

Earpits Syndrome in Children with Hearing Loss

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KEY WORDS Earpits syndrome; conductive hearing loss; mixed hearing loss; branchial fistulae; preauricular sinuses; consanguinity.

ABSTRACT Earpits syndrome is a very rare kind of syndrome deafness and inherited as autosomal dominant. A study was taken up to understand the prevalence of this syndrome in children below the age of 14 years with hearing loss. Out of 743 children with hearing impairment, earpits syndrome was observed in 2(0.27%) cases. Both the children were the products of consanguineous marriage. Mixed hearing loss was noticed in one case and conductive hearing loss in another case. However, no renal anomalies were detected in either of these cases.

INTRODUCTION

Earpits syndrome is one of the important forms of syndromic deafness which is autosomal dominant in nature. Cremers and Filkers van Noord (1980) who reported earpits syndrome described the characteristic features of the syndrome in detail. Preauricular pits, branchial fistulae and profound bilateral hearing loss are the most frequently expressed features of this syndrome. Slack and Phelps (1985) reported familial incidence of earpits syndrome. Islam and Habib (1995) described branchial cleft anomalies in earpits syndrome from Bangladesh. The other features of this syndrome include lacrimal duct stenosis, structural defects of the ear and renal anomalies. Studies carried out by Steel and Kros (2001) also reported, earpits syndrome as one of the important deafness syndrome among genetically inherited syndromes. Reports on association of syndromes with hearing loss are very limited from South-India and hence a study was taken up to understand the prevalence of this syndrome in children with hearing loss.

MATERIALS AND METHODS

The present study included 743 children below 14 years of age with hearing impairment. These subjects were selected from Govt.E.N.T. Hospital, Hyderabad and some schools for deaf children in and around the twin cities of Hyderabad and Secunderabad. The clinical diagnosis of these patients was made after a detailed examination by E.N.T surgeon and upon

confirmation of hearing loss, a detailed information regarding the proband's age and sex, occupation of parents, literacy, socio-economic status, prenatal, perinatal and postnatal history, family history, type of marriage and developmental history was collected from proband's parents and other available family members using a standard questionnaire. Various audiological tests like Pure Tone Audiometry (PTA), Brain Stem Evoked Response Audiometry (BERA) and Oto Acoustic Emissions (OAE) were carried out on the proband to know the degree and type of hearing loss.

RESULTS

743 children with hearing impairment below 14 years of age were analyzed out of which 2 (0.27%) children showed earpits syndrome. The expression of clinical features of earpits syndrome and the type and degree of hearing loss in both the cases are presented in table 1. Profound bilateral hearing loss was observed in both the cases. Mixed hearing loss was observed in one case with structural defects in outer, middle and

Table 1: Clinical Features of Earpits Syndrome

Clinical Features	Case-1	Case-2
Preauricular pits	+	+
Structural defect in outer ear	+	+
Structural defect in middle ear	+	+
Structural defect in inner ear	+	-
Branchial clefts	+	+
Renal anomalies	-	-
Degree of hearing loss	Profound	Profound
Type of hearing loss	Mixed	Conductive

inner ear, and in the other case, conductive hearing loss was observed with structural defects in only outer and middle ear. Preauricular pits and branchial clefts were seen in both of them. However no renal anomalies were seen in either of them.

DISCUSSION

Hearing Impairment, a most prevalent sensory handicap is seen in 1 in 750 children. Genetic factors play an important role in more than 50% of all the cases with hearing loss. Deafness is categorized as Syndromic deafness and isolated deafness depending on the presence or absence of associated manifestations from other organs (Tranebjaerg 2000). Approximately 30% of congenital cases associated with genetic factors are classified as Syndromic, of which ear pits deafness syndrome is one (Gorlin and Toriello 1995; Steel and Kros 2001). Earpits deafness syndrome is a rare autosomal dominant disorder. The clinical features of this syndrome include structural defects in the ear, branchial cleft, preauricular sinuses, lacrimal duct stenosis and renal anomalies. All the symptoms need not be expressed in all the cases that carry the gene for this syndrome. In the present study carried out on 743 children with hearing loss, two of them showed ear pits syndrome with preauricular pits, branchial clefts, structural defects in the ear and profound bilateral hearing loss. None of them were observed with renal anomalies. While one child had mixed hearing loss, the other child was observed with conductive hearing loss. Sellars and Beighton (1985) reported 3 children from Indian Stock with profound bilateral conductive deafness with variable malformation of external ear, congenital facial palsy and stapedial anomalies. However, Cremers et al. (1980) reported that the children with earpits syndrome can have any of the three kinds of hearing loss namely, sensorineural, conductive or mixed hearing loss.

Both of them were delivered by caesarian section and were the products of consanguineous marriage. No family history was seen in both the cases. Developmental delay was also not reported in both the cases, but in one of the cases, the mother of the child had high blood pressure and viral infection during pregnancy. Further studies are warranted to take up large scale screening programme of the hearing handicap to identify genetically inherited syndromes so that appropriate genetic counseling can be offered.

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