(International Print/Online Journal)

SJIF IMPACT FACTOR: 5.565
PUBMED-National Library of Medicine ID-101739732

ISSN (Print): 2209-2870
ISSN (Online): 2209-2862

International Journal of Medical Science and Current Research (IJMSCR)
Available online at: www.ijmscr.com
Volume 6, Issue 5 , Page No: 500-507
September-October 2023

IJMSCR

A Comparative Study Between Incidence Of Auditory Involvement Among Neonates Of Consanguineous And Non-Consanguineous Parents

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Type of Publication: Original Research Paper
Conflicts of Interest: Nil

Abstract

Introduction: Hearing is the deepest and most humanizing philosophical sense man possesses. 360 million people worldwide, suffer from disabling hearing loss. The estimated prevalence of childhood onset deafness is 2%, of which 50% of cases are thought to be inherited, i.e., of genetic/hereditary causes. Of these hereditary cases, approximately 30% are classified as syndromic and 70% as non-syndromic. Consanguineous marriage is a tradition which is commonly practiced among Asian, African and Latin American communities. At present, about 20% of world population live in communities with a preference for consanguineous marriage, which includes South India. Parental Consanguinity is highly associated with Autosomal recessive inheritance. There are 25% and 12.5% of gene sharing in second and third degree consanguinity respectively.(9)

Aim: To compare the incidence of auditory involvement among neonates born to consanguineous and non-consanguineous parents.

Objectives: 1. To compare the auditory involvement in neonates born to consanguineous and non-consanguineous parents. 2. To determine the association of the degree of consanguinity of parents on auditory function of neonate. 3. To detect permanent hearing impairment at the earliest possible time.

Materials and Methods:

Study Design: Prospective Observational Study.

Inclusion Criteria

Test Group - All neonates of consanguineous parents.

Control Group - Equal number of Neonates of non-consanguineous parents.

Exclusion Criteria: Children born with other congenital abnormalities like microtia, meatal stenosis, cleft palate, TORCHES CLAP, low birth weight, preterm babies, hyperbilurinemia requiring exchange transfusion, craniofacial abnormalities, syndromic illnesses, family history of deafness and exposure to ototoxic drugs were excluded from the study.

Materials and Methodology: The study was to compare the incidence of auditory involvement among neonates of consanguineous and non-consanguineous parents. The study sample selected by consecutive sampling within a specified period of time was subjected to transient evoked otoacoustic emissions (TEOAE) testing as the first level of hearing screening within 24-48 hours of birth. Neonates who failed the first level of hearing screening were subjected to a second level of hearing screening after 2-4 weeks by performing 2nd

TEOAE test. Brainstem evoked response audiometry (BERA) was performed in all those who failed the 2nd OAE testing, to confirm the hearing loss at the age of 2-4 weeks.

Results: In our study, the incidence of hearing impairment was 2.31% (n=3/130) and the incidence of hearing impairment in consanguineous parents was 4.62% (n=3/65). The incidence of hearing impairment in neonates on BERA evaluation was 2.31% (n=3/130), and all of them were born to consanguineous parents. There was no incidence of hearing loss in neonates born to non-consanguineous parents.

Conclusion: Although there was an association between hearing impairment and neonates born to consanguineous parents, it was not statistically significant. This might probably be due to the small sample size. Studies with larger sample size may be required to reveal the true magnitude of association between hearing impairment and consanguinity.

Keywords: Consanguinity, Transient evoked Otoacoustic emission, Brainstem evoked response audiometry, non-consanguinity

Introduction

Hearing is the deepest and most humanizing philosophical sense man possesses. 360 million people worldwide, suffer from disabling hearing loss. In India, 63 million people (6.3%) suffer from significant auditory loss. Four in every 1000 children suffer from severe to profound hearing loss. The estimated prevalence of childhood onset deafness is 2%, of which 50% of cases are thought to be inherited, i.e., of genetic/hereditary causes. Of these hereditary cases, approximately 30% are classified as syndromic and 70% as non-syndromic. About 400 named syndromes are associated with hearing loss, the associated hearing loss being quite variable – sensorineural or conductive, unilateral or bilateral, and progressive or stable. Among hereditary deafness, autosomal recessive inheritance predominates accounting for 75-80% of the cases followed by autosomal dominant inheritance in about 15-20% of the cases and X-linked and mitochondrial modes of inheritance in less than 1% of the cases.

Consanguineous marriage is a tradition which is commonly practiced among Asian, African and Latin American communities. At present, about 20% of world population live in communities with a preference for consanguineous marriage, which includes South India. Parental Consanguinity is highly associated with Autosomal recessive inheritance. There are 25% and 12.5% of gene sharing in second and third degree consanguinity respectively.[9] Hence, the current study was planned to determine the incidence of auditory involvement among neonates born to consanguineous parents.

Aim

To compare the incidence of auditory involvement among neonates born to consanguineous and non-consanguineous parents.

Objectives

1. To compare the auditory involvement in neonates born to consanguineous and non-consanguineous parents.
2. To determine the association of the degree of consanguinity of parents on auditory function of neonate
3. To detect permanent hearing impairment at the earliest possible time.

Materials And Methods

Study Design: Prospective Observational Study.

Study Period: November 2020 to October 2022

Inclusion Criteria

Test Group - All neonates of consanguineous parents.

Control Group - Equal number of Neonates of non-consanguineous parents.

Exclusion Criteria

Children born with other congenital abnormalities like microtia, meatal stenosis, cleft palate, TORCHES CLAP, low birth weight, preterm babies, hyperbilurinemia requiring exchange transfusion, craniofacial abnormalities, syndromic illnesses,

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family history of deafness and exposure to ototoxic drugs were excluded from the study.

Materials And Methodology

After obtaining Institutional Ethical committee approval and informed consent, study was conducted at Rajah Muthiah Medical College Hospital in the Departments of ENT and Department of Obstetrics & Gynaecology. The aim of the study was to compare the incidence of auditory involvement among neonates born to consanguineous and non-consanguineous parents. Each participant's demographic details was taken, and ear, nose and throat examination was done. Details were filled in the participant's proforma. After performing the otoscopy to rule out the blockage of external auditory canal by amniotic fluid, the study sample were subjected to Transient evoked Otoacoustic emission testing using a Madsen Accu Screen OAE & ABR Screener, which is a completely automated analysis system that gives a "PASS" or "REFER" result. Brainstem evoked response audiometry (BERA) was performed in all those who failed in 2nd OAE testing, to confirm the hearing loss at the age of 2-4 weeks.

Results And Observation:

Table: 1 Analysis of gender distribution

Gender	Neonates born to consanguineous parents		Neonates born to non-consanguineous parents		Total	
	n	%	N	%	n	%
Male	34	52.3%	36	55.4%	70	53.8%
Female	31	47.7%	29	44.6%	60	46.2%
Total	65	100%	65	100%	130	100%

Table: 2 Findings of Oto-acoustic emission testing (after 24 hours of birth) –screening 1.

OAE – I	Neonates born to Consanguineous parents		Neonates born to Non-Consanguineous parents	
	n	%	n	%
Pass	39	60%	45	69.2%
Refer	**26**	**40%**	**20**	**30.8%**
Total(n=130)	65	100%	65	100%

n- Number, % - Percentage

About 60% (n=39) of the neonates born to consanguineous parents and 69.2% (n=45) of the neonates born to non-consanguineous parents had passed the OAE-1st screening test. About 40% (n=26) of the neonates born to consanguineous parents and 30.8% (n=20) of the neonates born to non-consanguineous parents were referred for OAE-2nd screening.

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Table: 3 Findings of Oto-acoustic emission testing (at 2-4 weeks) – Screening 2.

OAE – 2	**Neonates born to Consanguineous parents**		**Neonates born to Non-Consanguineous parents**	
	n	**%**	**n**	**%**
Pass	17	65.4%	16	80%
Refer	**9**	**34.6%**	**4**	**20%**
Total (n=46)	26	100%	20	100%

n- Number, % - Percentage

About 65.4% (n=17) of the neonates born to consanguineous parents and 80% (n=16) of the neonates born to non-consanguineous parents had passed the OAE-2nd screening test. About 34.6% (n=9) of the neonates born to consanguineous parents and 20% (n=4) of the neonates born to non-consanguineous parents were referred for BERA.

Table: 4 Findings of brainstem evoked response audiometry (at 2-4 weeks of birth)

BERA	**Neonates born to Consanguineous parents**		**Neonates born to Non-Consanguineous parents**	
	n	**%**	**n**	**%**
Normal	6	66.7%	4	100%
Hearing Impairment	**3**	**33.3%**	**0**	**0**
Total (n=13)	9	100%	4	100%

n- Number, %- Percentage

Out of 13 neonates who underwent BERA testing, 66.7% (n=6) of neonates born to consanguineous parents had normal hearing and only 33.3% (n=3) of neonates born to consanguineous parents had hearing impairment. All three neonates with hearing impairment were females born to consanguineous parents. All the neonates born to Non-consanguineous parents had normal hearing, 100% (n=4).

Table: 5 Findings of BERA with degree of consanguinity

Degree of consanguinity	**BERA**				**Total n=9**	
	Normal		**Hearing impairment**			
	N	**%**	**n**	**%**	**n**	**%**
2nd degree	4	66.7%	**2**	33.3%	6	67%
3rd degree	2	66.7%	**1**	33.3%	3	33%

It is inferred from the above table that there were equal proportions (33.3%) of abnormality in BERA for neonates born to 2nd degree and 3rd degree consanguineous parents.

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Table: 6 Association of OAE screening with neonates born to consanguineous and non – consanguineous parents.

	Total	Pass		Refer		Chi – square (x2)	
		n	%	n	%	Value	P
Neonates born to consanguineous parents	65	56	87.2%	**9**	13.8%	2.13	0.144
Neonates born to non-consanguineous parents	65	61	93.8%	**4**	6.2%		
Total	130	117	90%	**13**	10%		

n- Number, % - Percentage, P –Probability

The association was not statistically significant, $X^2 = 2.13$, P=0.144(> 0.05).

Table: 7 Association of degree of consanguinity with OAE screening

Degree of consanguinity	Total	Pass		Refer		Chi – square (x2)	
		N	%	n	%	Value	P
2nd degree	23	17	73.9%	**6**	26.1%	4.471	**0.034***
3rd degree	42	39	92.9%	**3**	7.1%		
Total	65	56	86.2%	**9**	13.8%		

n- Number, % - Percentage, P –Probability, * - Significant

The association was statistically significant, $X^2 = 4.47$, P=0.034(< 0.05).

Table: 8 Results of neonates who were referred from OAE-2nd screening to BERA

Neonates who were referred from OAE-2nd screening to BERA	BERA				Total	
	Normal (False negative on OAE)		Hearing impairment (True positive on OAE)		n	%
n	n	%	n	%	13	100
13	10	76.9%	3	23.1%		

n- Number, % - Percentage

OAE screening had significant false negative rate of 76.9% and true positive rate of 23.1% when confirmed with BERA evaluation.

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Table: 9 Association of BERA with consanguinity

	Total	BERA				X2	
		Normal		Hearing impairment		Value	P
		n	%	n	%		
Neonates born to Consanguineous parents	9	6	66.7	3	33.3	1.73	0.188
Neonates born to Non-Consanguineous parents	4	4	100	-	-		
Total	13	100	66.7	3	33.3		

n- Number, % - Percentage, X^2 – Chi-Square, P –Probability,

The association was statistically not significant X^2 =1.73, P=0.188< 0.05.

Discussion

1) Association of Gender and auditory involvement

In our study, all 70 male neonates were found to have normal hearing status. Of the 60 female neonates, 57 neonates had normal hearing and 3 female neonates had hearing impairment. All 3 female neonates with hearing impairment were born to consanguineous parents. In a study conducted by Shakeel A et al (2018)**[(1)],** out of 13 neonates with hearing impairment, 7 were males and 6 were females. In our study, incidence of hearing impairment was only present in female gender. However, there were no statistical differences with respect to gender and hearing loss in all the above studies.

2) Otoacoustic emission

a) Incidence of hearing impairment in Oto-acoustic emission screening:

In our study, in the **OAE-1st screening (after 24 hours of birth)** out of 130 neonates, 84 (64.6%) had pass result and 46 (35.4%) had refer result.

On **OAE-2nd screening (at 2-4 weeks)**, Out of 46 neonates referred from OAE-1st screening, 33 (71.7%) had pass result and 13 (28.3%) had refer result.

In a study conducted by Shakeel A et al (2018)**[(1)]** Out of 1000 newborns screened, 17 newborns found to be positive on OAE-1st screening. On OAE-2nd screening which was performed after 1 week was found to be positive for 14 neonates.

b) Association of OAE with consanguinity :

On OAE-1st screening, 60% (n=39) of the neonates born to consanguineous parents and 69.2% (n=45) of the neonates born to non-consanguineous parents had passed the test. About 40% (n=26) of the neonates born to consanguineous parents and 30.8% (n=20) of the neonates born to non-consanguineous parents were referred for OAE 2nd screening.

On OAE-2nd screening, 65.4% (n=17) of the neonates born to consanguineous parents and 80% (n=16) of the neonates born to non-consanguineous parents had passed the test. About 34.6% (n=9) of the neonates born to consanguineous parents and 20% (n=4) of the neonates born to non-consanguineous parents were referred for BERA evaluation.

In a study done by Gonca Sennaroglu, Pelin Pistav Akmese[(3)], the parents of the 36 babies who were referred, 15 (41.7%) were born to consanguineous parents.

c) Association of OAE screening and Degree of Consanguinity:

In our study, in OAE screening there was significantly higher refer rate in neonates born to 2rd degree consanguineous parents 26.1%(n=6), when compared to neonates born to third degree consanguineous parents 7.1% (n=3). The association was statistically significant, X^2 = 4.47, P=0.034(< 0.05).In a study done by Gonca Sennaroglu, Pelin Pistav Akmese[(3)], in the parents of the 36 babies who

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were referred, 13 (36.1%) were first degree relatives and 2 (5.6%) were second degree relatives.

3) BERA:

a) Incidence of hearing impairment in BERA

In our study, 13 neonates who were referred from OAE-2nd screening were subjected to BERA. On BERA evaluation, 10 (76.9%) neonates had normal hearing and 3 (23.1%) neonates had hearing impairment. In a study conducted by Shakeel A et al (2018)**[(1)],** 14 neonates who were referred from OAE-2nd screening underwent BERA. 13 neonates had hearing impairment and 1 neonate had normal hearing.

b) Association of BERA on consanguinity

13 neonates who were referred from OAE-2nd screening underwent BERA. Of the 9 neonates born to consanguineous parents, 66.7% (n=6) had normal hearing and 33.3% (n=3) of had hearing impairment. All the neonates 100% (n=4) born to Non-Consanguineous parents had normal hearing. X^2 =1.73, P=0.188 (>0.05). Although this association was not statistically significant, there is a positive association between consanguinity and hearing impairment.

In a study conducted by Shakeel A et al (2018)**[(1)],** out of the 14 neonates who were referred for BERA, 13 neonates had hearing loss. Of which 8 neonates were born to consanguineous parents and 5 neonates were born to non-consanguineous parents.

C) Association of BERA and Degree of Consanguinity:

In our study, there were 65 neonates born to consanguineous parents. Out of 9 neonates who underwent BERA, 3 neonates had hearing impairment. Among them 2(67%) neonates were born to second degree consanguineous parents and 1 (33%) neonate was born to third degree consanguineous parents. In a study done by Bhagya V, Anshul Sharma (2021)[(8)], out of 763 hearing impaired children born to consanguineous parents, 543(71.2%) were born from second degree consanguineous marriage and 220 (28.8%) were born from third degree consanguineous marriage.

Both OAE and BERA showed increased refer rate and hearing impairment respectively in neonates born to second degree consanguineous parents as compared to neonates born to third degree consanguineous parents. This may be due to parental consanguinity which is highly associated with autosomal recessive inheritance.[(4)]

Genetics and Hearing impairment[(9)]:

Consanguinity has contributed significantly to the identification of mutated genes associated with Hearing Loss. Autosomal recessive forms of isolated deafness are higher than the world average in the Indian and Pakistani populations due to a preponderance of consanguineous marriage, caste barriers in marital relations, and assortative mating, whereas autosomal dominant, X-linked, and mitochondrial inheritance of deafness are less prevalent(Bhalla et al., 2009)[(10)].

According to National Deaf Children's Society-Position statement stated that 'Sometimes a gene mutation is a one-off that simply occurred by chance. This means that, although the hearing loss has a genetic cause, it has happened for the first time in that person and has not been inherited. However, it could be passed onto future generations.

Summary

1. In our study, the incidence of hearing impairment was 2.31% (n=3/130) and the incidence of hearing impairment in consanguineous parents was 4.62% (n=3/65).
2. The incidence of hearing impairment in neonates on BERA evaluation was 2.31% (n=3/130), and all of them were born to consanguineous parents. There was no incidence of hearing loss in neonates born to non-consanguineous parents. Although this association was not statistically significant, there is a positive association between consanguinity and hearing impairment.
3. In our study, among the neonates with hearing impairment who underwent BERA evaluation, 66.7% belonged to second degree consanguinity and 33.3% belong to third degree consanguinity. Therefore, there is an increased association between hearing impairment and degree of consanguinity.
4. In our study, there were 70 (53.8%) male neonates and 60 (46.2%) female neonates. On BERA evaluation, all 3 hearing impaired neonates were females born to consanguineous

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parents. There is no statistical significance between gender and hearing impairment.

5. OAE screening had significant false negative rate of 76.9% and true positive rate of 23.1% when confirmed with BERA evaluation.

Conclusion

Although there was an association between hearing impairment and neonates born to consanguineous parents, it was not statistically significant. This might probably be due to the small sample size. Studies with larger sample size may be required to reveal the true magnitude of association between hearing impairment and consanguinity.

Awareness needs to be created in society about the effects of consanguinity and hearing impairment. It is appropriate to create further awareness about genetic counselling and genetic testing to further curb the incidence of hearing impairment in neonates. Non-invasive screening tests like OAE and confirmatory test like BERA will go a long way to diagnose the hearing impairment in neonates at the earliest. Early rehabilitation is important for communication development, educational achievement and to ensure better quality of life.

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