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Prevalence of Orofacial Defects Due to Familial Aggregation, at Tertiary Care Centre in Chennai- A Cross Sectional Study

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Abstract

Both cleft lip and cleft palate are orofacial birth defects. A cleft lip may be just a small notch in the lip. It may also be a complete split in the lip that goes all the way to the base of the nose.A cleft palate can be on one or both sides of the roof of the mouth. It may go the full length of the palate .Though orofacial defects, such as cleft lip and cleft palate, are corrected by surgical methods, they do leave behind marks of their occurrence. This causes various complexity problems for patients in society. By doing this study, we will come to know incidence of orofacial defects by familial aggregation due to consanguineous marriages, i.e. marriages within relations. This can be further classified as first degree consanguineous marriage, that is marriage between immediate relations (eg. between brother and sister), then second degree consanguineous marriages, i.e. marriages between first cousins and so on. In this study, we will also come to know the extent of occurrence of these defects in family, i.e. we check if patient's siblings are also suffering from any kind of these defects. The motive of this study is to create public awareness in mind of people about consanguineous marriages and their effects in form of familial aggregation. Further, this study can be carried out by

checking the incidence of these orofacial defects in future generations and checking out rate of incidence of these defects when the patients married within relation and outside the relations. Moreover, by creating public awareness related to this issue, we can prevent future generations from these defects and prevent various kinds of complexity problems.

Keywords- *Orofacial Defects, Familial Aggregation, Consanguineous Marriages, Chennai*

INTRODUCTION

Cleft lip and cleft palate, which can also occur together as cleft lip and palate, are variations of a type of clefting congenital deformity caused by abnormal facial development during gestation. A cleft is a fissure or opening—a gap. It is the non-fusion of the body's natural structures that form before birth. Clefts can also affect other parts of the face, such as the eyes, ears, nose, cheeks, and forehead. In 1976, Paul Tessier described fifteen lines of cleft. Most of these craniofacial clefts are even rarer and are frequently described as Tessier clefts using the numerical locator devised by Tessier.[1] A cleft lip or palate can be successfully treated with surgery, especially so if conducted soon after birth or in early childhood.

If the cleft does not affect the palate structure of the mouth it is referred to as cleft lip. Cleft lip is formed in the top of the lip as either a small gap or an indentation in the lip or it continues into the nose. Lip cleft can occur as unilateral or bilateral. It is due to the failure of fusion of the maxillary and medial nasal processes during formation of the primary palate.Cleft palate is a condition in which the two plates of the skull that form the hard palate are not completely joined. Cleft palate occurs in about one in 700 live births worldwide.[2] Palate cleft can occur as complete or incomplete. When cleft palate occurs, the uvula is usually split. It occurs due to the failure of fusion of the lateral palatine processes, the nasal septum, and/or the median palatine processes during formation of the secondary palate.

Most children who have their clefts repaired early enough are able to have a happy youth and social life. Having a cleft lip or cleft palate does not inevitably lead to a psychosocial problem. However, adolescents with cleft lip or cleft palate are at an elevated risk for developing psychosocial problems related to self-concept, peer relationships and appearance. Though surgically corrected cleft lip or cleft palate may impact an individual's self-esteem, social skills and behavior. Also it is seen that self-concept may be adversely affected by the presence of a cleft lip or cleft palate, particularly among girls.[3]

Genetic factors contributing to cleft lip and cleft palate formation have been identified for some syndromic cases (such as Stickler's Syndrome can cause cleft lip and palate, joint pain, and myopia[4]

) but knowledge about genetic factors that contribute to the more common isolated cases of cleft lip or cleft palate is still patchy. Many clefts run in families, even though in some cases there does not seem to be an identifiable syndrome present,[5] possibly because of the current incomplete genetic understanding of midfacial development.

Environmental influences may also cause, or interact with genes to produce cleft lip and cleft palate. In humans, fetal cleft lip and other congenital abnormalities have also been linked to maternal hypoxia, as caused by few conditions like maternal smoking,[6] maternal alcohol abuse or some forms of maternal hypertension treatment.[7] Other environmental factors that have been studied include: maternal diet and vitamin intake, retinoids, anticonvulsant drugs, alcohol, cigarette use, nitrate compounds; organic solvents; parental exposure to lead; and illegal drugs (cocaine, crack cocaine, heroin, etc.).

Prevalence rates reported for live births for cleft lip with or without cleft palate and cleft palate alone varies within different ethnic groups. It caused about 4,000 deaths globally in 2010 down from 8,400 in 1990.[8] The highest prevalence rates for cleft lip and cleft palate are reported for Native Americans and Asians. Africans have the lowest prevalence rates.[9] Prevalence of "cleft uvula" has varied from .02% to 18.8% with the highest numbers found among Chippewa and Navajo and the lowest generally in Africans.[10][11]

Cleft lip and cleft palate may be complex, multifactorial, or polygenic, meaning they are likely associated with the effects of multiple genes in combination with lifestyles and environmental factors. Although these orofacial defects often cluster in families, they do not have a clear-cut pattern of inheritance. This makes it difficult to determine a person's risk of inheriting or passing on these disorders. In case of consanguineous marriages chances of inheriting affected genes increase than in normal marriages because there are high chances that both parents have defective gene in them, as they belong to same family and chances of passing them on to future generations increases.

So by doing this study, we have come to know incidence of orofacial defects caused by familial aggregation due to consanguineous marriages. We have also come to know the extent of occurrence of cleft lip and cleft palate in family, by checking whether patient's siblings suffered from similar defects.

The motive behind this study was to create public awareness about ill-effects of consanguineous marriages in form of cleft lip and palate, so that the patients suffering from these defects do not face any complexity problems in society even after surgical correction at young age.

AIMS AND OBJECTIVES

1. To study the prevalence of orofacial at tertiary care centre in Chennai

2. To find out prevalence of orofacial defects due to familial aggregation.
3. To find out if patient's siblings are affected by any orofacial defect.

MATERIAL AND METHODS

Sixty-three patients with cleft lip, cleft palate or both cleft lip and cleft palate, who had already undergone correction surgery, were detected through a survey in Tertiary Care Centre, in Chennai, Tamil Nadu, India and cross-sectional study was done between June 2013 and July 2013. Patients themselves or their parents (in case of minor patients) were the subjects for this study. Subjects who have been treated for cleft lip and cleft palate only (or both) in any Tertiary Care Centre in Chennai only were included in study. Subjects with orofacial defects other than cleft lip and cleft palate and subjects who had their treatment done in places other than Chennai were excluded from study.

A semi-structured and pre-coded questionnaire was used to collect data. Demographic details such as gender and other objective oriented details such as type of orofacial defect, extent of orofacial defect, occurrence of similar defects in parents and siblings and past history of orofacial defects in family were collected. After explaining the nature and procedure of the study to the candidate, informed consent was obtained from the participant.

The Fischer's Exact Test was used to determine differences between study and control groups. This test is employed mostly when sample size is smaller. Using this method deviation from null hypothesis was calculated.

A two sided study with sample proportion of 11% (0.11) and power 80% was done and alpha error was considered to be 5% for analysis of result from the data collected.

OBSERVATIONS AND RESULTS

Table 1: Descriptive data for the sample

Gender		
	Female	40
	Male	23
Parents Affected		
	Yes	9
	No	54
Defect Type		
	Cleft Lip	17
	Cleft Palate	16

	Both	30
Marriage Type		
	Consanguineous	35
	Non-consanguineous	28
Siblings Affected		
	Elder Sibling	6
	Younger Sibling	6
	Not Affected/No sibling	51

Out of 63 patients recruited, 40 were female and 23 male. 17 patients suffered from isolated cleft lip, 16 from isolated cleft palate, and 30 had both cleft lip and palate. 12 patients had either an elder or younger sibling affected the same condition. Summary of descriptive data is provided in Table 1.

On cross tabulation between type of marriage (consanguineous versus non-consanguineous) and the type of defect, 35 of the patients reported with consanguineous marriage. The data however was not statistically significant (P= 0.178). Table 2 provides a summary of the result

Table 2: Cross tabulation of marriage type versus defect

		Defect			
		Cleft Lip	Cleft Palate	Both	Total
Marriage	Consanguineous	9	12	14	35
	Non-consanguineous	8	4	16	28
	Total	17	16	30	63

On comparing the extent of sibling involvement, 12 out of the 63 patients had involvement of either an older or younger sibling (6 each). 6 of them had both cleft lip and cleft palate defect, 4 had isolated cleft lip and 2 had isolated cleft palate. The data was statistically insignificant (P= 0.710). Table 3 provides a summary of the result

Table 3: Siblings affected compared to defect type

		Defect			
		Cleft Lip	Cleft Palate	Both	Total
Siblings affected	Affected	4	2	6	12
	Not affected	13	14	24	51
Total		17	16	30	63

DISCUSSION

In this study we saw that mostly females were more affected with orofacial defects than males which were same as findings of study in Mississippi [23] and against findings of studies carried out in North Ireland [24] and South East of Scotland [25]. As this study was achieved only through questionnaire passed to patients, its results depend upon availability of affected patients. Mostly, they were females.

Since the lip and primary palate have distinct developmental origins from the secondary palate, clefts of these areas can be subdivided into cleft lip with or without cleft palate and isolated cleft palate in which the lip is not affected. We found that isolated cleft lip or isolated cleft palate had low incidence when compared with both cleft lip and cleft palate. This was similar to findings of studies by John C. Greene et Al [26] but in their studies mostly they concluded occurrence of isolated cleft palate more than cleft lip [25 26 27]. In my study report isolated cleft palate prevailed least and both cleft lip and cleft palate had maximum prevalence.

Marriages within relations i.e. consanguineous marriages are still prevalent in some places. In our study we enquired patients if their parents were relatives. Our results showed that familial aggregation due to consanguinity was also one of major cause of orofacial defects. This was similar to findings of studies by Sabbhag et Al[29] and Ravichandran K et Al[30] . But in most of studies considered importance was not given to consanguinity as factor responsible for orofacial defects.

When extent of these defects was seen in patient's siblings, we saw only in very few cases siblings were affected. From this we can see that when orofacial defects were passed on from one

generation to other, the defective genes responsible for defects are mostly suppressed and they become dominant in very few cases and cause these defects. But, we got a statistically insignificant data. But few studies like that by Lie RT.et Al[19] suggest that rate of recurrence of these orofacial defects increase by two times in siblings of affected persons.

CONCLUSIONS

This study was questionnaire based study, which mainly was dependent on patients having orofacial defects especially only cleft lip, cleft palate and both cleft lip and cleft palate. After completing this study, we could get appropriate conclusion for all our objectives.

We could conclude that females were more affected than males by these defects and when it came to recurrence of orofacial defects from previous generations we could conclude that in few cases it was inherited from previous generations. But, this study being a short term study does not give a scope to involve more generations in study due to its time constrain. This study can further be carried on by involving future generations of affected people and they can be checked if they gave birth to a child with orofacial defects. Also, we can check for children with orofacial defects if they were married within relations.

We also found out that in most of cases even if patient had siblings they were unaffected by any kind of orofacial defects. But, in cases of siblings being affected with orofacial defects, they mostly had same orofacial defect as that of patient.

Further, this study can be carried out by making it a prospective long term study, so that these patients who are already involved in study can be observed and we can check the rate with which recurrence of orofacial defects is there in patient's family.

SUMMARY

On basis of this study, we could mainly conclude that, in most of the cases, parents of affected person had consanguineous marriage. This study was mainly carried out to act as medium for creating public awareness in Chennai about disadvantages of consanguineous marriages i.e. marriages between relations. Also, on basis of oral enquiry, we came to know mostly these marriages take place for property reasons. So, we would like to conclude that consanguineous marriages are risky as they increase the chances of giving birth to child affected by any kind of congenital diseases, mostly orofacial defects. Though these defects are corrected surgically, they leave behind mark of their occurrence. This causes various complexity problems for the affected person. We came to know females were more prone to orofacial defects in region of Chennai. Many tertiary care centers were involved in this study. We could see that prevalence of both cleft lip and cleft palate was most and prevalence of isolated cleft palate was least. In cases where patients had siblings, mostly they were unaffected with any kind of orofacial defects. Even if patients were

affected, they also had same defect as that of patient.

This study overall provided conclusive result that mostly people who were married within relations gave birth to child having congenital birth defect, mostly orofacial defects. Therefore, prevalence of orofacial defects due to familial aggregation, at tertiary care centre in Chennai. A cross sectional study – was completed.

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