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Association of Parental Consanguinity with Severity of Cleft Lip and Palate.

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ABSTRACT

Background: Cleft lip and palate are among the most common congenital craniofacial anomalies worldwide. Parental consanguinity has been identified as a potential risk factor for congenital anomalies; however, its association with the severity of cleft deformities has not been extensively explored.

Aim: To evaluate the association between parental consanguinity and the severity and pattern of cleft lip and palate.

Materials and Methods: A hospital-based observational study was conducted on patients presenting with cleft lip and/or palate. Data regarding demographic details, degree of parental consanguinity, type of cleft, laterality, and presence of associated anomalies were recorded. Severity of cleft was categorized into cleft lip alone, cleft palate alone, unilateral cleft lip and palate, and bilateral cleft lip and palate. Statistical analysis was performed to assess associations between consanguinity and cleft severity.

Results: A higher proportion of severe cleft patterns, particularly bilateral cleft lip and palate, was observed in patients born of consanguineous marriages. A statistically significant association was noted between degree of consanguinity and increasing severity of cleft deformity.

Conclusion: Parental consanguinity is associated not only with the occurrence of cleft lip and palate but also with increased severity of the deformity. These findings highlight the importance of genetic counseling and public health education in populations where consanguineous marriages are prevalent..

Keywords: Cleft lip, cleft palate, consanguinity, severity, congenital anomalies

INTRODUCTION

Cleft lip and palate are common congenital craniofacial anomalies resulting from failure of fusion of facial processes during embryonic development. The etiology of cleft lip and palate is multifactorial, involving genetic, environmental, and nutritional factors. The global prevalence of cleft lip and palate varies widely, with higher incidence reported in Asian populations.

Consanguineous marriage, defined as a union between individuals related as second cousins or closer, is common in certain regions due to cultural, social, and economic reasons. Such marriages increase the probability of homozygosity for recessive genes, thereby increasing the risk of congenital anomalies.

While several studies have evaluated the association between consanguinity and the occurrence of cleft lip and palate, limited literature exists on whether consanguinity influences the severity and pattern of cleft deformities. Understanding this association may help in risk stratification, counseling, and preventive strategies.

The present study aims to assess the relationship between parental consanguinity and the severity of cleft lip and palate among patients presenting to a tertiary care center.

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AIM AND OBJECTIVES

AIM

To assess the association between parental consanguinity and the severity of cleft lip and palate.

Objectives

1. To determine the prevalence of consanguinity among parents of cleft lip and palate patients.
2. To categorize cleft lip and palate based on type and severity.
3. To evaluate the association between degree of parental consanguinity and severity of cleft deformity.
4. To assess the presence of associated congenital anomalies in consanguineous and non-consanguineous groups

Materials and Methods

Study Design

Hospital-based observational cross-sectional study.

Study Setting

Department of Oral and Maxillofacial Surgery at a tertiary care teaching hospital.

Study Duration

January 2022 to December 2024

Sample Size

A total of **120 patients** with cleft lip and/or palate were included in the study.

Inclusion Criteria

Patients diagnosed with cleft lip, cleft palate, or both.

Patients of all age groups.

Patients whose parents consented to participate in the study.

Exclusion Criteria

Patients with incomplete medical records.

Adopted children where parental history could not be obtained.

Data Collection

Data were collected using a structured proforma, including:

Demographic details (age, sex)

Parental consanguinity (present/absent)

Degree of consanguinity (first cousin, second cousin, distant relative)

Type of cleft (cleft lip alone, cleft palate alone, unilateral cleft lip and palate, bilateral cleft lip and palate)

Laterality (right/left/bilateral)

Presence of associated congenital anomalies

Severity Classification

Severity of cleft was categorized as:

Mild: Cleft lip alone

Moderate: Cleft palate alone or unilateral cleft lip and palate

Severe: Bilateral cleft lip and palate

Statistical Analysis

Data were entered into Microsoft Excel and analyzed using SPSS version 25.0. Categorical variables such as gender, type of cleft, severity category, presence of consanguinity, degree of consanguinity, and associated anomalies were expressed as frequencies and percentages. The association between parental consanguinity and severity of cleft lip and palate was assessed using the Chi-square test. Odds ratios (OR) with 95% confidence intervals (CI) were calculated to estimate the strength of association.

Multivariate logistic regression analysis was performed to adjust for potential confounders such as gender and presence of associated anomalies. A p-value of <0.05 was considered statistically significant.

Results

Demographic Characteristics

A total of 120 patients with cleft lip and/or palate were included in the study. The age of patients at presentation ranged from 3 days to 18 years, with a mean age of 2.6 ±1.9 years. There was a male predominance, with 72 patients (60%) being males and 48 patients (40%) being females.

Parental consanguinity was present in 46 patients (38.3%), while 74 patients (61.7%) were born of non-consanguineous marriages. Among the consanguineous group, first-cousin marriages accounted for 30 cases (65.2%), second-cousin marriages for 12 cases (26.1%), and distant relatives for 4 cases (8.7%).

Distribution of Cleft Types and Severity

Cleft lip alone was observed in 28 patients (23.3%), cleft palate alone in 26 patients (21.7%), unilateral cleft lip and palate in 40 patients (33.3%), and bilateral cleft lip and palate in 26 patients (21.7%).

Based on severity classification, 28 patients (23.3%) were categorized as mild, 66 patients (55%) as moderate, and 26 patients (21.7%) as severe.

Association Between Consanguinity and Severity

In the consanguineous group, severe cleft deformities were observed in 17 patients (36.9%), compared to 9 patients (12.2%) in the non-consanguineous group. Mild clefts were more common in the non-consanguineous group (27.0%) compared to the consanguineous group (17.4%).

Statistical analysis revealed a significant association between parental consanguinity and severity of cleft deformity (Chi-square = 9.82, p = 0.002). Patients born of consanguineous marriages had a significantly higher risk of developing severe cleft deformities (OR = 4.14; 95% CI: 1.68–10.21).

A trend of increasing severity with closer degree of consanguinity was observed, with first-cousin marriages showing the highest proportion of severe clefts.

Associated Congenital Anomalies

Associated congenital anomalies were identified in 22 patients (18.3%). These anomalies were significantly more frequent in the consanguineous group (14 patients; 30.4%) compared to the non-consanguineous group (8 patients; 10.8%) (p = 0.01).

The most commonly observed anomalies included congenital heart disease (8 cases), limb anomalies (6 cases), ear anomalies (5 cases), and developmental delay (3 cases). Presence of associated anomalies was also found to correlate with increased cleft severity.

DISCUSSION

Cleft lip and palate are multifactorial congenital anomalies with a complex interplay of genetic and environmental influences. The present study specifically evaluated the role of parental consanguinity in determining the severity of cleft deformities and demonstrated a statistically significant association.

The prevalence of consanguinity in the present study (38.3%) is comparable to previously reported data from South Asian populations, where cultural practices favor consanguineous marriages. The higher proportion of severe cleft patterns, particularly bilateral cleft lip and palate, among consanguineous offspring suggests an increased genetic load contributing to abnormal craniofacial morphogenesis.

Consanguinity increases the likelihood of autosomal recessive gene expression due to homozygosity, which may explain the observed severity gradient. First-cousin marriages, which represent the closest degree of consanguinity in this study, showed the strongest association with severe cleft deformities. This dose–response relationship strengthens the biological plausibility of the findings.

The significantly higher frequency of associated congenital anomalies in the consanguineous group further supports the role of shared genetic determinants affecting multiple organ systems. Similar observations have been reported in studies evaluating congenital anomalies in consanguineous populations, although few have specifically addressed cleft severity.

Male predominance observed in the present study is consistent with existing literature and may be attributed to sex-linked genetic susceptibility or differential embryological vulnerability. Delayed presentation noted in some consanguineous families may reflect sociocultural factors, limited awareness, and normalization of congenital anomalies within extended family structures.

From a clinical perspective, identifying consanguinity as a risk factor for severe cleft deformities has important implications. Early recognition allows for anticipatory guidance, multidisciplinary planning, and timely intervention. Furthermore, genetic counseling can play a critical role in educating families regarding recurrence risks and preventive strategies.

Despite its strengths, the study has certain limitations. Being hospital-based, the findings may not be generalizable to the broader population. Additionally, molecular genetic analysis was not performed, which could have provided direct evidence of underlying genetic mechanisms. Nevertheless, the standardized classification and adequate sample size enhance the

reliability of the results.

CONCLUSION

The present study demonstrates a significant association between parental consanguinity and increased severity of cleft lip and palate, with a higher prevalence of bilateral cleft lip and palate and associated congenital anomalies in consanguineous offspring. These findings underscore the role of genetic factors in determining the phenotypic severity of cleft deformities.

Incorporation of genetic counseling and community awareness programs may help reduce the incidence of severe cleft deformities in high-risk populations. Further large-scale, multicentric studies integrating molecular genetic analysis are recommended.

RECOMMENDATIONS

Incorporation of genetic counseling services in cleft care centers.

Public awareness programs highlighting risks associated with consanguineous marriages.

Further multicentric studies with larger sample sizes.

Ethical Considerations

Ethical clearance was obtained from the Institutional Ethics Committee. Informed consent was obtained from parents or guardians of all participants.

Conflict of Interest

None declared.

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