selected on the base of the presence of gross morphological changes in the bony labyrinth that could be visualized with today's imaging MDCT technology. The congenitally deaf ears showed no definite gross morphological changes and possible anomalies at the microscopic levels were not included in this study.

Eight slices multi-detector CT machine (Bright speed S, GE, USA) was used in assessment of 12 patients. Six patients were assessed using 64 multi-detector CT machine (Light speed, GE, USA). Before imaging, the patient was informed about the investigation and instructed not to move during scanning. The examination was performed under general anesthesia in 12 children. The patients were in supine position. A lateral scout view was taken and used for planning the axial images. Axial images were taken without any angulations (Tilt 0). The protocol was 120 mA, 130 kV, 0.6 mm slice thickness and head field of view (FOV). The scanning covered all components of the temporal bone.

All images were prospectively reconstructed at 0.6 mm with 0.4 mm overlap; using soft tissue and high-resolution bone filter. The reconstructed axial images were transferred to Advantage 4.4 GE, USA workstation for manipulation of data. Then multi-planar reformation (MPR) and minimum intensity projection (Min. IP) were generated in different planes. Using dedicated reconstructive software for the bony labyrinth, volume rendering images (VR) of the entire labyrinth were generated (Fig. 1).

MRI of the internal auditory canals was also performed confirming the presence or absence of cochlear nerve using 1.5 T magnet (Signa, General Electric Medical Systems, Milwaukee, WI, USA) using 8-channel head coil. The MRI protocol included steady state free precession technique (FIESTA sequence). Slide thickness was 3 mm with 0 interval. FOV was 23.0.

3. The classification system

The concept of aplastic–dysplastic labyrinthine anomaly based on that both aplastic and dysplastic congenital anomalies of the labyrinth are linked. One or more component(s) of the labyrinth may be aplastic and other(s) may be dysplastic. So all of the congenital anomalies of the labyrinth could be rearranged in a single spectrum with suggested term of aplastic–dysplastic labyrinthine malformation (ADLM).

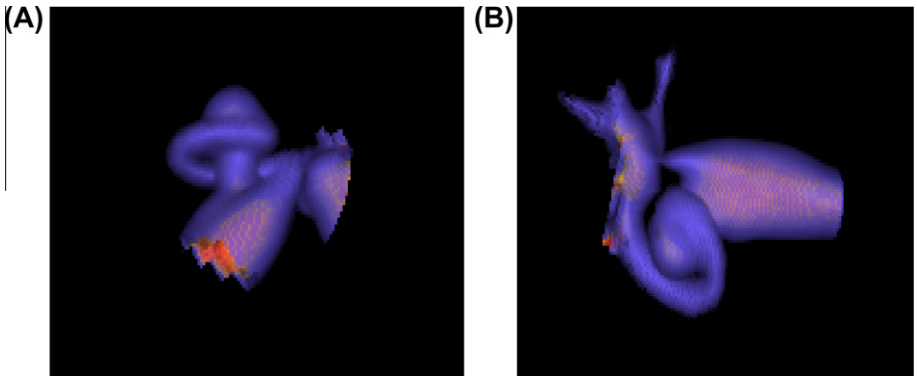

Fig. 1 (A and B) Volume rendering (VR) reconstruction of the cochlea, showing the turns of the normal cochlea from different angles.

Table 1 For simplicity each component of the labyrinth is given a numerical number as follows.

Number	Component of the labyrinth
1	Cochlea
2	Vestibule
3	Lateral semicircular canal
4	Superior semicircular canal
5	Posterior semicircular canal
6	Vestibular aqueduct

Each component of the labyrinth is given a numerical number as shown in Table 1. Letter A was given for aplasia and letter D was given for dysplasia. Letter N for the VIII nerve: N0 the nerve is present, N1 the nerve is absent. A(1–6) according to the aplastic component. D (1–6) according to the dysplastic component. N for the nerve.

3.1. Examples

A1 D2 N1 anomaly means that the cochlea is aplastic with cystic dysplasia of the vestibule. The VIII cranial nerve is absent.

D 3,4,5 N0 anomaly means that there is cystic dysplasia of all semicircular canals, The VIII cranial nerve is present.

4. Results

The patient's age ranged from 1 to 16 years. Among the 18 patients, two thirds (twelve patients) were females and the last third were males. All the male patients ($n = 6$) had unilateral involvement. Two thirds of the female patients had bilateral involvement. More than 44% ($n = 8$) of studied cases ($n = 18$) had bilateral involvement.

The cochlea was the most common component of bony labyrinth prone to aplasia appeared in 7 ears (26.9%) followed by semicircular canals (19.23%). The vestibule was the most common component of the bony labyrinth resistant to aplasia. It was aplastic in only one case (2 ears, 7.69%) showed bilateral A1–6 anomaly (bilateral total labyrinthine aplasia). On the other hand, the vestibule was the most common labyrinthine component that was prone to dysplasia. The vestibular dysplasia was noted in more than 92% of the studied ears, followed by lateral semicircular canal, cochlea, superior semicircular canal, and vestibular aqueduct which were involved in 80.77%, 61.54%, 46.15% and 23.08% respectively. The posterior semicircular canal dysplasia was seen in less than 12% of ears and it was the rarest (Table 2).

Table 2 Distribution of different congenital anomalies of the inner ear ($n = 26$ ears) according to the concept of ADLM.

Aplasia	Number of affected ears		Dysplasia	Number of affected ears	
	No.	%		No.	%
A1	7	26.92	D1	16	61.54
A2	2	7.69	D2	24	92.31
A3	5	19.23	D3	21	80.77
A4	5	19.23	D4	12	46.15
A5	5	19.23	D5	3	11.54
A6	2	7.69	D6	6	23.08

Cochlear aplasia (A1 anomaly) appeared in 7 ears (26.92% of the studied ears). Six of them had unilateral involvement. A case of bilateral A1 anomaly was recorded as a component of bilateral A1–6 anomaly (bilateral total labyrinthine aplasia or Michel deformity) (Fig. 2). In this study all ears with A1 anomaly ($n = 7$) showed absent cochlear nerve (N1 anomaly) as confirmed with MRI imaging. Otherwise, the cochlear nerve was present in all other cases except for a case of Patau syndrome showed unilateral A3 D1,2,4,5 N1 anomaly.

Aplasia of semicircular (SCC) canal affected the lateral, superior and posterior SCC with equal percent (19.23% of the affected ears). Total semicircular canal aplasia (A3,4,5 anomaly) appeared in one case of bilateral involvement. In this case, the anomalies were addressed as A3,4,5 D1,2 N0. Other congenital anomalies were also encountered as congenital heart disease. CHARGE syndrome was diagnosed in this case.

Dyspalsia of the inner ear presented morphologically as dysmorphic feature and cystic like dysplasia. The vestibule followed by lateral semicircular canals (D2 and D3 anomalies) were the most common compartments of the labyrinth showed dysplasia. Associated D2 and D3 anomalies presented in 20 ears (76.92 of the studied ears). The dysplasia of the posterior semicircular canal appeared only in 3 ears, in all of them dysplastic formation of all semicircular canals (combined D3, 4, 5 anomaly) was diagnosed (Fig. 3).

Dysplasia without any aplasia appeared in 16 ears (10 cases). All cases of aplasia were associated with dysplasia (8 cases) (Fig. 4). Only one female child presented with aplastic cochlea on the right side (right sided A1,4,5 D2,3 N1

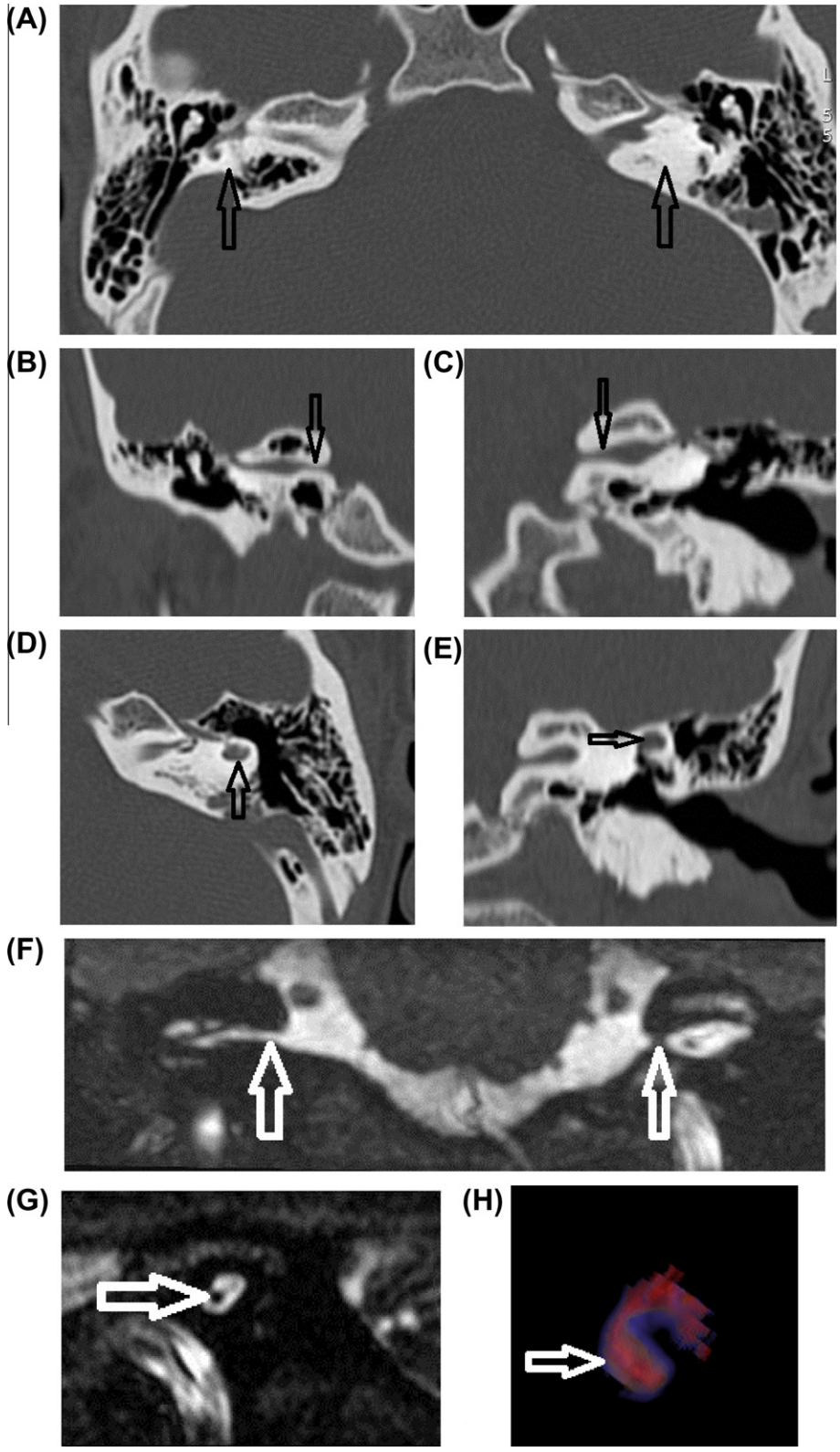

Fig. 2 A case of right sided A1–6 D0 N1 and left sided A1,3–6 D2 N1 anomalies (Michel deformity). A axial CT image showed absent labyrinth in both sides (arrows). (B and C) Coronal CT images showed stenotic (aplastic) internal auditory canals (IAC) bilaterally. (D and E) Axial and coronal CT images showed dysplastic rudimentary bud at the anatomical site of the left vestibule (arrows), also shown in H (VR image). F (axial) and G (sagittal oblique) thin MRI images (FIESTA) showed absent cochlear nerve while only facial nerve is present within the IAC (arrow in G).

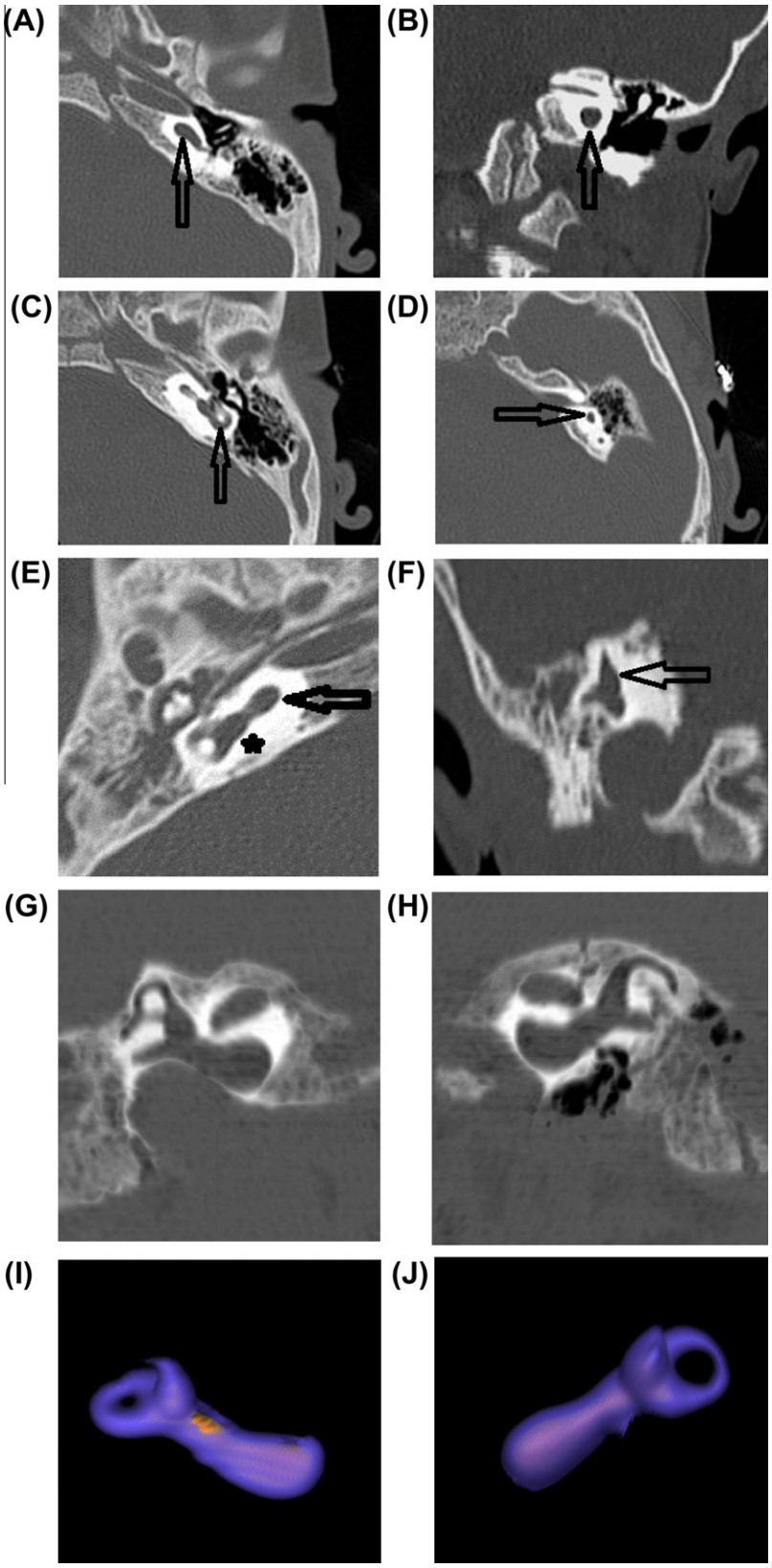

Fig. 3 Showing a case of bilateral D1,2,3,4 anomalies. Dysmorphic components of the labyrinth are shown, the cochlea (D1 anomaly, arrows in A,E), vestibule (D2 anomaly, arrow in B and star in E), lateral SCC (D3 anomaly arrow in C), superior SCC (D4 anomaly arrows in D,F). (A, C, D) Axial CT image for the left side. (B) Axial for left side. E and F axial and coronal for the right side. (G, H) Min. IP and I, J VR images of the entire labyrinth for the right and left sides, respectively.

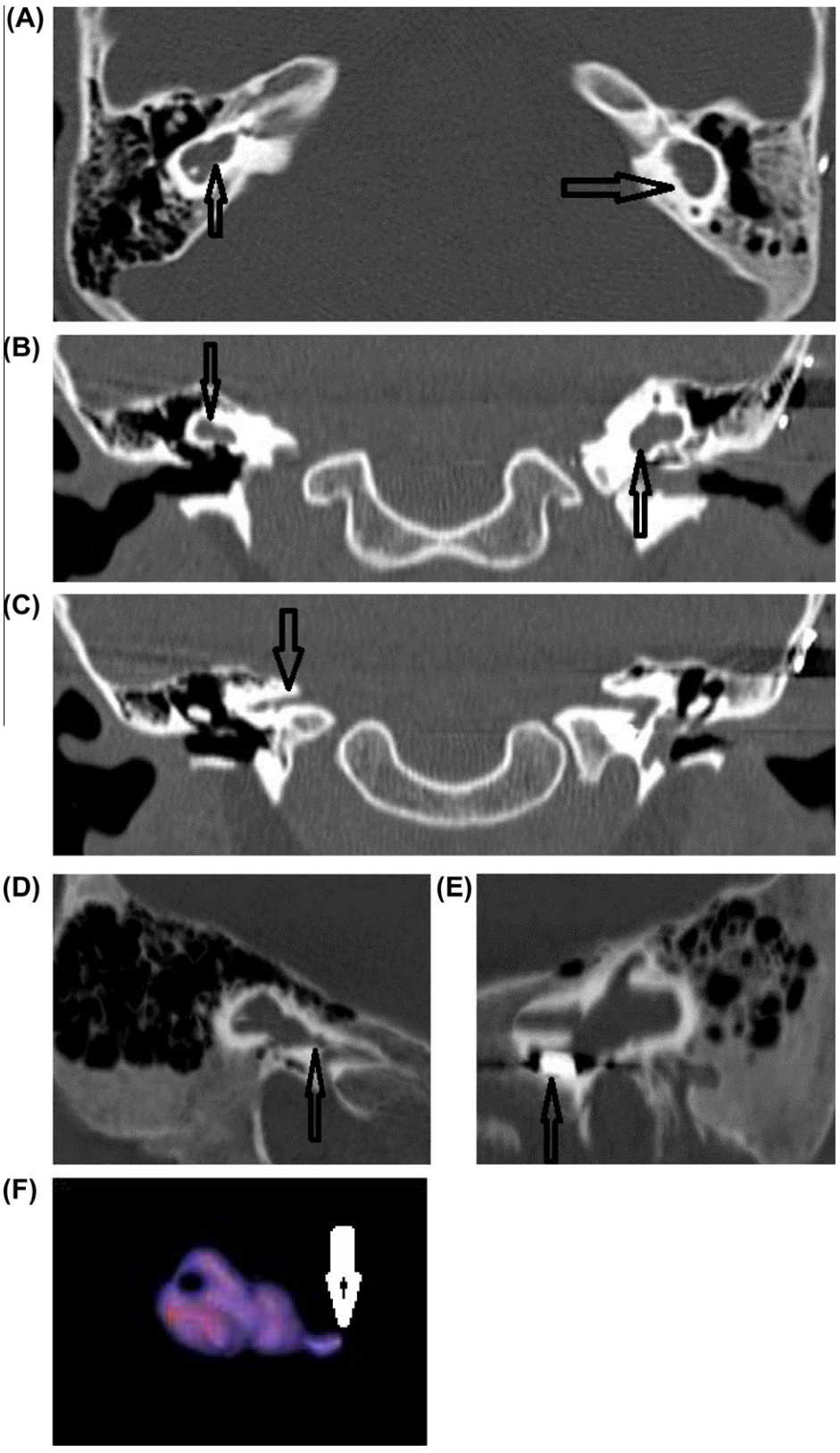

Fig. 4 A case of right sided A1,4,5 D2,3 N1 and left sided D1–4 N0 anomalies. (A and B) Axial and coronal CT images demonstrating dysplastic formation of the remaining portions of the labyrinth (arrows). (C) Coronal CT imaging showing stenotic (aplastic) right IAC (arrow) compared to the left side. (D) and (E) are Min. IP images of the labyrinth and F VR image of the right labyrinth. The absent cochlea (A1 anomaly) is shown by arrows in (D) and (F) with dysplastic rudimentary bud. This child had bilateral anomalies and left sided cochlear implant (arrow in E) was inserted considering that the contra-lateral (right) VIII cranial nerve was absent.

anomaly). On the left side only dysplasia was encountered (left sided D1,2,3,4 anomaly).

5. Discussion

In the present study, the congenital anomalies of the inner ear were more common in females. Among them, they tend to be bilateral. When bilateral involvement occurred, there was a tendency of bilateral symmetry but not necessarily mirror images.

The congenital morphological changes that involve the bony labyrinth could be summarized in either aplasia or dysplasia of one or more component of the labyrinth. The dysplasia will be present morphologically in the form of cystic dilatation or dysmorphic features. Always in the presence of aplasia, there is associated dysplasia, at least at the rudimentary bud of the aplastic component (Fig. 4). For example, the Mondini anomaly could be explained as cystic dilation of the cochlea with resultant disturbed partition (D1 anomaly). Other terms as common cavity and cystic cochleovestibular anomalies as well as the large vestibular aqueduct syndrome could be considered as cystic dysplastic malformation of one or more of the components of the labyrinth (D 1–6 anomaly). The change in size of the one or more of the components of the labyrinth is also associated with dysmorphic features.

Shelton (1989) (20) proposed that the aplastic internal auditory canal (IAC) revealed by CT did not contain a cochlear nerve and is a contraindication to cochlear implantation. The IAC is formed by inhibition of cartilage formation at the medial side of the otic vesicle. This inhibition requires the presence of vestibulo-cochlear nerve. In the absence of the nerve the canal will not be formed (21). Only the presence of the vestibulo-cochlear nerve allow the formation of the IAC, but survival and promotion of the nerve seems to require the presence of a growth factor from the otic vesicle (22). This could explain the close relationship between absent cochlea (A1 anomaly) and absent VIII nerve (N1 anomaly). Both facial and vestibulo-cochlear nerves share the same embryological precursor known as the facioacoustical primordium which separates into individual nerves at the 10–11 mm stage (At this stage, the cochlear primordium beginning to curl as well). The facial nerve, in spite of that, develops independently of the vestibulo-cochlear nerve and becomes caught in the otic vesicle cartilage formation. So, in the absence of the vestibulo-cochlear nerve, the IAC caliber becomes that of the facial nerve alone and that was defined as IAC aplasia (23).

When any component of the bony labyrinth is dysplastic, the vestibule has a high tendency to share. At the same time the most immune component for aplasia was the vestibule. A2 anomaly appeared in one ear sharing in A1–6 N1 anomaly (total labyrinthine aplasia or Michel deformity). The latter anomaly was bilateral and on the other side a small dysplastic vestibule was noted. The anomaly was addressed as A1,3–6 D2 N1 on the other side. The most common association among dysplastic components was the vestibule and lateral semicircular canals.

The introduction of the cochlear implantation surgery increased clinical and radiological interests in the topic of congenital anomalies of inner ear. The referring physician wants to know if the deaf patient has a formed labyrinth to implant or not. If there are any gross morphological changes that could complicate surgery as well as the presence or absence of the cochlear nerve.

Although there are many classifications systems proposed for these congenital anomalies (11–16), they depend on combination of different associated anomalies named after their first reporters or according to their combined morphology. Nowadays, there are anomalies that have been and continue to be documented with thin-section, high resolution multi-slice CT that do not fit neatly into single entity (10). On the other side, many reported congenital anomalies showed features belong to more than one entity (11–16). ADLM is a simplified numerical system for these anomalies. It could entail them with high degree of compliance. It gives a total idea to the referring physician about the status of the bony labyrinth in a short simplified numerical way.

6. Conclusion

The introduction of multi-slice CT imaging with its multiple 2- and 3-D reconstruction capabilities significantly helps in understanding of the gross morphological changes of the anomalies of the bony labyrinth. ADLM is a simplified numerical system that depends on congenital gross morphological changes that occur in the bony labyrinth as seen with today's imaging CT technology. It could entail the congenital anomalies of the inner ear with high degree of compliance. It gives a total idea to the referring physician about the status of the bony labyrinth in a short simplified numerical way.

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