

A Population Genetic Study of Cleft Chin in India

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ABSTRACT The paper reports data on cleft chin in 900 individuals from Kerala. The results of all surveys in India in which the cleft chin was observed have been tabulated. Gene frequency analysis has been attempted. Wide variations in gene frequencies were found for the gene *cl*. The major racial groups in India tend to follow definite trends. The trait varies from dimples to furrows, and is independent of age but reveals sex differences. A plea is made to collect more family, twin and population data.

Some persons have a distinct depression, dimple, fissure or cleft on the lower margin of the chin in the median sagittal plane. Such chins are also termed bilobed chins. The trait shows a great deal of variability in its mode of expressivity, from trace to Y-shaped furrow (fig. 1).

Although inadequate attempts have been made to establish the mode of inheritance of this trait, it appears that the trait is inherited as an autosomal dominant (Winchester, '60). Lebow and Sawin ('41) report a pedigree in which the trait is transmitted through four generations as a simple dominant. However, in the same pedigree, a carrier failed to express the trait. McKusick ('68) lists this trait as a good dominant (item no. 1166).

The incidence of this trait in various populations is hardly known. Singh and Malhotra ('71) report this trait among the Manipuris, and Shastree and Malhotra (unpublished) studied 616 Maharashtrian females. Three endogamous Gavda groups of Goa were studied by Malhotra ('71). Malhotra et al. ('71) demonstrated striking differences in the incidence of this trait in various populations.

The present paper attempts to study the following aspects of the trait: (1) a more complete analysis of the subjects studied; (2) age variation and sexual dimorphism; (3) gene frequency analysis; and (4) group variability as a guide to its use in inter-population studies.

METHODS AND MATERIALS

The 900 samples were randomly drawn from schools, colleges and villages on 100 unrelated males, age 9–50, from each of nine endogamous groups of Kerala, India. The chin of each individual was examined and the presence or absence of cleft was noted. Chi-square values have been calculated by using Woolf's G-tables ('57).

RESULTS AND DISCUSSIONS

Age variation. Since the frequency of individuals with cleft chin is very low and all the groups show a similar incidence the data from the nine endogamous groups have been pooled. The frequency of cleft chin by nine age classes is shown in table 1. Although the percentage of cleft chinned individuals varies from 4.84% to 12.12%, the differences are non-significant ($\chi^2 = 4.8$; d.f. = 8, $0.7 < p < 0.8$).

Malhotra ('71) studied three endogamous groups of Gavdas of Goa and found the trait to be independent of age (Hindu Gavdas $\chi^2 = 4.1$; d.f. = 4, $0.3 < p < 0.5$, Christian Gavdas $\chi^2 = 1.3$; d.f. = 4, $0.8 < p < 0.9$ and Nav-Hindu Gavdas $\chi^2 = 4.6$; d.f. = 5, $0.3 < p < 0.5$).

Sex differences. Singh and Malhotra ('71) studied this aspect of the trait in

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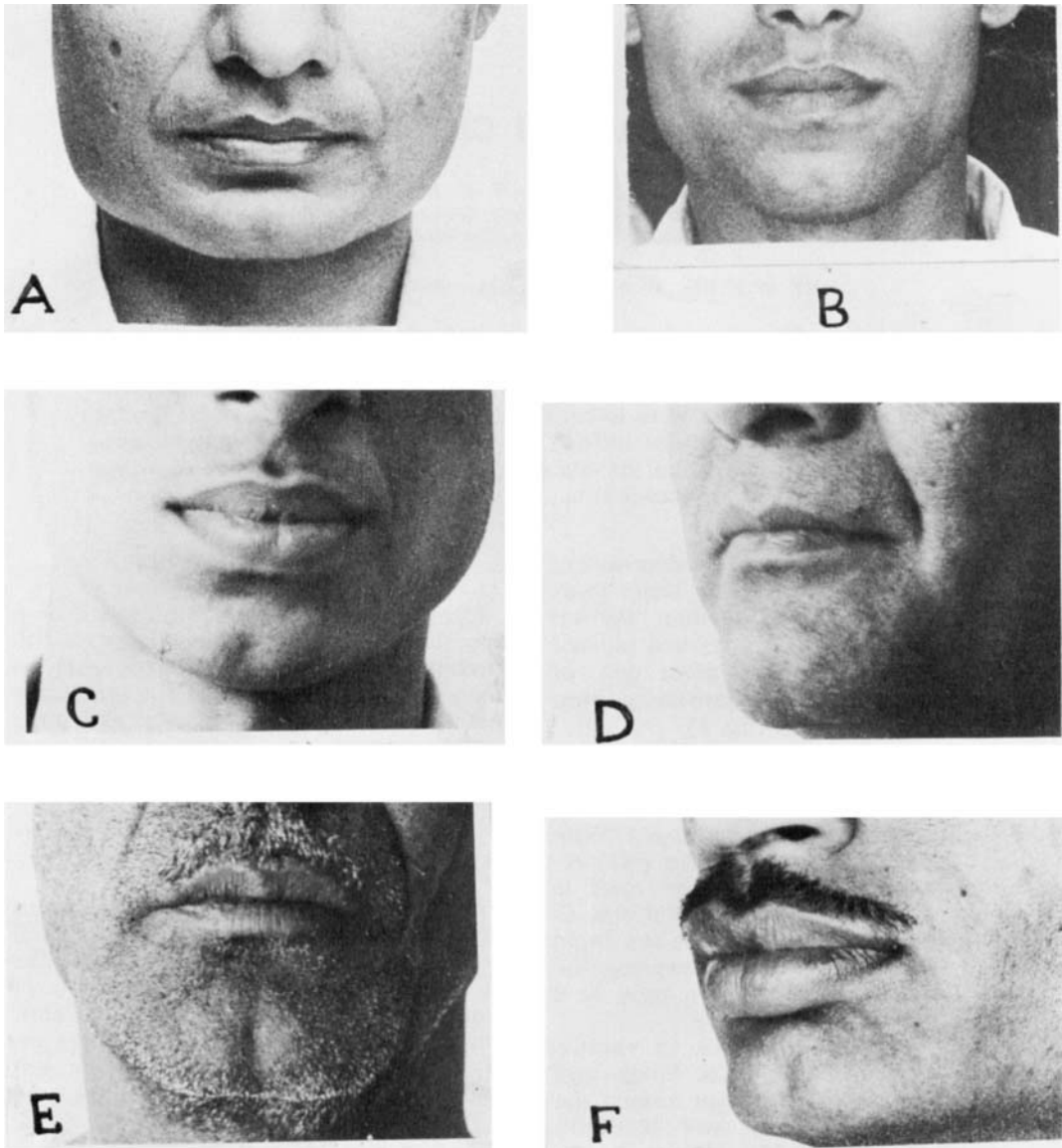


Fig 1 Various grades of clefts in the chin: A, trace; B, depression; C, dimple; D, medium vertical furrow; E, prominent vertical furrow, and F, Y-shaped fissure.

three population groups. While Korkus ($\chi^2 = 6.33$; d.f. = 1, $p < 0.2$) and Manipuri Brahmins ($\chi^2 = 17.63$; d.f. = 1, $p < 0.001$) showed significant sex differences, the Meitheis, a Mongoloid population, revealed homogeneous distribution ($\chi^2 = 1.17$, d.f. = 1, $0.2 < p < 0.3$).

Gene frequency analysis. Considering

the trait as a "good dominant," gene frequency can be estimated. Letting Cl and cl represent dominant and recessive alleles respectively three genotypes ($ClCl$, $Clcl$ and $clcl$) give rise to two phenotypes, cleft chin and absence of cleft. The gene frequency of allele cl has been calculated as $\sqrt{clcl}$.

TABLE 1

Frequency of cleft chinned individuals in various age classes (pooled data)

S. no.	Age classes (years)	Number tested	Present	%
1.	6-10	62	3	4.84
2.	11-15	223	16	7.17
3.	16-20	171	10	5.85
4.	21-25	112	7	6.25
5.	26-30	102	6	5.88
6.	31-35	81	8	9.88
7.	36-40	70	7	10.00
8.	41-45	46	5	10.87
9.	46-50	33	4	12.12
Total		900	66	7.33

Ethnic differences. The available phenotype and allele frequencies on this trait summarised in table 2 show the range of variability of this trait in populations of diverse origins. Data for cleft chin are reported for 22 populations of India (2,651 subjects) including the present study.

In the present series, people of Kerala, the range of variation of this trait is between 4% (Paraya) and 10% (Tiyya), with an average of 7.33%. An intra-series comparison reveals that the differences among the nine endogamous groups from Kerala are statistically non-significant (see table 3).

Table 2 shows that while the incidence of cleft chin among the males varies from 4% (Parayas) to 66.8% (Korkus), it ranges from 6% (Chitpavan Brahmins) to 84.6% (Korkus), among the females. On the three groups where data are available for both sexes the males have a higher incidence of cleft chin among the Manipuri Brahmins and Meithei, and the females among the Korkus. The pooled series mean for the females, however, shows a slightly higher incidence of cleft (19.76%) as compared to the males (16.47%).

A great deal of heterogeneity is observed among the different population groups (table 3). Since some populations have shown significant bisexual differences, comparisons have only been made between the males. Out of 105 pair comparisons significant difference above the level of 5% probability occurs in 55 combinations. The differences are highly significant ($p < 0.001$) in 29 instances. Shastree and Malhotra (unpublished) also reported striking

variations among 21 pairs out of 45 comparisons in the case of females.

Racially the data represent Caucasoids (Groups no. 10-12, 13-19, and 21), Mongoloids (Group no. 22), Australoids (Groups no. 1-9) and Australoid plus Mediterranean admixture (Group no. 20). Except for the Korkus, a Mundari-speaking tribe of Maharashtra, all are endogamous castes.

The gradient for the *cl* gene has been drawn in figure 2 to determine if different races in India form any meaningful pattern for this variation. It is noteworthy that both males and females of Korku, a tribal group, fall completely at one end of the gradient. Next in the ladder are various Caucasian castes, who also show quite a bit of variability. The Australoid group of castes cluster at the other end of the gradient. The Mongoloids represented by only one group occupy a position between Caucasoids and Australoids.

Although no far reaching conclusions can be drawn on the basis of the limited data at hand, the variation observed in the major racial groups tends to follow certain trends.

In view of the heterogeneity depicted by these groups and the apparent inheritance of the trait as a good dominant, we believe that this easily observable variation should be used as a marker and routinely recorded in population genetic surveys. Further family studies, however, are necessary to see whether the trait is due to a single gene and whether the variations observed in its manifestation are determined by multiple alleles, genes at differ-

TABLE 2
Incidence of *C1cl* phenotypes and gene frequencies among 22 Indian groups

No.	Population group	Abbrevi- ation	Sex	Sample size	Phenotypes				Allele frequencies			Source
					Cleft chin (Cl)		No cleft chin (cl)		Cl	cl		
					N	%	N	%				
Kerala												
1.	Ezhavas	EZ	M	100	7	7.00	93	93.00	0.0356	0.9644	Present study	
2.	Chovas	CHI	M	100	8	8.00	92	92.00	0.0408	0.9592	Present study	
3.	Tiyyas	TY	M	100	10	10.00	90	90.00	0.0513	0.9487	Present study	
4.	Thanta Pulaya	TP	M	100	8	8.00	92	92.00	0.0408	0.9592	Present study	
5.	Kana Pulaya	KP	M	100	6	6.00	94	94.00	0.0305	0.9695	Present study	
6.	Valluva Pulaya	VP	M	100	6	6.00	94	94.00	0.0305	0.9695	Present study	
7.	Paraya	PR	M	100	4	4.00	96	96.00	0.0202	0.9798	Present study	
8.	Vemparaya	VR	M	100	9	9.00	91	91.00	0.0461	0.9539	Present study	
9.	Sambavar	SR	M	100	8	8.00	92	92.00	0.0408	0.9592	Present study	
	Total			900	66	7.33	834	92.67	0.0374	0.9626	Present study	
Goa												
10.	Hindu Gavidas	HG	M	198	35	17.68	163	82.32	0.0927	0.9073	Malhotra ('71)	
11.	Christian Gavidas	CG	M	193	32	16.58	161	83.42	0.0867	0.9133	Malhotra ('71)	
12.	Nav-Hindu Gavidas	NH	M	196	32	16.33	164	83.67	0.0853	0.9147	Malhotra ('71)	
	Total			587	99	16.87	488	83.13	0.0883	0.9117	Malhotra ('71)	
Maharashtra												
13.	Deshastha Rigvedi	DR	F	100	8	8.00	92	92.00	0.0408	0.9592	Shastree and Malhotra ('71)	
14.	Brahmins											
	Deshastha Yajurvedi	DY	F	100	12	12.00	88	88.00	0.0619	0.9381	Shastree and Malhotra ('71)	
15.	Chitpavan Brahmins	CH	F	100	6	6.00	94	94.00	0.0305	0.9695	Shastree and Malhotra ('71)	
16.	Karhada Brahmins	KR	F	96	15	15.62	81	84.38	0.0814	0.9186	Shastree and Malhotra ('71)	
17.	Saraswat Brahmins	SA	F	60	18	30.00	42	70.00	0.1633	0.8367	Shastree and Malhotra ('71)	
18.	Chandraseniya Kayastha											
	Prabhu	CKP	F	60	14	23.33	46	76.67	0.1244	0.8756	Shastree and Malhotra ('71)	
19.	Maratha	MT	F	100	12	12.00	88	88.00	0.0619	0.9381	Shastree and Malhotra ('71)	
20.	Korku	KU	M	163	109	66.87	54	33.13	0.4244	0.5756	Malhotra, Bhanu, Singh and Shastree ('71)	
			F	52	44	84.62	8	15.38	0.6078	0.3922		
			T	215	153	71.16	62	28.84	0.4630	0.5370		
	Total			831	238	28.64	593	71.36	0.1553	0.8447		
Manipur												
21.	Brahmins	MB	M	59	23	38.98	36	61.02	0.2188	0.7812	Singh and Malhotra ('71)	
			F	56	4	7.14	52	92.86	0.0364	0.9636	Singh and Malhotra ('71)	
22.	Meitheis	MM	T	115	27	23.48	88	76.52	0.1253	0.8747	Singh and Malhotra ('71)	
			M	107	18	16.82	89	83.18	0.0880	0.9120	Singh and Malhotra ('71)	
			F	111	13	11.71	98	88.29	0.0604	0.9396	Singh and Malhotra ('71)	
			T	218	31	14.22	187	85.78	0.0738	0.9262	Singh and Malhotra ('71)	
	Total			333	58	17.42	275	82.58	0.0913	0.9087	Singh and Malhotra ('71)	
	Total			2651	461	17.39	2190	82.61	0.0911	0.9089		

M, Male; F, Female; T, M + F.

TABLE 3
Interpopulation Chi-square values for the 15 populations (males only) listed in table 2

Popula- tion groups	EZ	CHI	TY	TP	KP	VP	PR	VR	SR	KU	MB	MM	HG	CG	NH
EZ	—	0.07	0.58	0.07	0.08	0.08	0.87	0.27	0.07	103.16 ³	24.37 ³	4.86 ¹	6.93 ²	5.65 ¹	5.53 ¹
CHI			0.24	0.00	0.30	0.30	1.44	0.06	0.00	98.59 ³	22.22 ³	3.76 ¹	5.49 ¹	8.63 ²	4.24 ¹
TY				0.24	1.09	1.09	2.85	0.05	0.24	90.16 ³	18.47 ³	2.08	3.24	2.44	2.29
TP					0.30	0.30	1.44	0.06	0.00	98.59 ³	22.22 ³	3.76 ¹	5.49 ¹	8.63 ²	4.24 ¹
KP						0.00	0.42	0.65	0.30	108.01 ³	26.75 ³	6.17 ¹	8.62 ²	7.31 ²	7.06 ²
VP							0.42	2.10	0.30	108.01 ³	26.75 ³	6.17 ¹	8.62 ²	7.31 ²	7.06 ²
PR									1.44	232.38 ³	32.38 ³	9.66 ²	12.97 ³	11.38 ³	11.08 ³
SR									0.06	94.27 ³	20.27 ³	2.84	4.27 ¹	3.35	3.17
KU										98.59 ³	22.22 ³	3.76	5.49 ¹	8.63 ²	4.24 ¹
MB										13.82 ³	69.35 ³	9.73 ²	93.83 ³	97.61 ³	99.54 ³
MM													10.86 ³	12.16 ³	12.55 ³
HG													0.04	0.01	0.01
CG														0.08	0.03
NH															0.01

¹ $p < 0.05$. ² $p < 0.01$. ³ $p < 0.001$.

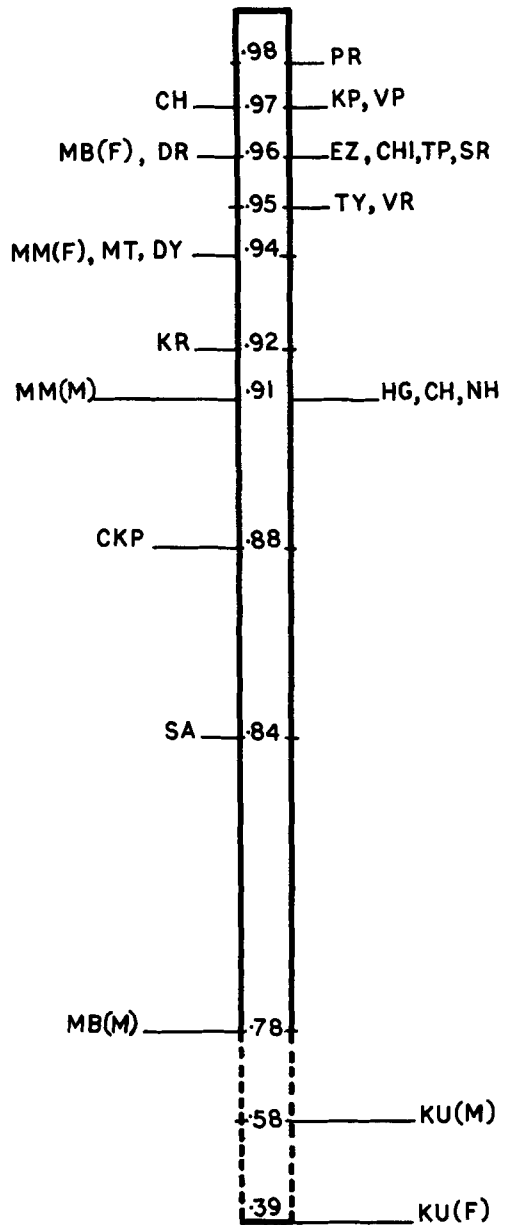


Fig. 2 "cl" gene gradient.

ent loci, or some environmental modification. We stress the need to collect more family, twin and population data.

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