doi:10.5368/aedj.2010.2.4.199-203.pdf

EFFECTS OF CONSANGUINEOUS MARRIAGES ON ORAL AND CRANIOFACIAL STRUCTURES: A STUDY ON DENTAL PATIENTS IN NORTH INDIA

[1] Lakshmayya Naidu D — [1] Professor, Department of Orthodontics and Dentofacial Orthopedics, Kotiwal Dental College and Research centre , Moradabad – Uttar Pradesh

[2] Srinivasa Raju M — [2] Professor *and Head,* Department of Oral medicine and Radiology, Teerthankar Dental College and Research centre, Moradabad– Uttar Pradesh

[3] Sumit Goel — [3] Senior Lecturer, Department of Oral medicine and Radiology, I.D.S.T Dental College, Modinagar– Uttar Pradesh

ABSTRACT

Background: Consanguinity describes a relationship between two people who share an ancestor, or share blood. These consanguineous marriages are a major risk to the health of offsprings till the extent that they can cause various craniofacial abnormalities, orofacial pigmentations and other abnormal birth defects. We conducted a study on patients visiting a dental college in North India to accesses the possible deleterious effects of consanguineous marriages on oral and craniofacial structures.

Methodology: The study group comprised of all the patients visiting outpatient department of Kotiwal Dental college and Research centre, Moradabad in North India from March 2009 to February 2010. 200 patients showed positive history of consanguinity. They were carefully evaluated and all the diagnostic information was noted.

Results: 66 out of 200 patients had systemic disorders like cardiovascular, pulmonary and renal disturbances. 56 patients had orofacial manifestations like orofacial pigmentations, craniofacial syndromes and occlusal abnormalities with or without systemic disorders.

Conclusion:The prevalence of consanguineous marriages is still high. Consanguineous marriages are a major risk to the health of offsprings till the extent that they can cause various craniofacial abnormalities, orofacial pigmentations and other abnormal birth defects. They increase the autosomal recessive conditions through the expression of recessive deleterious alleles, especially in the offspring of first degree cousins.

***KEY WORDS*: Consanguineous marriages, Oral and Craniofacial defects.**

INTRODUCTION

We are all connected to life. Every choice we make and every belief we hold exerts influence upon the whole of our life. And we live with the consequences of our choice. The human population has seen modern civilization and is still within family boundaries. One such familial-social bond is consanguineous marriage. Consanguinity means descent from a common ancestor; a consanguineous couple is usually defined as being related as second cousins or closer. The word derives from 'con'+ 'sanguine' – from the Latin, meaning 'of the same blood'[1,2] .

Around the globe consanguineous marriages have been practiced by many societies from time immemorial. Such marriages are favored by different populations usually bound to traditional customs, beliefs and to keep property in united form within the family. These are widely practiced in Asia, North Africa, Switzerland, Middle East, some parts of China, Japan and fishermen communities in Europe and America. Consanguineous marriage is widely favored in a large majority of the world's Islamic populations. One in two rural marriages in Tamil Nadu and Andhra Pradesh are consanguineous[1,2].

Hindus in India as a practice outlaw the consanguineous marriage by avoiding the same `gothra' or patrilineal relationship between the

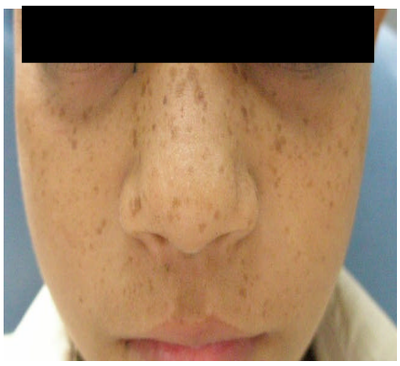

Fig.1. **Centrofacial lentinogenesis**

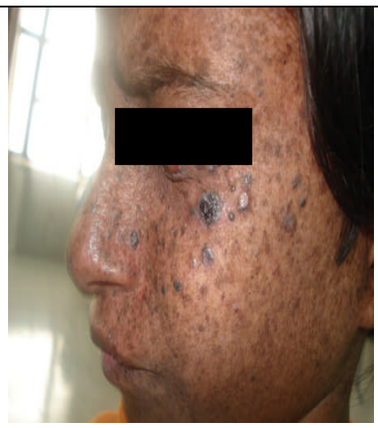

Fig.2.**Xeroderma pigmentosum**

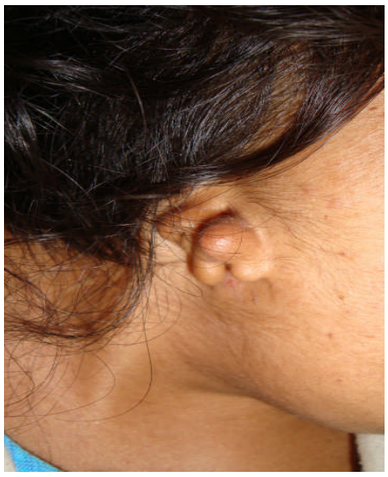

Fig.3. Nager syndrome with underdeveloped ear

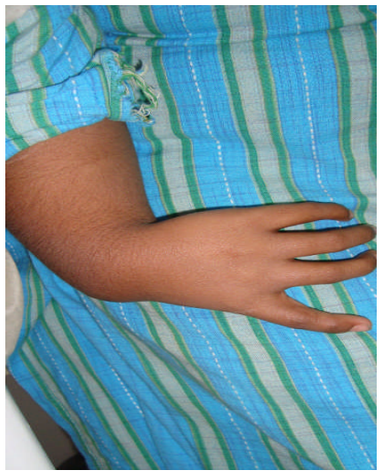

Fig.4. Nager syndrome with malformed hands

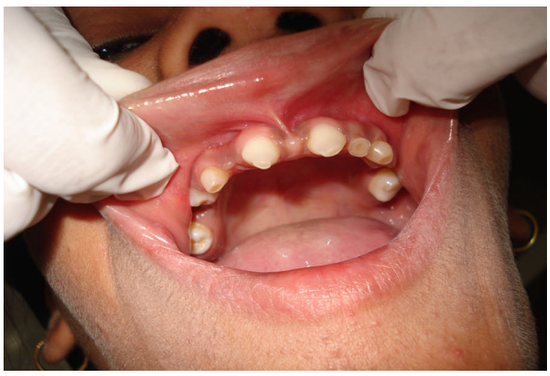

Fig.5.Nonsyndromic oligodontia

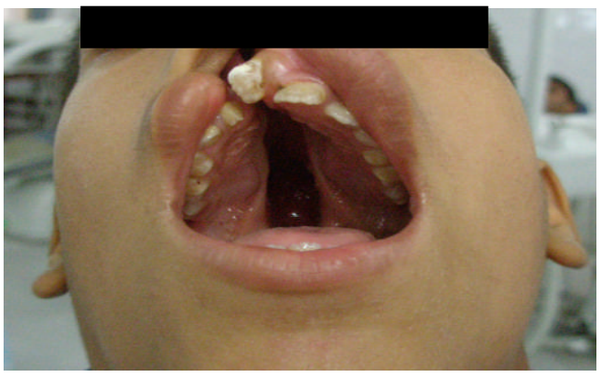

Fig.6. Cleft lip and palate.

probable bride and the groom. In some of the western countries including the United States, consanguinity closer than the first cousins are considered to be legally incest[3].

While assessing the consequences of consanguineous against non-consanguineous (non-blood related) marriages in health and disease, several scientific studies have shown that consanguinity leads to death of infants before, during or immediately after birth. There are increased incidences of birth defects including hydrocephalus, postaxial hand polydactyly, bilateral cleft lip and/or cleft palate, bipolar disorders, depression, reproductive disorders, sterility, infant mortality, child deaths, spontaneous abortions and stillbirths etc. Also there are reports indicating positive association between consanguinity and Down syndrome, also ventricular septal defect (VSD), atrial septal defect (ASD), atrioventricular septal defect (AVSD), pulmonary stenosis (PS) and pulmonary atresia (PA) , genetic diseases including blinding disorders, blood cancer (acute lymphocytic leukemia), breathing problems for children at birth (apnea), increased susceptibility to disease, blood diseases (hemophilia, α-thalassemia), cystic fibrosis, Chronic renal failure and Neonatal diabetes mellitus[1,2,3].

Studies accessing the consequences of these marriages on oral and craniofacial region are extremely rare. Therefore this study was conducted in a dental college of Northern India to evaluate the various oral and craniofacial abnormalities which can develop due to consanguinity.

Methodology

The study group comprised of all the patients visiting outpatient Department of Kotiwal Dental College and Research centre, Moradabad from March 2009 to February 2010. A special patient record sheet was formulated to record detailed patient's history, including family history of consanguinity, clinical findings and all necessary diagnostic investigations. The patients with positive history of consanguinity were carefully evaluated and all the diagnostic information was noted.

Results

Out of the examined, 200 patients showed a positive family history of consanguinity out of all the patients visiting the dental college. The data analysed showed that the prevalence of consanguineous marriages is still high. Muslims had a higher frequency than Hindus. Majority of consanguineous marriages were identified between first cousins. A 66 out of 200 patients had systemic disorders like cardiovascular, pulmonary and renal disturbances. A 56 patients had orofacial manifestations like orofacial pigmentations, craniofacial syndromes and occlusal abnormalities with or without systemic disorders. We recorded 3 siblings suffering from Xeroderma pigmentosum, 3 siblings having centrofacial lentinogenesis, 16 patients with craniofacial syndromes, 15 patients with malocclusion, 15 patients had cleft lip and palate and 4 patients had miscellaneous pathologies. **(Table 1)**

Table.1. showing various oral and craniofacial abnormalities identified in patients with positive family history of consanguineous marriages

Disorders	***Number of patients***
PIGMENTED LESIONS	
Centrofaial lentinogenesis	3 Siblings
Xeroderma pigmentosum	3 Siblings
CRANIOFACIAL SYNDROMES	
Mobius syndrome	1
Nagers syndrome	1
Hemifacial microsomia	3
Pierre Robins syndrome	3
Ectodermal dysplasia	6
Down syndrome	2
OTHERS	
Malocclusions	15
Non syndromic oligodontia	2
Enamel hypoplasia	2
Cleft lip / cleft palate	15

Discussion

Consanguineous marriages are major responsible risk factors for Bipolar disorders. It increases the autosomal recessive conditions through the expression of recessive deleterious alleles, especially in the offspring of first degree cousins. The population risk for any couple of having a child with a serious or lethal medical condition is around 2% (1 in 50). The excess risk for a couple who are

related as first cousins, in the absence of a known genetic disease in the family, is in the order of 3% (1 in 30). This fact often comes as a relief to couples who expect a significantly higher figure. The excess risk is as a result of autosomal recessive conditions arising due to homozygosity by descent – that is, the risk of a recessive mutation present in an ancestor being passed down 2 branches of the family, and coming together in the consanguineous marriage. It is thought that we all carry at least one mutated allele which would cause an autosomal recessive condition if present in two copies (homozygousity). If this mutant allele is passed down to both members of a consanguineous couple from a shared ancestor they will both be carriers for this condition, and will therefore have a 1 in 4 chance of having an affected child.[1,2,4,5]

The chance of both parents being carriers for a recessive condition is determined by how closely they are related, which means that the offspring risks can be minimised while retaining the social benefits of consanguinity if marriages occur between more distant relatives (e.g. second rather than first cousins).[4,5]

We diagnosed several cases as a consequence to consanguinity during our study period (Table 1) that included various craniofacial syndromes like Moebius syndrome, Nager syndrome, Pierre Robin syndrome; skin diseases like ectodermal dysplasia, oral and perioral pigmentations as well as other facial and dental abnormalities. Few of the important cases are described below

Centrofacial lentinogenesis; Centrofacial lentino-genesis is a pigmented disorder in which lentigenes (freckle like pigments which do not fade away even in winters) are distributed on the face around the nasal bridge and the lips. This pigmentation increases with age. The lentigenes appear as tanned macules measuring from 0.5 mm to 1 cm in diameter **(Fig.1).**

Xeroderma pigmentosum: Xeroderma pigmen - tosum is a rare disorder transmitted in an autosomal recessive manner and is characterised by pigmentary changes, photophobia, premature skin aging and malignant tumor development **(Fig.2)**.

Mobius syndrome: Moebius syndrome is a rare congenital disturbance which is described in most studies as congenital facial weakness combined with abnormal ocular abduction. Dysfunctions of other cranial nerves, orofacial malformations are commonly associated features but they are not necessary for the diagnosis making the syndrome extremely variable in its clinical manifestations. Nerves commonly affected in this syndrome are facial (in all cases), abducent (75% cases), hypoglossal (20% cases) and occulomotor (5% cases). Most cases are diagnosed during infancy. Facial and ocular symptoms are usually the presenting problems.

Nager syndrome: It is a rare craniofacial disorder involving underdevelopment of facial structures along with underdeveloped or missing thumb **(Fig.3 and Fig.4)**.

Hemifacial microsomia: It is the second most common birth defect after clefts and refers to underdevelopment of one side of the face along with maldevelopment of the ear.

Ectodermal dysplasia: Ectodermal dysplasia is a term used to describe a group of rare, inherited disorder characterized by dysplasia of tissues of ectodermal origin- primarily nails, teeth and skin and occasionally dysplasia of mesodermally described tissues. There is a classical triad of hypodontia, hypohydrosis and hypotrichosis.

Others: nonsyndromic oligodontia (**Fig.5),** mentally retarded child having enamel hypoplasia along with malocclusion, cleft lip and cleft palate. (**Fig.6)** were also noted

Genetic counseling

Genetic counseling is the process by which clients or their relatives, at risk of an inherited disorder, are advised of the consequences and nature of the disorder, the probabilities of developing or transmitting it, and the choices open to them in management and planning of their families, in an attempt to prevent, avoid or ameliorate the disorder. This has preventive, diagnostic, therapeutic and supportive value. Genetic counseling yields best results when done premarital or at least prior to

conception. A non-judgmental attitude towards consanguineous couples is essential on the part of the counselor, to establish good communication channels and to foster effective working relationships between the medical profession and communities where consanguineous marriages are prevalent [6-8].

CONCLUSION

Thus it is seen from previous studies and our case reports that consanguineous marriages are a major risk to the health of offsprings till the extent that they can cause various craniofacial abnormalities, orofacial pigmentations and other abnormal birth defects. They increase the autosomal recessive conditions through the expression of recessive deleterious alleles, especially in the offspring of first degree cousins. India needs to take a big leap in this direction as consanguineous marriages are more prevalent in our country. Adopting better translational research concept and intervention strategies help consanguineous couples reach informed and intelligent reproductive decisions, with which they have to live throughout their lives. The social benefits of consanguinity should not outweigh the biological damages; but it is sad that many in Indian community are ignorant about these facts.

References

1. Amudha S, Aruna N. Consanguinity and chromosomal abnormalities. Indian journal of human genetics 2005; 11: 108-110
2. Satyanarayana Rao TS. Consanguinity: still a challenge. Indian journal of psychiatry 2009; 51: 3-5
3. Brittles AH. Reproductive behaviour and health in consanguineous marriages. science 1991; 252: 789-91
4. Awadi AL Effects of consanguineous marriages on reproductive wastage. clinical genetics 2002; 24: 384-88
5. Hussain R. Sociodemographic correlates of consanguineous marriages in muslim population of India. J of biosocial science 2000; 32: 433-44
6. Brittles AH. Consanguinity and its relevance to clinical genetics. Clin Genet 2001; 60:89-98.
7. Nalini A, Gayathri N, Yasha TC, Ravishankar S, Urtizberea A, Huehne K, et al . Clinical, pathological and molecular findings in two siblings with giant axonal neuropathy (GAN): Report from India. Eur J Med Genet 2008; 51:426-35.
8. Puri RK, Khanna KK, Bhargava I, Balakrishnan S. Role of Consanguinity in Chromosomal syndromes. Indian J Med Res 1977; 65:859-64.

. **Corresponding Author**

Dr.Lakshmayya Naidu D
Professor of Orthodontics,
Kotiwal Dental College & research centre
Kanth Road,Moramustaqueem,
Moradabad – Uttar Pradesh
E mail:drdlnaidu@rediffmail.com