Sensorineural Hearing Impairment is a Common Feature of Consanguineous Marriage

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ABSTRACT

Introduction: Sensorineural hearing loss (SNHL) accounts for about 60% of all hearing loss. This is sometimes also called "Nerve deafness". The term "Sensorineural" is used to indicate that there is either a cochlear or an eight nerve lesion. The diagnosis of sensorineural hearing loss is made through audiometry, which shows a significant hearing loss without "The air-bone gap" that is characteristic of conductive hearing disturbances.
Among various risk factors described for deafness, consanguinity is an established high risk.
Aim of the Work: This work was carried out to study the prevalence of sensorineural hearing loss in offsprings of consanguineous marriage, who attended the Medical Genetics Center, Ain Shams University.
Patients and Methods: The study was performed on 950 children with congenital hearing loss.
Results: Consanguineous marriage was present in 71.2% of studied cases, 47.3% of these cases, parents were 1st cousin, in 36.7% parents were 2nd cousin, 16% had remote consanguinity. 28.8% of cases were the offspring of non-consanguinous marriage. Also, the results showed that 44.2% of cases had severe degree of sensorineural hearing loss (71-90 dBHL), 24.3% had profound hearing loss (>90 dBHL). Autosomal dominant inheritance (AD) hearing loss was detected in 40% of cases. Autosomal recessive inheritance hearing loss was discovered in 27% of cases, and in 18.7% of study cases the hearing loss was associated with genetic syndromes, where 88.8% of these cases were associated with Down syndrome.
Conclusion: The incidence of hereditary hearing impairement is commoner in developing counteries compared to developed counteries, so, prevention is essential to reduce the incidence of genetic hearing loss. Premarital and antenatal screening should be applied whenever possible, at least for those at risk of developing genetic diseases including hearing impairement.

Key Words:

Consanguinity; hearing disorders; preventive medicine.

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INTRODUCTION

Hearing is a complex process that consists of many steps.[1]

Sensorineural hearing loss is always due to damage to either the cochlea or the auditory nerve. Sound waves may reach the inner ear, but they are not transmitted successfully to the brain.[2]

The development of cochlea and hair cells is dependent on a genetic pathway called planar cell polarity (PCP) pathway. This pathway is involved in the formation of the polarized structure of the auditory sensory organ and regulates the embryonic development. Genetic disturbances disturb the pathway leading to congenital hearing loss.[3]

Sensorineural hearing loss presents more serious problems. It often cannot be cured. Fortunately, hearing aids can help restore some of the hearing loss.[4] Hearing impairment has debilitating effects on children as it can retard individual's language acquisition skills and impair the overall development.[5] The world wide prevalence of profound, congenital deafness is 11 per 10.000 children, and is attributable to genetic causes in at least 50% of cases.[6] In the developed countries about 60% of cases with deafness are reported to have a genetic origin.[7] Inheritance plays a major role in children with sensorineural hearing loss, with consanguinity being the major cause.[8] Among hereditary deafness, autosomal recessive inheritance predominates accounting for 80% of the cases followed by autosomal dominant inheritance in about 20% of the cases and X-linked and mitochondrial modes of inheritance in less than 1% of the cases.[9]

Consanguineous marriage is common among Asian, African, and Latin American Communities. The siblings of consanguineous marriages have a significantly higher incidence of autosomal recessive diseases including hearing impairment. Marriage within the family increase the risk of hearing impairment and other diseases.

AIM OF THE WORK

- To study the prevalence of sensorineural hearing loss (SNHL) due to consanguineous marriage among children who attended the Medical Genetic Center, Ain Shams University.
- To determine the inheritance pattern of SNHL among studied cases.
- To know the genetic causes of hearing loss among affected cases.

PATIENTS AND METHODS

This descriptive cross-section study was conducted during the period from (1 February 2006 to 31 December 2007) among children who attended the Medical Genetic Center, Ain Shams University. Nine hundred and fifty infants and preschool age children between ages 1 month to 6 years were included in the study.

Inclusion Criteria:

1. All infants and preschool age children at genetic risk who attend the outpatient clinic at the Medical Ge-

netics Center Ain Shams University.
2. Children with a positive family history of deafness.
3. Children with known risk factors for hearing impairment.

Exclusion criteria:
1. Children with positive past history of known risk factors (Hyperbilirubinemia, rubella, meningitis, encephalitis, etc...) were considered deaf as a result of these factors.
2. Children subjected to trauma or having tumor were excluded.

All patients were subjected to the following questionnaire after taking their parents written consent:

1- Personal History:
- Name, age, sex.

2- Full data about Prenatal, Neonatal and Postnatal history.

3- Developmental history.

4- Family history:
- Consanguinity of parents.
- Hearing loss
- Hearing and speech deficits.
- Exposure to know risk factors for hearing impairement [Infection (e.g. meningitis, rubella); birth trauma; ototoxic drugs; fever].

5- Family pedigree.

6- Complete physical and otologic examination was done.
- All participants had their own files in the Clinic for regular follow up visits.

The following investigations were routinely done:
- TSH for thyroid diseases causing hearing impairement (e.g. Pendred's disease).
- Thin layer chromatography (TLC) to exclude metabolic causes (e.g. in cases with positive family history of histinemia).

Special investigations for hearing assessment:
- ***Pneumatoscopy*** to rule out an effusion as the cause of conductive hearing loss.

- ***Immittancemetry*** [GSI 33]: to evaluate middle ear function which inform us about the threshold of hearing.

- ***Audiometry:***
  - For cases<3 years, Free Field Orbiter 922 Equipment was used.
  - For cases from 3–6 years, Medimate 602 Equipment (Play audiometry) was used.

Audiometry documents the degree and pattern of hearing loss.

- ***Auditory Brain Stress Response (ABR)*** [MK-12 Equipment] which describes the function of the auditory pathway up to the brain stem.

Otologic investigations were done at the auditory clinic of Ain-Shams University Hospital.

RESULTS

Table 1: Characteristics of the study group.

Sex	No.	Percent
Male	460	48.4%
Female	490	51.6%
Total	950	100%
Age category	**No.**	**Percent**
1 – 12 month	35	3.7%
13 – 24 month	100	10.5%
25 – 36 month	120	12.6%
37 – 48 month	210	22.1%
49 – 60 month	235	24.8%
61– 72 month	250	26.3%
Total	950	100%

Table 2: Degree of consanguinity of the parents of affected child.

Degree of consanguinity	No.	Percent
1st cousin	320	47.3%
2nd cousin	248	36.7%
Remote consanguinity	108	16%
Total	676	100%

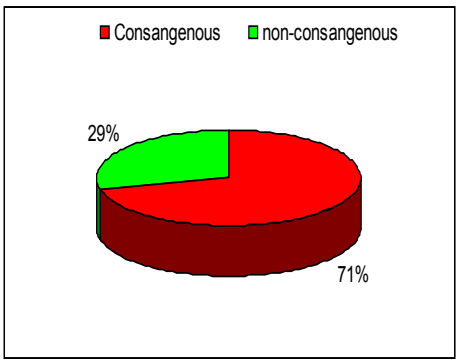

Fig. 1: Percentage of consanguinity among parents of affected children (950 index cases).

Table 3: Degree of sensorineural hearing loss (SNHL).

Degree	No.	%
Mild hearing impairment (> 25 – 40 dB HL)	49	5.2%
Moderate hearing impairment (41 – 55 dB HL)	110	11.6%
Moderate - Severe hearing impairment (56 – 70 dB HL)	140	14.7%
Severe hearing impairment (71– 90 dB HL)	420	44.2%
Profound hearing impairment (> 90 dB HL)	231	24.3%
Total	950	100%

Table 4: Inheritance pattern and causes of hearing loss in study cases.

Inheritance pattern	No.	%
* *Hearing loss inherited as:*		
• Autosomal dominant inheritance (AD)	308	40%
• Autosomal recessive inheritance (AR)	207	27%
• X-linked inheritance	257	33%
Total	772	100%
Causes of hearing loss:		
** *Hearing loss associated with genetic syndromes*		
• Down syndrome	158	88.8%
• Treacher collins	7	3.9%
• Crouzon syndrome	11	6.2%
• Alport syndrome	2	1.1%
Total	178	100%

N.B.:

* AD, AR, X-linked were detected in 81.3% ([772/950] of the study cases).

** Hearing loss associated with genetic syndromes were found in 18.7% ([178/950] of the study cases).

DISCUSSION

Consanguineous marriages have been practiced for hundreds of years in many parts of the world, so the effect of consanguinity on hereditary deafness has been studied and documented.[10]

Consanguinity is still a factor in the appearance of birth defects in developed countries.[11] Consanguineous marriage can vary quite widely between and within countries, religious and cultural factors play a major part in determining social attitudes and legal frameworks at local and national levels.[12]

Estimates of consanguinity ratios in different parts of Egypt ranged from 29 to 50%.[13]

Many authors have suggested that approximately one half of SNHL in children can be attributed to hereditary causes. So some authors suggest such marriage should be avoided as they make double the risk of infants with birth defects such as mental retardation, deafness and blindness when compared with an "Unrelated" marriage.[14]

The study showed that consanguineous marriage accounted for 71.2% of affected cases. Out of this 47.3% of affected children their parents were 1st degree consanguinity, and 2nd degree consanguinity was present in 36.7% of affected cases.

These results are similar to a study published by Zakzouk[15], which was conducted on 168 children with SNHL, and revealed that consanguineous marriage was responsible for (66.1%) of affected cases.

In this study, autosomal recessive hearing impairment was present in 207 (27%) children out of 772 children. Also the study showed that 308/772 (40%) of cases had hereditary deafness due to a single gene (AD inheritance), X-linked hearing loss was detected in 257 (33%) of affected cases.

Our results were in accordance with Northern et al.[16] who reported that consanguineous marriage increases the risk of transmission of polygenic inheritance.

The risk to subsequent siblings in this type of inheritance is higher when the parents are consanguineous than when they are unrelated. The effect of consanguinity on the development of childhood hearing impairment depends on the closeness of the relationship of parents. A marriage between first cousins poses a great risk, whereas a distant consanguinity has comparatively low risk of producing defective offspring.

In the current study, hearing impairment was detected in association with genetic syndromes in 178 cases (18.7% of the study cases), 158 of these cases (88.8%) were associated with Down syndrome, and 7 cases (3.9%) were associated with Treacher Collins syndrome.

Eleven cases (6.2%) were associated with Crouzon syndrome, while 2 cases (1.1%) were associated with Alport syndrome. A study published by Shott

et al.[17] showed that children with Down syndrome have a high incidence of hearing impairment.

Peterson et al.[18] found that persons with Crouzon syndrome also have a SNHL.

Prone et al.[19] found that patients with Treacher Collins syndrome exhibit hearing loss secondary to absent, small or unusually formed ear (Microtia) and commonly results from malformations of the middle ear, external ear canal abnormalities and symmetrically dysmorphic or absent ossicles in the middle ear space.

RECOMMENDATIONS

1. Via counseling, the role of consanguinity can be elucidated to the parents.
2. Early intervention in presence of the positive family history or previous risk factors of hearing loss is recommended.

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