A RARE CASE REPORT OF OCULOFACIAL DENTAL ANOMALIES IN PEDIATRIC AGE GROUP; NANCE-HOREN SYNDROME

SHUB NOOR SAEED, RABIA NAJAM, WAQAS, AMIR RASHID
Department of Pediatrics, Postgraduate Medical Institute / Ameer ud Din Medical College / Lahore General Hospital, Lahore

ABSTRACT
Nance-Horen Syndrome, a triad of characteristic facial, dental and ocular abnormalities along with intellectual impairment is a rare X linked genetic disorder[1-4]. Diagnosis is mainly based on clinical association of characteristic facial, ocular and dental anomalies along with mental retardation. In this case report, we presented a patient with Nance-Horen syndrome, who presented in OPD with post cataract excision corneal opacities along with non-ocular anomalies and similar presentation in two other male siblings. Pediatrician in developing country should know the significance of detailed systemic assessment of patients presenting with oculofacial anomalies to avoid missing important diagnosis like Nance-Horen Syndrome.

Keywords: Hutchinson teeth, Nance -Horen syndrome, Cataract dental syndrome.

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Correspondence to: Shub Noor Saeed,
Department of Pediatric, Postgraduate Medical institute/ Ameer-ud-Din Medical College/Lahore General Hospital, Lahore, Pakistan.

Email: shubnoor419@gmail.com

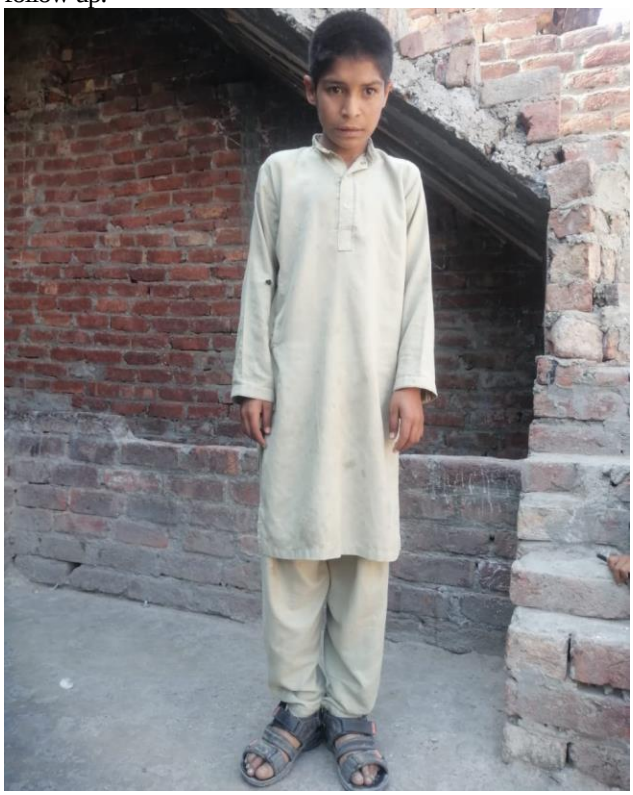
INTRODUCTION
Nance-Horen Syndrome first described by Nance and Horen in 1974 in different studies. It is an extremely rare x linked genetic disorder which has been reported in Caucasians, Chinese, Arabian, Indian, Turkish and Taunasian ethnic populations, however the accurate prevalence is not known[5-7] as many affected individuals go unrecognized or undiagnosed. As X linked genetic disorder with semi dominant transmission, the disease presents as ocular and non ocular features with variable expression in affected males while female carries shows mild or no symptoms at all[8]. NHS gene is located on Xp22.13. the protein product is a novel regulator of cell morphology and actin assembly and is expressed abundantly in brain, orofacial mesenchyme, lens and primordial teeth. To date ,33 different mutations have been reported[9-10]. One of such cases presented with clinical manifestations of Nance Horen syndrome in 3 affected male siblings of a family, while two female siblings have no clinical features at all.

CASE REPORT
In this study, we characterized the clinical features of Nance Horen Syndrome in 3 affected male siblings of a family while female siblings were unaffected. The youngest male sibling of the family aged 10 months, presented with post cataract excision corneal opacities. The other affected male siblings were of age 4 years and 10 years. All the affected males have history of bilateral congenital cataract, poor vision and nystagmus. Medical history revealed surgical enucleation of congenital cataract and intraocular lens implant in all 3 siblings, however postsurgical corneal opacities were found in 2 siblings. At the age of 6 years, the eldest sibling had glaucoma surgery for which both medical treatment and surgical treatment was done, however the patient still had

raised intraocular pressure of 45mmHg and he has best corrected visual acuity ranging from no light perception to 0.05. Other 2 male siblings have mildly raised intraocular pressure.

Extra ocular findings include dental malocclusion, hutchinson incisors, mulberry molars and poor dental hygiene. One of the siblings had increase number of folds in the ears and eldest sibling had an elongated face. Developmental delay and intellectual impairment was found in all 3 siblings with different severity. however, no skeletal abnormality was noted.

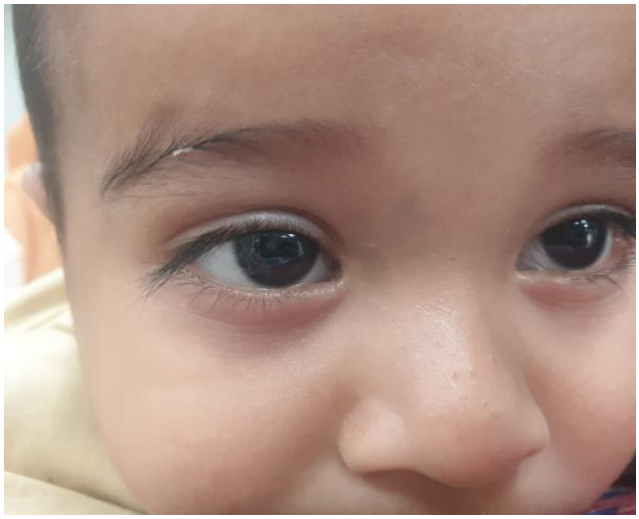
The history, clinical features and radiological findings is this case were consistent with diagnosis of Nance Horen Syndrome. Congenital syphilis was ruled out due to absence of other features of the disease apart from hutchinson incisors and mulberry molars as well as with negative serological report. However, genetic testing, which is diagnostic was not available in our country. Management was planned according to a multidisciplinary approach of a team of ophthalmologists, orthodontists, general pediatrician and developmental experts and it was directed towards specific problems. Short term treatment plan involved excision of corneal opacities, correction of refractive error, preventive resin restoration and root canal. Long term treatment plan consisted of education in a special institute for ocular and intellectual impairment and counseling regarding follow up.



Eldest sibling with an elongated face and decrease IQ level

Middle sibling with post cataract surgery lens implant

Hutchinson teeth


post cataract extraction corneal opacities


Hutinsons incisors


Unaffected female siblings

DISCUSSION

Congenital cataract is leading cause of blindness in childhood, and 50% of these are inherited which may be a part of multisystem disease.one of such genetic diseases is Nance Horen Syndrome which is an x linked genetic disorder comprising of ocular and dental anomalies, facial dysmorphism and intellectual impairment, hence the name cataract dental syndrome or Mesioden- Cataract Syndrome[8,11].

Typical ocular NHS features include Bilateral congenital cataract, nystagmus, visual impairment, strabismus, microcornea and glaucoma. In 89% of cases ocular abnormalities require surgery and visual prognosis remain poor. Facial dysmorphism is in form of long face, anteverted nares, mandibular prognathism, large ears and increase number of folds in ears. Dental anomalies include hutchinson incisors, supernumeries, dental agenesis, lotus flowers or mulberry molars, abnormal calcification of pulp and incisor fractures. Intellectual impairment, low IQ and developmental delay is found in one third of the patients with autistic spectrum disorder features reported in one of the study[2,14]. As the disease has variable phenotypical manifestations even in siblings of same family, so a detailed and careful examination of facial, dental, ocular and neurological system is required for accurate diagnosis. In one of the study, y sutural posterior lens opacities and microcornea with dental anomalies were found in 100% of female carrier[12,13]. Intellectual impairment rarely occurs in females.

Early prenatal diagnosis can be achieved in high-risk families and post natal genetic testing is also available. A multidisciplinary approach including clinical and molecular genecists, dentists, maxillofacial surgeons, ophthalmologists and neurologists is required for management[14,16]. Management is directed toward treating the subjective problems of the patients and regular long term follow up.

CONCLUSION

Orofacial and ocular anomalies can lead to diagnosis of inherited and congenital syndromes. This case demonstrates the role of oral health professionals and ophthalmologists in diagnosing rare genetic syndrome by carefully evaluating the child for non ocular manifestations with the help of pediatricians. This case report not only highlighted a rare diagnosis of Nance Horen Syndrome but also lead to the inference of different phenotypical presentations of disease.

DECLARATION OF PATIENT CONSENT

The authors of the study certify that informed written consent was obtained from the parents prior to the case report regarding sharing of his/ her children's images and clinical reports. However, it was reassured that names of

patients would not be published and identity would be kept concealed.

ETHICAL APPROVAL

The study was approved by the Ethical Review Committee of Postgraduate Medical Institute / Ameer-ud-Din Medical College/Lahore General hospital, Lahore via Research No. 00-112-21 Dated: April 27, 2021.

REFERENCES

1. Subasioglu A, Savas S, Kucukyilmaz E, Kesim S, Yagci A, Dundar M. Genetic background of supernumerary teeth. Eur J Dent. 2015;9(1):153–158.
2. Bixler D, Higgins M, Hartsfield Jr J. The Nance-Horan syndrome: A rare Xlinked ocular-dental trait with expression in heterozygous females. Clin Genet. 1984;26:30–35.
3. Nance EW, Warburg N, Bixler D, Helveston EM. Congenital X-linked cataracts, dental anomalies and brachymetacarpalia. Birth Defects Orig Artic Ser X. 1974;4:285–295.
4. HORAN BM, Billson FA. X-linked cataract and hutchinsonian teeth. Aust Paediatr J. 1974;10:98–102.
5. Hong N, Chen YH, Xie C, Xu BS, Huang H, Li X, et al. Identification of a novel mutation in a Chinese family with Nance-Horan syndrome by whole exome sequencing. J Zhejiang Univ Sci B. 2014;15(8):727–734.
6. Li A, Li B, Wu L, Yang L, Chen N, Ma Z. Identification of a novel NHS mutation in a Chinese family with Nance-Horan Syndrome. Curr Eye Res. 2015;40(4):434–438.
7. Ramprasad VL, Thool A, Murugan S, Nancarrow D, Vyas P, Rao SK, et al. Truncating mutation in the NHS gene: phenotypic heterogeneity of Nance-Horan syndrome in an asian Indian family. Invest Ophthalmol Vis Sci. 2005;46(1):17–23.
8. Nance-Horan Syndrome. National Organization for Rare Disorders (NORD). 2014; http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/1055/viewAbstract. Accessed 9/15/2015.
9. Nance-Horan syndrome. Orphanet. September 2007; http://www.orpha.net/consor/cgi-bin/ Disease_Search.php?lng=EN&data_id=931. Accessed 9/15/2015.
10. Nance-Horan syndrome. OMIM. December 5, 2015; http://omim.org/entry/302350. Accessed 9/15/2015.
11. Horan M, Billson F. X-linked cataract and hutchinsonian teeth. Aust Paediatr J. 1974;10:98–102. [Google Scholar]
12. Nance-Horan Syndrome. National Organization for Rare Disorders (NORD) 2011. [Last accessed on 2017 Dec 22]. Available from: https://www.rarediseases.org/rare-diseases/nance-horan-syndrome/
13. Toutain A Nance-Horan syndrome. NORD Guide to Rare Disorders. Philadelphia, PA: Lippincott Williams & Wilkins; 2003. pp. 654–655. [Google Scholar]
14. 14.Toutain A. Nance horan syndrome. Orphanet Encyclopedia. 2003:1–4. [Google Scholar]
15. Toutain A, Ayrault AD, Moraine C. Mental retardation in Nance-Horan syndrome: Clinical and neuropsychological assessment in four families. Am J Med Genet. 1997;71:305–314. [PubMed] [Google Scholar]
16. Reches A, Yaron Y, Burdon K, Crystal-Shalit O, Kidron D, Malcov M, et al. Prenatal detection of congenital bilateral cataract leading to the diagnosis of Nance-Horan syndrome in the extended family. Prenat Diagn. 2007;27:662–664. [PubMed]

AUTHOR'S CONTRIBUTIONS

SNS: Manuscript Writing, Introduction, Discussion
RN: References
WA: Case writing
AR: Proof reading