IMAGING PROFILE OF THE EAR IN HEARING LOSS PATIENTS IN HOSPITAL UNIVERSITI SAINS MALAYSIA

DR ROHAIZAM JAPAR @ JAAFAR

DISSERTATION SUBMITTED IN PARTIAL FULFILLMENT OF THE REQUIREMENTS FOR THE DEGREE OF MASTER OF MEDICINE (OTORHINOLARYNGOLOGY – HEAD AND NECK SURGERY)

UNIVERSITI SAINS MALAYSIA

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Once again and never ending, appreciating you with regards,
ROHAIZAM

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LIST OF ABBREVIATIONS

3D	3-Dimensional
AVCN	Anterior Ventral Cochlear Nucleus
BSER	Brainstem Evoked Response
CT	Computed Tomography
DCN	Dorsal Cochlear Nucleus
FASE	Fast Asymmetric Spin-echo
HRCT	High-resolution Computed Tomography
HR-MRI	High-resolution Magnetic Resonance Imaging
HUSM	Hospital Universiti Sains Malaysia
IAM	Internal Acoutic Meatus
L	Left
MPR	Multi-Planar Rendering
MR	Magnetic Resonance
MRI	Magnetic Resonance Imaging
PTA	Pure Tone Audiometry
R	Right
USM	Universiti Sains Malaysia
VCN	Ventral Cochlear Nucleus

IMAGING PROFILE OF THE EAR IN HEARING LOSS PATIENTS IN HOSPITAL UNIVERSITI SAINS MALAYSIA

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Introduction: Hearing impairment is a major disability. The otologic assessments together with high-resolution CT images were able to obtain precise diagnostic profile of ear malformations. These details are essential for further management of patients.

Objective: This study was conducted to investigate the detailed anatomical profile of the ears in hearing loss patient in Hospital Universiti Sains Malaysia by high-resolution CT scan of the temporal bone.

Methodology: This was a cross-sectional study of high resolution CT scans of temporal bone from 1st January 2008 to 31st May 2014 in HUSM. The study sample consisted of all patients with hearing loss who had undergone HRCT in HUSM and fulfilled the study criteria.

Results: In this study, which consisted of 55 patients, 40% of patients showed some form of abnormality on the HRCT temporal bone with 36.4% with outer ear abnormality, 9.0% had middle ear abnormality, 40.1% noted to have inner ear abnormality and 13.6% with multiple areas of abnormality. In term of laterality, 54.5% were observed to have bilateral diseases, 27.3% on right ear and the remaining 18.2% had it on the left ear. The mean vestibular aqueduct diameter was 0.08 ± 0.09 cm (R) and 0.06 ± 0.04 cm (L), lateral semicircular canal diameter 0.10 ± 0.03 cm (R) and 0.10 ± 0.02 cm (L), vestibule diameter 0.30 ± 0.06 cm (R) and 0.31 ± 0.05 cm (L), bone width between the lateral vestibular wall and the inner wall of the lateral semicircular canal diameter 0.35 ± 0.07 cm (R) and (L) and internal acoustic meatus diameter 0.40 ± 0.08 cm (R) and 0.41 ± 0.08 cm (L). Patients with external ear abnormality were observed to have normal to severe hearing loss, patients with middle ear abnormality had mild to severe hearing loss and patients with inner ear abnormality had normal to profound hearing loss. The most common inner ear abnormalities in this study were enlarged vestibular aqueduct and cochlear ossifications.

Conclusion: This study showed that HRCT temporal bone could detect ear abnormality up to 40% of patients with hearing loss and most common abnormality would be enlarged vestibular aqueduct and cochlear ossification. The severity of hearing loss was higher in patients with inner ear abnormality.

Professor Dr. Dinsuhaimi Sidek: Supervisor

Dr Rohaizan Yunus: Co-Supervisor

Associates Professor Dr. Suzina Sheikh Ab. Hamid: Co-Supervisor

ABSTRAK

PROFIL IMEJ TELINGA PESAKIT YANG BERMASALAH PENDENGARAN DI HOSPITAL UNIVERSITI SAINS MALAYSIA

Pengenalan:

Masalah pendengaran adalah suatu kecacatan besar. Pemeriksaan telinga dan penilaian tomografi berkomputer (CT) beresolusi tinggi mampu untuk mendapatkan profil kecacatan telinga yang tepat. Profil telinga ini adalah penting untuk merawat pesakit.

Objektif:

Kajian ini dijalankan untuk mengkaji profil anatomi telinga pesakit di Hospital Universiti Sains Malaysia dengan tomografi berkomputer (CT) beresolusi tinggi.

Metodologi:

Ini merupakan kajian keratan rentas berkaitan tomografi berkomputer (CT) beresolusi tinggi tulang temporal dari 1hb Januari 2008 to 31hb Mei 2014 di HUSM. Sampel terdiri daripada semua pesakit dengan masalah pendengaran yang menjalani HRCT di HUSM dan memenuhi kriteria kajian.

Keputusan:

Kajian ini terdiri daripada 55 orang pesakit, 40% daripada mereka menunjukkan anomali telinga pada penilaian tomografi berkomputer (CT) beresolusi tinggi di mana; 36.4% menunjukkan abnormal telinga luar, 9.0% abnormal telinga tengah, 40.1% abnormal telinga dalam dan 13.6% adalah

kombinasi. Sebanyak 54.5% menunjukkan masalah di kedua-dua belah telinga, 27.3% hanya di telinga kanan dan 18.2% di telinga kiri. Diameter "vestibular aqueduct" adalah 0.08 ± 0.09 cm (R) dan 0.06 ± 0.04 cm (L), diameter saluran separuh bulat lateral adalah 0.10 ± 0.03 cm (R) dan 0.10 ± 0.02 cm (L), diameter vestibular adalah 0.30 ± 0.06 cm (R) dan 0.31 ± 0.05 cm (L), diameter lebar antara dinding vestibular dan dinding saluran separuh bulat lateral adalah 0.35 ± 0.07 cm (R) dan (L) dan diameter meatus auditori adalah 0.40 ± 0.08 cm (R) dan 0.41 ± 0.08 cm (L). Pesakit yang mempunyai masalah telinga luar mempunyai tahap pendengaran normal sehingga teruk, masalah telinga tengah mempunyai tahap pendengaran sedikit sehingga teruk dan masalah telinga dalam mempunyai tahap pendengaran normal sehingga sangat teruk. "Vestibular aqueduct" yang besar dan osifikasi koklea merupakan abnormaliti telinga dalam yang paling kerap ditemui dalam kajian ini.

Kesimpulan:

Kajian in menunjukkan bahawa penilaian tomografi berkomputer beresolusi tinggi (HRCT) mampu untuk mendapatkan profil kecacatan telinga sehingga 40%. Saluran vestibular yang besar dan ossifikasi koklear merupakan abnormaliti yang paling kerap ditemui. Keterukan tahap pendengaran yang tinggi adalah dalam kalangan pesakit yang mempunyai masalah telinga dalam.

ABSTRACT

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Conclusion:

This study showed that HRCT temporal bone could detect ear abnormality up to 40% of patients with hearing loss and most common abnormality would be enlarged vestibular aqueduct and cochlear ossification. The severity of hearing loss was higher in patients with inner ear abnormality.

CHAPTER 1

INTRODUCTION

The emergence of new technique for hearing preservation and rehabilitation has been a challenge over years. Knowledge of human ears, morphology and morphometric study is very important for an otologist to plan the surgery as well as predict the outcome of surgery and hearing status. Early diagnosis and correction of hearing loss will maximize speech perception, thus attained normal language (Agha *et al.,* 2014).

Hearing impairment is a major disability. Early detection can prevent the developmental disabilities in speech, language and congnition (Pajor and Jozefowicz-Korczynska, 2008; Paul, 2011). Hearing loss as a result from auditory system malformation may arise from morphologic abnormalities of external ear canal, the middle ear or the inner ear and various combinations. Malformation of the external, middle and inner ear can be caused by genetic defects and acquired factors. Genetic defects for example chromosomal abnormality, mutations and polygenic inheritance or acquired factors for example drugs, nutritional deficiencies, viral infections or maternal influences can cause hearing loss (Kiefer *et al.,* 2010). Embryogenesis anomalies also depend on the timing of the developmental arrest (Zarandy and Rutka*,* 2010).

Malformation of the outer and middle ear is often combined, whereas most of inner ear malformations occur independently. Malformations of outer and middle ear can be graded as (1) minor auricular malformations which is defined as small

auricular cartilage with good middle ear and mastoid pneumatization, (2) middle grade auricular malformations defined as microtia, stenosis or atresia of the ear canal with normal or slightly reduced middle ear and mastoid pneumatization and ossicular malformation, and (3) severe auricular malformation defined as auricular aplasia, external auditory canal atresia, middle ear and mastoid missing pneumatization and missing ossicles (Kiefer *et al.,* 2010).

Malformations of inner ear varies such as (1) Michel deformity which has total aplasia of bony and membranous labyrinth with no inner ear development, (2) cochlear aplasia or hypoplasia, (3) Mondini's variant such as dysplasia of bony and membranous labyrinth, (4) abnormal cochlear turns, (5) incomplete partition and semicircular canal abnormality, (6) enlarged vestibular aqueduct syndrome and (7) malformation of vestibule or cochlear nerve (Berrettini *et al*, 2005, Kiefer *et al*, 2010). A patient with sensorineural hearing usually has inner ear anomalies, and it was readily detected in about 20-30% of patient radiogically (Ozeki *et al.,* 2009; Vossough, 2003; Zarandy and Rutka, 2010).

Kiefer *et al* (2010) also stated that the incidence of malformations of external and middle ear is 1:20 000 to 1:10 000 and the incidence of malformations of inner ear is 1:80 000 in non-syndromic children. In addition to that, the incidence of severe hearing loss of any origin is 1:1000 while congenital sensorineural hearing loss is 1:4 000.

Assessment of auricular deformity and hearing evaluation for newborn is very important. Early detection of any abnormality can be intervened as early as

possilble. In Malaysia, studies conducted by Elango *et al* (1992) and Elango (1993), the unknown aetiology of deafness was about 20% out of 165 subjects and the prevalence of hearing loss in primary school children is 5.8% while the prevalence of middle ear disorders was 7.2%.

The otologic assessment together with HRCT and magnetic resonance images make it possible to obtain precise diagnostic profile of inner ear malformations. The most common ear anomaly is enlargement of vestibular aqueduct that account about 1 to 7% and it is the most frequent morphogenetic cause of hearing loss in children (Berrettini *et al.,* 2005; Pinto *et al.,* 2005). The enlargement of endolymphatic sac and duct is always present in patients with Pendred syndrome (Berrettini *et al.,* 2005; Kiefer *et al.,* 2010).

As for our population, there were no previous studies that determine the inner ear abnormality particularly enlarged vestibular aquaduct syndrome as well as other abnormality since the last 5 years. This study was conducted to investigate the detailed anatomy of the ears, in hearing loss patient in Hospital Universiti Sains Malaysia by HRCT scan. These details are essential for further management of such patients.

CHAPTER 2

LITERATURE REVIEW

2.1 Anatomy

The basic anatomy of the ear is conveniently described in three parts. Each part of the ear is important to understand further discussion in this study. This chapter will deal with the anatomy of external, middle and inner ear, as well as the ultrastructure of the ear.

2.1.1 Anatomy of external ear

The external ear consists of auricle or pinna, external auditory canal and the tympanic membrane (Figure 1.1). The pinna projects at a variable angle from the side of the head and have some function in collecting sound. Its angulation helps in sound localization (Alberti, 2001; Wright and Valentine, 2008). It is made from folds of resilient yellow elastic cartilage, which give its characteristic shape (Sinnatamby and Last, 2006). It is also a defining feature of the face, with shape and size varies with age, sex and ethnic origin (Alexander *et al.,* 2011).

The external auditory canal measures about 24 mm along its posterior wall from the concha to the tympanic membrane. Its outer part is directed upwards, backwards and medially while the inner part is directed downwards, forwards and medially. It is divided into two parts: (1) the lateral cartilaginous part is about 8 mm long and (2) the medial bony part about 16 mm (Wright and Valentine, 2008). Two constrictions in the external auditory canal; one at the osteocartilagenous junction and the other, the narrowest part called the isthmus, about 5 mm from the tympanic

membrane.

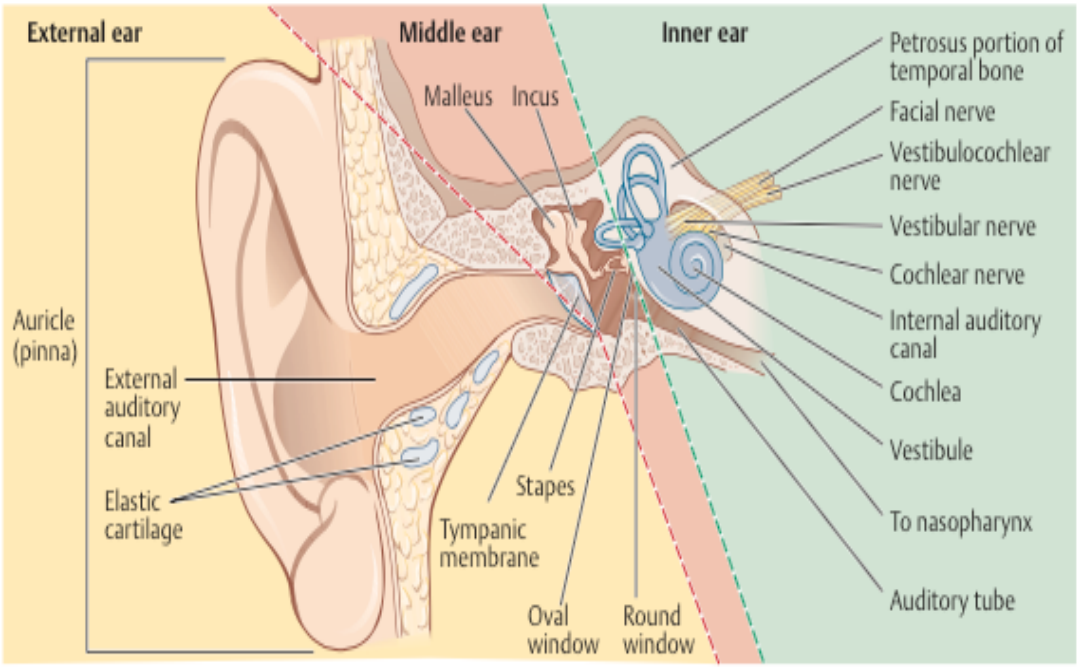

Figure 1.1: Coronal cross-section of the external, middle and inner ear (Adunka & Buchman, 2010).

2.1.2 Anatomy of middle ear

The middle ear consists of the tympanic cavity, auditory tube and the mastoid air cell system (Figure 1.1). The tympanic cavity is an irregular, air-filled space within the temporal bone, laterally is the tympanic membrane and the osseous labyrinth medially. It houses the ossicles and the tympanic segment of the facial nerve.

The tympanic membrane forms the majority of the lateral wall of the tympanic cavity. It is oval in shape, about one cm diameter, forming an angle of about 55° with the floor of the meatus. Its longest diameter from posterosuperior to anteroinferior is 9-10 mm, while perpendicular to this the shortest diameter is 8-9 mm (Alberti, 2001; Wright and Valentine, 2008).

The medial wall of tympanic cavity is the first turn of the cochlear, called the promontory (Figure 1.2). The horizontal ridge for the facial nerve, lateral semicircular canal, oval and round window runs above it. The roof of tympanic cavity is the tegmen tympani and the floor is the bony plate covers the jugular bulb. Its anterior wall has two openings, the auditory tube and canal for tensor tympani and the lower part is form by the posterior wall of carotid canal. The posterior wall is deficient above which has the aditus, which lead to the tympanic antrum. The pyramid located below it and projects into the tympanic cavity (Sinnatamby and Last, 2006).

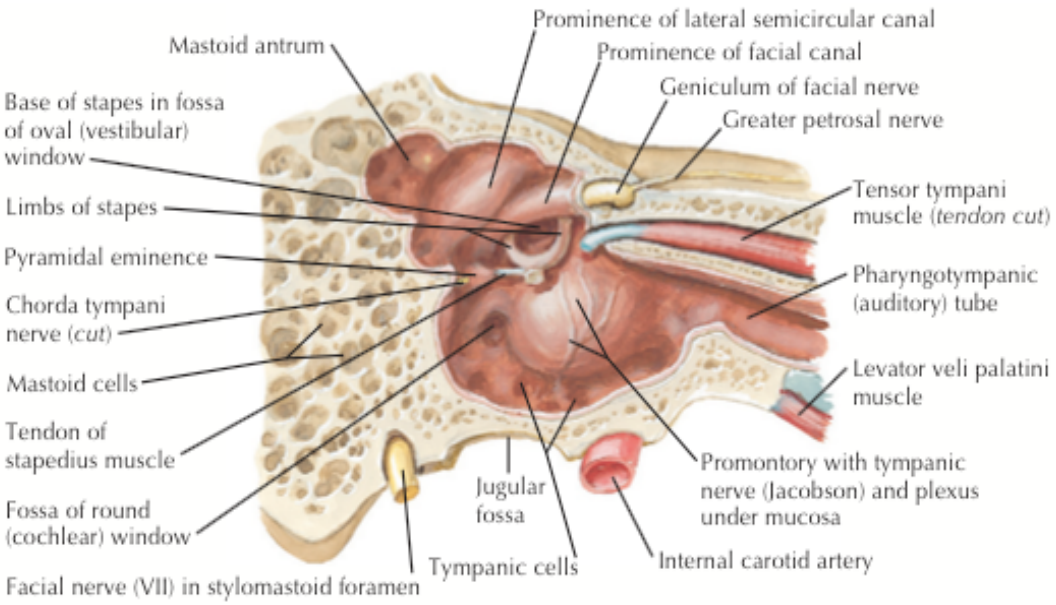

Figure 1.2: Lateral view of medial wall of tympanic cavity (Netter, 2012).

The tympanic cavity also can be divided into three compartments: the epitympanum, mesotympanum and hypotympanum. The epitympanum or attic, lies above the level of the malleolar folds and is separated from the mesotympanum and hypotympanum by mucosal membranes and folds. The hypotympanum lies below

the level of the inferior part of the tympanic sulcus and is continuous with the mesotympanum above. It contains the ossicles (malleus, incus and stapes), muscles (stapedius and tensor tympani), chorda tympani and the tympanic plexus (Wright and Valentine, 2008).

The malleus is club-shaped ossicle with handle embedded in the tympanic membrane, directed upwards. The malleus head articulates with the incus at incudomalleolar joint. The incus runs backwards and its long process attached to the stapes. The footplate of the stapes covers the oval window, an opening into the vestibule (Alberti, 2001).

The auditory tube connects the middle ear with the nasopharynx (Figure 1.3). It is about 36 mm in length, runs downwards at 45° and turned forwards and medially. The bony lateral third arises from the anterior wall of the tympanic cavity while the medial two-thirds is cartilaginous, (Wright and Valentine, 2008). Patients with congenital syndromes and anomalies have a higher incidence of eustachian tube dysfunction (Miura *et al.,* 2002).

The mastoid antrum is an air-filled sinus within the petrous part of the temporal bone. It connects the middle ear by the aditus. The roof of the mastoid antrum and mastoid air cell space form the floor of the middle cranial fossa, while the medial wall relates to the posterior semicircular canal (Wright and Valentine, 2008).

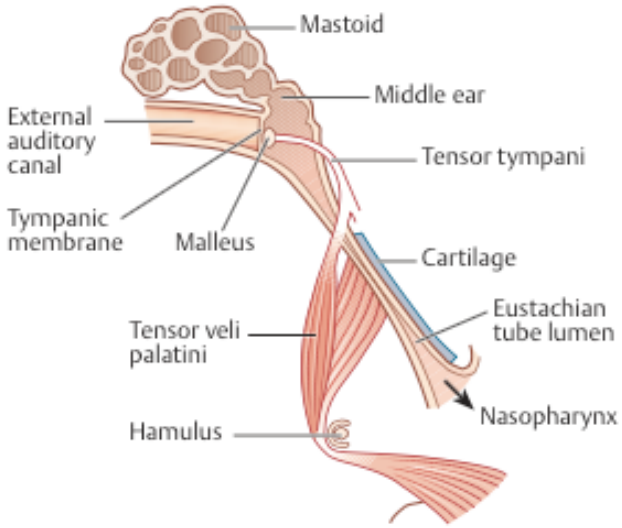

Figure 1.3: Schematic diagram cross-section of the eustachian tube (Adunka & Buchman, 2011).

2.1.3 Anatomy of inner ear

The inner ear is a complex important organ of hearing and balance. It develops independently of the middle and external ears but somehow it is interconnected by the stapes suprastructure giving continuity to the auditory pathway. It attain in part its adult size at 23 weeks intrauterine with changes in shape until 39 weeks, therefore it is practically full adult size at birth (Sinnatamby and Last, 2006; Kiernan, 2006; Richard *et al.,* 2010).

The inner ear consists of a bony and a membranous part (Figure 1.4). The membranous labyrinth is filled with endolymph while the space between membranous and bony labyrinths is filled with perilymph. The bony labyrinth consists of three parts, which are the vestibule, the semicircular canals and the cochlea while the membranous labyrinth consists of cochlear duct, the utricle and saccule, three semicircular ducts, and endolymphatic duct and sac (Sinnatamby and Last, 2006; Vossough, 2003).

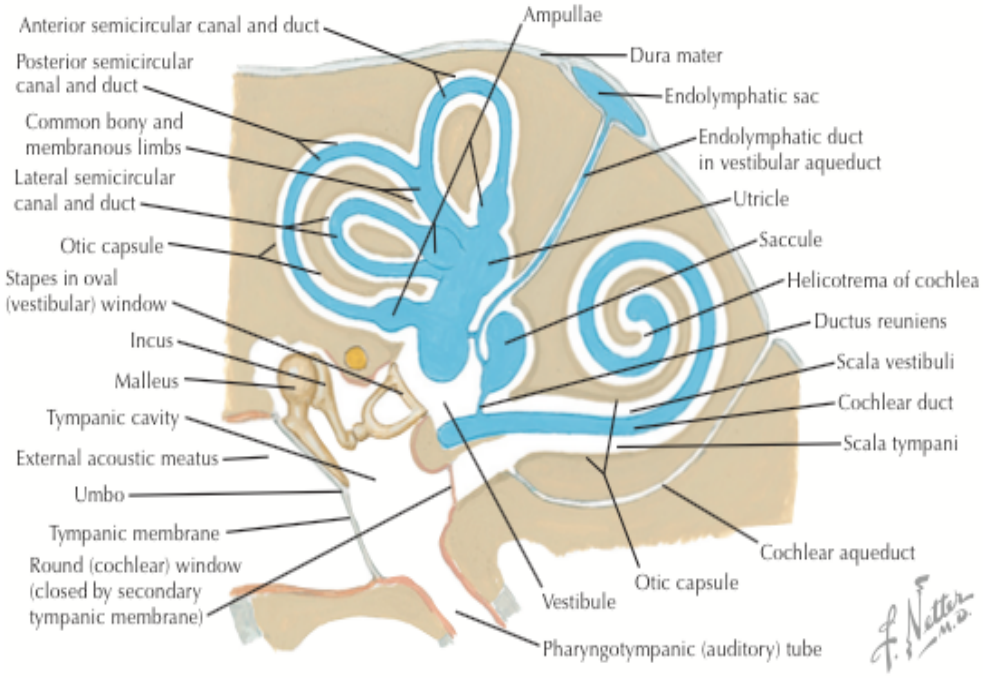

Figure 1.4: Schematic diagram of the bony and membranous labyrinth (Netter, 2012).

Vestibule is the central chamber of the labyrinth. There are two recesses on its medial wall, a spherical recess, which lodges the saccule and an elliptical recess, which lodges the utricle. The opening of vestibular aqueduct lies below the elliptical recess passes the endolymphatic duct. There are five openings of semicircular canals at the posterosuperior part of vestibule. The lateral wall abuts on the middle ear behind the promontory while the medial wall abuts on the internal acoustic meatus (Sinnatamby and Last, 2006).

There are three semicircular canals, the superior in a vertical plane across the long axis of petrous bone convexity upwards, posterior also in vertical plane across the long axis of petrous bone but convexity backwards and lateral that placed 30 degree

off the horizontal plane convexity backwards (Sinnatamby and Last, 2006). They lie at right angles to each other. Each one of the canal is two-thirds of a circle measured about 20 mm, with 1mm in caliber except the dilated end called the ampulla with the caliber of 2 mm (Sinnatamby and Last, 2006). The ampullated end opens independently into the vestibule and the non-ampullated ends of posterior and superior canals unite to form a common channel called the crus commune. Thus, the three canals open into the vestibule by five openings.

The cochlea is a coiled tube making 2.5 to 2.75 turns round a central pyramid of bone called the modiolus (Figure 1.5). The base of modiolus is directed towards internal acoustic meatus and transmits nerves and vessels to the cochlea. The cochlear volume is about 0.2 ml and contains about 30 000 hair cells take place for transduction while about 19 000 nerve fibres transmit the signal to and from the brain (Alberti, 2001). The osseous spiral lamina winding around the modiolus divides the bony cochlea and gives attachment to the basilar membrane. The bony cochlea contains three compartments: (1) scala vestibuli, (2) scala tympani and (3) scala media.

The scala vestibuli and scala tympani are filled with perilymph and communicate with each other at the apex of cochlea through helicotrema. It acts as a pressure equalizing mechanism at frequencies below the audible range (Alberti, 2001). Scala vestibuli is closed by the stapes footplate which separates it from the air-filled middle ear while the scala tympani is closed by secondary tympanic membrane.

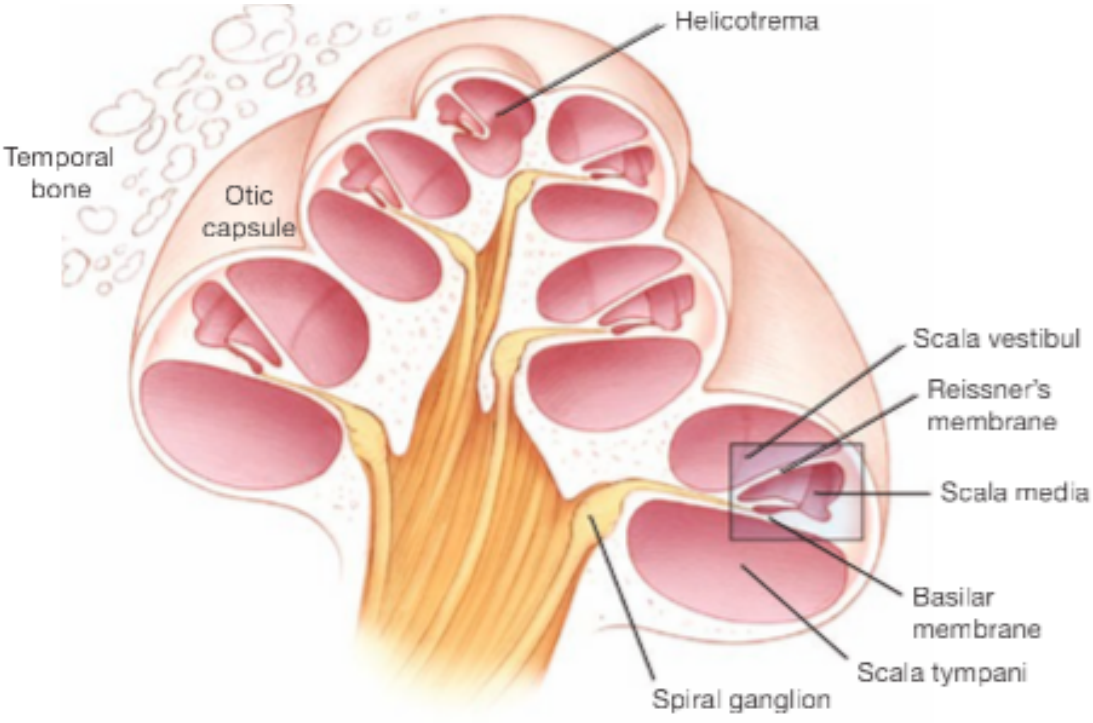

Figure 1.5: Cross-section of the cochlear (Gulya, 2010).

The membranous labyrinth (Figure 1.6), also known as the cochlear duct or the scala media is a triangular blind-coiled tube on cross-section. Its three walls are formed by: (1) the basilar membrane, which supports the Organ of Corti, (2) the Reissner's membrane which separates it from the scala vestibuli, and (3) the stria vascularis, which contains vascular epithelium produces endolymph. The cochlear duct is connected to the saccule by ductus reuniens. The length of basilar membrane increases from the basal coil to the apical coil. Therefore, the higher frequencies are heard at the basal coil while lower at the apical coil.

The utricle lies in the posterior part of bony vestibule while the saccule lies anterior to it, opposite the stapes footplate. The utricle receives five openings of three semicircular ducts and connected to the saccule through utriculosaccular duct. It then becomes the endolymphatic duct and its terminal dilated end is the

endolymphatic sac, which lies between the dura on the posterior surface of the petrous bone.

There are three semicircular ducts correspond to the three bony canals. They open in the utricle. The ampullated end of each duct contains a thickened ridge of neuroepithelium called crista ampullaris. However, in this study, the organ of balance will not be discussed any further.

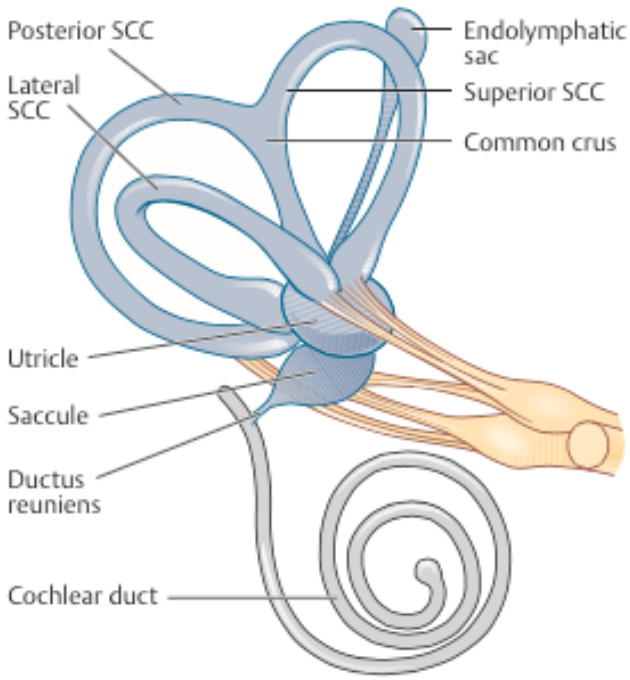

Figure 1.6: Lateral view of the membranous labyrinth (Adunka & Buchman, 2010).

2.1.4 Ultrastructure of the ear

The organ of Corti is the sensory organ of hearing situated repetitively on the basilar membrane called organ of Corti-basilar membrane complex, (Furness & Hackney, 2008). The important components of the organ of Corti are the hair cells, the supporting cells, the tunnel of Corti and the tectorial membrane (Figure 1.7).

The hair cells are important receptor cells of hearing and transduce sound energy into electrical energy. It consists of two types of cell, the inner and the outer hair cells. The inner hair cells form a single longitudinal row while outer hair cells are arranged in three or four rows. Inner hair cells are richly supplied by afferent cochlear fibres and important in the transmission of auditory impulses. Outer hair cells mainly receive efferent innervation from the olivary complex and are concerned with modulating the function of inner hair cells (Furness & Hackney, 2008). These two cells are separated by two rows of pillar cells to form the tunnel of Corti, which contains cortilymph.

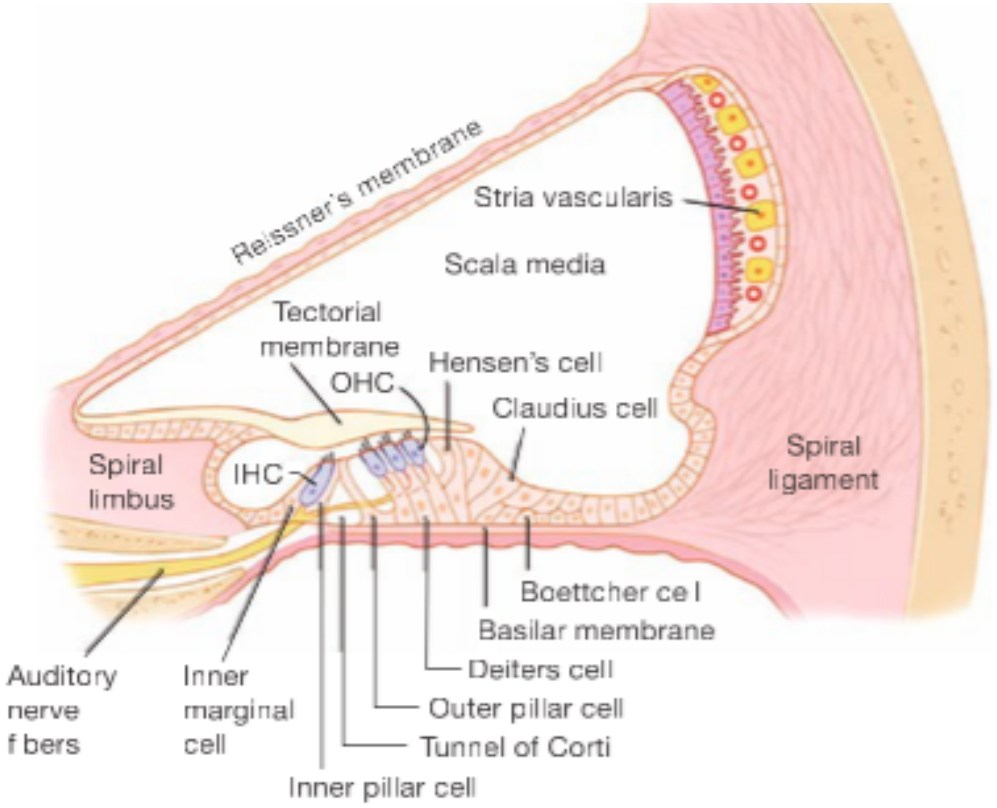

Figure 1.7: Microscopic anatomy of the cochlear (Gulya, 2010).

The supporting cell includes the cup-shaped body of the Deiters' cells, the Hensen cells which lie outsite the Deiters' cells and a single layer of cuboidal Claudius's cells which cover the reminder of the basilar membrane (Furness & Hackney, 2008). Lastly, the tectorial membrane, which overlies the organ of Corti consists of gelatinous matrix with delicate fibres. Stimulus is produce by shearing force between the hair cells and tectorial membrane.

In conclusion, the ear consists of a mechanoelectrical transduction mechanism. The sound conduction has two parts, the outer ear consisting the pinna and ear canal, and the middle ear consisting the tympanic membrane; and the inner ear, or the cochlear transduces vibration into a neural signals, which is then taken along the auditory pathway to the central processing where it is perceived as sound (Alberti, 2001; Furness & Hackney, 2008). Therefore, the morphological properties of each and every component are important contribution for a normal sound perception.

2.2 Physiology

The basic knowledge of sound properties is important to understand the auditory system performances. Generally, the audible sound is ranging from 16 and 32 Hz to somewhere between 16 000 and 20 000 Hz (Alberti, 2001). As head act as a barrier between two ears, it reflects the sound off and reduces the amplitude drastically. Its maximum effect is in the horizontal plane and 90° to the side. Thus, a reflection from the head, pinna and the external ear resonances is to add 15-20 dB to the sound pressure (Pickles, 2008).

With this matter, the external ear, which includes the pinna and external auditory meatus, is important for the first stage of hearing and localization. The pinna and concha act like a trumpet, concentrates the sound to the external auditory meatus. The amplification of sounds can be between 3 and 4 kHz adding to the sensitivity of the ear at this frequency (Alberti, 2001). The most important resonance arising in the concha, adding about 10 dB and the resonance at the tympanic membrane, which adds another 10-12 dB, and other resonances increase the sound pressure at other frequencies.

There is a certain impedance mismatching with an energy transfer between two mediums from air to the tympanic membrane. Therefore, the middle ear will couples sound energy to the cochlea to match the impedance of the air to cochlear fluids as well as produces a differential pressure between the oval and round window. This is important for the cochlear fluids movement to start the acoustic transduction and transmission (Pickles, 2008).

There are two major mechanisms have been identified in the middle ear in order to overcome the impedance of the tympanic membrane to the much higher impedance of the cochlear. The first principle is the area of the tympanic membrane is larger than the stapes footplate by 18.75:1 ratio. The area of tympanic membrane is about 60 mm^2 compare to the stapes footplate is about 3.2 mm^2. Therefore, the collected forces of the tympanic membrane are concentrated on a smaller area of the stapes footplate. The second mechanism is the lever action mechanism where the handle of malleus is longer than the long process of the incus than by 4.4:1 ratio. This action will increase the impedence ratio at the

stapes (Pickles, 2008). Both mechanisms are about to overcome the loss due to the air/fluid interface.

The function of the inner ear is to transduce vibration into nervous impulses via travelling wave. The wave travels along the cochlea fires at a rate of just less than 200 times per second, diminished rapidly as it continues to move towards the apex. The nervous impulses are carried along the 8^{th} nerve from the cochlea to the brain stem (Figure 1.8). Sound level information is based upon the place of stimulation on the basilar membrane and by the rate of nerve firing (Alberti, 2001).

The fibres from each auditory nerve split, some passing to one side of the brain, others remaining on the same side relays at the inferior colliculus, medial geniculate body then to synapse at the auditory cortex at Brodmann's area 41 where the sounds are processed (Alberti, 2001; Furness and Hackney, 2008).

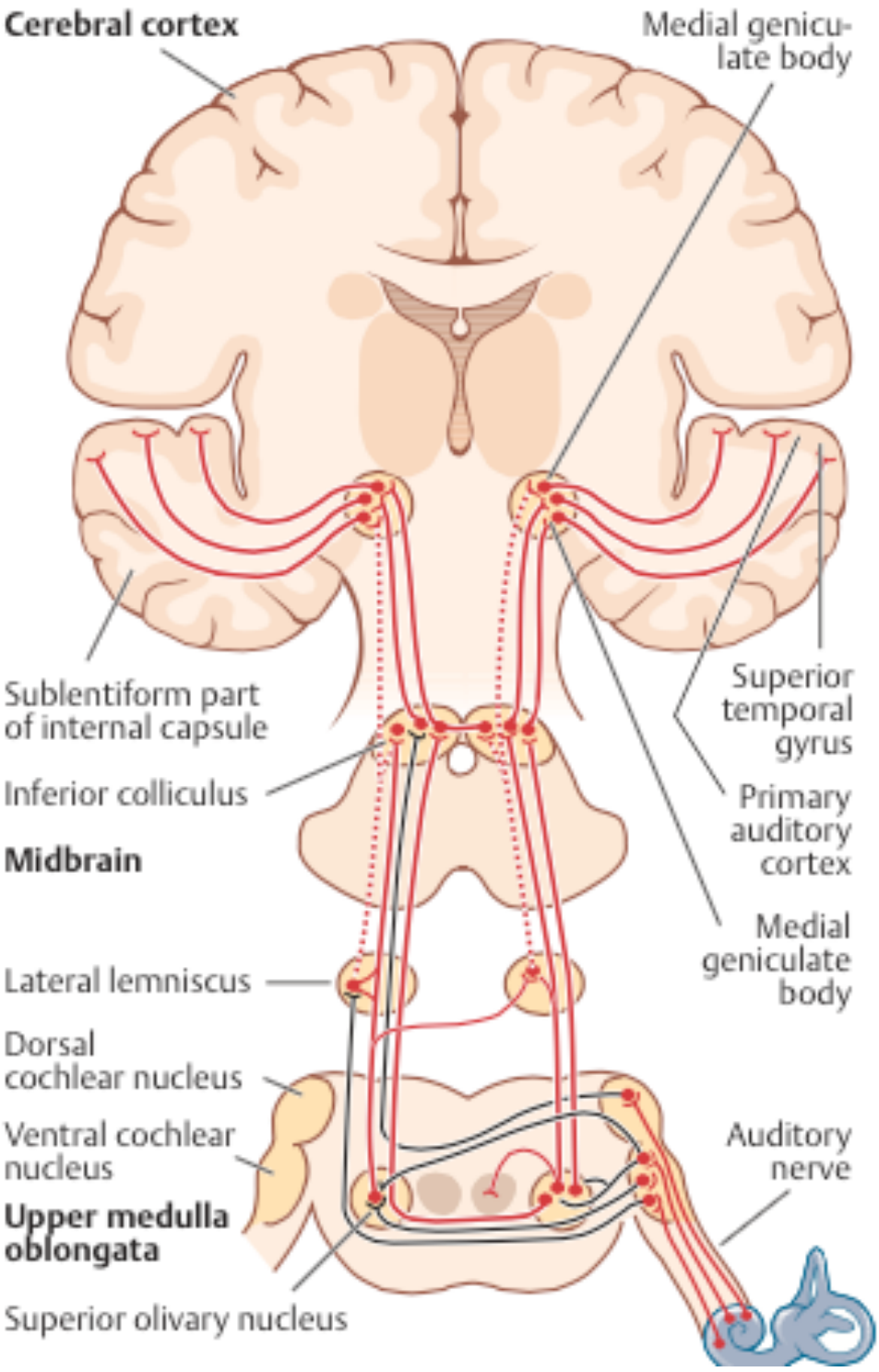

Figure 1.8: Schematic diagram of the auditory pathway (Adunka & Buchman, 2011).

2.3 Congenital deformity of the ear

Congenital ear deformity accounts for more than fifty percent of hearing loss. About 75% are non-syndromic while the remaining is hereditary. Autosomal recessive inheritance and mutation of GJB2 gene that responsible for a gap junction protein in the inner ear to maintain the endocochlear potential are the most common findings (Kiefer and O'donoghue, 2010).

Generally, the more severe the anomaly, the poorer the hearing will be. However, the aetiology of hearing loss sometimes difficult to establish because it is multifactorial (Pajor & Jozefowicz-Korczynska, 2008).

This topic will be discussed about the congenital deformity of the ear, as well as important syndromes associated with hearing impairment.

2.3.1 Congenital deformity of the external ear

Malformation of the external ear occur in 5 per 10 000 births (Wiznitzer *et al.,* 1987). As the auricle is developed from neural crest mesenchyme and epithelial tissue of the first and second branchial arches, and the external ear by those of the first branchial groove, any arrested or abnormality in the development will cause deformity of the external ear (Wiznitzer *et al.,* 1987).

Both external and middle ear deformity causes conductive hearing loss. The external and middle ear anomalies are classified into three groups:

(1) Minor aplasia where the pinna is normal or with a minor deformity. The external auditory meatus is narrow but the tympanic membrane is functional and normal.

There may be ossicular fixation, usually at the stapes.

(2) Major aplasia where there is microtia (Figure 1.9) with external ear canal atresia and fixation of an abnormal malleus and incus. The stapes is usually normal.

(3) Major aplasia/atresia where the external ear may be atretic, small tympanic cavity, sclerosed mastoid and risk of cochlear dyplasias.

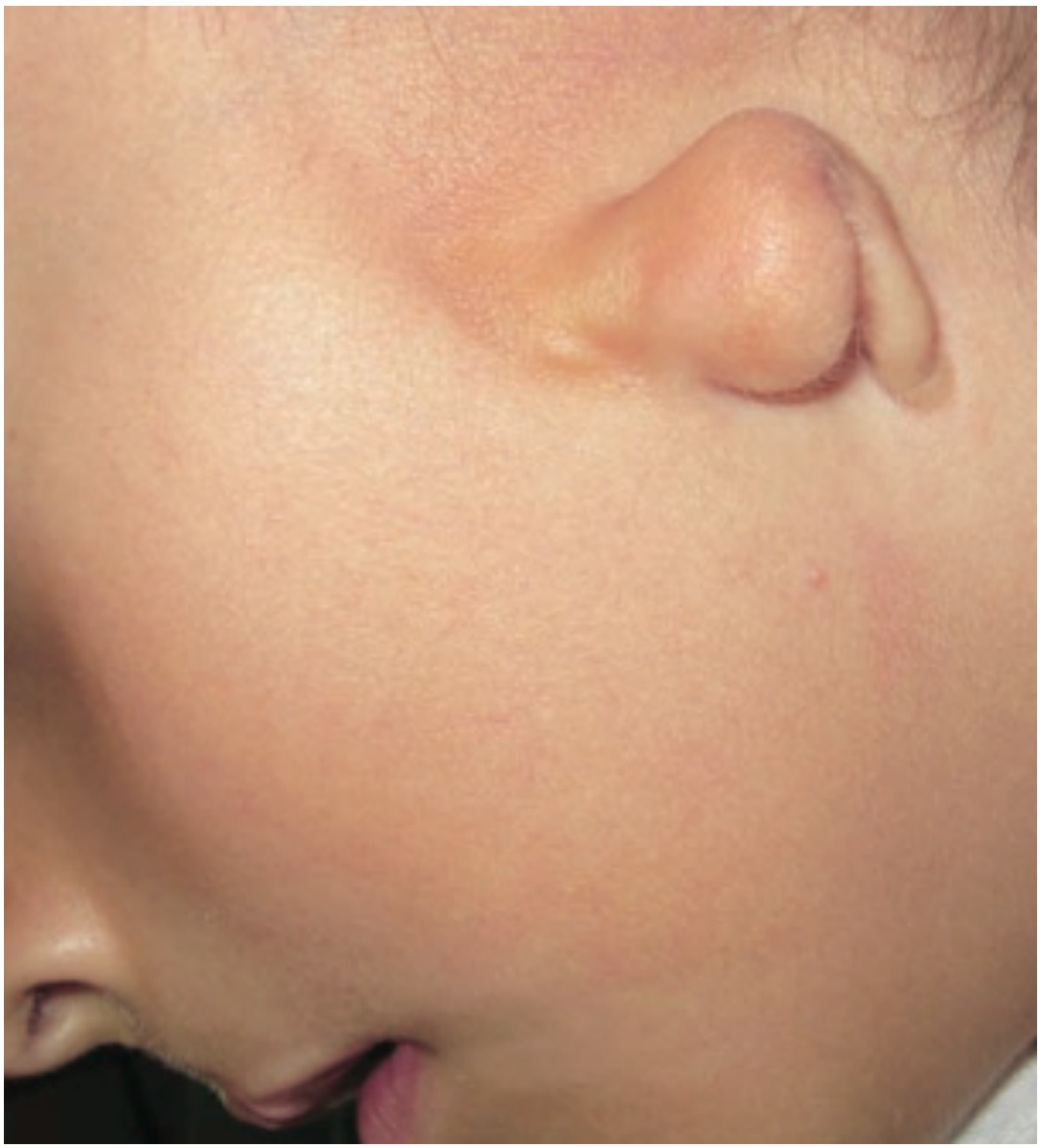

Figure 1.9: A lateral view demonstrating microtia (Gopen and Bhuta, 2013).

2.3.2 Congenital deformity of the middle ear

Congenital deformities of the middle ear are rare. Developmetal arrest of the endoderm of the dorsal division of the first pharyngeal pouch causes middle ear cavity and Eustachian tube defects (Wiznitzer *et al.,* 1987). Congenital deformity of the middle ear causes conductive hearing loss however, patient with cochlear otosclerosis usually have mixed hearing loss (Shin *et al.,* 2001). Elango *et al.* (1994), stated that middle ear disorders for example middle ear effusion and otitis media per se is more prevalent in our country.

Middle ear malformation can be summarized into five categories, which are (1) isolated stapedial ankylosis (2) stapedial ankylosis with other ossicular anomalies (3) ossicular anomalies with mobile stapes (4) oval/round window dysplasia (5) incudostapedial disconnection (Maroldi *et al*, 2001).

2.3.3 Congenital deformity of the inner ear

Congenital anomalies of hearing apparatus usually associated with vestibular anomalies. Aplasia and/or dysplasia of the lateral semicircular canal are the most commonly encountered anomalies of the inner ear as it is the last single structure to be formed during embryogenesis (Ozeki *et al.,* 2009; Yukawa *et al.,* 2008). Lateral semicircular canal anomaly usually associated with severe degree of sensorineural hearing loss (Ozeki *et al.,* 2009).

Classification of congenital malformation of the inner ear is based on embryogenesis (Arnoldner *et al.,* 2004). Aplasia and/or dysplasia of the cochlear comprises:

(1) Michel deformity, which is extremely rare, constitutes only one percent of osseues inner ear malformation (Vossough, 2003). It is believes that there is developmental arrest during the fourth week of intrauterine life, causing the cochlear and vestibule to form a common cavity. The semicircular canal or ducts can be normal or malformed and the internal auditory canal is normal (Arnoldner *et al.,* 2004; Casselman *et al.,* 2001). It causes profound sensorineural deafness (Arnoldner *et al.,* 2004; Casselman *et al.,* 2001).

(2) Cochlear aplasia or hypoplasia occurs when there is arrested development of the cochlear bud during the fifth and sixth week intrauterine while the semicircular canal and vestibule are preserved (Arnoldner *et al.,* 2004; Casselman *et al.,* 2001). It represents about 15% of cochlear malformation (Vossough, 2003). The hearing threshold is not more than 53 dB (Arnoldner *et al.,* 2004).

(3) Mondini dysplasia. It was first describe by Carlo Mondini as a shortened cochlear and lacking an interscalar septum in its apical turn. The pathology of embryogenesis occurs during the seventh week of gestation. It is characterized by a small cochlear with only one and a half turns; no interscala septum and dilatation of the vestibule and vestibular aqueduct and the semicircular canals may be absent. Residual hearing can be appreciated, especially in the high tones with average threshold were about 75 dB (Vossough, 2003; Arnoldner *et al.,* 2004; Kadom & Sze, 2010; Nakashima *et al.,* 2012).

(4) Bing-Siebenman dysplasia where there is maldevelopment of the membranous labyrinth, however the bony labyrinth is normal.

(5) Scheibe dysplasia or also called cochleosaccular dysgenesis where the bony labyrinth is normal, but there is aplasia and hypoplasia of the stria vascularis. Hearing loss is severe.

2.3.4 Syndromes associated with hearing loss

There are many syndromes associated with congenital deafness. With the advances in genetic basis of hearing loss, gene coding and maping gives rise to the possibility of gene transfer. Syndromic deafness gene maping is easier compare to non-syndromic deafness, which relies on the number family members who bear the trait. Currently, at least 30 syndromic and over 120 non-syndromic loci have been discovered to cause hearing loss (Hildebrand *et al.,* 2007). Therefore, genetic councelling is recommended to patients and family with syndromes (Albert and Connell, 2008).

This chapter is to discuss some of the common genetic syndromes emphasize on ENT-related features, classified by autosomal dominant and autosomal recessive trait as well as trisomies. Here are those that we consider to be most important.

2.3.4.1 Autosomal recessive

1. Pendred syndrome characterized by sensorineural hearing loss associated with thyroid pathology. An enlargement of the endolymphatic sac and duct is always present in those patients (Nakashima *et al.,* 2012).

2. Usher syndrome characterized by retinitis pigmentosa and profound sensorineural deafness. There is absent of vestibular function (Albert and Connell, 2008).

3. Jervell and Lange-Nielson syndrome has abnormality in short arm of chromosome 11, characterized by prolonged Q-T interval on ECG and profound sensorineural deafness with absent vestibular function (Albert and Connell, 2008).

4. Refsum disease characterized by retinitis pigmentosa, cerebellar ataxia, peripheral neuropathy and sensorineural hearing loss.

5. Branchio-oto-renal syndrome due to branchial arch anomalies, presented with renal dysplasia and a mixed hearing loss.

2.3.4.2 Autosomal dominant

1. The gene of large vestibular aqueduct syndrome is mapped at PS locus, 7q31, characterized by the presence of an abnormally large vestibular aqueduct. It is associated with fluctuating, progressive sensorineural hearing loss. It is the most common morphogenetic cause of hering loss in children, with prevalence about 55.9% (Naganawa *et al.,* 1999; Vossough, 2003; Berrettini *et al.,* 2005; Hassanein *et al.,* 2005).

Large vestibular aqueduct syndrome maybe associated with syndromic or non-syndromic sensorineural hearing loss such as Pendred and brachio-oto-renal syndrome and often associated with other congenital inner ear anomalies, such as enlarged semicircular canal, modiolar deficiency or cochlear hypoplasia (Naganawa *et al.,* 1999; Berrettini *et al.,* 2005). The morphology varies from funnel, filiform or tubular-shaped (Marques *et al.,* 2007).

2. Waardenburg syndrome is the most common type of syndromic autosomal dominant hearing loss characterized by telecanthus, pigment disorder (20% have a white forelock and 45% heterochromia iridis) and sensorineural hearing loss. It is believed to have the abnormality mapped at the long arm of chromosome 2. Four types have been discussed in the literature; type 1 and 2, there is mutation in the PAX3 gene, type 2 has mutation in the MITF genes and type 4 has mutation in EDNRB, EDN3 and SOX10 gene (Albert and Connell, 2008). The hearing loss is believed to be caused by dysplasia of the membranous labyrinth (Yukawa *et al.,* 2008).

3. Treacher-Collins syndrome (Figure 1.10) is characterized by hypoplasia of the malar bones, maxilla and mandible. There are multiple, external and inner ear abnormalities for example microtia, canal atresia, ossicular malformation and sensorineural hearing loss (Albert and Connell, 2008).