Case Report

A family with Heimler's syndrome: Fifth case report in world literature!

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Abstract

Hearing loss due to genetic causes has a reported prevalence of 1 in 1000 births Among these 15- 30% are associated with other abnormalities , although a small number are associated with oro dental disorders. Syndromic hearing loss contributes to 30% of all genetic causes. There are greater than 500 syndromes associated with deafness. We report a pair of siblings along with their father who exhibited. findings similar to those described in Heimler's syndrome namely sensorineural hearing loss , amelogenesis imperfecta and nail abnormalities. Our findings support the theory of autosomal dominant inheritance. To our knowledge, there have been only four case reports of Heimler's syndrome till date. We present this case for its rarity in World literature and as the first case in Indian literature!

Keywords: Amelogenesis imperfecta, Sensori- neural hearing loss, Beau's lines.

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INTRODUCTION

Ectodermal dysplasia is a term used to describe a number of conditions that are present at or shortly after birth in which two or more of the bodies ectodermal structures (example: hair, teeth, nails, sweat glands) fail to develop or grow properly. There are two hundred different types of ectodermal dysplasia described in medical literature. The incidence of its occurrence is about seven cases per ten thousand births. In early 1980s two doctors, Friere-Maia and Pinheiro classified ectodermal dysplasias into;

Type A: Pure ectodermal dysplasia

Type B: Ectodermal dysplasia syndrome

Type A consists of congenital abnormalities of two or more ectodermal structures numbered as (1) hair (2) teeth (3) nails 4) sweat glands. Type B Ectodermal dysplasia are defined as having an inherited abnormality in one of the four major structures mentioned above plus one or more abnormalities in other ectodermal structures such as ears, lips or skin. The features may include:

Hair: Fine sparse and light coloured or fragile, wavy or coiled.

Teeth: Pointed or peg shaped having enamel defects with susceptibility to cavities and decay.

Nails: Thin and brittle.

Sweat Glands: Reduced in number which lead to difficulty in coping with increase body temperature.

Skin Dry: Thin, scaly, cracked susceptible to bleeding and infection.

Ectodermal dysplasias occur due to two hundred abnormalities in genes that control the normal development of ectodermal structures. Most of the ectodermal dysplasias are diagnosed clinically. Unfortunately there is no cure or corrective treatment for any form of ectodermal dysplasias. Fortunately the syndromes are usually non progressive and generally do not affect over all lifespan. Hemliers syndrome is type B ectodermal dysplasia in which Amelogenesis Imperfecta, Sensori Neural Hearing loss and Beaus lines were reported.[1]

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CASE REPORT

A sixteen year old boy presented to ENT outpatient department with complaints of bilateral hearing loss since the age of seven years. On examination he had normal tympanic membranes, bilateral severe sensori neural hearing loss with yellowish discolouration of teeth. On enquiring about oral hygiene he narrated that his father and youngest sister had similar yellowish discoloration of teeth. The whole family was examined and two siblings along with father were found to be affected. A Dental examination of the family was done which was diagnosed as Amelogenesis Imperfecta (enamel hypoplasia) affecting permanent dentition in father and son whereas it was found to be affecting primary and permanent dentition in youngest sibling. All three members exhibited peg shaped teeth and dental caries. They also had dry scaly skin at places cracked with bleeding and crust formation. The eldest boy presented with a combination of bilateral moderately severe sensori – neural deafness, amelogenesis imperfecta and Beaus lines on nails of fingers and toes. His youngest sister eleven years old also had bilateral severe to profound sensorineural deafness since childhood with amelogenesis imperfecta , Beau's lines and Idiopathic guttate hypomelanosis along with bilateral renal calcification. Both the siblings were born at term following normal pregnancy with birth weight of 2.5 and 3kg. None of them had any major illness and all developmental milestones were achieved within normal limits. The father of these siblings aged fourty two years had bilateral moderately severe sensori neural hearing loss since childhood with amelogenesis imperfecta, attrision of teeth and Beau's lines and idiopathic guttate hypomelanosis. There was no history of intellectual impairment in any of the affected members nor history of consangious marriage of parents. The mother was normal where as his younger sister aged fourteen years was apparently normal except for bilateral renal calcification. Biochemical investigations revealed high levels of calcium in all three siblings along with their father. Illustrations - 1 to 4.

Figure 1:

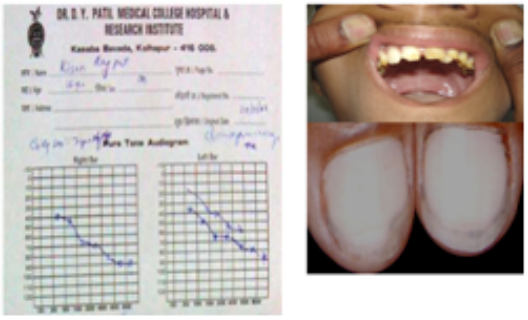

Figure 2:

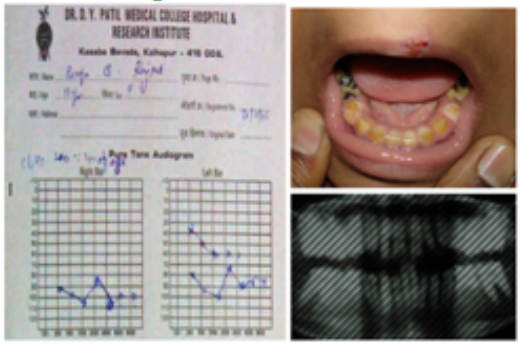

Figure 3:

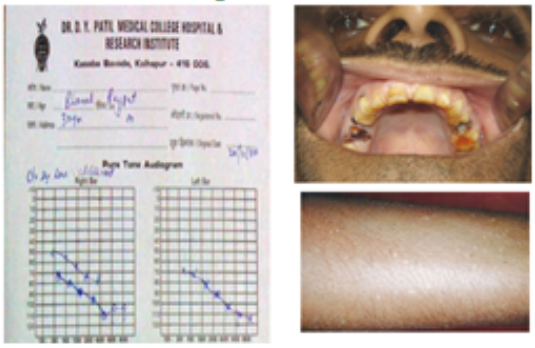

Figure 4:

Figure 1: Family with affected father and two siblings (tick-marked)

Figure 2: Sibling –1: Audiogram showing bilateral moderately severe sensori – neural deafness, amelogenesis imperfecta and Beaus lines on finger nails.

Figure 3: Sibling –2: Audiogram showing bilateral severe to profound sensori – neural deafness, amelogenesis imperfecta with carious teeth and OPG showing attrision of teeth.

Figure 4: Father of Siblings - Audiogram showing bilateral severe sensori – neural deafness, amelogenesis imperfecta with carious teeth and forearm skin showing guttate hypomelanosis.

Table 1: Summary of all five case reports

Word literature Review	1st case 1991 Heimler *et al*		2nd case 1999 Tischkoowitz *et al*	3rd case 2003 Pollack *et al*		4th case 2006 Ong *et al*		5th case 2007 A.A.Mohite *et al*		
	Sibling 1	Sibling 2	Single	Sibling 1	Sibling 2	Identical Twin 1	Identical Twin 2	Father	Sibling 1	Sibling 2
Sensorineural hearing loss	Bilateral	Bilateral	Unilateral	Bilateral	Bilateral	Bilateral	Bilateral	Bilateral	Bilateral	Bilateral
Diagnosed at	18 months	2 ½ years	7 Years	1st year	1st year	3 years	3 years	Child hood	7 years	7 years
Primary dentition	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Enamel hypoplasia
Permanent dentition	Enamel hypoplasia	Enamel hypoplasia	Enamel hypomineralisation	Enamel hypoplasia	Enamel hypoplasia	Enamel hypoplasia	Enamel hypoplasia	Enamel hypoplasia	Enamel hypoplasia	Enamel hypoplasia
Nail defect	Punctate Leukonychia Beau's lines	Punctate Leukonychia Beau's lines	Beau's lines	Absent	Absent	Leukonychia Beau's lines	Leukonychia Beau's lines	Punctate Leukonychia Beau's lines	Punctate Leukonychia Beau's lines	Punctate Leukonychia Beau's lines
Skin – hypomelanosis	Absent	Absent	Absent	Absent	Absent	Present	Present	Present	Present	Present
High serum Calcium	-	-	-	-	-	-	-	+	+	+

DISCUSSION

The combination of sensorineural hearing loss, amelogenesis imperfecta and nail abnormalities was first reported by Heimler *et al*[2]. They described two siblings who had profound bilateral sensorineural hearing loss, amelogenesis imperfecta of permanent dentition and Beau's lines. Inheritance of this syndrome was postulated to be autosomal recessive which does not hold true in our report. Our case report favours autosomal dominant inheritance. This can be explained on the following basis. Identification of genes and mutation analysis in families segregating recessive and dominant inheritance have revealed unique behaviour of several mutant alleles. It has been documentated that a gene expressing recessive inheritance in some families was implicated in other families segregating autosomal dominant inheritance. TMC1 is example of one such gene, that was identified in several recessive families from India and Pakistan, as well as in an American large kindred segregating dominant inheritance.[3] This phenomenon where mutations within the same gene found to result in a variety of clinical phenotypes with different modes of inheritance has been postulated to be the result of differential behaviour/effect of the mutant alleles. The second case was reported by Marc Tischkowitz *et al* in 1999[4] with findings similar to Heimlers syndrome. Pollack *et al* in 2003[5] reported a pair of siblings who exhibited sensorineural hearing loss and eamel hypoplasia. However nail findings of Beau's lines and leuconychia were absent in their cases.This was the third case of Heimlers syndrome. The fourth case was reported by Ong K R *et al* in 2005 [6] was identical twin girls with sensorineural deafness ,enamel and nail abnormalities. We report the fifth case in which father and two siblings were affected with similar findings of profound bilateral sensorineural hearing loss, enamel hypoplasia and nail findings of Beau's lines as in Heimlers Syndrome. Heimler *et al* suggested that the syndrome could be the result of a single gene affecting derivatives of the ectodermal tissue because the abnormalities describe have a common embryological origin in the ectoderm.

Sensorineural hearing loss is probably due to a defect of cells of the Organ of Corti which are of ectodermal origin. Amelogenesis Imperfecta is classified according to the predominant clinical and radiographic appearance of the enamel defect and on the mode of inheritance of the trait. Beau's lines are transverse lines across the nails that can arise from severe illness such as sepsis, acquired immunodeficiency syndrome, bullous pemphigoid and can also be physiological in menstruating women. Idipathic guttate hypomelanosis are multiple a symptomatic depigmented macules usually seen on lower limbs and sometimes on upper limbs and trunk. It is supposed to be a type of guttate vitiligo. Our case also emphasizes the importance of thorough examination of the skin and nails in patients presenting with a combination of impaired hearing and dental pathology. Table (1)

CONCLUSION

For several individuals with Ectodermal dysplasia healthcare may be needed from dentist, plastic surgeons, paediatricians, dermatologists and many other personnel. In the future it may be possible to improve some forms of ectodermal dysplasia when the affected individual is still an embryo within its mothers womb but such goals are still confined to research laboratories at present.

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