

DENTISTRY IN PROFOUND CHILDHOOD DEAFNESS IN SOUTH AFRICA

LAWRENCE STEPHEN¹, SEAN SELLARS², PETER BEIGHTON³

Abstract

The dental care of children with profound childhood deafness is influenced by difficulties in communication. In addition, a number of genetic conditions in which deafness is a significant component have additional features which may create special needs in dental management.

During the past three decades we have examined more than 4000 children with severe hearing loss in special schools for the deaf. Our experience in the diagnostic categorisation and the dental management of deaf children is reviewed in this article.

Key Words: Deafness, Dental, Genetic, Oro-facial

Introduction

Children with congenital or early-onset profound hearing loss are severely handicapped, and highly sophisticated education is necessary. The condition is relatively common and it has been estimated that one in every thousand neonates will have severe deafness (Arnos 1994). In Southern Africa 25 special schools for the deaf provide for the needs of profoundly deaf children, and over the years, more than 4000 of these young people have been examined and investigated (Sellars and Beighton 1983; Gardner et al 1997). It has emerged that in general they have the same spectrum of acquired dental problems as in their unaffected peers. Routine dental care is necessarily influenced, however, by the hearing loss and difficulties in communication. In addition, there are several genetic and multifactorial disorders in which deafness is a component, and in which the

syndromic manifestations pose specific dental problems.

Children with profound hearing loss are encountered in conventional dental practice and in some instances dental practitioners provide a regular service for children at the special schools. On a basis of our experience in this field, we have reviewed the aetiology of profound childhood deafness, with emphasis on genetic disorders, and discussed the dental management of the deaf child.

Aetiology Of Profound Childhood Deafness

The aetiology and implications of profound childhood deafness have been outlined in the book "Genetics and Otology (Beighton and Sellars 1982).

Profound deafness is conventionally categorised as "perceptive" or "conductive". The former type, which is also termed "sensori-neural", results from dysfunction in the inner ear, auditory nerve connections or in the brain. Conductive deafness is caused by abnormalities in the middle ear or external auditory meatus. This categorisation is crucial for appropriate otological management.

If deafness develops in childhood, after speech has been established, the hearing loss is termed "post-lingual"; if the onset of deafness is pre or post-natally, in the period prior to normal speech development, it is termed "pre-lingual". It follows that children who never hear cannot develop normal speech.

Profound childhood deafness as encountered in the special schools in South Africa can be grouped into broad diagnostic categories in the following approximate proportions:

Acquired – due to exogenous factors	30 - 40%
-------------------------------------	----------

¹ Professor of Dentistry, Faculty of Dentistry, University of the Western Cape, Cape Town, South Africa

² Emeritus Professor of Otorhinolaryngology, Faculty of Health Sciences University of Cape Town, Observatory 7925, Cape Town, South Africa

³ Emeritus Professor of Human Genetics, Division of Human Genetics, Faculty of Health Sciences, University of Cape Town, Observatory 7925 Cape Town, South Africa

From: UWC/UCT Special Dental Genetic Clinic
Red Cross Children's Hospital, Cape Town

Address for correspondence:
Emeritus Professor P Beighton, Division of Human Genetics,
Faculty of Health Sciences, University of Cape Town,
Observatory 7925, Cape Town, South Africa
Tel: +27-21-406530
Email: Peter.Beighton@uct.ac.za



Figure 1: Waardenburg syndrome. A girl with the characteristic white forelock and eyes of a striking ultramarine colour. Patchy depigmentation of the forehead and right eyebrow are evident.



Figure 2: Treacher Collins Syndrome. Mandibular and maxillary hypoplasia are associated with severe dental malalignment. The external ears are malformed.

Genetic non-syndromic deafness	10 - 20%
Genetic syndromic deafness	5 - 10%
Cryptogenic – cause unknown	30 - 40%
Multifactorial deafness syndromes	5%

Considerable geographical variation in these figures results from socio-demographic and genetic factors (Sellars et al 1977; Beighton et al 1991).

1. Acquired Deafness

In about 40% of all children with profound deafness, the hearing loss is the consequence of damage to the hearing mechanism by exogenous factors:

1. Prenatal

- maternal rubella
- maternal drug therapy – quinine, thalidomide
- maternal alcohol

1.2. Perinatal

- birth trauma or anoxia
- kernicterus

1.3. Postnatal

- Meningo-encephalitis - viral, bacterial or tuberculous
- Otitis media – recurrent severe
- Trauma
- Drug therapy – aminoglycosides, quinine

2. Genetic Non-Syndromic Deafness

A significant proportion of profoundly deaf children are members of an affected family in which a clear cut pattern of genetic transmission is evident, in the absence of any additional syndromic manifestations. At the molecular level, this group of conditions is highly heterogeneous.

- 2.1 Autosomal Dominant (AD)
- 2.2 Autosomal Recessive (AR)

- 2.3 X-linked (XL)
- 2.4 Mitochondrial

3. Genetic Syndromic Deafness

In a number of genetic syndromes, profound deafness is associated with additional manifestations which permit clinical diagnosis. The majority of these conditions are rare, but those which are likely to be encountered in dental practice are listed below. Their mode of genetic transmission and estimates of their frequency within profound childhood deafness as a whole are listed below:

3.1 Waardenberg Syndrome	AD	2-5%
3.2 Treacher Collins syndrome	AD	1-2%
3.3 Usher Syndrome	AR	2%
3.4 Pendred Syndrome	AR	1%
3.5 Branchio-oto-renal syndrome	AD	1%
3.6 Uncommon genetic deafness syndromes		1%

4. Cryptogenic Deafness

In approximately 40% of profoundly deaf children the cause is unknown. In these children there is no relevant case history and no physical abnormalities. Unrecognisable ear or brain infection, undiagnosed maternal rubella and non-syndromic familial deafness all contribute to this category. With the application of molecular genetic technology, it is becoming evident that a significant proportion of children with "cryptogenic" deafness have an autosomal recessive genetic disorder (Petit 1996). In these circumstances there is a risk of recurrence in siblings of an affected child.

5. Multifactorial Deafness Syndromes

A few well-defined disorders which can cause profound childhood deafness have a multifactorial aetiology. Deafness in these disorders may be moderate rather than severe; the most



Figure 3: Sclerosteosis. A child with a bulky mandible, dental malalignment and deafness, due to progressive bone overgrowth.



Figure 4: Sclerosing Bone Dysplasia. Lateral view of the skull in a sclerosing bone dysplasia. Tooth extraction can be very difficult in this group of conditions.

common of these conditions are listed below.

- 5.1 Goldenhar syndrome (hemifacial microsomia; oculo-auriculo-vertebral dysplasia).
- 5.2 Wildervanck syndrome (cervico-oculo-acoustic dysplasia). Several forms of deafness which are essentially the result of exogenous agents probably also involve a genetic predisposition.
- 5.3 Rubella Embryopathy
- 5.4 Foetal Alcohol syndrome
- 5.5 Streptomycin ototoxicity

Clinical Manifestations Of Genetic Deafness Syndromes

The genetic deafness syndromes which are likely to be encountered in special schools for the deaf are outlined in this section. Although oro-facial and dental involvement is not a major feature of all of these conditions, they are well-known in the sphere of genetic deafness, and may therefore come to the attention of the attending dental surgeon.

1. Waardenburg syndrome

The Waardenburg syndrome (WS) is a striking disorder in which pigmentary disturbance of the hair, skin and eyes is associated with perceptive deafness (Figure 1). In special schools for the deaf, the frequency of the WS ranges from 2% to 5%. The characteristic but variable manifestations are hypertelorism, a white forelock, heterochromia of the irides, patchy dermal hypopigmentation and depigmentation of body hair (Winship and Beighton 1992). Synophrys and dystopia canthorum are additional manifestations which contribute to the characteristic facial appearance. Aganglionic megacolon is an inconsistent concomitant which can cause bowel dysfunction. About 13 % of affected individuals have profound perceptive hearing loss, due to abnormalities in the inner ear. The condition is transmitted as an autosomal dominant trait and it is relevant that affected family members do not necessarily have hearing loss.

2. Treacher Collins Syndrome

The Treacher Collins syndrome (TCS), also termed “mandibulo-facial dysostosis” is characterised by hypoplasia of the mandible and zygoma, macrostomia, colobomata of the lower eyelids, down-sloping palpebral fissures and variable malformations of the external ears and the ossicles of the middle ear (Figure 2). The manifestations are very variable, and the facial appearance can range from severe disturbance to virtual normality. Involvement of the middle ear leads to conductive deafness; accurate diagnosis is crucial as this form of hearing loss may be improved by surgery.

The TCS is an autosomal dominant trait, with variable severity from generation to generation. Not all affected persons are deaf; nevertheless, a reflection of the frequency of the condition is that fact that children with the condition are encountered in virtually every special school for the deaf.

Involvement of the mandible and maxilla frequently leads to crowding or misplacement of the teeth. A high or cleft palate can exacerbate this problem. If the mandibular hypoplasia is severe, major reconstructive surgery may be necessary for functional and cosmetic reasons.

3. Usher Syndrome

The Usher syndrome manifests as profound perceptive deafness, which is present at birth due to cochlear hypoplasia. By mid-childhood, night blindness and constriction of the visual fields become evident, due to retinitis pigmentosa. The visual problems progress and the affected person eventually becomes

both blind and deaf. Education is focussed on preparation for a life with these overwhelming double handicaps. Inheritance is autosomal recessive and considerable non-allelic heterogeneity has been demonstrated.

The condition is encountered in the majority of special schools for the deaf, and apart from a positive family history, the diagnosis may be suspected when reading problems develop in the affected child.

There are no specific dental manifestations, but the visual disturbance can compromise dental hygiene with consequent caries and periodontitis. Routine dental care is based upon cognisance of the communication difficulties posed by absence of both hearing and sight.



Figure 5: Osteogenesis imperfecta. An affected mother and child with frequent fractures and blue sclerae which typify the disorder. Deafness and dentinogenesis imperfecta are variable concomitants.

4. Pendred Syndrome

The Pendred syndrome manifests as severe congenital sensorineural deafness with enlargement of the thyroid gland which develops in late childhood. The condition results from an inborn error of thyroxin metabolism, and it is inherited as an autosomal recessive trait. Diagnostic confusion can arise due to the physiological thyroid enlargement which sometimes occurs at puberty or in geographical regions where dietary iodine is deficient. The thyroid enlargement may be massive, requiring surgical reduction

5. Branchio-oto-renal syndrome

The Branchio-oto-renal syndrome presents with abnormalities of the embryological branchial arch, notably fistulae in the side of the neck and pits anterior to the auricles. Hearing loss may be conductive or sensorineural and very variable in severity. Malformation of the auricles and middle ears is equally inconsistent. Structural renal abnormalities may lead to eventual renal failure. The condition is a well established autosomal dominant trait.

In the dental context, the palate may be narrow, with a pronounced overbite and malalignment of the teeth. In the older child, renal status may impact upon dental management.

6. Other Genetic Deafness syndromes

Childhood deafness is a component of numerous uncommon genetic syndromes. In some, the deafness is profound while in others the hearing loss may be mild. Conditions in this category which have been occasionally encountered in the special schools for children with profound or partial deafness in South Africa are listed below:

- 6.1 Acrofacial dysostosis
- 6.2 Crouzon syndrome
- 6.3 Apert syndrome



Figure 6: Dentinogenesis imperfecta. The teeth are translucent, discoloured and brittle.

- 6.4 Deafness-Multiple lentigenes syndrome (LEOPARD syndrome)
- 6.5 Alport syndrome
- 6.6 Alstrom syndrome
- 6.7 Refsum syndrome
- 6.8 Osteopetrosis
- 6.9 Sclerosteosis (Figures 3 and 4)
- 6.10 Osteogenesis imperfecta (Figures 5 and 6)
- 6.11 Jervell surdocardiac syndrome
- 6.12 Dwarfing skeletal dysplasias; SED congenita; Kniest syndrome
- 6.13 Mucopolysaccharidoses (notably MPS II, Hunter syndrome)
- 6.14 Stapes fixation, with congenital facial palsy and external ear malformation (Sellars syndrome)

7. Uncommon Genetic Deafness syndromes and Dental Anomalies

The combination of deafness with abnormalities of the teeth and periodontal tissues is a feature of a few genetic disorders. These conditions are very rare and as yet none of them have been documented in the special schools for the deaf in South Africa.

- 7.1 Lacrimoauriculodentodigital syndrome
- 7.2 Otodental dysplasia
- 7.3 Dental enamel dysplasia, brachydactyly, deafness
- 7.4 Johansson-Blizzard syndrome (absence of permanent teeth, pancreatic and thyroid dysfunction, growth retardation, perceptive deafness)
- 7.5 Gingival hyperplasia and progressive perceptive deafness

Multifactorial Deafness Syndromes

1. Goldenhar syndrome

The Goldenhar syndrome, also known as hemifacial microsomia and oculo-auriculo-vertebral dysplasia, is a variable but well recognised disorder which is encountered in most special schools for the deaf. The syndrome components, which are usually unilateral, comprise conductive deafness, facial asymmetry, malformed external ears and middle ear structures, colobomata of the upper eyelids and epibulbar dermoids. Spinal involvement includes fusions, hemivertebrae and spina bifida. Cardiac and other visceral abnormalities are present in a small proportion of affected persons.

Although the condition is occasionally familial, the majority of individuals with the condition are sporadic and the genetic pathogenesis remains uncertain.

A variety of oro-dental problems have their origins in

asymmetrical hypoplasia of the jaws. In particular, asymmetry of the maxilla and mandible leads to dental malocclusion, while hypoplasia of the mandibular ramus and temporo-mandibular joint can cause difficulty in mastication and articulation. The facial musculature, tongue and palate may also be hypoplastic and dysfunctional, while asymmetrical macrostomia can result from clefting at the corner of the mouth.

2. Wildervanck syndrome

The Wildervanck syndrome, also termed cervico-oculo-acoustic dysplasia presents as the Klippel-Feil syndrome (i.e. fusion of cervical vertebrae) plus mixed or perceptive deafness and occasionally unilateral Abducens (6th) cranial nerve palsy. Cleft palate is a variable component which can lead to dental crowding and malalignment. The genetic pathogenesis is uncertain.

3. Rubella Embryopathy

Rubella (German measles) is a mild febrile illness which manifests with a transient rose red macular rash and enlarged lymph glands in the posterior cervical triangle. Although this viral infection is little more than a minor inconvenience to the affected person, the consequences can be devastating to the unborn child. Foetal damage in surviving neonates can involve the brain, heart, auricles and the hearing mechanism.

In a child with perceptive deafness, the diagnosis of rubella embryopathy can be suspected on a basis of a combination of external ear malformation and structural cardiac abnormalities. A high palate, long fingers and maxillary hypoplasia may represent additional diagnostic features, although this association has not been substantiated.

Rubella embryopathy is frequently cited as the cause of hearing loss in children in the special schools for the deaf. In many instances, this diagnosis is suggested with hindsight by parents or carers seeking a reason for the child's hearing deficit and without objective evidence, counselling and prognostication is circumspect.

Malalignment and crowding of the teeth may be associated with a high palate, but otherwise, there are no specific dental concomitants. The possible presence of a structural cardiac defect is a crucial factor for dental anaesthesia and the assessment of cardiac status is mandatory in any deaf child in whom the diagnosis of rubella embryopathy is suspected. Equally, as in any child with a structural cardiac lesion, antibiotic cover during dental procedures is mandatory.

4. Foetal Alcohol syndrome

The Foetal alcohol syndrome is a spectrum of developmental abnormalities which result from maternal ingestion of alcohol

during pregnancy. This condition is present in as many as 5% of all scholars in some communities in the Cape and represents a major socio-medical problem.

Children with the foetal alcohol syndrome (FAS) have a low birth weight, slight stature, blepharophimosis and absent nasolabial folds; the latter manifest as a smooth upper lip. Intellectual dysfunction which compromises scholastic achievement is the most important component of the FAS. Hearing loss is present in a small proportion of affected children, but in view of the very high frequency of the disorder in the Cape, the FAS is encountered in the special schools.

Dental hygiene is frequently sub-optimal in affected children, with consequent periodontitis and caries. Dental management is influenced by the intellectual dysfunction.

5. Aminoglycoside Ototoxicity

The aminoglycoside group of medicinal agents are potentially ototoxic and maternal therapy during pregnancy can damage the hearing of the foetus.

In the past, gentamycin was utilised for the treatment of external burns, with consequent hearing loss; until recently deaf children with extensive scarring due to burns were regularly encountered in the special schools for the deaf in South Africa.

Tuberculosis is a major health problem in South Africa where treatment is compromised by the emergence of strains of the causative bacterium which are resistant to several therapeutic agents. Streptomycin is a well established therapeutic modality, but in high doses is ototoxic, causing perceptive hearing loss. Undue sensitivity to conventional doses of streptomycin can be transmitted by mitochondrial inheritance; this familial condition is present in the Cape, and poses a potential threat to the hearing mechanism of persons with tuberculosis (Gardner et al 1997). The incidence is unknown, but pre-treatment screening would be an appropriate approach.

Dental Management in Profound Childhood Deafness

The following factors warrant consideration in the dental management of deaf children:

1. Deafness and speech problems may result from communication difficulties especially in regard to local or general anaesthesia, and to the dental procedures to be undertaken. The involvement of a teacher or carer who can communicate with the deaf child may be essential, especially if sign language is used.

2. Intellectual deficit can result from kernicterus, cerebral palsy and metabolic disorders. If severe, mental impairment may overshadow the hearing loss and necessitate a modified dental approach. Some children with brain damage have epilepsy; hyperplasia of the gums may result from anti-epileptic therapy.

3. In certain genetic deafness disorders, such as the Treacher Collins syndrome, involvement of the face (mandible and maxilla), palate, dentition and gingival tissues frequently requires highly complex maxillo-facial, orthodontic and dental management. In these circumstances, diagnostic precision and awareness of the potential range of oro-facial manifestations is of value in planning the protocol for ongoing dental management.

4. Systemic syndromic manifestations such as cardiac involvement may impact on long term dental management, as in rubella embryopathy. Awareness of the potential for complications of this type in some genetic deafness disorders facilitates appropriate comprehensive health care.

References

1. Arnos KS. Hereditary hearing loss. *N Engl J Med* 331:469-470, 1994.
2. Sellars S, Beighton G, Horan F, Beighton P. Deafness in Black children in Southern Africa. *S A Med J* 51: 309-312; 1977.
3. Gardner JC, Goliath R, Viljoen D, Sellars S, Cortopassi G, Hutchin T, Greenberg J, Beighton P. Familial streptomycin ototoxicity in a South African family: a mitochondrial disorder. *J Med Genet* 34:904-906, 1997.
4. Beighton P, Sellars SL. *Genetics and Otology*. Churchill Livingstone, Edinburgh 1982
5. Sellars S, Beighton P. Childhood deafness in Southern Africa. *J Laryngol & Otol* 97: 885-889; 1983.
6. Beighton P, Viljoen D, Winship I, Beighton G, Sellars S. Profound childhood deafness in Southern Africa. *Ann NY Acad Sci* 630: 290-291, 1991.
7. Petit C. Genes responsible for human hereditary deafness: symphony of a thousand. *Nature Genet* 14:385-391, 1996.
8. Winship I, Beighton P. Phenotypic discriminants in the Waardenburg syndrome. *Clin Genet* 41: 181-188, 1992.
9. Gardner J, Beighton P, Sellars SL. The genetics of childhood deafness. *SAMJ* 87(12):1661-2, 1997.

Acknowledgements

We are appreciative of Greta's long term involvement in the investigation of children with profound hearing loss and for her participation in the preparation of this document.

Our investigations were supported by the South African Medical Research Council and the National Research Foundation.