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TASTE SENSITIVITY TO PTC AND COLOUR BLINDNESS IN ESTONIANS

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ABSTRACT

The article discusses the genetic characterisation of Estonians on the basis of the traits of PTC and colour blindness in Estonian population samples from various parts of Estonia.

The taste sensitivity to PTC was studied in 2559 Estonian school-children from 24 localities and of colour blindness in 4300 males from 33 localities. Results: The frequency of nontasters was 25.4% on average; *t* gene frequency (50.4% for Estonians) varied between 36% and 65%. PTC nontasters frequency is higher on West-Estonian islands and in Western Estonia compared to other parts of Estonia. The frequency of colour blindness *cb* gene (5.3% for Estonians) is higher in North, North-East and East Estonia (6–8%); its mean frequency on the rest of the territory is lower. The genetic diversity in various traits seem to be a trace of the historical development of the Estonian nation.

***Key words*:** *PTC, colour blindness, population genetics, Estonians*.

INTRODUCTION

Taste sensitivity to phenylthiocarbamide (PTC) and colour blindness belong to those physiological traits that directly prove genetic diversity of people concerning the perception of the world; they are highly informative genetic markers in the studies of human diversity.

Determing taste sensitivity to PTC begun as early as in 1931, after the chemical substance was synthesized by chemist A. L. Fox. On describing the taste of PTC it appeared that the people tested were divided into two groups; most people tasted it bitter, but to others it was

without taste. So it was possible to discover polymorphism in the taste sensitivity to PTC, and to divide the human population into "tasters" and "nontasters" [4].

Later the ability to taste the PTC was found to be inherited as a simple Mendelian dominant trait, and nontasting is a simple recessive characteristic, but there are references that the threshold of taste sensitivity is higher in heterozygotes than homozygotes [8]. For studying taste sensitivity, a standard method using serial dilutions of PTC was developed. It appeared that the "tasters" varied greatly according to their sensitivity to taste PTC; everybody has their own threshold (the lowest concentration of PTC where the bitter taste appears), and taste sensitivity in populations has a bimodal distribution, one mode – "tasters", the other – "nontasters" [7].

Recent genetic PTC studies have revealed that the ability to taste PTC or not is conveyed by a single gene that codes for a taste receptor on the tongue. The PTC gene, TAS2R38, was discovered in 2003 [15]. Phenotypic variance in PTC sensitivity is accounted for the presence of just two common alleles: a tasting allele and a non-tasting allele, and the frequencies of these alleles in human populations correspond well to frequencies estimated from phenotype data [21].

Inability to clearly identify different colours of the spectrum is widely known as colour blindness. An individual with normal colour vision is capable to distinguish all the primary colours and blend them into different tones of colours. In the case of partial colour blindness, an individual does not distinguish clearly, in most cases, between red and green colour. These colour vision deficiencies are named protanomaly and deuteranomaly, but the inability to perceive red and green colours – protanop and deuteranop. Red-green colour blindness is a sex-linked trait; the corresponding genes are situated in the X chromosome, but normal colour vision is dominant in relation to colour blindness. Therefore, colour blindness is expressed mainly in men. Red and green colour pigments are present at the tip of the long arm of the X chromosome Xq28 [17].

In the distribution of colour blindness and PTC taste sensitivity, regional and racial differences occur. Colour blindness is more frequent among the Caucasoid peoples than among the Mongoloids [9]. PTC nontasers appear more frequently among Caucasoid populations. In Mongoloid populations, as Japanese and Chinese, nontasters are con-

siderably rare and, vice versa, tasters are found there much more frequently [18].

These data have also been referred to in studies of Estonians, and erroneous statements about the occurrence of PTC nontasters in Estonia have been made; the corresponding studies still have to be started [19]. Population genetic studies of 10 polymorphic systems (blood groups, PTC, colour blindness) of the Estonian population have been studied by the author of the present paper, and genetic analyses have been given earlier [11, 12, 5:570, 6:622]. The aim of the current study is to give an overview of the diversity of taste sensitivity to PTC and colour blindness among Estonians.

MATERIAL AND METHODS

The material used in the present study was gathered by the author mainly during the anthropological expeditions of the Institute of History of the Academy of Sciences in the 1970s and 1980s. The subjects were of Estonian descent; their parents and grandparents came from the same district. Tests were carried out on both schoolchildren and grown-up men. In the case of taste sensitivity to PTC, only the schoolchildren's material (2559 individuals from 24 regional locations) was used to avoid possible errors in the case of older people, and so that the data would be wholly comparable all over Estonia. The ability to detect the bitter taste of PTC was tested using 15 concentrations of PTC solutions, following the technique of Harris and Kalmus [7]. The concentrations of the solutions, in boiled tap water, were obtained by means of the formula $2.6 \times 2\text{-}n$ g/l (where n is the number of the solution), whereby in the case of the strongest concentration n=0, in the weakest – n=15. Tasting was started from the solution with the lowest concentration (no.15). Between every different solution, pure water was given. The threshold for each subject was the lowest concentration at which he was able to distinguish the PTC solution from pure water. Approximately 2% of schoolchildren did not even feel the bitter taste of the strongest solution (n=0). These people are marked with a negative sign in Fig.1. The distribution of the thresholds shows a typical bimodal shape (Fig.1). The threshold for schoolchildren was solution no. 5. The subjects who felt the taste of PTC solutions no. 15–6 were regarded as "tasters", of no. 5–0 as "nontasters".

Colour blindness was studied among 4300 males from 33 districts. The colour vision test for red-green colour deficiency among men and schoolboys was carried out by using polychromatic tables by J. Rabkin [24]. The test was conducted in daylight inside a room, avoiding direct sunlight.

RESULTS AND DISCUSSION

As regards to PTC tasting ability, the data on Estonians exibit a bimodal distribution of the threshold (Fig. 1). Among the 2559 individuals studied, 74.6% were tasters, 25.4% did not feel the bitter taste of PTC. The percentage and gene frequency of nontasters is given in Table 1. The frequency of nontasters of PTC, *t* gene, varies in Estonian different local samples between 36% and 65%, with the mean frequency 50.4%) (Table 1, Fig. 2). In the West Estonian islands the *t* gene frequency is higher (60%) in comparison to the other parts of Estonia. It is also comparatively high in some other westernmost parts of West and South-West Estonia (56–57%). In the East Estonian region (in Alatskivi), in the area between Lake Võrtsjärv and Lakes Peipsi-Pihkva (Pskov) and in some locations of inner Estonia, the frequency is lower (40–53%) (Fig.2).

Frequency of nontasters of PTC in Estonians is lower in comparison with the Finns; however, on the West Estonian islands and on the West Estonian coast, the percentage of nontasters of PTC is higher, being similar to that of other Finno-Ugric peoples, such as Komis, Maris, Hungarians but also non-Finno-Ugric peoples like Lithuanians, Russians, Swedes, et al. (Table 1, Table 2). Frequency of *t* gene in Latvians from Varakļāni is more similar to that of West-Estonian islands, from Alūksne to that of the South-East Estonian population. The frequency of nontasters is somewhat lower in Lapps from Inari and Kola Peninsula. The frequency of *t* gene is lower in the Mongoloid peoples – in Evenks 24%, Chinese 21%, etc. At that, taste sensibility to PTC is much higher in Mongoloid peoples who distinguish even a very weak solution (no.28) from pure water [25].

Table 1. Frequency of PTC nontasters (% and *t*-gene)

Population group	N	Phenotype		Genotype	
		n	%	*t*	σ
1. Kuressaare	116	38	32,76	.5724	.0381
2. Orissaare	113	47	41.59	.6449	.0359
3. Muhu	103	35	33.98	.5829	.0400
4. Haapsalu	110	36	32.73	.5721	.0391
5. Lihula	107	21	19.63	.4431	.0433
6. Tõstamaa	75	21	28.00	.5292	.0489
7. Audru	80	20	25.00	.5000	.0484
8. Pärnu-Jaagupi	99	31	31.31	.5596	.0416
9. Märjamaa	100	15	15.00	.3875	.0461
10. Kehra	100	22	22.00	.4690	.0441
11. Kunda	109	28	25.69	.5069	.0413
12. Iisaku	154	34	22.08	.4699	.0356
13. Järva-Jaani	83	13	15.66	.3957	.0504
14. Suure-Jaani	101	43	42.57	.6525	.0377
15. Alatskivi	92	17	18.48	.4299	.0471
16. Elva	96	21	21.88	.4677	.0451
17. Võnnu	67	15	22.39	.4732	.0538
18. Põlva	224	59	26.34	.5132	.0286
19. Värska	106	17	16.04	.4005	.0445
20. Meremäe	123	34	27.64	.5257	.0383
21. Valga	98	13	13.27	.3642	.0470
22. Abja	97	20	20.62	.4541	.0437
23. Viljandi	104	18	17.31	.4161	.0446
24. Kilingi-Nõmme	102	32	31.37	.5601	.0410
1–24 in total	2559	650	25.40	.5040	.0085

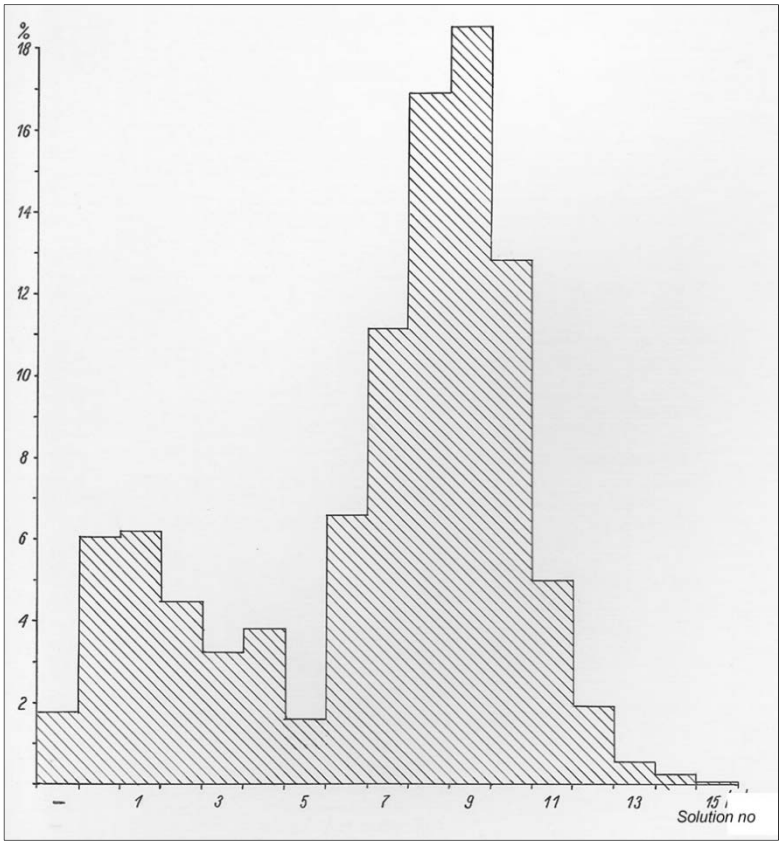

Fig. 1. Taste sensitivity to PTC.

Table 2. Frequency of PTC nontasters in various populations (% and *t*-gene)

Population	N	%	*t*-gene	Sources
Estonians	2559	25.4	.504	[11]
Finns	811	35.0	.592	[14]
Finns (Helsinki)	202	29.2	.541	[1]
Finns (Oulu)	761	22.1	.470	[2]
Lapps (Inari)	184	15.8	.396	[23]
Lapps (Kolta)	149	36.2	.602	[23]
Lapps (Kola Peninsula)	124	12.5	.353	[28]
Vepsians	176	28.4	.533	[11]
Komi	302	36.8	.606	[23]
Mari	321	26.5	.514	[23]
Hungarians	401	32.2	.567	[3]
Hungarians (Transcarpathian)	203	31.5	.562	Author's data
Latvians (Alūksne)	109	24.8	.498	[11]
Latvians (Varakļāni)	109	43.1	.657	[11]
Lithuanians	163	31.9	.565	[26]
Belarusians	2694	35.6	.597	[26]
Russians	245	32.1	.567	[22]
Swedes (Åland)	124	33.1	.575	[13]
Swedes	509	33.8	.581	[20]
Kyrgyz	640	19.6	.443	[22]
Evenks	137	5.8	.241	[25]
Chinese	239	4.6	.214	[16]

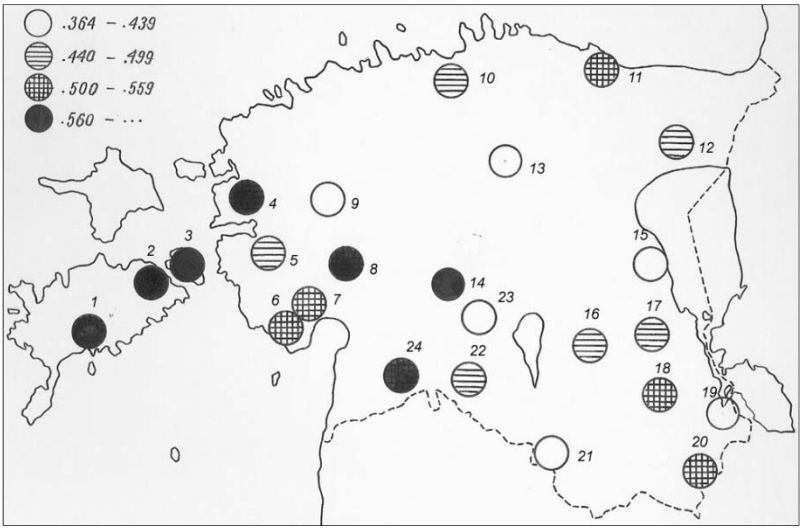

Fig. 2. Distribution of nontaster (*t*) gene.

Colour blindness. In Estonia, the colour blindness has been studied among male population (grown-up men and schoolboys). For testing colour blindness in Tallinn schools, 7.17% of schoolboys from 293 tested were found to be colour blind; among them – deuteranomaly 3.1%, protanamaly 1.35%, deuteranop 1.36% and protanop 1.36% [10]. These data correspond well with the earlier data by R. I. Serebrovskaya (in 235 males 3.0% of deuteranomaly and 1.3% of protanomaly) [27].

Table 3 shows the percentage of incidence of colour blindness. The frequency of colour blindness varies in different Estonian local samples between 2% and 9%, with the average 5.3%. The frequency is higher in East and North-East Estonia (6–8%). The frequency of *cb* gene in West Estonia is comparatively high as well (6,2%) and its frequency decreases towards South-East Estonia (Fig. 3). The whole South-East is characterised by low occurrence of colour blindness (on average 4.3%). It is low on the West Estonian islands and in Central Estonia as well.

Table 3. Frequency of colour blindness

Population	N	Phenotype		Genotype	
		n	%	*cb*	σ
1.Saaremaa, Muhu	382	14	3.66	.0366	.0096
2.Hiiumaa	102	5	4.90	.0490	.0214
3.Haapsalu	210	13	6.19	.0619	.0166
4.Lihula	158	9	5.70	.0570	.0184
5.Tõstamaa	43	4	9.30	.0930	.0443
6.Audru, Pootsi	178	11	6.18	.0618	.0180
7.Pärnu-Jaagupi	132	8	6.06	.0606	.0208
8.Rapla	182	8	4.40	.0440	.0152
9.Keila	137	8	5.84	.0584	.0200
10.Tallinn	293	21	7.17	.0717	.0151
11.Kehra	75	5	6.67	.0667	.0288
12.Rakvere	99	8	8.08	.0808	.0274
13.Kohtla-Järve	99	5	5.05	.0505	.0220
14.Iisaku	147	10	6.80	.0680	.0208
15.Väike-Maarja	100	8	8.00	.0800	.0271
16.Paide	94	2	2.13	.0213	.0149
17.Suure-Jaani	56	3	5.36	.0536	.0301
18.Põltsamaa	101	4	3.96	.0396	.0194
19.Jõgeva	65	5	7.69	.0769	.0330
20.Alatskivi	55	4	7.27	.0727	.0350
21.Elva	91	4	4.40	.0440	.0215
22.Otepää	99	3	3.03	.0303	.0172
23.Võnnu	37	2	5.40	.0540	.0372
24.Põlva	295	15	5.08	.0508	.0128
25.Antsla	102	5	4.90	.0490	.0214
26.Võru	103	5	4.85	.0485	.0212
27.Värska	49	3	6.12	.0612	.0342
28.Petseri	102	2	1.96	.0196	.0137
29.Meremäe	210	8	3.81	.0381	.0132
30.Valga	105	6	5.71	.0571	.0226
31.Abja, Karksi	196	14	7.14	.0714	.0184
32.Viljandi	100	3	3.00	.0300	.0171
33.Kilingi-Nõmme	103	5	4.85	.0485	.0212
1–33 in total	4300	230	5.35	.0535	.0034

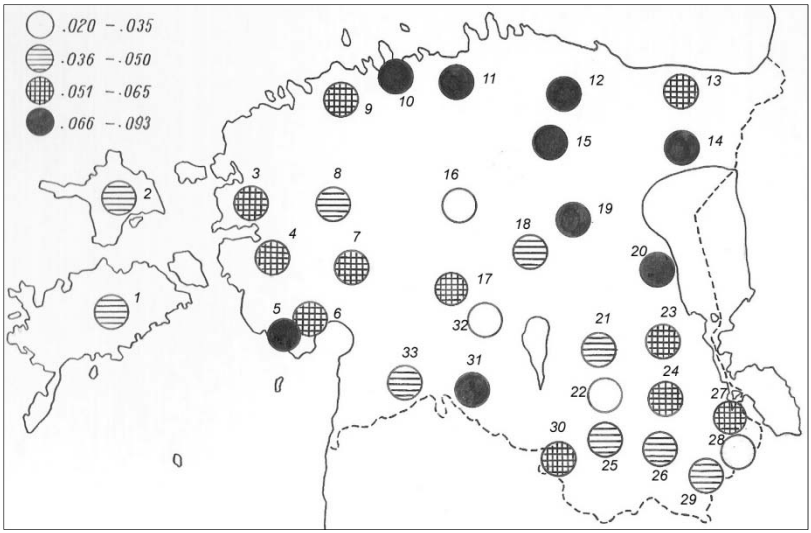

Fig. 3. Distribution of colour blindness (*cb*) gene.

In the distribution of colour blindness, regional and racial differences occur. In Europoid populations colour vision deficiency is higher than in Mongoloids [9]. In comparison with some other European populations, the mean percentage of Estonians' colour blindness is comparatively low (Table 4). Frequency of colour blindness in Latvians in Alūksne and Varakļāni is similar to that of Central and South-West Estonians. On the territory of Northern Eurasia the mean frequency is 5.5%, varying in the limits of 0.0%–24.0% [6:303]. Territory with the frequency below the mean (0.0%–4.3%) embraces the central and north-eastern regions of Siberia. Towards north and south and southwest the frequency increases. The highest frequency of colour blindness is found on the territory of Belarus with a nucleus in Grodnensk region – 24.1% [6:303].

Table 4. Frequency of colour blindness in various populations

Population	N	%	Sources
Estonians	4300	5.3	[11]
Estonians	235	6.2	[27]
Latvians (Alūksne)	90	4.4	[11]
Latvians (Varakļāni)	76	4.0	[11]
Vepsians	103	7.8	Author's data
Hungarians (Transcarpathia)	201	4.5	Author's data
Norwegians	9049	8.0	[9]
Scots	464	7.8	[9]
Germans	1000	7.5	[9]
American Indians	392	2.0	[9]
Congo Blacks	929	1.7	[9]

Thus, the survey showed genetic heterogeneity in the traits of taste sensitivity to PTC and colour blindness in Estonians; the greatest genetic differences were observed in the West-East direction (as in the other genetic characteristics of polymorphic systems [11,12]). On the West Estonian islands and on the West Estonian coast the percentage of nontasters of PTC is higher than anywhere else in Estonia. Frequency of colour blindness is higher in North, North-East and East Estonia; its mean frequency on the rest of the territory is lower. The genetic diversity in various traits seem to be a trace of the historical development of the Estonian nation.

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