



## Assessment of role of consanguinity in formation of cleft lip and cleft palate

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### ABSTRACT

Consanguinity is considered a significant factor in autosomal recessive diseases; it has also been associated with congenital anomalies such as hydrocephalus, polydactylia and Cleft Lip and Palate deformities. The risk of congenital conditions is higher in subjects born of first degree consanguineous parents compared with those of non-consanguineous marriages. The aim of this study is to evaluate the prevalence of consanguinity with formation of cleft lip/ cleft palate formation in Tamil, Telugu population. This is a retrospective study. The details of 86,000 patient records were reviewed and analysed, out of which 76 patients who had undergone surgical treatment for cleft lip and cleft palate deformities between June 2019 to march 2020 were included in this study. The details like age, gender, family history and cleft diagnosis were evaluated and entered in SPSS and analysed through a chi-square test. It was observed that 11% of patients reported with history of consanguineous marriages which was associated with formation of cleft lip/palate. Within the limits of the study, it was concluded that consanguinity had a significant role in formation of cleft lip/cleft palate.



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### INTRODUCTION

The cleft is a fissure or opening occurring during the embryonic development due to the failure of parts to fuse. The clefts in the craniofacial region

or anatomical distortions of the cranium and face with deficiencies or excess of tissues which involves both hard and soft tissues which may also need various surgical treatments ranging from simple extraction of teeth which may lead to Dry socket as a common complication to an extensive procedure like maxilla mandibular orthognathic surgeries with osteotomies which may lead to lot of blood loss and in some rare cases, cleft patients may be associated with some rare carcinomas (Williams, 1998; Jesudasan *et al.*, 2015; Christabel *et al.*, 2016; Rahman, 2017b; Marimuthu *et al.*, 2018). The cleft results in disfigurement and damage to tissues. It affects psychological development and causes a financial burden to the family of the affected. The incidence of cleft in Indian subcontinent is approximately between 27,000 and 33,000 Cleft per year. (Mossey and Little, 2009; Kumar, 2017; Packiri, 2017). The

aetiology of clefting is multifactorial. It includes both environmental and genetic factors. (Murray, 1399; Patil, 2017). The role of genetic factors in aetiology of cleft lip/cleft palate is important and it is associated with 400 different syndromes. In India, consanguineous marriages are not uncommon due to prevailing cultural practises (Abhinav et al., 2019).

Consanguinity is an identified factor that increases risk of transmission of genetic disorders. Consanguinity is a cultural practice which could have an epigenetic role in the development of congenital abnormalities (Aljohar et al., 2008; Kumar and Sneha, 2016; Rahman, 2017a). Consanguineous marriages are a common social norm in many society worldwide where marriages are between close blood related persons varying from first cousins to uncle-niece relations (Rahman, 2017a). About 10.4% of couples in the global population are reported to be related as the second cousins or closer, a similar relationship holds for progeny couples (Bittles and Black, 2010b; Kumar and Sneha, 2016; Patturaja and Pradeep, 2016).

Consanguinity is commonly practised in Arab countries (50%) South India (10.5%), China, North India, Latin America, Japan, South Europe (10%) (Bittles and Black, 2010a) several studies report significant association between consanguinity and congenital anomalies (Kulkarni and Kurian, 1990; Mehndiratta et al., 2007; Jain et al., 2019). The objective of this study was to assess the association between consanguinity and formation of cleft lip and cleft palate.

## MATERIALS AND METHODS

### Study setting

This study is a university setting. Study conducted in Saveetha dental College., predominantly. The pros of the study include, flexibility of the study and less time consumption. The cons of the study include – it is limited to a certain population – 76. Patients who underwent cleft surgery were included for the study. Approval was obtained from the institutional ethical committee [IEC]. The ethical approval code is SDC/SIHEC/2020/DIASDATA/0619-0320. Two examiners were involved in the study.

### Sampling

It is a retrospective study. Data was collected from June 1, 2019 to March 31, 2020. Totally 86,000 casesheets were reviewed out of which 76 patients had undergone cleft surgery. Cross verification of data for error was done by the presence of an additional reviewer. Simple random sampling was done to minimize the sampling bias. Tamil and Telugu

population was included.

### Data collection/Tabulation

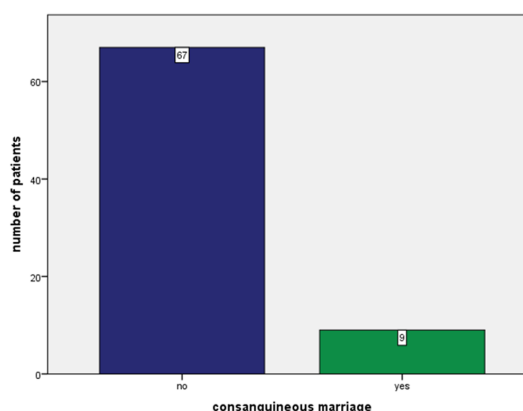
Data or family history of patients who had undergone cleft lip and cleft palate surgery was collected from initial to last in chronological order. Data verification was done based on the age and family history (consanguineous marriage). Data was entered in excel sheet in a methodical manner and was imported to SPSS. Incomplete or uncensored data was excluded from the study.

### Analytics

IBM SPSS 2.0 Software was used for data analysis. Independent variables include – age, gender and dependent variables include cleft anomaly, consanguineous marriage history. Descriptive and inferential statistics was used. Descriptive statistics include the frequency of distribution patients-age, gender and history of consanguineous marriage and inferential test includes chi-square test.

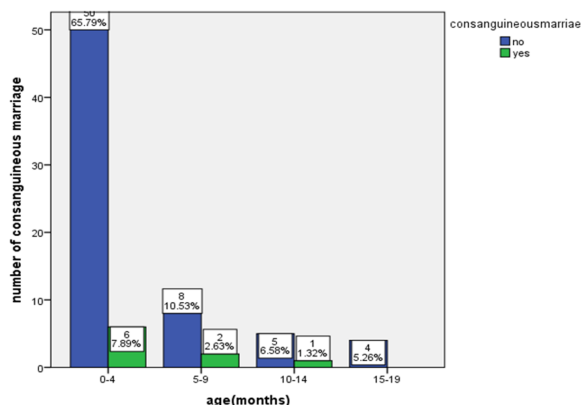
## RESULTS AND DISCUSSION

While observing frequency of consanguineous marriage, it was seen that out of 78 patients, 11.5% give a history of consanguineous marriage.(Figure 1). Blue bar depicts no consanguineous marriage and green bar represents presence of consanguineous marriage. From the graph it is evident that the incidence of consanguineous marriage among cleft patients reported in this study is very low [11.5%].

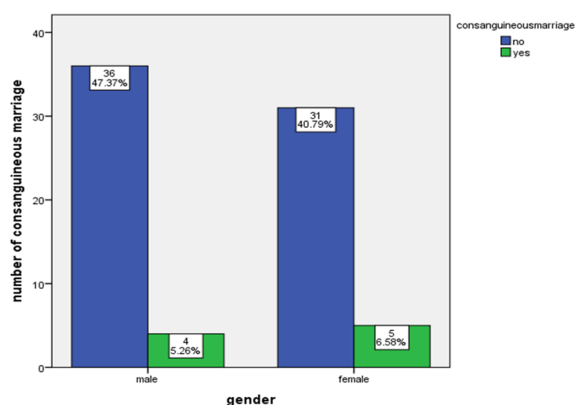


**Figure 1: Bar chart showing frequency of consanguineous marriages in children with cleft.**

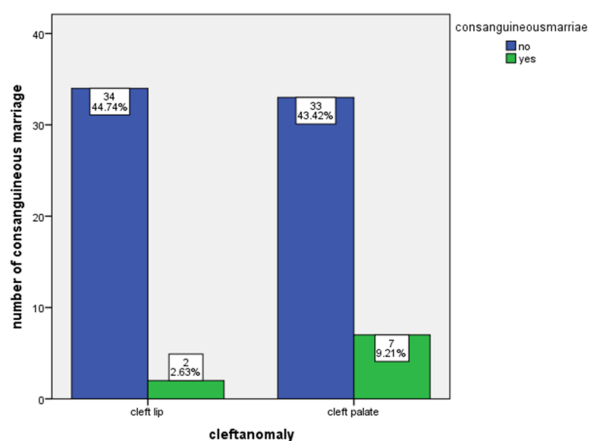
Coming to the age group, the maximum number of cleft anomalies with a history of consanguineous marriage is 0 to 4 months (7.89%)(Figure 2). X-axis represents age groups in months and Y-axis represents the number of consanguineous marriages. Chi square test shows no significant associa-



**Figure 2:** Bar chart represents the association of age of children with cleft anomaly.



**Figure 3:** Bar chart represents the association of gender of children with cleft anomaly.



**Figure 4:** Bar chart represents the association of consanguineous marriage with children with cleft anomaly.

tion, Pearson chi square- 1.377,  $p=0.711$  ( $p>0.05$ ) which is found to be statistically non significant. Even though, it is statistically not significant, the incidence of history of consanguineous marriages among cleft patients was found to be common among 0-4 years age group.

In the distribution of age of patients with cleft lip/cleft palate, it was seen that 0 to 4 months age group has maximum number of cleft patients (7.89%) followed by 4 to 9 months (2.63%). History of consanguineous marriage is found to be maximum in 0 to 4 months age group (7.89%).

(Figure 3) shows the gender predilection of patients with cleft lip and cleft palate shows that females (6.58%) were prevalent to consanguinity than male (5.26%) patients. X-axis represents gender and Y-axis represents the number of consanguineous marriages. Chi square test shows no significant association, Pearson chi square- 0.274,  $p=0.600$  ( $p>0.05$ ) which is statistically non-significant. Even though it is statistically not significant, the incidence of history of consanguineous marriages among cleft patients was found to be common among females.

From association of cleft anomaly with consanguineous marriage, it is observed that consanguinity was prevalent in cleft palate (9.21%) than cleft lip (2.63%) (Figure 4). X-axis represents gender and Y-axis represents the number of consanguineous marriages. Chi square test shows no significant association, Pearson chi square- 2.589,  $p=0.108$  ( $p>0.05$ ) which is statistically non significant. Even though, it is statistically not significant, the incidence of history of consanguineous marriages among cleft patients was found to be common among cleft palate patients than in cleft lip patients.

In this study, we observed that there was association between consanguineous marriage and formation of cleft lip and cleft palate. A consanguineous marriage is the marriage between blood relatives. These blood relatives have at least one common ancestor. In some parts of the world, consanguineous marriage is common. In India, the incidence of consanguineous marriage is between 5% and 60%. In the Indian subcontinent, uncle and niece marriages are commonly seen (Turnpenny and Ellard, 2007).

The present study is a study from the South Indian population as the cases were available only from Tamil and Telugu population from South India. Indian population has a heterogeneous origin, so the study population of the particular geographic area can be more relevant than the general population. The syndromic cases of clefts were excluded as it has a different aetiology.

In a Saudi Arabian study, to investigate the influence of consanguinity in nonsyndromic oro-facial cleft, demonstrated that consanguinity was higher in parents of Cleft palate cases than cleft lip patients (Alamoudi *et al.*, 2014). This finding was in accordance with the finding of the present study where 11.5% gave the history of consanguineous marriage.

This study differs from a study done in China where they could not show any association of consanguinity with birth defects (Zhang *et al.*, 2012).

The results would have been more relevant if each cleft group is compared with a matched control group which is the limitation of the study. The findings of the study can be used to create awareness on risk factors of consanguinity. This study has a future scope where different degrees of consanguinity can be studied and its association to cleft lip/palate.

## CONCLUSION

Within the limitations, this study shows that the consanguinity is moderately associated with occurrence of clefts. The awareness about consanguinity and occurrence of genetic abnormalities should reach the individuals to avoid the further complications.

## Conflict of Interest

The authors declare that they have no conflict of interest for this study.

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