Chapter 4: The causes of deafness

David A. Adams

It is often difficult to identify accurately the cause of deafness in a child because exposure to a known pathogen, such as rubella, does not necessarily imply responsibility for the deafness. Nevertheless, the clinician must not be deterred from making a thorough search for the cause. Fraser (1976) pointed out that this search is not of purely academic interest, but does have considerable practical implications. Parents naturally want to know the cause of deafness in their child and often this is one of the first question they ask. Awareness of the causes of deafness helps to identify high risk groups and is therefore useful in assisting early detection. It also assists in the planning of programmes for prevention or reduction in the size of the problem.

Classification

A good history, examination and full audiological investigation will usually permit classification of the deafness, whether conductive, sensorineural mixed or non-organic.

Confusion may occur when attempts are made to classify deafness according to whether it is congenital or acquired. In this chapter the following form is used.

*Congenital disorders causing or predisposing to deafness*

1. Genetic, with anatomical abnormalities of external or middle ear
   - (a) deafness present at birth
   - (b) hearing probably normal at birth, deafness begins in childhood
2. Non-genetic, due to disease affecting the developing embryo or fetus
3. Other congenital disorders predisposing to development of deafness during childhood.

*Perinatal causes of deafness*

*Acquired disorders causing deafness*

**Conductive deafness**

*Table 4.1* summarizes the conditions in which the hearing loss is mainly conductive. Many other syndromes present with external or middle ear deformity although the hearing loss is usually mixed or predominantly sensorineural. These are discussed later.
**Congenital disorders causing conductive deafness**

**Down’s syndrome (trisomy 21)**

Down’s syndrome is a common disorder occurring in 1 in 600 of all live births. The incidence has been estimated at 1 in 1000 births in mothers under the age of 25 years and 1 in 100 births in mothers aged 40 or older. The affected individual usually has an extra number 21 chromosome. The characteristic facies makes the disorder easily recognizable.

Children with Down’s syndrome present multiple problems to the otologist. Cunningham and McArthur (1981) estimated that as many as 50% of Down’s syndrome children with hearing loss passed the normal childhood screening tests as carried out by local authorities. Maurizi et al (1985) concluded that middle ear pathology is more common than might be expected on a purely clinical basis and that objective tests including evoked response audiometry are essential for reliable evaluation. The present author has found this technique to be of limited value if there is only a slight conductive loss, since it is often difficult to determine if there is a response present at near normal threshold levels.

**Table 4.1 Causes of conductive deafness**

(1) **Congenital disorders**

(a) Genetic, with abnormality of external or middle ear

(i) Deafness present at birth

- Down’s syndrome
- Crouzon’s disease
- Marfan’s syndrome
- Treacher Collins syndrome
- Pierre Robin syndrome
- achondroplasia
- Duane syndrome
- Apert’s syndrome
- otopalatodigital syndrome

(ii) Deafness appearing in childhood

- osteogenesis imperfecta
- otosclerosis

(b) Congenital disorders predisposing to secretory otitis media or infection (see Table 4.2)

(c) Miscellaneous disorders (see Table 4.3)

(2) **Acquired disorders (see Table 4.4)**

These children are very susceptible to repeated upper respiratory tract infections, including sinusitis and otitis media, both infected and secretory (with effusion). In one series, 60% of children with Down’s syndrome, examined during the summer months, were found to have secretory otitis media (Schwartz and Schwartz, 1978). Down’s syndrome is associated with narrow external auditory canals, making it difficult to insert ventilation tubes.
In addition, there may be ossicular chain abnormalities, usually of the stapes. Balkany et al (1979) found that 40% of a group of Down's syndrome children had a conductive deafness not due to infection or secretory otitis media. Exploratory surgery in 17 of these children revealed congenital ossicular malformations or destruction probably as a consequence of previous chronic infection.

There may also be an underlying sensorineural deafness, a short cochlea being the commonest reported finding.

It is therefore important to re-assess hearing thresholds after insertion of ventilation tubes as residual hearing loss is a major additional handicap in these children (Cunningham and McArthur, 1981).

Crouzon's disease (craniofacial dysostosis)

As with most of the hereditary causes of conductive deafness this is inherited as an autosomal dominant trait. Affected children have hypoplasia of the mandible and maxilla with a parrot-beak nose. There is usually skull deformity (craniostenosis) and exophthalmos.

Associated with the syndrome may be stenosis or atresia of the external auditory canal. The tympanic membrane may be absent and the malleus fused to the bony wall of the epitympanum. Other features include a deformed stapes, often fused to the promontory, and a narrow round window niche. A conductive hearing loss is present in one-third of children with Crouzon's disease.

Marfan's syndrome

This is inherited as an autosomal dominant trait. Affected children are tall, often with scoliosis and have long fingers and toes. Other features include hypotonic muscles, a tendency for lens dislocation and cardiac problems, especially aortic aneurysm. Deafness is a rare finding (Konigsmark and Gorlin, 1976).

Treacher Collins syndrome (mandibulofacial dysostosis)

The features of this autosomal dominant trait are confined to the head. The commonest feature of the syndrome is hypoplasia of the malar bones and maxilla. There is an antimongoloid slant to the palpebral fissures. The mandible is usually hypoplastic.

There may be deformities of the pinna, usually microtia, with stenosis or atresia of the external auditory canal. The tympanic membrane may be replaced by a bony plate. The ossicular chain can have a variety of malformations and, in some cases, the middle ear cleft is absent. Tensor tympani and stapedius muscles are often absent. Inner ear abnormalities, if present, would appear to be confined to the vestibular labyrinth (Schuknecht, 1974).

McKenzie (1958) proposed that the basic defect in Treacher Collins syndrome is a temporary deficiency in the blood supply to those structures which develop from the first arch cartilage.
In spite of their appearance the distribution of intelligence in these children would appear to be similar to that in the normal population (Fisch, 1981).

**Pierre Robin syndrome**

This is considered to be an autosomal dominant trait, although, in some cases, it may be due intrauterine disease during the first trimester. The features of this syndrome include cleft palate, hypoplasia of the mandible, glossoptosis, congenital dislocation of the hip and club foot. There may be mental retardation associated with either microcephaly or hydrocephalus.

The external ears may be cup-shaped and appear to be low set because of the hypoplastic mandible. The middle ear cleft may be absent, or there may be thickening of the stapes footplate and crura. Inner ear deformities include abnormal communications between the middle and apical turns of the cochlea, a poorly developed modiolus or a narrow internal auditory canal.

The audiogram shows a conductive deafness, but in cases with inner ear abnormalities the hearing loss is mixed.

**Achondroplasia (dwarfism)**

Although this is inherited as an autosomal dominant trait, about three-quarters of cases may be due to fresh mutation. The incidence rises with increasing parental age.

The main effects are on the skeletal system. There is slow growth of cartilage and delayed endochondral ossification. The result is stunted growth, with disproportionately short limbs and a large head with prominent forehead and depressed nasal bridge.

In the middle ear, the ossicles may be fused to the bony margins of the middle ear cleft. The cochlea may be deformed. The hearing loss, if present, is usually conductive as a result of the middle ear abnormality and also of a predisposition to secretory otitis media.

**Duane syndrome (cervical oculoacoustic dysplasia)**

The affected children with this autosomal dominant syndrome have a very short neck, congenital paralysis of the sixth cranial nerve, and enophthalmos with conductive deafness.

Abnormalities of the external ear include microtia and atresia of the external auditory canal. In the middle ear, the ossicles may be fused and not connected to the oval window. The oval window may be closed by a membrane. Some children have a mixed hearing loss.

**Apert's syndrome (acrocephalosyndactyly)**

This is occasionally inherited as an autosomal dominant trait, although most of the cases are thought to be the result of fresh mutation with a high mutation rate related to advancing parental age. These children have a high, tower skull and flat forehead.
(acrocephaly). There is maxillary hypoplasia with a high-arched cleft palate and saddle nose. The fingers and toes are fused (syndactyly).

The audiogram shows a flat conductive loss of varying degrees. Surgical exploration has demonstrated congenital fixation of the stapes footplate.

**Otopalatodigital syndrome**

This X-linked trait is characterized by bossing of the frontal and occipital bones. There is hypertelorism, hypoplasia of the mandible and cleft palate. The fingers are short and clubbed. Most cases show mild mental retardation.

The pinnae are low set and small. There is a conductive deafness due to abnormalities of the ossicular chain.

**Osteogenesis imperfecta**

The association of fragile bones, blue sclerae and conductive deafness is known as the syndrome of van de Hoeve and de Kleyn. Not all children with osteogenesis imperfecta have blue sclerae and not all have deafness. There would appear to be two distinct forms of the disorder: osteogenesis imperfecta congenita (autosomal recessive) and osteogenesis imperfecta tarda (autosomal dominant).

The basic defect of osteogenesis imperfecta seems to be that collagen does not mature properly, giving a faulty framework for the hydroxyapatite crystals deposited during ossification.

The congenital form is usually lethal, often *in utero*, with skull fractures being the commonest cause of death. In osteogenesis imperfecta tarda the deafness may begin soon after puberty. Morrison (1979) reported a series in which the onset of deafness started as early as 6 years and as late as 51 years, with a peak in the third decade. The deafness is conductive initially, although some cases develop a mixed loss. Schuknecht (1974) summarized the findings of several authors, noting that the disease was characterized by the presence of new soft vascular bone in the region of the oval window, resembling that found in otosclerosis. Bergstrom (1977) reported deformity of the stapes.

It has been argued that otosclerosis is a localized form of osteogenesis imperfecta as they share many common features. Shea and Postma (1982), however, reported that the results of surgery in a group of patients with osteogenesis imperfecta were not as good as those obtained in otosclerotic ears. This, with the earlier age of onset of deafness in osteogenesis imperfecta, would suggest that the two conditions are at least clinically distinct.
Otosclerosis

This is a disease of uncertain aetiology. In many cases it is inherited as an autosomal dominant trait with variable penetrance (Sando, Suehiro and Wood, 1983).

Deafness does not usually begin until puberty and is, on average, later than that in osteogenesis imperfecta. Cawthorne (1955) reported that 70% of patients with clinical otosclerosis first noticed their hearing loss between the ages of 11 and 30 years. In another large series of 610 patients, in whom the deafness began before 18 years of age, the average age of onset of deafness was surprisingly low at 11.5 years (Robinson, 1983). One-half of the patients in this series had a family history of otosclerosis.

There is general agreement that the deafness in children is conductive, with normal or good inner ear function. The results of stapedectomy in children are good, but it should be remembered that middle ear infection is common in this age group. The additional risk to the cochlea which this might present may be avoided by fitting a hearing aid until the child is older.

**Congenital disorders predisposing to secretory otitis media or infection (Table 4.2)**

**Cystic fibrosis (mucoviscidosis)**

Cystic fibrosis is the commonest autosomal recessive disease in the UK, occurring in approximately 1 in 2000 births. The precise nature of the defect is unknown. It affects both mucus and non-mucus secreting glands. The nasal airway, sinus ostia, eustachian tube and middle ear are blocked by viscid mucus. There is also involvement of the salivary glands, bile duct and intestine with fat malabsorption and impaired digestion.

**Table 4.2 Congenital disorders predisposing to secretory otitis media or infection**

<table>
<thead>
<tr>
<th>Cystic fibrosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Immotile cilia syndrome</td>
</tr>
<tr>
<td>Cleft palate</td>
</tr>
<tr>
<td>Immune deficiency disease</td>
</tr>
</tbody>
</table>

Children with this disease are susceptible to secretory otitis media and middle ear infection. In addition, these children are often treated with potentially ototoxic antibiotics in high dosage and are therefore also at risk of developing a sensorineural deafness.

The diagnosis is established by the finding of an increased concentration of sodium in the sweat to above 60 mmol/L.

**Immotile cilia syndrome**

In this rare disease there is impairment of the normal ciliary mechanism of respiratory tract mucosa. The clinical spectrum of disease varies from children with a chronic cough to the sinusitis, bronchiectasis and situs inversus of Kartagener's syndrome. These children are
very susceptible to secretory otitis media. The diagnosis is confirmed by electron microscopy of biopsies of upper respiratory tract mucosa.

**Cleft palate**

The incidence of cleft lip-palate is 1 in 500-750 live births (Rood and Stool, 1981). Otitis media, especially the secretory type is common. Paradise, Bluestone and Felder (1969) found that virtually all children with cleft palates under the age of 20 months had otitis media which was usually secretory in nature.

Two factors may be responsible for this. First, regurgitation of irritant food and fluids around the eustachian tube orifice will cause oedema and obstruction. Second, there is usually some degree of eustachian tube dysfunction associated with failure of the tube to open properly on swallowing. There is no midline anchorage in the unrepaired cleft palate and this prevents the tensor palati muscle exerting sufficient force on the eustachian tube orifice to open it.

Rood and Stool (1981) found that the incidence of secretory otitis media reduced after palatal repair and also with advancing age. Repair of the cleft palate does not, however, always solve the eustachian tube problem. Scarring may inhibit movement of the tensor palati. Furthermore, infracture of the pterygoid hamulus is sometimes used to relieve tension on the palatal repair. This in itself disturbs the functional opening of the eustachian tube.

**Immune deficiency disease**

Disorders of the immune system predispose to infection of various body systems, including the middle ear.

*Miscellaneous congenital causes of conductive deafness* *(Table 4.3)*

**Isolated malformations of the external and middle ears**

These, and their management, are discussed in Chapter 5.

**Table 4.3 Congenital disorders with conductive deafness - miscellaneous conditions**

Isolated malformations  
Congenital cholesteatoma  
Rhabdomyosarcoma  
Fibrous dysplasia  
Goldenhar's syndrome

**Congenital cholesteatoma**

True congenital (primary) cholesteatoma may be due to an epithelial rest left behind as the otic cyst sinks in from the surface of the developing embryo. More recently, Aimi (1983) postulated that most congenital cholesteatoma occurred near the tympanic isthmus of
the middle ear, at the junction of first and second branchial arches. This would suggest that the origin of the cholesteatoma was related to migration of external canal ectoderm into the middle ear at an early stage of development, perhaps because of the failure of the inhibitory function of the tympanic ring.

Derlacki and Clemis (1965) outlined three criteria for diagnosis:

(1) development behind an intact tympanic membrane
(2) no history of ear infections
(3) the lesion must arise from inclusion of squamous epithelium during embryonic development.

It must not be forgotten, however, that cholesteatoma, limited to the middle ear and mastoid behind an intact tympanic membrane, may be due to epidermal ingrowth followed by healing of a perforation of the tympanic membrane (Schuknecht, 1974).

Congenital cholesteatoma may be classified according to the site of origin (Schuknecht, 1974):

(1) petrous apex
(2) middle ear and mastoid
(3) external auditory canal.

The lesion behaves in the same way as acquired cholesteatoma, with enlargement of the cyst and bony erosion. The spectrum of symptoms ranges from conductive deafness to facial paralysis and the intracranial complications of the disease. In some cases the hearing may be normal because sound is conducted from the tympanic membrane through the cyst to the stapes footplate.

McDonald, Cody and Ryan (1984) reported a series of 21 patients considered to have congenital cholesteatoma. This group represented 2% of all the cholesteatomata considered by the authors, suggesting that congenital cholesteatoma may not be as rare as previously supposed. Conductive deafness was the commonest presenting symptom in 18 patients and in 13 of those the tympanic membrane was described as opaque, white or had a cyst visible through it. The disease was confined to the middle ear and mastoid in 20 patients, in the other patient there was extension to the petrous apex.

Rhabdomyosarcoma

Rhabdomyosarcoma, although rare, is the commonest type of malignant neoplasm arising in the soft tissues of the head and neck in children (Chasin, 1984).

The tumour probably originates from a primitive skeletal muscle cell (the myoblast) or from a mesenchymal stem cell. Those related to the ear present with a friable mass in the external auditory canal. There is discharge, bleeding, conductive deafness and occasionally facial paralysis.
The prognosis is poor, although treatment with a combination of radiotherapy, chemotherapy and surgery may give some benefit.

**Fibrous dysplasia of bone**

A comprehensive review of this disorder was provided by Nager, Kennedy and Kopstein (1982). It is a disease of unknown aetiology. There are two types - monostotic and polyostotic. In monostotic disease a single bone is involved with the skull or face being the site in about 10% of cases. If the lesion involves the temporal bone, it may present as a slowly progressive, hard, painless swelling in the mastoid or squamous portion. Temporal bone disease usually becomes evident during childhood with progressive conductive deafness, increase in size or change in shape of the temporal bone, and progressive obliteration of the external auditory canal (Nager and Holliday, 1984).

Polyostotic fibrous dysplasia, if associated with café-au-lait spots and precocious sexual development, is known as McCune Albright syndrome. In polyostotic disease, both temporal bones may be affected.

The radiological features vary with the amount of fibrosis and calcification, with areas of radiotranslucency adjacent to areas of increased bony density.

The complications include exposure of dura, predisposition to acquired cholesteatoma and cranial nerve involvement.

Hereditary fibrous dysplasia is very rare. Adams and Kerr (1983) described a unique family, many members of which have polyostotic fibrous dysplasia of bone. In this family the disorder would appear to be inherited as an autosomal dominant trait with variable penetrance. Deafness is an early symptom in affected children and is purely conductive. The tympanogram shows a very high compliance in most cases. The hearing loss is progressive and eventually becomes mixed. Seven of the patients have had exploratory middle ear surgery with the commonest finding being the replacement of the long process of the incus by fibrous tissue. There is at present considerable debate as to the nature of the disease in this family. The histology has some features in keeping with a diagnosis of Paget's disease, although the clinical and biochemical findings are quite different.

**Goldenhar's syndrome (oculoauriculovertebral dysplasia)**

The aetiology of this condition is unknown, although it is probably not hereditary. Lesions of the eye include a cleft upper lid, dermoids and defects of the extraocular muscles. There may be auricular appendages, microtia and atresia of the external auditory canal. There is often unilateral hypoplasia of the mandible with hemivertebrae and club foot.

Approximately 50% of cases have a conductive deafness due to the external ear abnormalities.
**Acquired disorders causing conductive deafness**

The conditions acquired during childhood which cause hearing loss are summarized in Table 4.4. Otitis media, whether suppurative or secretory, is the commonest cause of deafness in childhood. These disorders are discussed in Chapters 13 and 12 respectively.

**Otitis externa**

In children, acute (infected) otitis externa and eczematous otitis externa are the commonest forms of this disorder.

Swimming in chlorinated pools predisposes to the condition. The irritation of the skin of the external canal makes the child scratch and the subsequent trauma allows the skin to become infected.

Some children develop an allergy to acrylic or silicone earmoulds. This can be successfully overcome by using non-allergic moulds made from vulcanite, although these are expensive.

**Table 4.4 Acquired causes of conductive deafness**

Inflammation
- otitis externa
- acute (suppurative) otitis media
- chronic (suppurative) otitis media
- acute secretory otitis media
- chronic secretory otitis media

Trauma
- Foreign body
- Wax

Conductive deafness is not a feature of otitis externa unless the external canal is blocked by debris or oedematous skin.

**Trauma**

Conductive deafness can be caused by direct or indirect trauma to the ear. Direct trauma is from a foreign body perforating the tympanic membrane or a longitudinal fracture of the petrous temporal bone.

Many parents are obsessed with the need to remove wax manually from a child's ear using cotton buds. The child's head may jerk during this manoeuvre and the object may be driven through the tympanic membrane. There may be damage to the ossicular chain and on occasions to the cochlea. Perforation of the tympanic membrane also occurs during clumsy attempts to syringe an ear.

Head injuries, as the result of accidents involving traffic or falls at home, are common in the young. The child's skull is more deformable than that of an adult and will often dent
without fracture (Pond fracture). The sutures have not united and fissure fractures may persist as separated sutures. As in adults, fractures of the temporal bone are classified relative to the axis of the petrous portion. Longitudinal fractures are commonest (80% of cases) and are associated with a blow to the side of the head. The fracture line usually spares the cochlea and the deafness tends to be conductive in nature but may be sensorineural or mixed. There is often bleeding from the ear if the skin of the external canal is lacerated and the tympanic membrane torn. In other cases there may be a haemotympanum or cerebrospinal fluid behind an intact tympanic membrane. The ossicular chain may be damaged by dislocation or fracture.

The hearing loss in a child may not be noticed until some time after the injury. The child may be unconscious, or admitted to a paediatric unit and not complain of deafness. The ear must not be cleaned or syringed to obtain a better look at the tympanic membrane since this might introduce infection.

Radiology is difficult in the young as they tend to be restless and uncooperative. In some cases, fractures of the temporal bone are not visible on X-ray.

Most cases will settle spontaneously with healing of the perforation and the hearing will return to normal assuming there is no damage to the ossicular chain or cochlea. Cholesteatoma, as a result of entrapment of squamous cells in the fracture line is a rare complication (Freeman, 1983).

Indirect trauma occurs as the result of a slap on the ear, an explosion or to barotrauma. Children are more likely than adults to have eustachian tube dysfunction and may experience problems when flying, usually during descent.

**Foreign body**

Children often present with a foreign body in the external auditory canal and in many cases it is an incidental finding.

Foreign bodies may be of two types: those which are hygroscopic (peas, beans, paper) and those which are not (beads, gravel). A foreign body will only cause deafness if it completely occludes the ear canal or is pushed through the tympanic membrane during attempts to remove it.

Most are easily removed by syringing, although this must be avoided if the foreign body is hygroscopic. If the object is in the outer one-third of the ear canal it can often be removed using a hooked probe. This is not always easy since children are much more likely to move about with subsequent risk of damage to the tympanic membrane. It is best to try once only and if unsuccessful to arrange removal of the foreign body under general anaesthesia.

**Wax**

The ear's self-cleansing mechanism will usually keep the external ear free of wax. The use of cotton buds may cause impaction of wax deep in the external canal.
Children who use hearing aids often have excessive wax in the ear canal. This must be removed on each visit to the clinic as it may block the earmould. In a profoundly deaf child, with high-powered aids, accumulation of wax in the ear canal can cause feedback and limit the useful output of the aid.

**Sensorineural deafness**

Sensorineural deafness in children may result from the various known congenital or acquired disorders summarized in Table 4.5. In published series the incidence of deafness with cause unknown may be as high as 50%.

Four patterns of pathological abnormality of the cochlea have been described in patients with sensorineural deafness.

*Michel dysplasia* is the most severe, with total absence of the labyrinth, perhaps as a result of failure of the otic vesicle to separate from the neural ridge.

*Mondini dysplasia* affects the cochlea and semicircular canals. The cochlear duct is reduced to the basal coil only. The organ of Corti may be absent or reduced to a mound of undifferentiated cells. This type of dysplasia is seen in the Klippel-Feil and Pendred's syndromes. It may be visible on polytomography. Alexander (1904) described this dysplasia in association with auditory nerve involvement.

In *Bing-Siebenmann dysplasia*, the bony labyrinth is normal with underdevelopment of the membranous part.

*Scheibe (cochleosaccular) dysplasia* is the least severe and is thought to be present in about 70% of cases of congenital deafness. The stria vascularis has alternating areas of aplasia and hyperplasia. The organ of Corti is rudimentary and the hair cells sparse or absent. The saccule is collapsed. The utricle and semicircular canals are normal. It has been identified in Waardenburg’s, Usher’s and Refsum’s syndromes and also in rubella deafness.

**Genetic disorders with deafness present at birth**

A fuller discussion of the genetics of deafness is given in Chapter 3. Holmes (1977) described hereditary deafness, *without any other abnormality*, which could be any combination of the following:

1. Inherited as a dominant, recessive or X-linked trait
2. Slight or profound
3. Affecting low, middle or high frequencies
4. Present at birth, or developing during childhood
5. Progressive or stable.
Klippel-Feil syndrome (brevicollis)

The aetiology of this condition is uncertain, although some cases would appear to be the result of an autosomal recessive trait. It is much commoner in females than males.

The external ear may have microtia with preauricular appendages and atresia of the external auditory canal. Middle ear manifestations include deformity of the incudostapedial joint or stapes and fusion of the short process of incus to the floor of the attic. The cochlea is short and there may be distortion of the internal auditory meatus. Most have a sensorineural loss, although it may be mixed.

Turner's syndrome (gonadal aplasia)

These patients have an abnormal genetic constitution, with an XO pattern. It is present in 1 in 5000 live births. The external ears are low set, with large lobes. The mastoid air cell system is poorly developed and there may be abnormalities of the stapes. There is some debate as to whether or not the disorder is associated with sensorineural deafness, although cases have been reported. Anderson et al (1969) stated that, in their series, 64% of patients had a sensorineural deafness with a bilaterally symmetrical loss in the mid-frequency range. A conductive or mixed loss, was present in 22%; perhaps, in some cases, as a consequence of repeated attacks of otitis media.

Fanconi's syndrome

This autosomal recessive condition presents with congenital anaemia, skin pigmentation, skeletal deformities and mental retardation. The hearing loss appears to affect the high frequencies first, and is slowly progressive.

Pili torti

In this autosomal recessive disease dry, brittle hair is associated with sensorineural deafness.

Usher's syndrome

Inherited as an autosomal recessive trait, this is an association of retinitis pigmentosa with progressive sensorineural deafness. These children may also have vertigo and epilepsy.

Pendred's syndrome

This is inherited as an autosomal recessive trait. A congenital defect in thyroxine synthesis eventually causes goitre, which usually becomes obvious between the ages of 5 and 10 years. The sensorineural deafness is severe to profound, is said to be present at birth, and is certainly present by 6 months. This condition may not be diagnosed in the first child until 8-10 years of age when the goitre appears. The diagnosis will then be made much earlier in subsequent siblings.
**Congenital hypothyroidism (cretinism)**

The cause of the hearing loss in this condition is different from that in Pendred's syndrome (Fisch, 1981). The detection of partial deafness, sensorineural or mixed, is often made difficult by associated mental or physical abnormalities. Objective assessment is usually necessary. Fisch pointed out that this cause of deafness might be prevented by effective screening of neonates for hypothyroidism.

**Waardenburg's syndrome**

This is an autosomal dominant trait. Of those affected, 20% have a white forelock, 45% have irides of different colours or have different colours in one iris (heterochromia iridis) and 90-95% have lateral displacement of the medial canthi. This, not the white forelock, is the most common finding and gives a wide appearance to the bridge of the nose. The hearing loss may be moderate or profound, unilateral or bilateral. If the hearing loss is partial it may affect the low rather than the high frequencies.

**Jervell and Lange-Nielsen syndrome**

One-half of affected children with this autosomal recessive disorder, die before the age of 20 years. The deafness is bilateral and severe to profound. It is associated with abnormalities of the electrocardiograph, in particular a prolongation of the Q-T interval.

**Genetic disorders with deafness developing after birth**

Various authors have reported examples of hereditary sensorineural deafness with no other abnormality, in which the hearing appears to be normal at birth with a gradual onset of deafness, which may progress, occurring during childhood (Konigsmark and Gorlin, 1976; Holmes, 1977; Creamers, 1979).

**Alport's syndrome**

There is debate as to the aetiology of this disorder. The renal lesion may be inherited as a partially X-linked dominant trait (Fisch, 1981). Children present with haematuria and albuminuria within the first decade. Males are much more seriously affected than females and often die before 30 years of age.

In approximately 50% of patients a high frequency sensorineural deafness begins around the age of 10 years. This loss usually progresses to become severe. Ruber (1985) suggested that a renal lesion must be excluded in all adolescents with a newly found, progressive sensorineural deafness.

**Renal tubular acidosis**

This is a rare autosomal recessive disorder with only 23 cases reported in the world literature (Takanobu et al, 1984). The present author has seen one child, thought to be the first in Ireland. The child's deafness was first noticed at 3 years of age and was found at that time
to be a flat, moderate to severe sensorineural deafness. His most recent audiogram shows a profound loss, worse for the high frequencies.

**Refsum's disease**

Retinitis pigmentosa with peripheral neuropathy and cerebellar ataxia are the features of this autosomal recessive disorder. Sensorineural deafness usually starts between the ages of 10 and 20 years and is asymmetrical in some cases.

**Cogan's syndrome**

The aetiology of this is unknown, although it has been suggested that it is an autoimmune disease and, as such, is a localized manifestation of polyarteritis nodosa (Stephens, Luxon and Hinchcliffe, 1982). There is non-syphilitic interstitial keratitis with sensorineural deafness and vertigo. It usually first manifests in adolescence with sudden onset of vertigo, tinnitus and rapidly progressive deafness. Treatment with high doses of steroids may halt the deterioration in hearing.

**Norrie's syndrome**

In this X-linked recessive disorder there is progressive blindness with, in some cases, mental retardation. Progressive sensorineural deafness is present in about one-third of patients.

**Non-genetic disorders: deafness due to intrauterine disease**

These conditions, sometimes referred to as the embryopathies, are common and often preventable causes of congenital sensorineural deafness. The best known of these are the maternal infections which may be transmitted to the fetus across the placenta, through the cervix or at the time of birth.

**Rubella**

This is the commonest identifiable cause of congenital sensorineural deafness in children (Martin, 1982). Deafness occurs in about one-third of rubella children. Affected children may also have microcephaly with mental retardation, eye lesions including cataracts and retinitis, abnormalities of the cardiovascular system and lower limb deformities.

There is a mistaken belief that deafness only occurs if infection is within the first trimester. Hardy (1973) pointed out that infection with rubella at any stage in the pregnancy can cause deafness: infection at 0-8 weeks - 86% of children born with deafness, 9-12 weeks - 85%, 13-20 weeks - 53%, 21-35 weeks - 20%.

The virus enters the mother either through the nose or mouth and is transmitted through the placenta to the fetus. The maternal infection may be subclinical in about 40% of cases. Deafness is sometimes the only abnormality.

There is seasonal variation in the incidence of rubella (Martin, 1982). The numbers of children with rubella deafness born in December and January are much greater than those
born in the summer months. This is not due to a seasonal variation in birth rate. It would appear that children conceived in March and April are more at risk of rubella than at any other time of year.

Hemenway, Sando and McChesney (1969) described the abnormal findings in the ear. There may be abnormalities of the stapes or cartilaginous fixation of the stapes footplate. The child's middle ear may contain fetal mesenchyme. The cochlea and saccule have Scheibe-type dysplasia.

The sensorineural hearing loss is usually severe to profound. If moderate it may progress to a severe loss (Fraser, 1976). Fisch (1981) described the typical hearing loss as flat, affecting all frequencies more or less evenly, although it may be trough-shaped with a maximum loss for the middle frequencies. The hearing loss is often asymmetrical and may even be unilateral. Occasionally, the deafness is of the mixed type. Some children may have normal peripheral hearing with central auditory imperception.

The diagnosis of rubella is often made on clinical grounds. It is possible to culture the virus from throat swabs or samples of stool or urine up to the age of 6 months. Persistence of IgG antibody after the disappearance of maternal IgG indicates congenital infection. Rubella specific IgM is present in the infected child for about the first 6 months after birth.

Martin (1982) pointed out that eradication of rubella would abolish one-fifth of all congenital sensorineural deafness. The policy in the UK at present is to offer vaccination to all girls between 10 and 14 years of age and also to screen all women at antenatal clinics. There are several problems with this policy. For it to be successful in abolishing congenital rubella there would have to be almost a 100% uptake in the target population. This is known not to be the case. In addition, the vaccine would need to be 100% effective (Begg and Noah, 1985). At present no check is made on girls after vaccination. Around 40% of babies damaged by rubella are first born. Antenatal screening for rubella is therefore too late since the fetus may already be infected (Kudesia et al, 1985). These authors advocated a change in policy with testing before and after vaccination to ensure a primary response. This technique would also distinguish women who were protected by the vaccine from those with antibodies to the natural virus.

Cytomegalovirus

There is controversy as to the importance of cytomegalovirus in the aetiology of congenital sensorineural deafness. Fraser (1976) thought that it was a relatively minor cause of hearing loss. More recently, several authors have suggested that the importance of cytomegalovirus as a cause of congenital deafness has been underestimated. Bergstrom (1977) stated that 10 times as many children were born with cytomegalovirus infection as with rubella. Pappas (1983), in a review of children with subclinical infection, found that cytomegalovirus is the most common viral agent causing sensorineural deafness in children. It may well be that many cases of congenital sensorineural deafness in the 'cause unknown' group are due to cytomegalovirus.

There are two clinical types of the infection. The systemic infection (10% of patients) is obvious at birth or in the neonatal period. This has a much worse prognosis with the child
often being severely handicapped. Children with focal infection (90% of patients) appear to be normal at birth and, in other words, have subclinical disease.

Pappas (1983) summarized the pathological findings. Cells with intranuclear inclusion bodies were found in Reissner's membrane and the stria vascularis. Immunofluorescent techniques demonstrated viral antigens among the inner ear cells including the organ of Corti and neurons of the spiral ganglion. Cytomegalovirus infection can cause destruction of both cochlear and labyrinthine structures.

The hearing loss is usually severe to profound and bilateral, but may, in a few cases, be unilateral (Saigal et al, 1982; Pappas, 1983).

If the disorder is suspected at birth the virus can be cultured from urine samples. This is only useful during the first few weeks of life. Serological tests which show either a rising titre of IgG antibody or the presence of cytomegalovirus-specific IgM will confirm the diagnosis.

**Toxoplasmosis**

The causative organism of this infection is *Toxoplasma gondii*. The disease is much less common than either rubella or cytomegalovirus infection. The condition is usually subclinical at birth but may eventually manifest itself with progressive blindness because of chorioretinitis. Some children present with hepatosplenomegaly and jaundice. In some, cerebral calcification may result in epilepsy or hydrocephalus.

Kelemen (1958) reported the pathological findings in two children. Both had calcium deposits in the stria vascularis and spiral ligament.

The diagnosis may be made by injecting material from a lymph node or cerebrospinal fluid into mice and examining the brain for calcification 4-6 weeks later. The Sabin-Feldman dye test and the indirect fluorescent antibody test will confirm the presence of infection. Radiology of the skull can be useful if there is focal calcification of the brain.

**Congenital syphilis**

Deafness caused by congenital syphilis may begin in childhood, although in most cases the onset occurs between the ages of 25-35 years (Karmody and Schuknecht, 1966). In early onset disease the infection is severe and often fatal. The ear symptoms are overshadowed by systemic disease. In the later onset or tardive form, deafness with vertigo and tinnitus may be presenting symptoms. There may be profound unilateral deafness.

In the middle ear there may be thickening of the malleus with fusion of the malleus head and incus. There is osteitis of the temporal bone with mononuclear leucocytic infiltration. Obliterative endarteritis and hydrops are found, resulting in degeneration of the cochlear and vestibular end organs.
Herpes simplex

Congenital deafness has also been attributed to infection with herpes simplex virus, although specific reports are not available (Veltri et al., 1981). These authors demonstrated, histopathologically, infection of the labyrinthine sensory cells.

Ototoxic drugs

The effect of these drugs on the fetal cochlea is discussed in the section dealing with acquired causes of deafness.

Irradiation

Irradiation can cause deafness in adults. It is sometimes quoted as a potential cause of congenital sensorineural deafness. The present author cannot find any reports in the literature in which this relationship has been established.

Ultrasound

Pye, Knight and Arnett (1984) described cochlear hair cell damage in guinea pigs due to ultrasound of 12.5 kHz. Ultrasonic scanning, using frequencies of 3.5 or 5 MHz, is a commonly employed technique in obstetric practice. At present, there is no evidence that this is harmful to the fetal ear.

Maternal diabetes

Fraser (1976) stated that the role of maternal diabetes as a cause of congenital deafness had not been established. Gratz, Pollack and Zimmerman (1981) described the radiological findings in two children of unrelated, insulin-dependent mothers. In each case there was hypoplasia of the internal auditory meatus. The cochlea, vestibule and semicircular canals were radiologically normal.

Perinatal disorders causing sensorineural deafness

Perinatal mortality has decreased dramatically over the last two decades. This may mean that more children survive with handicaps such as deafness. The main risk factors in the perinatal period are summarized in Table 4.6.

<table>
<thead>
<tr>
<th>Table 4.6 Perinatal causes of sensorineural deafness</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypoxia</td>
</tr>
<tr>
<td>Hyperbilirubinaemia</td>
</tr>
<tr>
<td>Preterm delivery and low birthweight</td>
</tr>
</tbody>
</table>

Hypoxia

Hall (1964) described the otopathological findings in neonatal asphyxia, including a decrease in cell numbers in the cochlear nuclei. The cochlea appeared histologically normal.
A review of the literature would suggest that perinatal asphyxia only rarely causes a hearing loss. D'Souza et al (1981) found a sensorineural hearing loss in one child from a group of 26 with a history of severe perinatal asphyxia. Significantly, one-third of these children had speech and language defects. Karjalainen et al (1982) examined the hearing in 20 known cases of severe placental insufficiency and intrauterine hypoxia. There was no evidence of sensorineural deafness in any of these children.

**Hyperbilirubinaemia**

The best known cause of this condition is Rhesus disease, although it may also be found with other blood group incompatibilities, hereditary spherocytosis and liver immaturity. The incidence of deafness caused by hyperbilirubinaemia has fallen, presumably as a result of increased awareness of the problem and the availability of exchange transfusions.

Uziel, Maort and Pujol (1983) used hyperbilirubinaemia in the Gunn rat as an experimental model of this condition. Functional and morphological studies showed no cochlear abnormality, although evoked response audiometry suggested the presence of a defect in the brainstem auditory pathways of these rats. Schuknecht (1974), on the other hand, believed the lesion to be in the cochlea in man.

Hyperbilirubinaemia may cause kernicterus (bilirubin encephalopathy). In this condition, 20-40% of affected children have sensorineural deafness. The hearing loss is bilateral and predominantly high frequency in type.

**Low birthweight and preterm children**

Preterm delivery (before the end of the thirty-seventh week) and low birthweight (weighing less than 2500 g) are usually concomitant conditions and are therefore best considered together.

These infants have a higher incidence of hearing loss than normal (Fraser, 1976; Abramovich et al, 1979; Minoli and Moro, 1985). There are several reasons for this. They are more likely to have suffered episodes of hypoxia or acidosis. In addition, these children have immature metabolic functions and kernicterus can result from smaller increases in serum bilirubin levels than in mature neonates. There is also the possibility that the deafness and low birthweight are concomitantly caused by the same factor, for example rubella.

In the immediate postnatal period these children spend a variable amount of time in intensive care units in noisy incubators. This is discussed later in this chapter. They are very prone to life-threatening infections and are given antibiotics which are potentially ototoxic.

Some experimental animals seem to have a 'critical period' during which structural and functional development of hearing occurs (Uziel, 1985). Deafness is likely to result from exposure to noxious agents at different times during this critical period. There is no evidence at present that there is a similar critical period in humans.
In summary, it is often difficult to ascertain the causative agent in these children because of the number of potential risk factors. It is possible that these factors exert a synergistic effect on the auditory system.

**Acquired disorders causing sensorineural deafness**

These conditions are summarized in Table 4.7. Extension of middle ear infection to cause cochlear damage is discussed in Chapter 14.

**Table 4.7 Acquired conditions causing sensorineural deafness**

<table>
<thead>
<tr>
<th>Infections</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>complication of otitis media</td>
<td></td>
</tr>
<tr>
<td>viral labyrinthitis</td>
<td></td>
</tr>
<tr>
<td>Immunization</td>
<td></td>
</tr>
<tr>
<td>Autoimmune deafness</td>
<td></td>
</tr>
<tr>
<td>Meningitis</td>
<td></td>
</tr>
<tr>
<td>Ototoxic drugs</td>
<td></td>
</tr>
<tr>
<td>Trauma</td>
<td></td>
</tr>
<tr>
<td>Metabolic disease</td>
<td></td>
</tr>
<tr>
<td>Neoplastic disease</td>
<td></td>
</tr>
</tbody>
</table>

Various different viral agents, in addition to those already discussed, have been identified as pathogenic in the ear. These include mumps, measles, herpes simplex, herpes varicella-zoster and influenza viruses.

Davis (1982) provided the first experimental evidence for viraemic spread to the ear and it seems likely that this is the common route by which the viruses reach the auditory system. Labyrinthitis may also be caused by extension of infection from the meninges.

**Mumps**

The deafness in mumps is usually of sudden onset, occurring within the first week of infection, although its recognition may be delayed. In some cases the deafness may follow subclinical infection. The loss may affect the high frequencies only but, more commonly, is profound. It is usually unilateral, but may on occasions affect both ears.

**Measles**

Schuknecht (1974) reported the incidence of deafness with measles as the suspected cause to be about 4-10% of populations of deaf children. The hearing loss tends to be bilateral and moderate to severe.

The pathological findings include degeneration of the organ of Corti, spiral ganglion and vestibular sensory cells.
Reye's syndrome

This acute, and sometimes fatal, illness usually starts during recovery from a viral illness especially influenza and varicella. More recently it has been linked to the use of aspirin in children. Clinically the child's condition deteriorates with vomiting, lethargy or irritability. In severe cases cerebral oedema will progress to coma and death. Rarey et al (1983) described the pathological features in a 2-month old child with Reye's syndrome. The inner hair cells of the organ of Corti were damaged more severely than the outer cells with various degrees of degeneration of non-sensory epithelial cells lining the cochlear duct. Similar lesions were found in the vestibular end organs.

The present author is not aware of any published reports of auditory function in children who have recovered from Reye's syndrome; those seen in Belfast to the present have normal hearing.

Immunization

Tetanus immunization and antitoxin are known to cause peripheral neuropathies in some patients. In a review of the literature Mair and Elverland (1977) identified nine cases in which deafness occurred 2-10 days after tetanus immunization or tetanus antitoxin. These authors pointed out that it is extremely difficult to be certain that the two events are related, although in the absence of other aetiological factors a cause-effect relationship may be assumed. The present author has seen one child in which triple vaccination was followed within 2 days by a 'flu-like illness and severe bilateral sensorineural loss.

There are no reports in the literature of deafness after diphtheria or polio immunization.

Autoimmune sensorineural hearing loss

Immunological destruction of the auditory and vestibular systems is a recognized feature of many diseases (Stephens, Luxon and Hinchcliffe, 1982; Brookes, 1985; Naclerio, 1985).

Damage may be caused in several ways. Immune complexes lodge in the microcirculation of the ear causing obstruction and hypoxia in the distal tissues. Complement fixation may cause a vasculitis with subsequent inflammatory response. It is also possible that there is an inappropriate direct immune reaction against cells derived from the neural crest.

Brookes and Newland (1986) presented eight cases, one a child of 11 years, with deafness and evidence of circulating immune complexes. Plasma exchange, thought to remove the immune complexes, was of marginal benefit in restoring hearing in about one-half of the cases, although in some, relief was only temporary.

Meningitis

The most frequent cause of acquired deafness in childhood is meningitis (Martin, 1982). Rahko et al (1984) presented the audiological and vestibular findings in 219 cases.
These authors considered that deafness was due to labyrinthitis following spread of infection through the cochlear aqueduct, internal acoustic meatus or endolymphatic duct. It would appear, in some cases, that either the onset or detection of deafness may be delayed for up to 6 months after the illness.

About 10% of children with meningitis will develop some degree of hearing loss. In some children the initial hearing loss in bacterial meningitis will recover within 6 months (Munoz et al, 1983).

*Haemophilus influenzae* is responsible for about 45% of cases of bacterial meningitis, especially in children aged 2 months to 4 years. Nylen and Rosenhall (1979) published a series of 97 children treated for this illness. Fifteen had moderate to severe sensorineural deafness, in six of these the loss was unilateral. In many other children there were minor abnormalities on audiometric testing.

*Neisseria meningitidis* causes 15-25% of bacterial meningitis. It is probably the most dangerous with respect to hearing loss, causing about 50% of all deafness from meningitis (Rahko et al, 1984).

*Streptococcus pneumoniae* is estimated to cause about 20-25% of cases of meningitis. It is often accompanied by acute otitis media (Schuknecht, 1974).

Streptococcal and staphylococcal meningitis are much less common than the above. Tuberculous meningitis is associated with a high incidence of hearing loss and other neurological deficits.

Nadol (1978) considered the findings in 304 patients thought to have viral meningitis. The causative virus was identified in only one-sixth of the group and included mumps, measles, herpes simplex and varicella-zoster viruses. None of these patients had a sensorineural hearing loss.

**Ototoxic drugs**

The potential ototoxicity of many drugs is well recognized, the two most important groups being the 'loop' diuretics and the aminoglycoside antibiotics. A comprehensive summary of the pharmacology of these drugs and pathological lesions produced was provided by Harper (1982).

Animal research has demonstrated that aminoglycosides will cause intrauterine cochlear damage (Uziel, 1985). There also appears to be interspecies and interstrain variability. In humans, there are surprisingly few reports of deafness due to the administration of aminoglycosides either during pregnancy or childhood (Abramovich et al, 1979; Crifo et al, 1980). It would appear that neonates and older children have reduced risks from the ototoxic effects of these drugs. Children with cystic fibrosis often receive prolonged treatment high doses of these drugs. Crifo et al (1980) found only one such child, in a group of 30, with a bilateral slight high frequency loss assumed to be due to gentamicin.
Similar findings were reported in 53 children with cystic fibrosis who were given tobramycin. Only one developed a transient high frequency loss (Thomsen and Friis, 1979).

Erythromycin is commonly used in children with a history of penicillin allergy. Schweitzer and Olson (1984) presented a case report in which pharmacological doses were used. The patient developed a hearing loss 5 days after erythromycin was first given. On stopping the drug the hearing improved, although the high frequency loss persisted. A survey of the literature revealed a further 32 cases with a reversible high frequency loss.

This apparent decrease in ototoxicity in children must not be allowed to induce a feeling of complacency. Bernard (1981) demonstrated alterations in the brainstem evoked potentials of preterm babies due to conventional doses of aminoglycosides. As with adults, the serum peak and trough levels must be carefully monitored, especially in children with renal disease.

**Trauma**

The effects of trauma on the cochlea are fully discussed in Volumes 2 and 3.

A blow to the head, sufficient to render a child unconscious can cause cochlear concussion with a fracture of the temporal bone (Schuknecht, 1974). Transverse fractures of the petrous temporal bone are associated with damage to the cochlea or auditory nerve. In children, the deafness may not be noticed for some time after the injury. Rupture of the round or oval window may be caused by sudden violent exercise and is predisposed to by anatomical abnormalities (Pashley, 1982). Surgical trauma, even after minor procedures such as myringotomy may damage the cochlea.

The effects of noise exposure on the adult ear are well known. Most women continue to work for the first few months of their pregnancy. Mothers' abdominal and uterine walls will provide protection from noise. Szmeja et al (1979) found that, when the mother was exposed to 100 dB noise, there was a change in fetal heart rhythm and also fetal movements. These may reflect distress. The effects of excessive environmental noise on the ears of the fetus are unknown.

Many neonates spend their first few days or weeks of life in incubators in constant noise levels of 60-80 dB. Added to this is the 5-25 dB generated by other life-support equipment such as ventilators, humidifies and monitors. Medical and nursing staff will often stimulate apnoeic babies by striking the side of the incubator. This can cause impulse signals up to 140 dB SPL (Bess, Peek and Chapman, 1979).

Animal experiments have demonstrated outer hair cell loss in neonatal guinea pigs and rats exposed to noise (Douek et al, 1976; Uziel, 1985). In human neonates, however, the noise levels currently found in incubators do not seem to cause a hearing loss (Schulte and Stennert, 1978; Abramovich et al, 1979).

Children and adolescents live in a self-induced noisy environment. Portable stereo radiocassette players with headphones can generate noise intensity levels potentially hazardous to human ears (Catalano and Levin, 1985). Much has been written about the potential dangers
of rock concert music and there are many reports of temporary threshold shifts in both musicians and the audience. Ruben (1985) pointed out that an adolescent with a seemingly insignificant high frequency loss may become severely handicapped in middle age due to the additive effects of industrial noise exposure and other causes of sensorineural deafness.

Ménière's disease

This is extremely rare in children and presents a similar clinical picture to that found in adults.

Metabolic disease

Disorders of the microcirculation are common in diabetes mellitus and it is quoted as a cause of deafness. Seiger et al (1983), in a survey of the literature, found conflicting views. One explanation of this was the heterogenicity of the different groups studied. These authors presented the findings in a group of 51 insulin-dependent diabetic children. None had evidence of deafness. This may have been due to many factors including short duration of the illness and lack of sensitivity in the auditory tests used.

Neoplastic disease

Acoustic neuroma (schwannoma) may be present at birth, but only become clinically obvious in later life. There are very few reported cases of acoustic neuroma in children.

Leukaemia may affect the temporal bone in two ways (Schuknecht, 1974). Leukaemic infiltrates may be found in middle ear mucosa and perilymph spaces. Haemorrhage may cause sudden deafness, usually with dizziness.

Sudden deafness

Children rarely complain of sudden loss of hearing. Tieri et al (1984) suggested the following as possible causes:

(1) infection - mumps, measles, meningitis, varicella
(2) trauma - concussion, fractures of temporal bone, perilymph fistula
(3) idiopathic - the mechanism may be vascular with spasm, thrombosis, embolism or haemorrhage causing cell anoxia and death.

Mixed deafness

The conditions which cause a mixed deafness are summarized in Table 4.8. Otosclerosis in children is usually associated with conductive deafness only.

Earpits-deafness syndrome

Slack and Phelps (1985) presented a description of four families and a review of the literature. The condition is characterized by unilateral or bilateral auricular deformities in 75% of cases. These are preauricular pits or appendages with unilateral or bilateral branchial
fistulae or cysts in 50% of affected children. The ossicular chain may be abnormal and there is distortion of the basal turn of the cochlea. Two of the patients in this series had ossiculoplasties with no improvement in hearing. This confirmed the findings of other otologists dealing with this condition.

**Osteopetrosis (Albers-Schönberg disease)**

This may be inherited in a dominant form (benign) or as a recessive trait (clinically malignant). There is abnormal bone growth with failure of reabsorption of calcified cartilage and persistence of primitive bone. The bony labyrinths and ossicles consist of dense calcified cartilage. The mastoid is usually not pneumatized. These patients present to otologists with mixed deafness and recurrent facial nerve palsy.

**Table 4.8 Causes of mixed deafness**

1. Congenital abnormalities, deafness present at birth, earpits-deafness syndrome

2. Congenital abnormalities, deafness occurring in childhood
   - osteopetrosis (Albers-Schönberg disease)
   - histiocytosis X
   - mucopolysaccharidosis

3. Acquired disease
   - infection.

**Histiocytosis X (Langerhans cell histiocytosis)**

The three forms of this disease, eosinophilic granuloma, Hand-Schüller-Christian disease and Letterer-Siwe disease, are probably different expressions of the same basic disorder.

Eosinophilic granuloma is the mildest form. There is localized skeletal destruction which, if present in the temporal bones, is manifest by swelling over the mastoid process with otorrhoea and granulations in the external auditory canal. There is a mixed hearing loss and occasional facial paralysis.

Hand-Schüller-Christian disease often presents with a triad of symptoms: diabetes insipidus, exophthalmos and osteolytic lesions of the cranium. Temporal bone involvement produces a similar clinical picture to that in eosinophilic granuloma.

The most severe form of histiocytosis X is found in Letterer-Siwe disease. This is usually fatal.

A review of histiocytosis X was provided by De-Marino et al (1985). There is proliferation of abnormal histocytes. The ear manifestations in children may mimic those of chronic otitis media with mastoiditis, but are resistent to the usual treatments. The diagnosis is made by histological examination of a biopsy, taking care to obtain a sample of tissue deep to surface granulations. Treatment uses a combination of surgery, radiotherapy and chemotherapy.
Mucopolysaccharidoses

The best known of these are Hurler's (type 1) and Hunter's (type 2) syndromes. There is abnormal metabolism of intracellular high molecular weight carbohydrates. Most of the mucopolysaccharidoses are autosomal recessive traits except for Hunter's syndrome which is X-linked.

Fisch (1981) noted that all affected children examined by him had a hearing loss. In many of the children the hearing loss was conductive, although it was sometimes superimposed on a moderate high frequency sensorineural deafness to give a mixed loss. Schuknecht (1974) indicated that deafness is not always present in these children but, if present, is of the mixed type.

Schachern, Shea and Paparella (1984) presented the findings from the temporal bones of three patients with Hurler's syndrome. They included otitis media, residual mesenchyme in the round window niche, partial occlusion of the middle ear and basophilic concretions in the stria vascularis. Other reports note the absence of the incudostapedial joint and obliteration of both oval and round windows with fibrous tissue invading the otic capsule.

Infection

This is the commonest cause of mixed deafness in children and is discussed elsewhere.

Non-organic deafness (psychogenic deafness)

There are three types of this condition.

Functional (hysterical) deafness

This is apparent deafness in the absence of a pathological process affecting the auditory pathway. The deafness is a product of the subconscious. It is estimated that functional deafness is responsible for about 5% of all audiological clinic attendances. It would appear to be very uncommon under the age of 5 years.

It may be a reaction to stress, especially if the child is not doing well at school and the parents' expectations are unrealistically high. In some cases it is a means of identifying with another member of the family who has a hearing problem.

The deafness may be moderate to severe with evidence of other psychological disturbances such as mutism, tremors, aggressive or withdrawn behaviour. The child's voice is usually unaltered with no deterioration in the quality of speech. These children often give different serial audiograms with better speech discrimination scores than would be expected from the pure-tone readings. Clinically the child's hearing is usually much better than the audiogram would suggest. This group must be differentiated from those children who seem to have difficulty in understanding what is involved in pure-tone audiometry.
Malingering

In this type there is intention on the part of the child to deceive. This is rare in children as most are not sophisticated enough to maintain the pretence for long and there is rarely the motivation for financial gain as sometimes seen in adults.

Organic deafness with psychogenic overlay

Children with true ear disease occasionally appear to be much deafer than can be explained by the pathology.

In all three types of non-organic deafness, objective tests, including evoked response audiometry, will reveal the true hearing thresholds.

These children present difficult management problems. It is important to stress to parents the need to avoid accusing the child of feigning a hearing loss. Attempts should be made to look for areas of conflict at home or school. This will often mean referral to a child psychologist or psychiatrist. These children must not be issued with hearing aids for fear of reinforcing their 'deafness'.

Taylor (1979) discussed the reasons for the differences in prevalence rates between various studies (Table 4.9). In retrospective studies serological data are not available, access to accurate hospital records is not always possible and sample groups differ. True comparisons are therefore difficult to make.

Table 4.9 Different prevalence rates of deafness between various studies

<table>
<thead>
<tr>
<th>Causes</th>
<th>Percentage of cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic</td>
<td>24-39</td>
</tr>
<tr>
<td>Embryopathies (mainly rubella)</td>
<td>6-24</td>
</tr>
<tr>
<td>Perinatal</td>
<td>6-23</td>
</tr>
<tr>
<td>Unknown</td>
<td>25-45</td>
</tr>
</tbody>
</table>

A more recent multicentre study of EEC children with a hearing loss of 50 dB or worse was reported by Martin (1982). In this study rubella was responsible for 20% of deafness in children in the UK. Deafness was identified as having a genetic basis in about 12% of cases, caused by perinatal anoxia or jaundice in 10% and in 40% of cases the cause was unknown.

The large size of the 'cause unknown' group is a feature of all reported series. Much discussion has ensued as to possible aetiologies in this group. At present, it is generally assumed that most of these are the result of recessive genes or to gene mutations. Many of the others are caused by undiagnosed intrauterine infection or to the effects of other unrecognized cochlear pathogens. Barr (1982) underlined the marked interspecies differences in response to thalidomide. This suggests an interaction between genetic factors and exogenous pathogens in the causation of deafness. It would seem that a genetically deficient auditory pathway is more susceptible to external agents. A better understanding of the
processes causing deafness, together with appropriate and early investigation of the deaf child, should reduce the size of the 'cause unknown' group.