Original Research Paper

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Otorhinolaryngology

"ASSOCIATION OF FAMILY HISTORY AND CONSANGUINITY WITH PERMANENT HEARING LOSS."

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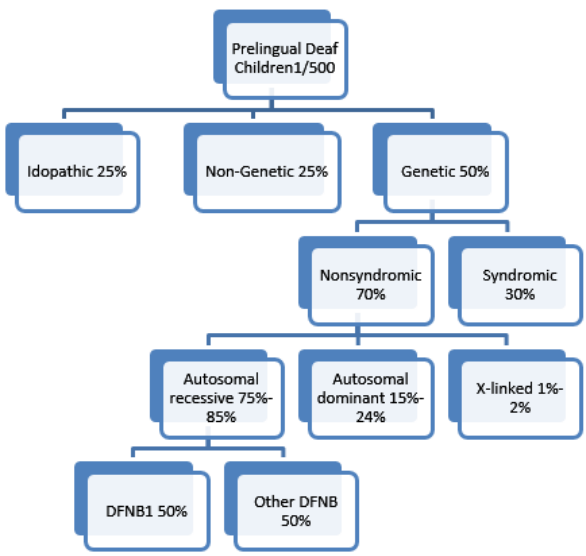
ABSTRACT Knowing the association of family history of hearing loss and consanguinity with congenital hearing loss provides important clues to prevent the occurrence in subsequent generations. A case control study was designed on 30 children with permanent hearing loss and 30 children with normal hearing from Feb.2014 to Dec.2015.The case control ratio was 1:1. Detailed history was taken from the parents regarding family history and consanguinity. 36.7% of cases had a positive family history. 56.7% of cases had parents with consanguinity. Family history and consanguinity seem to be important risk factors of congenital hearing impairment, both in isolation and in combination.

KEYWORDS : . Family history, Consanguinity, childhood permanent hearing loss

INTRODUCTION:

About 1 in 1000 babies born in India are profoundly deaf (>/= 90 dB in better ear). Most children (50%) with permanent hearing loss have a genetic etiology.

CAUSES OF CHILDHOOD DEAFNESS



- SYNDROMIC HEARING IMPAIRMENT: is associated with malformations of external ear or other organs or with medical problems involving other organ systems.

1. Autosomal Dominant Syndromic Hearing Impairment : Waardenburg syndrome, Branchiootorenal syndrome
2. Autosomal Recessive Syndromic Hearing Impairment : Usher syndrome, Pendred syndrome,Jervell and Lange-Nielsen Syndrome, Biotinidase deficiency,Refsum disease
3. X Linked Syndromic Hearing Impairment : Alport Syndrome, Mohr-tranebjaerg Syndrome
4. Mitochondrial Syndromic Hearing Impairment

- Non-Syndromic Hearing Impairment : has no associated visible abnormalities of external ear or any related medical problems. But can be associated with abnormalities of middle ear or inner ear.
- Non genetic causes: include maternal infections such as CMV, rubella, syphilis and perinatal factors such as preterm, low birth weight, etc.
- There is a strong association of family history and consanguinity with permanent hearing loss in infants.

DEGREE OF FAMILY RELATIONSHIPS:

- FIRST DEGREE: parent, siblings and children. They share half of their genes
- SECOND DEGREE: grandparents, grandchildren, aunts, uncles, nephews and nieces. They share quarter of their genes.
- THIRD DEGREE: second degree cousins share 1/32 of their genes. First degree cousins share 1/8th of their genes.

In 1853, William Wilde observed that a high rate of consanguinity resulted in an increased risk of having hearing impaired children

In 1880, Arthur Hartmann published his research data on the genetics of hearing impairment and concluded that direct transmission (autosomal dominant) and indirect transmission (autosomal recessive) patterns existed.

In 1887, Adam Pulitzer established that the indirect route of inheritance in pedigrees having first generation common ancestors resulted in multiple affected siblings with deaf children a few generations later.

In 2000, Colin Mathers, et al stated that the prevalence rate for congenital hearing impairment in the world was 0.112%.

In 2009, G.padma, P.V.Ramchander, U.V.Nandur and T.Padma studied GJB2 and GJB6 mutations found in Indian probands with congenital hearing impairment.

In 2013, Heramba Ganapathy Selvarajan, et al did a case control study on 420 infants with permanent hearing impairment and normal hearing. Family history and consanguinity was seen in 18.6% and 39.5% of hearing impaired group. The combinations of the two risk factors are seen in 10% of hearing impaired group whereas only 0.5% had in control group.

MATERIALAND METHODS:-

Prospective study of 60 patients; 30 of them had permanent hearing loss and 30 had normal hearing. A case control study was carried out in age group of 6 months to 5 years in MGM Medical College and hospital, Kamothe, Navi Mumbai from February 2014 to December 2015. The case control ratio was 1:1. Alternate sampling method was used in the hospital for selecting control group. Parent interview was carried out to collect the information of family history of hearing impairment and consanguineous marriage.

The collection of data comprised of the following:-

- A detailed clinical history regarding age at which the hearing

disability was suspected and any associated history of speech impairment was taken
- A three generation family history with attention to other relatives with hearing loss was also obtained.
- ENT examination
- General examination to rule out anamolies of syndromic deafness
- Audio logical evaluation:
- Behavioral observational audiometry (0 to 7 months)
- Distraction test (6 to 24 months)
- Performance test (2 to 5 years)
- PureToneAudiometry {PTA} (>3 Years)
- BERA {Brainstem evoked response audiometry}
- OAE {oto acoustic emission}

OBSERVATION AND RESULTS:
- Age (p>0.05) and gender (p>0.05) were not significant factors in permanent hearing loss.
- Socioeconomic status also had no significant association with permanent hearing loss.(p>0.05)
- There was significant association between language development and hearing loss (p< 0.05)

Table no 1: Association between language development and hearing loss

Language

			BERA		Total
			Controls	Cases	
Language	Delayed	Count	29	28	57
		%	96.7%	93.3%	95%
	Absent	Count	1	2	3
		%	3.3%	6.7%	5%
Total		Count	30	30	60
		%	100%	100%	100%

Chi-square = 0.351, DF = 1, p = 0.045, Significant

- High significant association between family history and hearing loss (p<0.01)

Table no 2: Association between family history and hearing loss

			BERA		Total
			Controls	Cases	
Family History	Positive	Count	0	11	11
		%	0.0%	36.7%	18.3%
	Negative	Count	30	19	49
		%	100%	63.3%	81.7%
Total		Count	30	30	60
		%	100%	100%	100%

Chi-square = 13.469, DF = 1, p = 0.001, Highly Significant

Significant association between consanguinity and hearing loss (p<0.05)

Table no 3: Association between consanguinity and hearing loss

			BERA		Total
			Controls	Cases	
Consanguinity	Second Degree Consanguinity	Count	2	5	7
		%	6.7%	16.7%	11.7%
	Third Degree Consanguinity	Count	6	12	18
		%	20%	40%	30%
	Negative	Count	22	13	35
		%	73.3%	43.3%	58.3%
Total		Count	30	30	60
		%	100%	100%	100%

Chi-square = 5.601, DF = 2, p = 0.042, Significant

No significant association between BOA and BERA (p>0.05)

Significant association between OAE and BERA (p<0.001)

Table no.4: Association between hearing assessment by OAE and BERA

			BERA		Total
			Controls	Cases	
OTOACOUSTIC EMISSION	present	Count	22	9	31
		%	73.3%	30%	51.7%
	Absent	Count	8	21	29
		%	26.7%	70%	48.3%
Total		Count	30	30	60
		%	100%	100%	100%

Chi-square=11.279, DF=1, p=0.001, Highly Significant

DISCUSSION:

This is a comparative study on 60 patients between age group of 6 months to 5 years who attended our ENT OPD. They were from all socioeconomic groups.30 of them were with permanent hearing loss and remaining 30 were with normal hearing. The prevalence of consanguinity and family history of hearing loss in these 2 groups were assessed. There is no predilection for a particular age group. Male to female ratio was 1:1.Both cases and controls were immunized, had normal APGAR scores and had more or less equal NICU admission history. Almost 93.3% of cases with hearing loss had delayed language development, while 10% had delayed social development. Almost 36.7% of cases had positive family history of hearing loss.

In this study, 16.7% of cases had parents who were related to the second degree. Another 40% were of third degree. Remaining 43.3% had no history of consanguinity. The most common factor of detection is still parental suspicion. Hence the low rates of presentation in neonatal age group. All the parents and available family members were counseled against consanguineous marriage and advised to undergo genetic testing where feasible.

CONCLUSION:

The findings of this study reveal that family history and consanguinity are important risk factors for sensorineural hearing loss in children, both in isolation and in combination. Genetic counseling before marriage is a preventive measure that can solve this problem.

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