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TONGUE PIGMENTATION IN MAN

Ethnic Studies and Further Pedigrees *

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SUMMARY

A genetic study has been carried out on tongue pigmentation in man, based on 10 pedigrees and 39 Indian populations. It is concluded in favour of a genetic basis of the trait, which would be inherited as an autosomal recessive.

Based on 3 pedigrees and data on 406 families, tongue pigmentation has been recently suggested as a diallelic autosomal recessive character in human populations (Rao 1970*a*, *b*, and *c*). Rates of incidence of the trait in some Indian populations were also reported, both for adults (Rao 1970*b*) and newborn babies (Rao and Bose 1970). These rates were found to be much higher among adults than the newborn. It was then inferred that either the intensity of the colour pigmentation in the newborn is too dilute to be detected with the naked eye, or the recessive gene responsible for the trait may fail to penetrate in the recessive homozygotes occasionally. Concerning the latter possibility, it was found (Rao and Ghorai 1970) that the penetrance value of the recessive allele was very high. Thus, tongue pigmentation was suggested as an autosomal trait with rare failure of penetrance of the recessive allele.

In this article, we present 7 more pedigrees from India, and population data on 39 Indian populations including the 8 published earlier (Rao 1970*b*). This series of 39 populations includes 4 tribes, 18 caste groups, and 17 heterogeneous mixtures of castes and tribes. Detailed study of these pedigrees is suggestive of only autosomal inheritance, as was proposed earlier. An attempt is also made to study the distribution of this trait in the Indian populations, and the possible relation between the trait and skin colour, a problem raised by Davis (1968).

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MATERIAL AND ANALYSIS

Pedigree Study

In all, 10 pedigrees are reported here, which include the 3 already published ones. The latter are presented in Fig. 1 (from Rao 1970*a*) and Figs. 2 and 3 (from Rao 1970*b*), while the 7 new pedigrees are presented in Figs. 4 to 10. Description of the first 3 pedigrees may be found in the original publications.

Figures 4 to 9 run over four generations each; Fig. 10 runs over five living generations. These 7 pedigrees were compiled at Waltair, an eastern coastal town of Andhra Pradesh, India, and the former 3 pedigrees were from the state of Kerala in India. The populations from where these pedigrees were collected are shown in Table II. The 7 new pedigrees presented here come from rather backward communities where a reasonable degree of illegitimacy is known to exist. Out of the old 3 pedigrees also, the same problem arises in case of Fig. 2 (Rao 1970*b*).

SYMBOLS USED IN THE PEDIGREES

□	Male	N.S.	Not seen
○	Female	N.A.	Not available
■ ●	Presence of tongue pigmentation (personal examination)	Numbers below the individual	Corresponding ages
◪ ◔	Presence of tongue pigmentation (hear-say information)	?	Examined, but doubtful
D	Dead	↗	Propositus

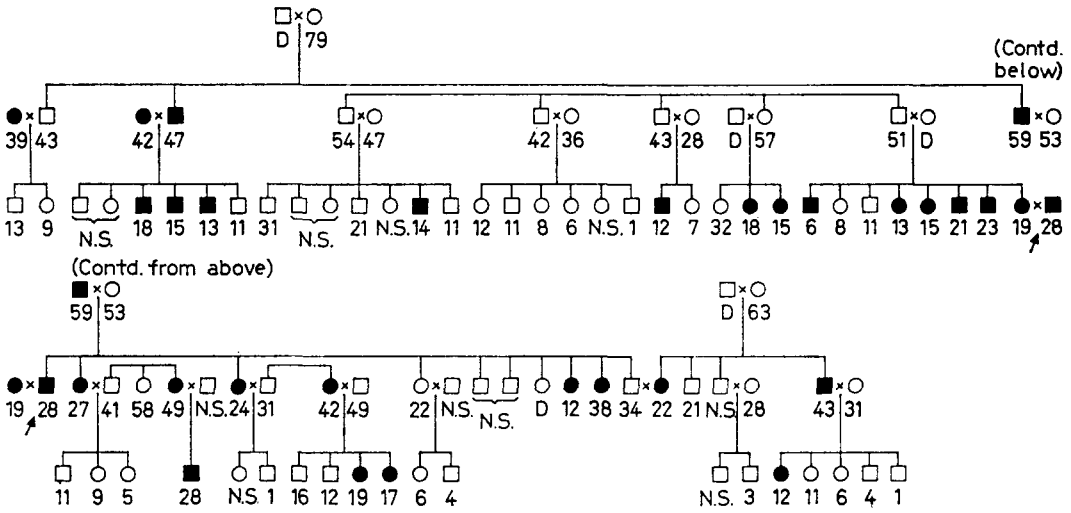

FIG. 1. [Courtesy of *Human Heredity*, 20: 9]

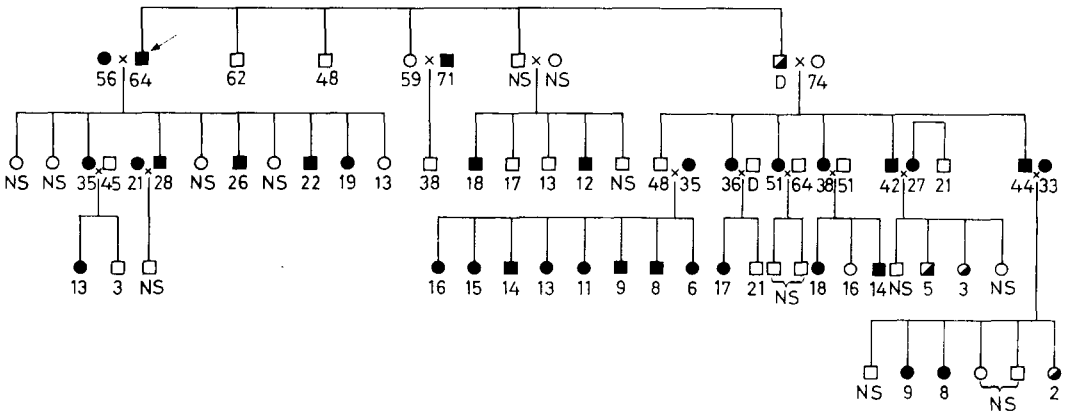

FIG. 2. [Courtesy of *Human Heredity*, 20: 592]

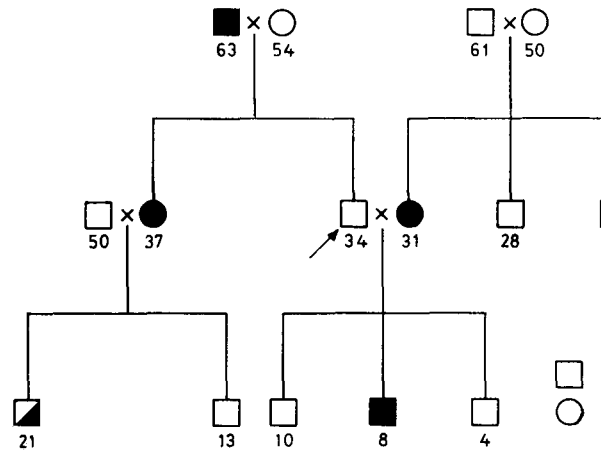

FIG. 3. [Courtesy of *Human Heredity*, 20: 592]

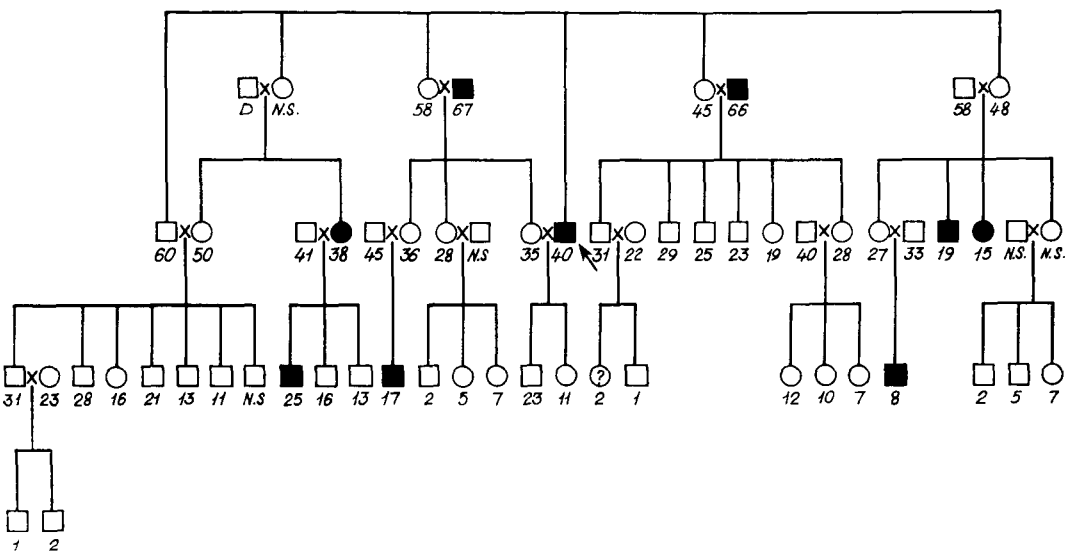

FIG. 4.

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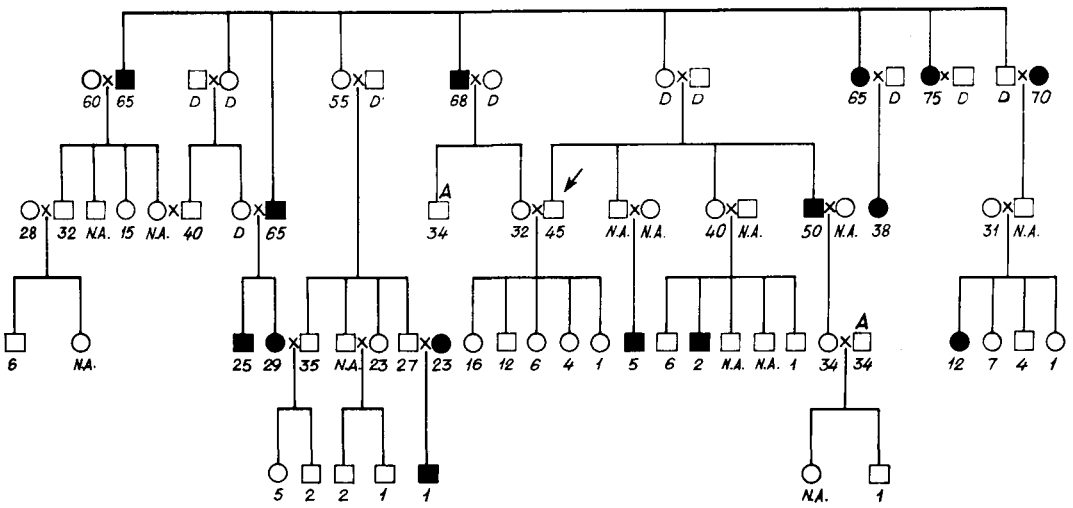

FIG. 5

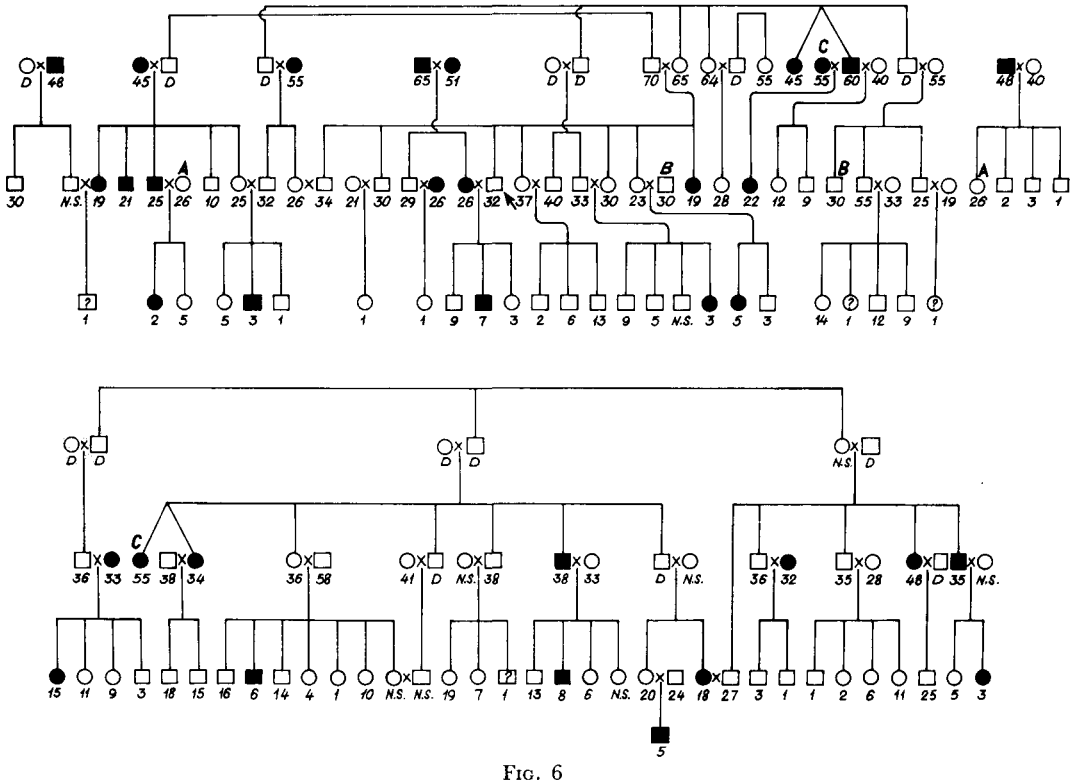

FIG. 6

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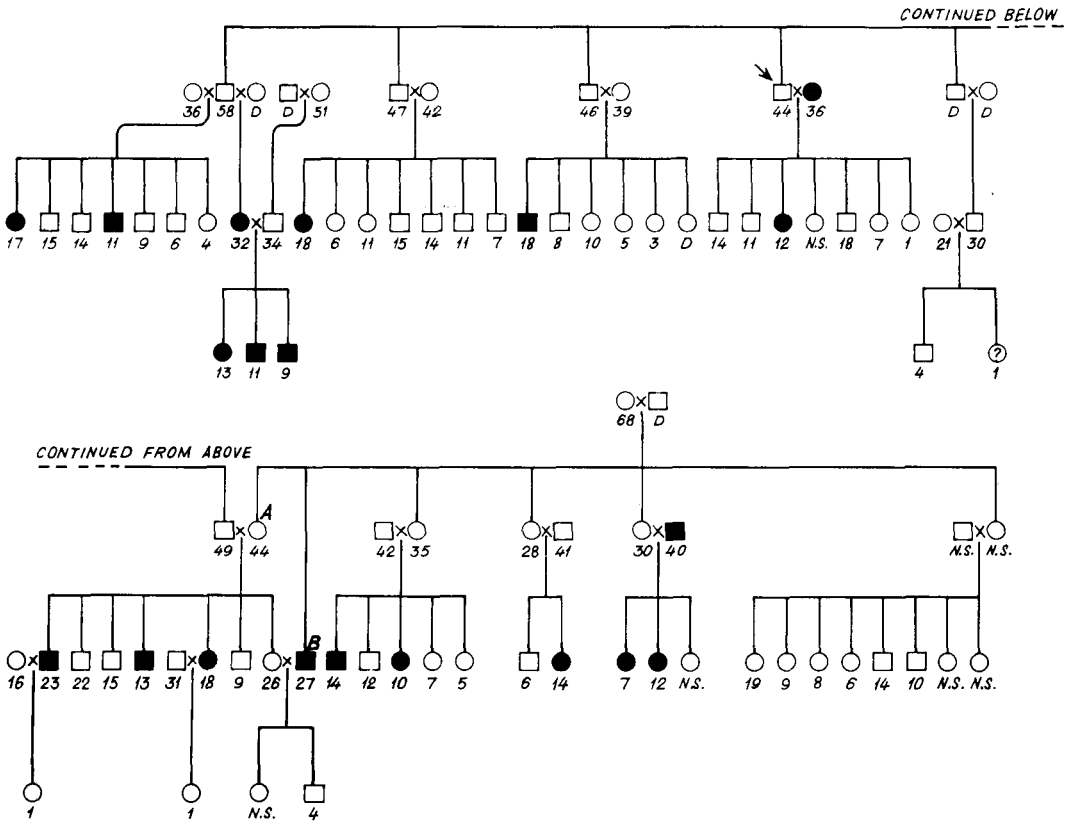

FIG. 7

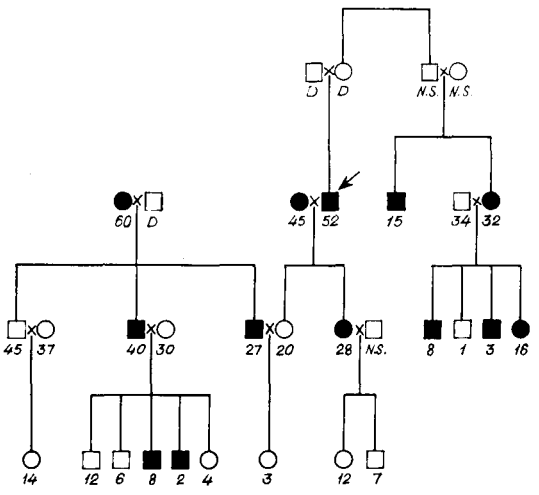

FIG. 8

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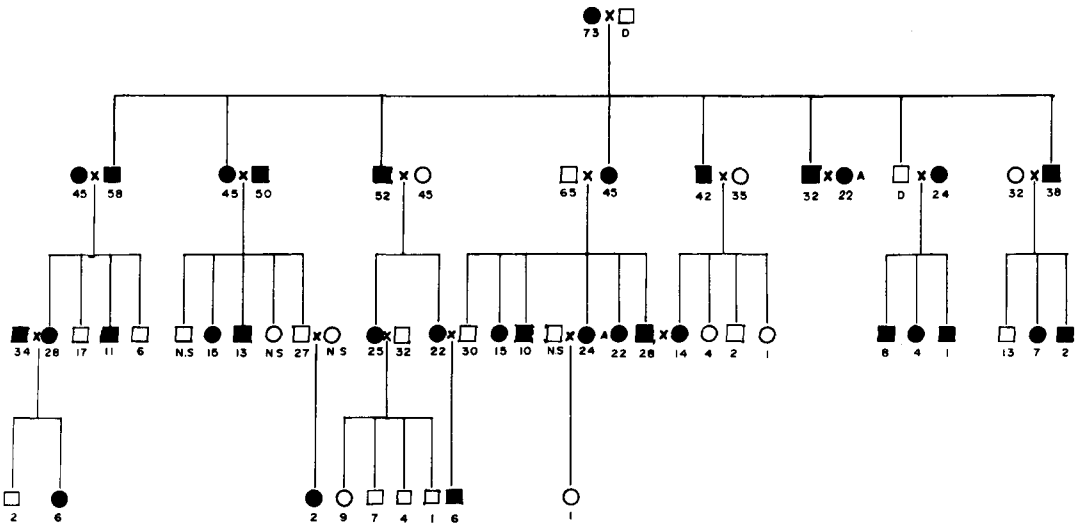

FIG. 9

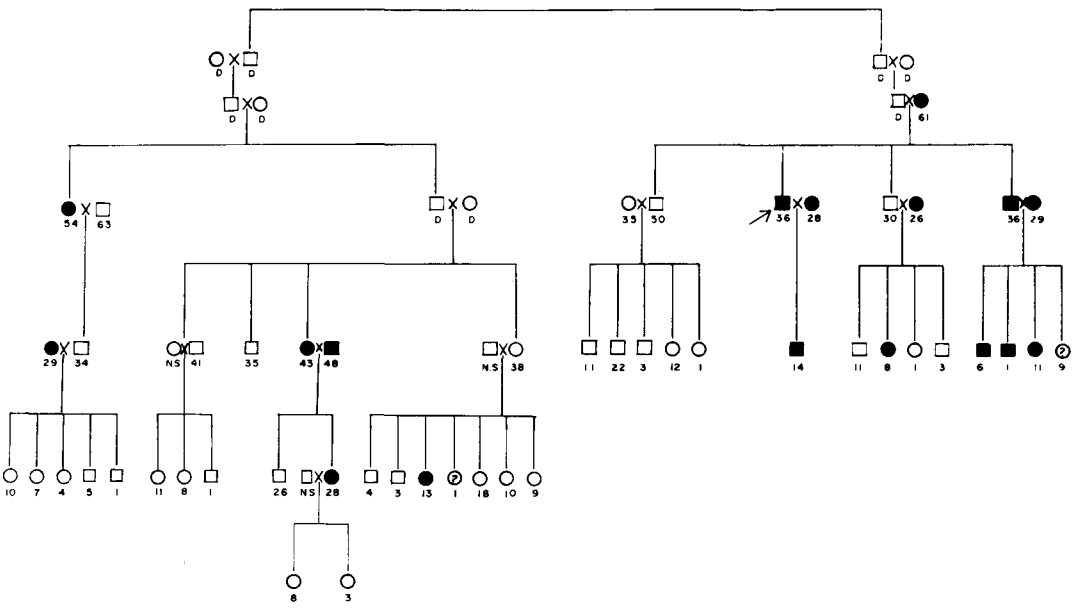

FIG. 10

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Population Study

In all, 39 Indian " populations " were studied for the distribution of tongue pigmentation, including the 8 published earlier (Rao 1970*b*). Though many of these populations are endogamous groups, quite a few of them represent heterogeneous groups with mixed caste affiliations.

Altogether, we have studied 22 endogamous groups and 17 " mixed " populations. Of the 22 endogamous groups, 18 are caste groups and 4 are tribes. All the relevant information about these 39 populations is summarised in Table I in two parts, (*a*) dealing with the endogamous groups and (*b*) dealing with the 17 mixed populations. The latter 17 populations are presented separately in Table I(*b*) because, either the endogamous identity is unknown or due to smallness of the sample sizes several endogamous groups from the same geographical region are grouped together to constitute one " population ".

In Table I(*a*) and (*b*), all the populations coming from economically backward communities have been identified with asterisk marks (*) along with the serial numbers, since most of such communities suffer from malnutrition, and that malnutrition may give rise to transient discolouration of the tongue similar to " tongue pigmentation " (Rao 1970*d* and *e*). Thus, the rates of incidence for such populations may have been enhanced in Table I. In the description of the 39 populations we shall refer to each population by the serial number (cf. Table I).

For the sake of consistency of observation, all the sample subjects were examined for the trait by one of the authors (D.C.R.) during 1968-70. The 6 populations studied from Maharashtra (nos. 1 to 5 and 23) were represented in our samples by school-going children at Poona, whose ages ranged from 5 to 17 years. The 2 populations from West Bengal (6 and 7) were represented by household adults at Calcutta, all above 20 years of age. Again, school-going children at Jeypore constitute the samples for 8, 9, 10, 24, and 25 from Orissa, all between 7 and 19 years of age (mostly around 12 years). Populations 31 and 34 were represented by household adults at Waltair, Andhra Pradesh, whose ages ranged from 19 to 58 years. The remaining samples from Andhra Pradesh caste groups (11 to 18, 26 to 30, and 33) were obtained from the students of the Andhra University, Waltair, whose ages ranged from 17 to 25 years. Household adults from Madras city constitute the samples for 35 and 36, whose ages ranged between 21 and 67 years. Samples from Kerala (37 to 39) came from school-going students at Ernakulam, their ages ranging from 6 to 16 years (mostly around 11 years). The 2 tribes of Orissa studied here are Gadaba and Parja (19 and 20). Samples for these two tribes were collected from and around Jeypore town, under Koraput district. They were all above 20 years of age. The 2 tribes of Andhra Pradesh studied here are Jatapu and Savara (21 and 22). These tribes were studied in the agency areas of Srikakulam district (around G. L. Puram), who were all adults. It may be noted that the tribes studied here (19 to 22 and 32) are mostly confined to the districts of Srikakulam and Koraput and that these two districts are adjacent to one another.

As a first step, the possible hereditary importance of tongue pigmentation is studied by comparing the rates of incidence in the pedigrees with those obtained through population studies [Table I(a)] for the same underlying populations. This analysis is clearly set out in Table II, where n_1 stands for the number of individuals in a pedigree who were personally examined for the trait; the rate of incidence in a pedigree (r_1) was computed by treating the doubtful cases (marked by ? in the pedigrees)

TABLE I

(*a*) DISTRIBUTION OF TONGUE PIGMENTATION IN SOME INDIAN ENDOGAMOUS GROUPS

No.	State	Population	Sex	Sample size n	Incidence rate $r(\%)$	Variance s^2
1	Maharashtra	Parsi	M	146	8.22	5.15
2	Maharashtra	C.K.P.	M	108	9.26	7.78
3	Maharashtra	Brahmin	M	571	13.13	1.99
4	Maharashtra	Maratha	M	832	17.07	1.69
5	Maharashtra	Mali	M	35	20.00	45.70
6	West Bengal	Brahmin	M+F	104	9.62	8.36
7	West Bengal	Kayastha	M+F	164	18.29	9.11
8	Orissa	Brahmin (Holua)	F	89	23.60	20.26
9	Orissa	Brahmin (Danua)	F	60	10.00	15.00
10	Orissa	Karan	F	42	16.67	33.07
11	Andhra Pradesh	Brahmin (Niyogi)	M	68	17.65	21.37
12	Andhra Pradesh	Brahmin (Vaidiki)	M	88	17.04	16.06
13	Andhra Pradesh	Vysya	M	37	13.51	31.58
14	Andhra Pradesh	Kamma	M	73	23.29	24.47
15	Andra Pradesh	Velama	M	13	23.08	136.56
16	Andhra Pradesh	Naidu	M	25	20.00	64.00
17	Andhra Pradesh	Kapu	M	44	29.54	47.30
18	Andhra Pradesh	Reddy	M	30	10.00	30.00
19*	Orissa	Gadaba	M+F	60	43.33	40.92
20*	Orissa	Parja	M+F	69	30.43	30.68
21*	Andhra Pradesh	Jatapu	M+F	160	37.50	14.65
22*	Andhra Pradesh	Savara	M+F	107	31.79	20.26

(*b*) DISTRIBUTION OF TONGUE PIGMENTATION IN SOME INDIAN POPULATIONS (MIXED)

23	Maharashtra	Others	M	260	15.77	5.11
24	Orissa	Sarua & Kottigiya Brahmin	F	18	5.56	29.17
25	Orissa	All	M	73	8.22	10.33
26	Andhra Pradesh	Other Brahmin	M	34	17.65	42.75
27	Andhra Pradesh	Kshatriya + Yadava + Sali	M	31	22.58	56.39
28	Andhra Pradesh	Christian	M	36	30.56	58.95
29*	Andhra Pradesh	Scheduled Caste	M	28	32.14	77.89
30	Andhra Pradesh	Unknown	F	61	19.67	25.90
31	Andhra Pradesh	Unknown	M	35	20.00	45.70
32*	Andhra Pradesh	Konda Dora + Jatapu Dora	M	25	24.00	72.96
33*	Andhra Pradesh	Harijan & Girijan	M	18	22.22	96.02
34	Andhra Pradesh	Unknown	F	35	28.57	58.37
35	Madras	Unknown	M	79	11.39	12.78
36	Madras	Unknown	F	79	26.58	24.70
37	Kerala	Christian	M	268	19.78	5.90
38	Kerala	Hindu	M	231	24.24	7.95
39	Kerala	Others	M	32	12.50	34.22

* Economically backward communities.

and the unexamined but pigmented cases (indicated by partial dark shade in the pedigrees) as normal ones; s_1^2 corresponds to the (sample) variance of the rate of incidence; n_2, r_2, and s_2^2 have similar meaning for the corresponding population study.

Out of the total of 9 pedigrees analysed in Table II, 5 show significantly higher rates of incidence than those at the population level, thereby providing good evidence to suspect some genetic basis for the trait. Also, following Cochran (1954), one can combine the results of all the 9 contingency tables (2×2) given in Table II by computing the overall statistic,

$$\gamma = \frac{\Sigma w(p_1 - p_2)}{\sqrt{\Sigma wp(1-p)}}$$

where, being $p_i = r_i/100$, p is the overall proportion of tongue pigmentation (pedigree and population data combined) for each contingency table,

$$w = n_1 n_2/(n_1 + n_2)$$

and the summations are taken over all the 9 contingency tables. This statistic was proposed by Cochran to be tested as a normal deviate, and a good discussion on its performance can be found in Radhakrishna (1965). We get the value of this statistic as 6.59 which exhibits a significantly higher rate of incidence in the pedigrees than in the population studies.

This overall test provides even stronger evidence in favour of a genetic basis for the trait than the individual tests of Table II (P is nearly 0). Further, since we identified only two phenotypes, viz. pigmented and normal, there is enough reason to think in terms of only a single-locus hypothesis for the trait. Also, we shall confine ourselves to a diallelic system, though multiallelic systems cannot be ruled out.

Thus, let the two alleles be A and a. In what follows, we shall refer to only Figs. 4-10. Discussion on Figs. 1-3 may be found in Rao (1970*a*, *b*). It is abundantly clear from the pedigrees that a Y-linked or a sex-limited hypothesis cannot be supported as a possible genetic hypothesis for the inheritance of the trait.

As an alternative, let us consider an X-linked recessive hypothesis (a being the recessive allele). It is then clear that all the sons should be affected (pigmented, X_aY) and all the daughters should be normal (X_AX_a) from each mating of the type "affected female $\times$ normal male" ($X_aX_a \times X_AY$). But all the pedigrees provide evidence against such an expectation, thereby totally rejecting X-linked recessivity.

Similarly, under X-linked dominance, we expect all the sons to be normal (X_aY) and all the daughters to be affected (X_AX_a) in each mating of the type "normal female $\times$ affected male" ($X_aX_a \times X_AY$). Neither this expectation is realised in the pedigrees.

Next, let us consider an autosomal dominant hypothesis. Under this, all the children out of "normal $\times$ normal" ($aa \times aa$) matings are expected to be normal (aa). This is not supported by the pedigrees, though there are some individual families in the pedigrees that go to the credit of such a hypothesis.

Finally, we observe that all the pedigrees strongly support an autosomal recessive

hypothesis, though some observations are contrary to such a hypothesis: there are several " affected × affected " (*aa* × *aa*) matings in the pedigrees, and we examined 25 children out of such matings (Figs. 4-10) with the result that 9 turned out to be normal (*AA* or *Aa*) children. Though this proportion turns out to be very high (nearly 1/3), such " exceptional " cases may be disposed of on some grounds like illegitimacy (which is believed to be very high in these pedigrees) or some variation in the genic action (like incomplete penetrance for the recessive allele). Concerning the former possibility, it should be admitted here that no serological tests could be carried out. The latter possibility (incomplete penetrance) is known to play some role though not to a great extent (Rao and Ghorai 1970). Thus, we conclude that the most probable genetic hypothesis for the inheritance of tongue pigmentation is diallelic autosomal recessivity, with rare failure of penetrance for the recessive allele.

TABLE II
COMPARISON OF INCIDENCE RATES OBTAINED THROUGH PEDIGREE AND POPULATION STUDIES

Population studied	Pedigree study				Population study			χ_1^2
	Figure no.	n_1	r_1	s_1^2	n_2	r_2	s_2^2	
Hindu (Kerala)	1	78	38.46	30.34	231	24.24	7.95	5.28
Hindu (Kerala)	2	49	67.35	44.88	231	24.24	7.95	35.18
Brahmin (Madras)	3	15	26.67	130.38	158[a]	18.99	9.74	0.42
Sali (Andhra Pradesh)	4	54	16.67	25.72	31[b]	22.58	56.39	0.42
Kapu (Andhra Pradesh)	5	46	32.61	47.77	44	29.54	47.30	0.10
Jalari (Andhra Pradesh)	6	122	27.05	16.17	—	—	—	—
Vysya (Andhra Pradesh)	7	80	25.00	23.44	37	13.51	31.58	2.40
Yadava (Andhra Pradesh)	8	26	50.00	96.15	31[b]	22.58	56.39	4.93
Kapu (Andhra Pradesh)	9	51	62.74	45.84	44	29.54	47.30	11.83
Reddy (Andhra Pradesh)	10	51	33.33	43.57	30	10.00	30.00	7.40

[a] Corresponds to a mixed sample from Madras (no. 35 in Table I).
[b] Corresponds to a mixed sample from Andhra Pradesh (n. 27 in Table I).

Concerning the distribution of the trait, the rates of incidence (r) and the corresponding variance (s^2) are presented in Table I. These rates of incidence may be compared for any two populations (testing for the equality) by using the statistic,

$$(r_1 - r_2)^2/(s_1^2 + s_2^2)$$

which follows χ^2 distribution with 1 *DF*. A glance at Table I reveals a great deal of variation in the incidence rates, thus suggesting it as an anthropological marker in human populations. Also, one discovers from Table I several clusters or homogeneous groups of populations. For example, all the four tribes (nos. 19 to 22) tend to cluster around a single point.

Finally, we make an attempt here to investigate whether or not there is any association between the rate of incidence and the skin colour, a problem that was raised by Davis (1968). To do this, we consider 4 population groups from Table I(*a*) in the order of decreasing intensity of fairness of skin colour. They are the Parsis, West Bengal Brahmins, Andhra Pradesh Brahmins (pooled over Vaidiki and Niyogi), and all the four tribes (together). A comparison of the incidence rates among these 4 population groups (4 × 2 contingency table) yields $\chi^2 = 65.35$ (3 *DF*, *P* is nearly zero). This provides strong evidence in favour of an association between the incidence rate and skin colour. It is also clear that the incidence rates are higher for the dark skinned populations than those for the fair skinned. And, since the skin colour is controlled by the amount of melanin pigment, it appears that tongue pigmentation and melanin are closely associated, if not the same. We hope to be able to report our findings on this aspect shortly.

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RIASSUNTO

È stato condotto uno studio genetico sulla pigmentazione della lingua nell'uomo, sulla base di 10 alberi genealogici e di 39 popolazioni dell'India. Si conclude in favore di una base genetica di tale carattere, il quale sarebbe trasmesso con un meccanismo recessivo autosomico.

RÉSUMÉ

Une étude génétique sur la pigmentation de la langue chez l'homme a étée conduite sur la base de 10 arbres généalogiques et de 39 populations indiennes. L'on conclut en faveur d'une base génétique de ce caractère, dont la transmission aurait lieu avec un mécanisme autosomal récessif.

ZUSAMMENFASSUNG

Eine Erbforschung über die Pigmentierung der Zunge beim Menschen wurde anhand von 10 Stammbäumen und 39 Bevölkerungen in Indien vorgenommen. Man schliesst daraus, dass eine Erbbasis mit autosomem rezessiven Erbgang besteht.

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