Original Article

Establishing Ancestry through Pedigree of a Village with High Prevalence of Hearing-impaired

Sunil Kumar Raina, Munish Saroch[1], Geetika Yadav[2], Ashok Bhardwaj

Departments of Community Medicine and [1]Otolaryngology, Dr. RPGMC, Kangra, Himachal Pradesh, [2]Division of NCD, Indian Council of Medical Research, New Delhi, India

Abstract

Introduction: Population-based surveys into the cause of deafness have consistently shown that a high percentage (50%) of childhood deafness can be attributed to genetic causes. The flip side to genetic testing in population surveys is the operational difficulties associated with carrying it out. The present study was therefore planned with the aim of establishing ancestry through pedigree of a village with high prevalence of hearing-impaired. **Materials and Methods:** A two-stage study design was used to conduct this study. The first stage involved a house-to-house survey to identify hearing-impaired whereas the second stage involved clinical examination for hearing impairment. **Results:** Of a total of 2522 individuals studied, 80 were identified as hearing-impaired yielding a crude prevalence of 3.17%. The pedigree analysis of the whole village revealed a common ancestry. **Discussion:** In the late 20th century, deaf-mutism became a subject of debate and social isolation for Dadhkai villagers. A highlight of Dadhkai has been that its surroundings are not deaf-friendly as is expected. Consequently, as intermarriage flourished, the village community increasingly started resembling each other. This could have led to increase in autosomal recessive inheritance of deafness. Similar studies such as evaluation of six patients from two generations from a large sibship of Turkish ethnicity with double consanguinity in the family support the evidence. **Conclusion:** Flourishing of intermarriage and thereby consanguinity may be the reason behind high number of hearing impaired in this village. Therefore population based genetic counseling may be the key to prevent the same in future.

Keywords: Ancestry, hearing-impaired, pedigree, village

Introduction

Population-based surveys into the cause of deafness have consistently shown that about 50% of childhood deafness can be attributed to genetic causes.[1] The surveys have also pointed out that they may not be identified in a considerable proportion of individuals.[1] Molecular work in this regard has identified genetic causes as common. However, there is a fair degree of heterogeny in genetics as a cause.

Proven genetic testing techniques allow for accurate identification of genetic cause. It also allows for conducting counseling of the deaf and help family planning. Other important use in carrier testing among relatives provides essential information about environmental risk factors.[2] However, the flip side to genetic testing in population surveys is the operational difficulties associated with carrying it out. The present study was therefore planned with the aim of establishing ancestry through pedigree of a village with high prevalence of hearing-impaired.

Access this article online

Quick Response Code:

Website: www.jisha.org

DOI: 10.4103/jisha.JISHA_15_16

Materials and Methods

Study area

The study was conducted in Dhadkai village of Doda district of Jammu and Kashmir. District Doda is about 160 km from Jammu city (the capital city of Jammu and Kashmir state) situated in the middle and outer Himalayas. The district has a hilly terrain with area of 8912 km^2 and of population of 409,936. Doda district situated at an elevation of 1107 M (3632 ft) is the third largest district in Jammu and Kashmir in terms of area and falls geographically between 32° 53' and 34° 21' North latitude and 75° 1' and 76° 47' East longitude. It is administratively divided into four Tehsils (administrative units): Doda tehsil, Bhadarwah tehsil, Thathri tehsil, and Gandoh tehsil.

Address for correspondence: Dr. Sunil Kumar Raina, Department of Community Medicine, Dr. RPGMC, Tanda, Kangra, Himachal Pradesh, India.
E-mail: ojasrainasunil@yahoo.co.in

How to cite this article: Raina SK, Saroch M, Yadav G, Bhardwaj A. Establishing ancestry through pedigree of a village with high prevalence of hearing-impaired. J Indian Speech Language Hearing Assoc 2017;31:1-4.

Study population

The study was conducted on 2522 individuals of Dhadkai village (often called "The Village of Silence") of tehsil Gandoh of district Doda of Jammu And Kashmir.[3] Dhadkai is about 70 km from Doda city and had a land area of <3 square miles.

It is located over a hillock connected to Mainland by a footbridge. There is no road link to the village. The village is mainly inhabited by schedule tribe community of Muslim religion, referred to locally as Gujjars. Gujjars are transhumant agropastoralists.

Transhumant agropastoralism is the seasonal migration of livestock and humans from one agro-ecological zone to the other and back, from an established permanent home base.

Study design

The study was conducted in two phases. Phase 1 included visit to the homes of all residents (2522) of Dadhkai village of district Doda by a team of field investigators. No one refused to participate in the study. The team of field investigators visited all individuals residing in Dadhkai and individuals migrated to Plains of Punjab (as part of their migration). During the visits by the investigators, all individuals were administered a screening instrument (structured questionnaire) specifically developed to identify hearing-impaired individuals. In case of children < 10 years, the screening instrument was administered in front of a parent or a guardian. The entire questionnaire for the study was filled after taking written consent of participant from the individual or from his/her parent/guardian.

The screening instrument was field tested to make it culturally and linguistically acceptable to the local population. The instrument captured details on:

1. Hearing impairment among all the residents of village Dadhkai
2. Sociodemographic profile of all families screened.

The field investigators were all locals, well versed with sociogeography of the study area. Being locals, the field investigators were well versed with local sign language. For the purpose of this study, they were trained in administration of the questionnaire and in identifying individuals suspected for hearing-impaired. The training was carried out for 3 weeks in a tertiary care center under the supervision of an otolaryngologist (MS) and two public health specialists (SKR and AKB).

As part of phase 2, all individuals identified as deaf on screening questionnaire were examined by a clinical team comprising an ENT specialist and an audiologist. The clinical examination included a (1) physical examination for possible causes of hearing loss, such as earwax or inflammation from an infection, (2) general screening tests, (3) tuning fork tests, and (4) audiometer tests. The audiometer test included pure tone audiometry to identify hearing threshold levels, enable determination of the degree, type, and configuration of a hearing loss. The clinical examination was carried out to confirm hearing impairment.

Further, the village is covered under the disability pension scheme, and the village Sarpanch (headman) has a record of all hearing-impaired individuals. The records were obtained from the Sarpanch to account for any case missed during the conduct of the study.

Pedigree formation

Pedigree history of the entire village was prepared with their consent and after ensuring confidentially. This was done to locate the ancestry of affected (deaf) cases in the families and in understanding the disease inheritance pattern. This also helped us in defining consanguinity/social relationships between the family members. For the preparation of pedigree, we approached members of different families and initiated them into developing an understanding about deafness prevalent in the village and the role of pedigree development. During pedigree formation, we went up to four to seven generations back depending on the knowledge of their ancestry. In this exercise, the information obtained from one member of the family was cross-checked with other members of family and from those who were associated with that particular family either through ancestry or from marital link.

This ensured accuracy in formation of their pedigree chart. In this way, the pedigree of the whole village was formed and then linked up. The family ancestries were further linked in families who shared common ancestry.

Results

Of a total of 2522 individuals studied, 80 were identified as hearing-impaired yielding a crude prevalence of 3.17%. The youngest male and female hearing-impaired cases were of 1 and 3 years old, respectively. The oldest male and female hearing-impaired cases were of 55 and 72 years old, respectively.

The prevalence of hearing impairment was highest among the younger age group of <15 years with 48 (61%) individuals belonging to this age group.

The pedigree analysis of families of 47 hearing-impaired individuals has been reproduced in the current manuscript. The first family in Figure 1a had eight deaf members in the present (V) generation of which five were females. Their parental generation (IV) had two consanguineous marriages, and seven members of the generation were hearing-impaired. The four members of the grandparent generation (III) were hearing-impaired with no consanguineous marriages. The generations preceding this (I and II) reported no consanguineous marriage and no hearing-impaired member. In the second family [Figure 1b], the third generation had nine hearing-impaired members of which five were male. The preceding parental generation (II) had three hearing-impaired members all of whom are dead and one consanguineous marriage. In the third family [Figure 1c], out of nine children in the third generation, eight cases were hearing-impaired and one hearing-impaired member was in the second generation

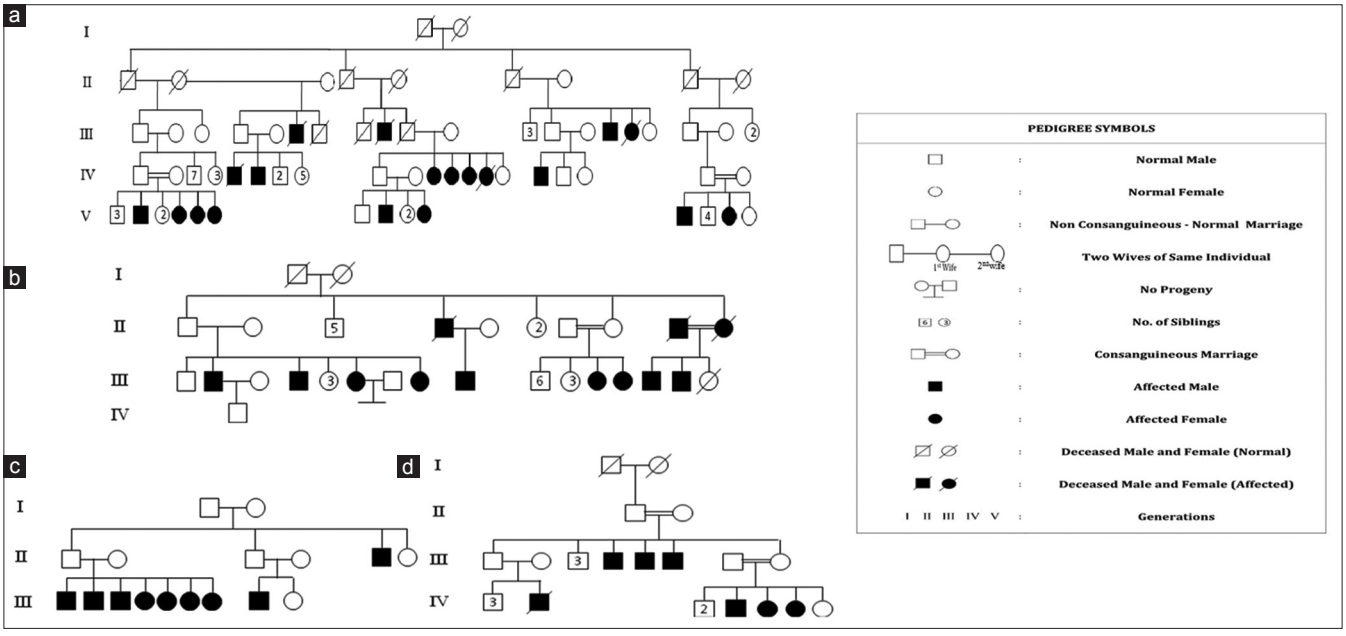

Figure 1: Pedigree diagram of four families (a to d) of Dhadkai village

with no history of consanguineous marriage. The fourth family [Figure 1d] reported four hearing-impaired children in the current generation (IV), of which one had died and three hearing-impaired were present in the preceding generation (III). History of consanguineous marriage was present in both second and third generation.

The pedigree analysis of Dhadkai population shows that the deafness runs in skip generations, i.e., in an autosomal recessive way. There is a history of consanguineous marriages between the couples who were related to each other either from paternal or maternal side.

Discussion

Hearing impairment is broadly categorized into syndromic or nonsyndromic. Syndromic cases make up the minority of all inherited deafness but diagnosed more accurately due to additional features of the syndromes.

Nonsyndromic types are classified on the basis of mode of inheritance. This population showed deafness in skip generation in an autosomal recessive way. Both the partners must have been carriers in order for a child to have deafness; a child who inherits the gene from one parent will be a carrier. Abnormal genetic makeup accounts for approximately 50% of permanent childhood deafness.[4] In the late 20th century, deaf-mutism became a subject of debate and social isolation for Dadhkai villagers. Dadhkai sign language is commonly used by hearing residents as well as deaf ones till date. This has allowed the deaf residents to smoothly integrate into society. A highlight of Dadhkai has been that its surroundings are not deaf-friendly as is expected. Consequently, as intermarriage flourished, the village community increasingly started resembling each other. This could have led to increase in autosomal recessive inheritance of deafness. Similar studies such as evaluation of six patients from two generations from a large sibship of Turkish ethnicity with double consanguinity in the family support the evidence.[5] Shafique *et al*. tested a panel of thirty unrelated consanguineous Pakistani families for autosomal recessive inheritance pattern, where 60% of congenital hearing impairment is attributed to consanguineous marriages and suggested genetic counseling of families to inform couples about the risk of their offspring to be hearing-impaired.[6] The probable inheritance pattern of autosomal recessive has also been discussed a large inbred Brazilian pedigree.[7]

The purpose of any medical research is to apply the findings to clinical use. The prerequisite for treating deafness will certainly be early and accurate diagnosis. Groups such as Dadhkai village with strong family histories of deafness with any related syndromes and premature newborns should be targeted for screening in the first instance. Molecular techniques can scan for genetic defect at several levels.[8] Early diagnosis of hearing impairment in children would be essential if medical and surgical interventions were to stand a good chance of preserving some hearing ability.

However, the attitude of patients and their families and their views on such intervention must be respected. The deaf community has its own culture, and there has been anecdotal evidence that deaf couples do not necessarily want their children to be able to hear if it means that they will be excluded from the deaf community to which their parents belong.[9] Still understanding genetic etiology can provide valuable clues of prognosis (i.e., whether losses will worsen), optimal intervention (e.g., hearing aids, cochlear implantation, and sign language), and the risk of hearing loss recurrence in future children and other family members.[10-13]

Conclusion

Intermarriage and resulting consanguinity may be the reason behind high number of hearing impaired in this village. Population based genetic counseling may be the key to prevent the same in future.

Limitation

It is important to identify the genetic makeup or the mutated genes involved. Without the genetic test, it is inappropriate to comment on the cause of the disease.

Financial support and sponsorship

The study has been funded by a research grant from the Indian Council of Medical Research.

Conflicts of interest

There are no conflicts of interest.

References

1. Morton NE. Genetic epidemiology of hearing impairment. Ann N Y Acad Sci 1991;630:16-31.
2. Bitner-Glindzicz M. Hereditary deafness and phenotyping in humans. Br Med Bull 2002;63:73-94.
3. Razdan S, Raina SK, Pandita KK, Razdan S, Nanda R, Kaul R, *et al.* Inbreeding as a cause for deafness: Dadhkai study. Indian J Hum Genet 2012;18:71-4.
4. Marazita ML, Ploughman LM, Rawlings B, Remington E, Arnos KS, Nance WE. Genetic epidemiological studies of early-onset deafness in the U.S. school-age population. Am J Med Genet 1993;46:486-91.
5. Morava E, Kühnisch J, Drijvers JM, Robben JH, Cremers C, van Setten P, *et al.* Autosomal recessive mental retardation, deafness, ankylosis, and mild hypophosphatemia associated with a novel ANKH mutation in a consanguineous family. J Clin Endocrinol Metab 2011;96:E189-98.
6. Shafique S, Siddiqi S, Schraders M, Oostrik J, Ayub H, Bilal A, *et al.* Genetic spectrum of autosomal recessive non-syndromic hearing loss in Pakistani families. PLoS One 2014;9:e100146.
7. Lezirovitz K, Pardono E, de Mello Auricchio MT, de Carvalho E Silva FL, Lopes JJ, Abreu-Silva RS, *et al.* Unexpected genetic heterogeneity in a large consanguineous Brazilian pedigree presenting deafness. Eur J Hum Genet 2008;16:89-96.
8. Smith SD, Kimberling WJ, Schaefer GB, Horton MB, Tinley ST. Medical genetic evaluation for the etiology of hearing loss in children. J Commun Disord 1998;31:371-889.
9. Pau H. Molecular otology, development of the auditory system and recent advances in genetic manipulation. In: Gleeson M, editor. Scott Brown's Otorhinolaryngology, Head and Neck Surgery. 7th ed. Great Britain: Edward Arnold; 2008. p. 811-20.
10. Wu CC, Lee YC, Chen PJ, Hsu CJ. Predominance of genetic diagnosis and imaging results as predictors in determining the speech perception performance outcome after cochlear implantation in children. Arch Pediatr Adolesc Med 2008;162:269-76.
11. Kabatova Z, Profant M, Simkova L, Groma M, Nechojdomova D. Cochlear implantation in malformed inner ear. Bratisl Lek Listy 2009;110:609-13.
12. Black J, Hickson L, Black B, Perry C. Prognostic indicators in paediatric cochlear implant surgery: A systematic literature review. Cochlear Implants Int 2011;12:67-93.
13. Brown KK, Rehm HL. Molecular diagnosis of hearing loss. Curr Protoc Hum Genet 2012. DOI: 10.1002/0471142905.hg0916s72