

Genetic Correlations in the Distribution of Lip Patterns in Parents of Children with Cleft Lip and Palate

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ABSTRACT

Introduction: Cheiloscopy, the study of lip prints, offers a novel approach in genetic research, particularly in understanding the inheritance of cleft lip and palate (CL/P). Lip prints, unique like fingerprints, are influenced by genetic factors and may provide critical insights into the hereditary components of CL/P.

Materials and Methods: This study, conducted in the Dept. of Public Health Dentistry, SPDC, Sawangi, Wardha, India, included parents of children with confirmed CL/P. Using the cellophane method, lip prints were collected non-invasively. The patterns were analyzed according to Suzuki and Tsuchihashi's classification. Data analysis was performed using SPSS with chi-square and logistic regression methods, focusing on significance levels below 0.05.

Results: The study revealed significant insights into the genetic correlations of CL/P. The average age of participants was 32.28 years, with a balanced gender distribution. The prevalence of cleft types showed both cleft lip and palate as most common. Analysis indicated Type II lip pattern as the most prevalent across genders and quadrants. Notably, gender-specific variations in lip patterns were observed. A significant prevalence of whorl patterns was noted, suggesting their potential as a genetic marker for CL/P, although no gender-based differences were statistically significant.

Conclusion: The study underscores the potential of lip print analysis as a non-invasive tool for understanding CL/P genetics. The significant prevalence of certain lip patterns, especially Type II and whorl patterns, suggests their relevance as genetic markers, highlighting the need for further research in this promising field.

Keywords: Cleft lip and palate, genetic marker, lip prints, whorl pattern

INTRODUCTION

Cheiloscopy, the examination of lip prints, has generated considerable attention in the realm of genetic research, specifically in comprehending the hereditary tendencies of cleft lip and palate (CL/P).^{1,2} Lip prints are a distinct feature of the lips that is similar to fingerprints.^{3,4} Lip prints, like fingerprints, have distinct characteristics that are unique to each individual. These formations are caused by the lines and furrows on the surface of the lip and are thought to be influenced by genetic factors. As a result, examining these patterns provides a novel approach to understanding the hereditary components of cleft lip and palate.⁵

In order to comprehend the epidemiology and etiology of any congenital abnormality, precise phenotyping is very important. This is because the ability to identify effects is diminished when different groups are considered as a single entity. Despite the fact that clefts of the lip and palate can manifest in a variety of ways, they are typically put into the category of qualitative characteristics.⁶ Furthermore, there is a wealth of evidence that suggests that the spectrum is actually more complex and ought to include a variety of sub-clinical phenotypic characteristics that have been reported in both

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people who have chronic low back pain and their relatives who are "unaffected" by the condition.⁷

Analysis of lip print patterns in parents who have children with CL/P has been given a high priority in the research that is currently being conducted. Whether or not

there are distinct lip print patterns that are associated with an increased likelihood of having children with these disorders is the question that will be investigated in this study. The researchers hope that by analyzing these patterns, they will be able to construct more accurate predictive models for the inheritance of CL/P.^{8,9} These findings have the potential to serve as a non-invasive screening tool in genetic counselling and are able to consistently indicate a higher risk of CL/P. With this information, medical professionals would be able to identify individuals or couples who are at risk and provide them with guidance and support based on accurate information. Understanding of the genetic markers that are associated with CL/P can be of assistance in the development of specific preventative measures, which may result in a reduction in the incidence of these problems.^{10, 11}

It is possible that this research will lead to significant improvements in the early detection and treatment of medical conditions, which will ultimately result in improved planning and control methods, particularly in the areas of prenatal and pre-conception healthcare. Furthermore, the information that was gleaned from this study is of utmost significance for the field of genetic counselling, particularly for families who have a history of CL/P or who have had a child affected by the condition in the past.

Table 1: Demographic information of participants under the study

Demographic Information	
Age (Mean±SD)	32.28± 7.28
Sex (M/F)	78/72
Type of Cleft	
•Cleft Lip	22.67 %
•Cleft Palate	18.67 %
•Both	58.67 %

Table 2: Prevalence of different lip pattern as per lip quadrant

Type of Lip pattern	1st Quadrant (URL)		2nd Quadrant (ULL)		3rd Quadrant (LLL)		4th Quadrant (LRL)	
	Male	Female	Male	Female	Male	Female	Male	Female
Type I	13 (16.7%)	27 (37.5%)	13 (16.7%)	18 (25.0%)	22 (28.2%)	23 (31.9%)	31 (39.7%)	22 (30.6%)
Type I'	25 (32.1%)	5 (6.9%)	18 (23.1%)	9 (12.5%)	13 (16.7%)	6 (8.3%)	12 (15.4%)	13 (18.1%)
Type II	36 (46.2%)	33 (45.8%)	38 (48.7%)	38 (52.8%)	38 (48.7%)	34 (47.2%)	34 (43.6%)	28 (38.9%)
Type III	1 (1.3%)	6 (8.3%)	1 (1.3%)	2 (2.8%)	3 (3.8%)	9 (12.5%)	1 (1.3%)	9 (12.5%)
Type IV	0 (0.0%)	1 (1.4%)	0 (0.0%)	1 (1.4%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
Type V (Whorl Pattern)	3 (3.8%)	0 (0.0%)	8 (10.3%)	4 (5.6%)	2 (2.6%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
Total	78 (100.0%)	72 (100.0%)	78 (100.0%)	72 (100.0%)	78 (100.0%)	72 (100.0%)	78 (100.0%)	72 (100.0%)
p-value	0.00	0.283	0.108	0.040				

RUL- Upper Right lip; ULL- Upper left lip;LLL- Lower left lip; LRL- lower right lip

MATERIALS AND METHODS

The study was conducted in the Dept. of Public Health Dentistry, SPDC, Sawangi, Wardha, India, after obtaining ethical approval from institutional ethics committees, parents of children with confirmed cleft lip and palate were included while parents of Children with other craniofacial anomalies or syndromes, and those who have undergone lip surgery were excluded. Based on power of previously published article 150 samples were need to reject the null hypothesis of identifying lip pattern among parents of cleft lip and parent patient. Simple random sampling method was used to select samples in this study.¹²

The process of data collection was carried out using methods that were both standardized and non-invasive throughout. For the purpose of obtaining the lip prints of the subjects, the cellophane method was successfully utilized. There was no strain placed on the lips of the subjects because they were positioned in such a way that allowed them to relax their lips. The application of a dark-coloured lipstick to the lips was accomplished with a single motion, and disposable cotton buds were utilized to ensure that the lipstick was distributed evenly across the lips. It was instructed to everyone that they should gently press their lips against a piece of folded white bond paper that was placed between their lips. It was folded in half to make the paper. Lip prints were visualized with the help of a magnifying lens, and after that, they were analyzed and classified in accordance with the classification system that Suzuki and Tsuchihashi (1970) proposed, which is still widely utilized in the modern world.¹³

Statistical Package for the Social Sciences version 20.0 was utilized in order to perform the analysis on the data that was collected (SPSS, IBM). Research was conducted using the Chi-square and logistic regression methods. The significance of the P value was determined to be less than 0.05. (confidence interval of 95 percent was taken).



RESULTS

Intriguing insights into the possible genetic correlations of cleft lip and palate (CL/P) conditions were uncovered through the examination of lip print patterns in parents of children who were diagnosed with CL/P. The findings from the comprehensive examination of lip print characteristics are presented in this section, along with a statistical analysis that aimed to establish any significant correlations between particular lip print patterns and the types of CL/P that were observed in the participants of the study.

Table 1 contains demographic information that indicates that the average age of the people who participated in the study was 32.28 years old. The standard deviation of the participants' ages was 7.28 years, which indicates that there was a moderately varied age range among the participants. A total of 78 males (M) and 72 females (F) were present, making the gender distribution almost exactly equal. In terms of the different types of clefts that were observed, the majority of the cases, which accounted for 58.67 percent of the total, were of both cleft lip and palate, which indicates that this is the most common type of cleft in the group that was being studied. Cleft lip alone was present in 22.67 percent of the cases, while cleft palate alone was slightly less common, accounting for 18.67 percent of the cases. Both of these conditions were present in the cases.

The table 2 represents a detailed analysis of the prevalence of different lip patterns across four quadrants of the lips in both male and female participants. Significantly, Type II lip pattern emerges as the most prevalent across all quadrants and genders, with the highest representation in the 3rd quadrant (LLL) for males (48.7%) and in the 2nd quadrant (ULL) for females (52.8%). Type I shows a notable gender disparity, being more common in females, particularly in the 1st quadrant (URL) with 37.5%. Interestingly, Type I is significantly more prevalent in males in the 1st quadrant (32.1%) compared to females (6.9%). Type III and Type V (Whorl Pattern) are relatively rare, with Type IV being virtually absent in the sample. The statistical significance (p-value) indicates a notable difference in lip pattern distribution between genders, particularly in the 4th quadrant (LRL) with a p-value of 0.040, suggesting gender-specific variations in lip print patterns.

Table 3 presents data on the presence of a whorl pattern as a potential genetic marker for cleft lip and palate, analyzed across male and female participants. The whorl pattern was observed in a significant proportion of the study population, with 66.7% of males and 75.0% of females exhibiting this pattern, resulting

Table 3: Presence of genetical marker of cleft lip and palate

Variable	Gender		Total	P-value
	Male	Female		
Whorl pattern	52 (66.7%)	54 (75.0%)	106 (70.7%)	0.174
Whorl pattern in more than one quadrant	23 (29.5%)	21 (29.2%)	44 (29.3%)	0.555

in a combined prevalence of 70.7%. Additionally, the presence of a whorl pattern in more than one quadrant of the lips was found in approximately 29% of both genders (29.5% in males and 29.2% in females). However there was no significant difference observed as per gender.

DISCUSSION

The study's discussion on the importance of whorl patterns in lip prints as prospective genetic markers for cleft lip and palate (CL/P) revealed a significant prevalence of these patterns in both males and females, indicating their potential as universal markers for CL/P. This discovery is consistent with prior studies on dermatoglyphic patterns, but it is notable for the absence of major disparities across genders. This emphasizes the intricate nature of genetic markers in CL/P and underscores the necessity for additional research.

The current study reveals that Type II lip pattern is the most common in all quadrants and genders. It is particularly widespread in the 3rd Quadrant (LLL) for males, with a representation of 48.7%. For females, the highest representation of Type II lip pattern is in the 2nd Quadrant (ULL), with a percentage of 52.8%. However, the lip print pattern observed in the Rajasthan population consistently exhibited a type III pattern, regardless of whether the upper lip, lower lip, or both lips were analyzed. The research undertaken by Sivapathasundharam et al., Augustine, and Ramandeep S. Narang et al. have yielded comparable conclusions. Additionally, the researchers found that the type III pattern was the most common.¹⁰

Whorl lip print patterns can arise on either the upper or lower lip. An upper lip whorl groove pattern is a singular circular arrangement of grooves that is positioned precisely on the midline of the upper lip. Lower lip whorls are symmetrical circular patterns located either to the left or right of the midline of the lower lip. Hirth and his colleagues in Germany made the initial discovery of whorls in lip print patterns over thirty years ago as part of their comprehensive investigation on the genetics and diversity of such patterns.¹⁴

Hirth et al.¹⁴ noted an elevated occurrence of whorls on the lower lip in non-syndromic CL/P patients and their relatives. They hypothesized that these observed whorls in families with clefts could potentially indicate a milder manifestation of lower lip pits or fistulae. This was an observed phenomenon. If it is indeed accurate that families affected by CL/P exhibit a greater prevalence of whorl lip prints, then it is plausible to consider these prints as a component of an extended range of observable characteristics associated with nonsyndromic CL/P. Whorls, when associated with clefting, may serve as a valuable clinical indicator for predicting the probability of recurrence in families with CL/P. This assessment takes into consideration both their subtle morphological traits and their familial background.¹⁵

Our study, like the Egyptian study, discovered a majority of Type II irregular patterns and an unusual Type V pattern, which serves as a genetic indicator for the inheritance of cleft lip (palate) deformity. The study identified Type V (whorl pattern) as a genetic marker for the transmission of cleft lip (palate)



deformity, and observed that Type I' and Type II were more prevalent.^{13,16} Furthermore, they determined that it functioned as a genetic indicator for persons in good health, as no Type III pattern was observed. Nevertheless, we also discovered a Type III pattern, which contradicts their findings in this regard.^{14,16}

CONCLUSION

According to the findings of our study, the most common lip pattern across all lip quadrants and genders is Type II. Furthermore, the study discovered the whorl pattern in both genders, which was observed in a significant portion of the study population. However, the lack of significant gender differences in whorl pattern prevalence and presence in multiple quadrants suggests a more complex genetic interplay. While the findings are encouraging, they also highlight the need for additional research to confirm these patterns as reliable markers for CL/P and to investigate their broader implications in genetic counselling and clinical practice.

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