«ИЗУЧЕНИЕ ПОЛИМОРФИЗМА Q-ГЕТЕРОХРОМАТИНОВЫХ РАЙОНОВ ХРОМОСОМ В ПОПУЛЯЦИЯХ, ПРОЖИВАЮЩИХ В РЕСПУБЛИКЕ КАЗАХСТАН».

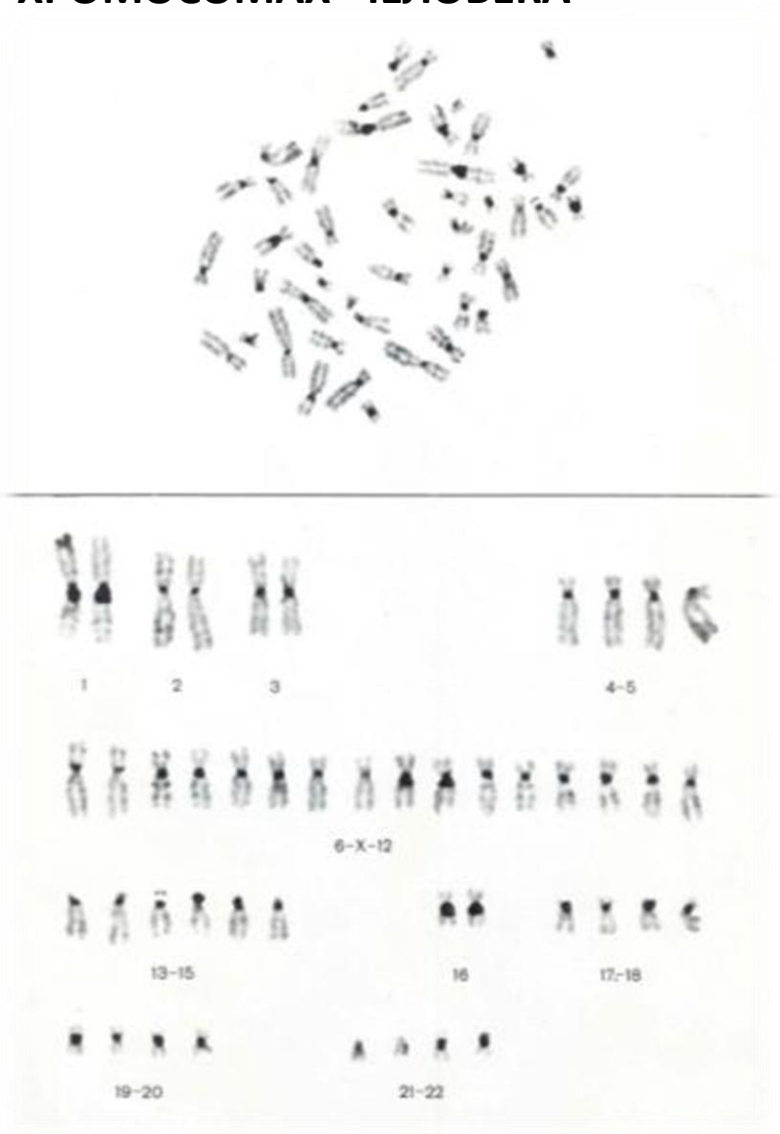
Расшифровка генома (физическая карта) показала, что гены составляют всего лишь 1.5 - 2% ДНК хромосом человека, остальные ДНК представляют собою бессмысленные последовательности молекул азотистых оснований, то есть не содержат никакой генетической информации

Инициаторы Проекта «Геном человека» ожидали обнаружить 100 000 - 140 000 генов. Оказалось, что на самом деле наш геном содержит всего лишь около 30 000 генов. Эта цифра, действительно, очень скромна она в два раза больше, чем у плодовой мушки и чуть меньше, чем у риса, который имеет около 40 000. В смысле количестве генов, мы занимаем место рядом с *Arabidopsis*, сорного растения, имеющего в своем геноме примерно 26 000 генов.

Полное картирование генома человека показал, что 98% ДНК хромосом представляют собою последовательности молекул азотистых оснований, не содержащие никакой генетической информации. Из них около 20% формируют структуры, видимые под оптическим микроскопом, и получили называние гетерохроматин.

В настоящее время мы умеем различить два вида гетерохроматина. Первый вид гетерохроматина получил название C-гетерохроматин (от слово – constitutive heterochromatin) и он имеется в геноме у всех высших эукариот, включая человека.

С – ГЕТЕРОХРОМАТИНОВЫЕ РАЙОНЫ ИМЕЮТСЯ НА ВСЕХ БЕЗ ИСКЛЮЧЕНИЯ ХРОМОСОМАХ ЧЕЛОВЕКА



Другой вид гетерохроматина называется Q-гетерохроматин (от слово Quinacrine – акрихин), с помощью которого выявляется этот вид гетерохроматина.

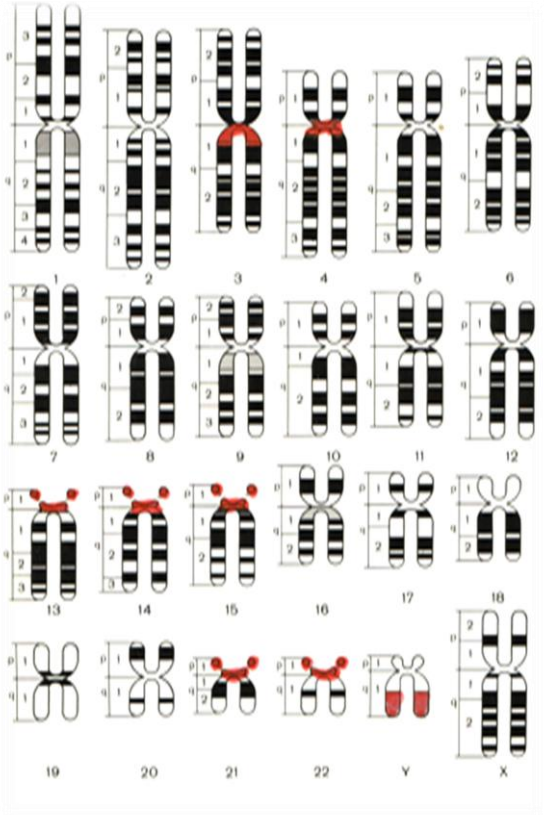
Q-гетерохроматин имеется в геноме только трех высших приматов:

человек, шимпанзе и горилла.

В кариотипе человека Q- гетерохроматин *можно обнаружить* только 7 парах аутосом: 3, 4, 13, 14, 15, 21, 22 и на Y хромосоме (обозначены красным цветом)

Q – ПОЛИМОРФНЫЕ ЛОКУСЫ

ХРОМОСОМ ЧЕЛОВЕКА



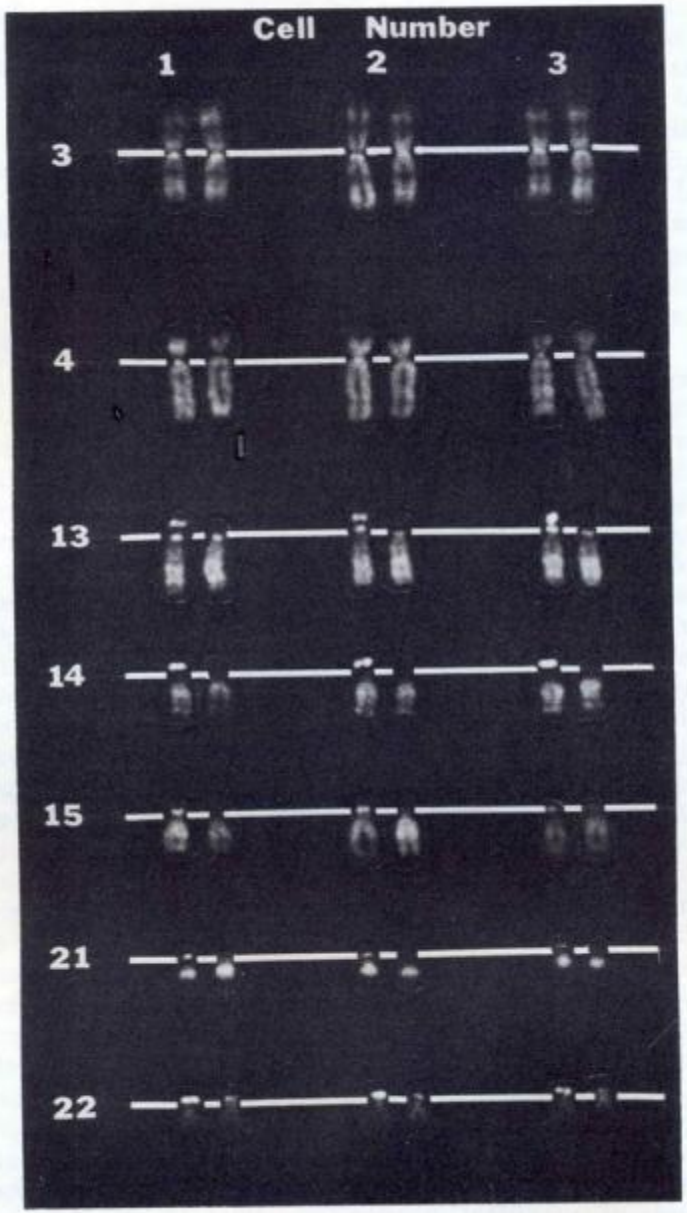
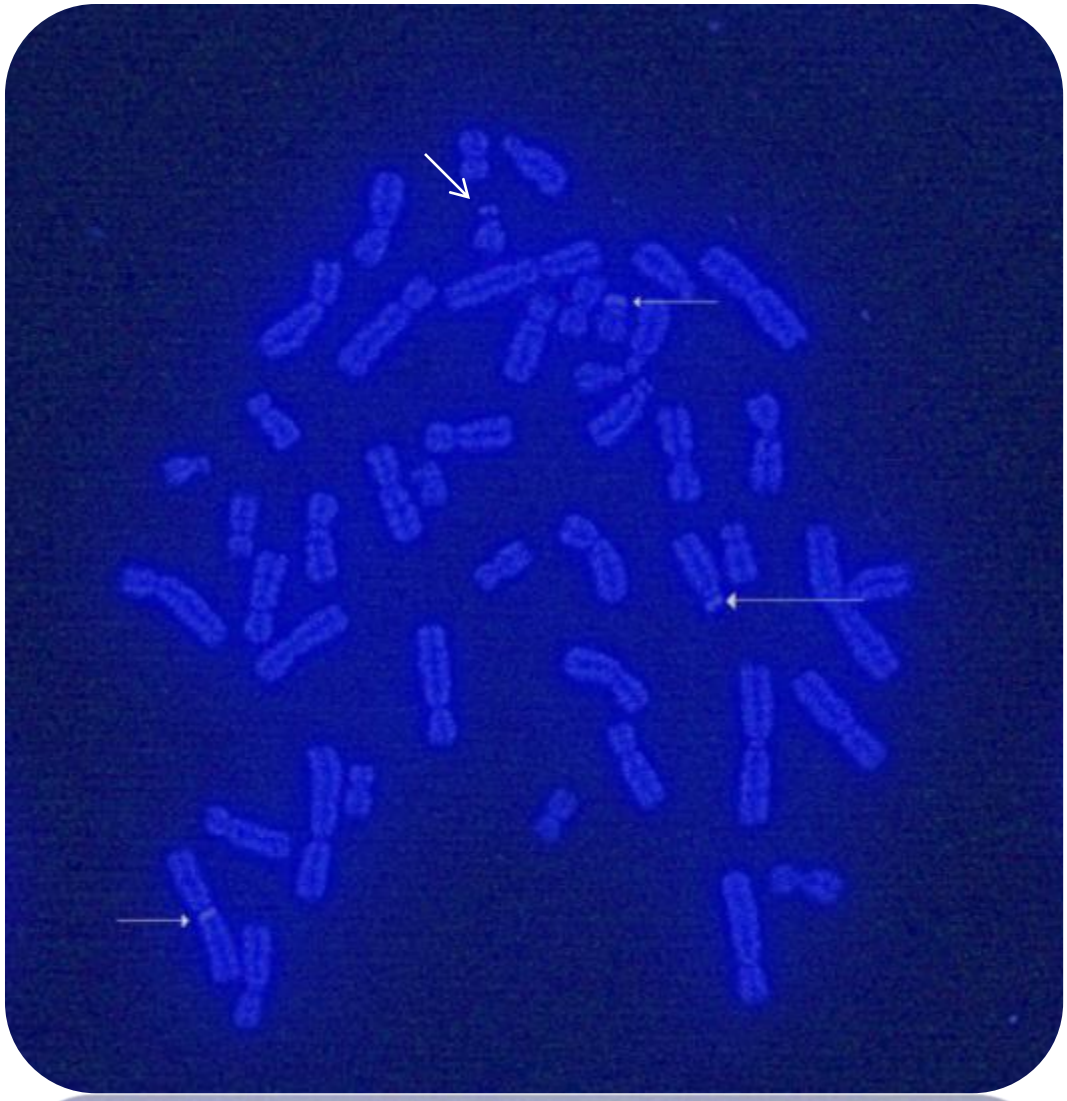


Figure 7.1. Heteromorphic regions as demonstrated by the QFQ technique.

Q – ГЕТЕРОХРОМАТИНОВЫЕ РАЙОНЫ ХРОМОСОМ (УКАЗАНЫ СТРЕЛКАМИ)



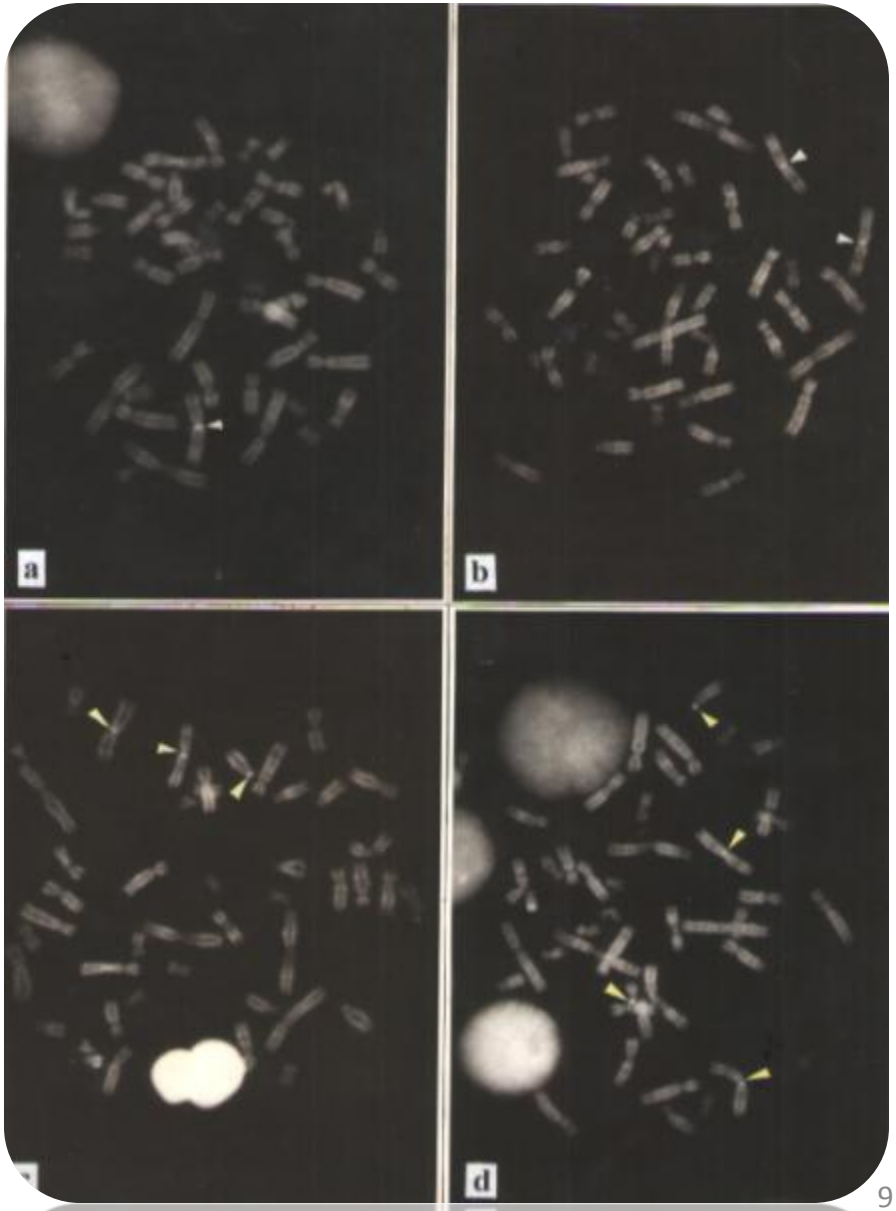
КОЛИЧЕСТВО

Q – ГЕТЕРОХРОМАТИНОВЫХ

РАЙОНОВ (Q-ГР) ХРОМОСОМ У

ИНДИВИДОВ В ПОПУЛЯЦИИ

КОЛЕБЛЕТСЯ ОТ 0 ДО 10

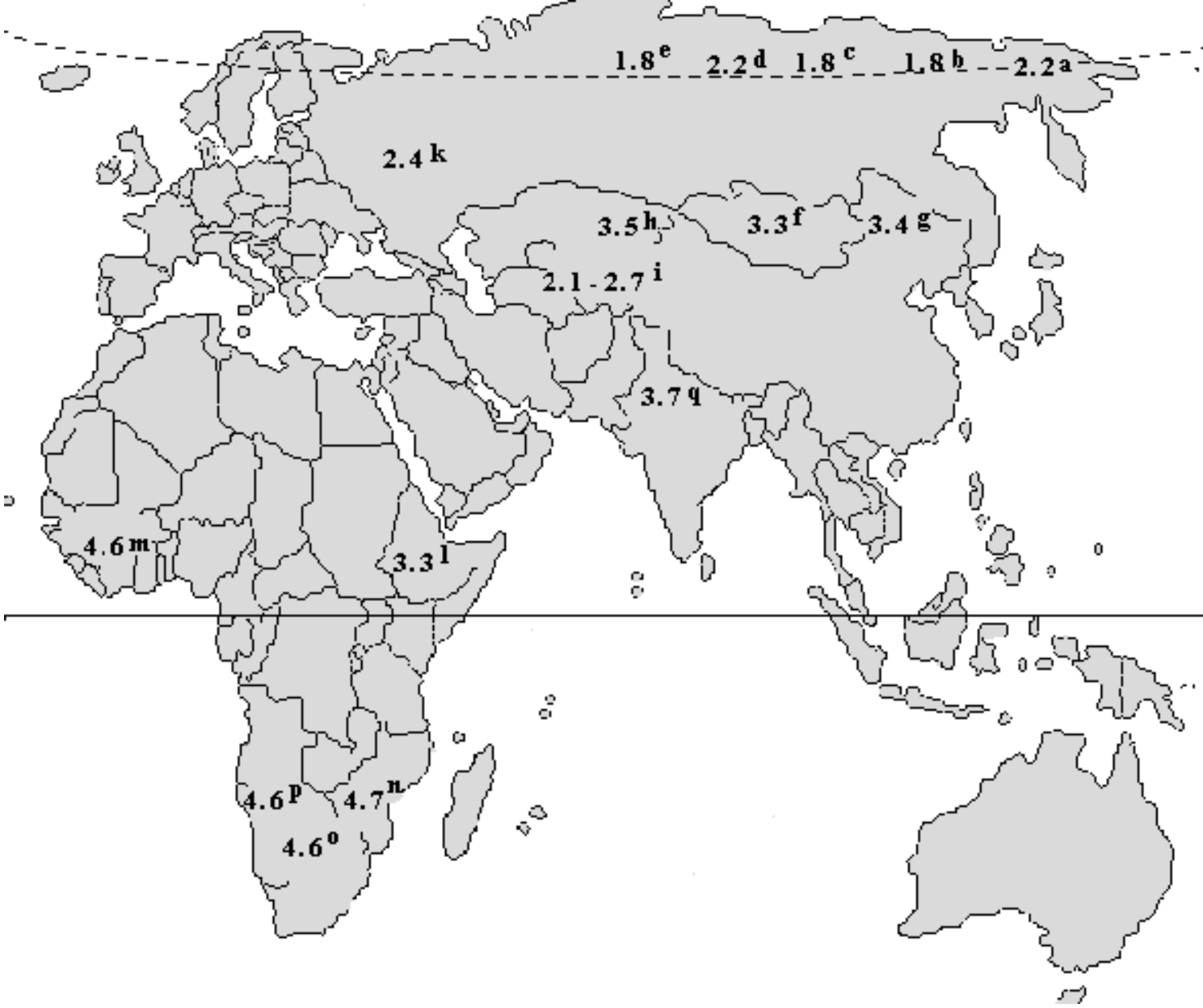


Distribution of the number and mean number of Q-HRs per individual in populations living in Eurasia and Africa.

Number of Q-HRs	1	2	3	4	5
0	57	58	55	10	
1	116	123	114	13	1
2	141	175	186	51	21
3	56	99	136	85	54
4	13	51	69	58	78
5	2	14	32	49	79
6			9	24	55
7			2	3	29
8				4	6
9					3
10					1
Mean number of Q-HRs	1,63±0,05	2,01±0,05	2,31±0,06	3,49±0,09	4,68±0,08

1 – mountaineers (n=385); 2 – Northern Mongoloids (n=520); 3 – Highland Mongoloids(n=603); 4 – Stepper Mongoloids (n=297); 5 – Lowland Negroids (n=327).

The mean number of Q-HRs per individual in the native populations of Eurasia and Africa.



a = Chukchi of Chukotsk (n = 132); b = Yakuts of Yakut ASSR (n = 127); c = Selkups of eastern Siberia (n = 90); d = Nenets of eastern Siberia (n = 117); e = Khants of eastern Siberia (n = 54); f = Mongolians of the MPR (n = 72); g = Chinese of northern China (n = 124); h = Kazakhs of southern Kazakhstan (n = 101); i = Kirghiz of Pamir and Tien Shan (n = 603); k = Russians of Bishkek (n = 200); l = Ethiopians of Ethiopian uplands (n = 52); m = Guinea-Bissau Negroes (n = 13); n = Mozambique Negroes (n = 148); o = Zimbabwe Negroes (n = 34); p = Angola Negroes (n = 132); q = Indians of northern India (n = 58).

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C- and Q-band polymorphisms in the chromosomes of three human populations

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SUMMARY

The incidence of the various classes of C-band and Q-band polymorphisms on ten pairs of chromosomes in the human complement have been investigated in three Scottish populations, two from the mainland (newborn and 14 year olds) and one, of individuals over 65 years, from an island in the Outer Hebrides. Although there is an overall similarity between the populations, there are some differences, especially with the island group. For all populations, over 90% of the C-band size variants fall within the medium class. Rather more variation is found in the Q-band intensity polymorphisms: the island population appears to have fewer Brilliant and Intense variants than do the other two groups, 2·9 per person as compared to 4·2 and 3·9 for the newborn and 14 year olds respectively; this may be an age difference rather than a population difference.

The authors wish to thank Dr A. J. Keay and Dr J. Syme for access to population A; Dr R. C. MacNair for access to population B; the staff of the Registry of Abnormal Karyotypes, M.R.C. Clinical and Population Cytogenetics Unit, for their work on population C; the technicians of the Cytogenetics Section, MRC Clinical and Population Cytogenetics Unit, and Mrs R. Hill for expert assistance.

Distribution of the number and mean number of Q-HRs per individual in Kyrgyz subjects of different age

Number of Q-HRs	Populations									
	I		II		III		IV		V	
	Neonates		7 to 19 years old		20 to 39 years old		40 to 59 years old		60 years old and over	
	(n=145)		(n=317)		(n=112)		(n=67)		(n=23)	
0	4		21		7		8		3	
1	19		70		20		13		4	
2	23		105		41		20		11	
3	38		71		19		14		2	
4	37		39		16		10		3	
5	16		9		7		2			
6	5		2		2					
7	3									
Total	458		706		270		145		44	
Mean number of Q-HR	3.16	0.13	2.23	0.07	2.41	0.13	2.16	0.16	1.91	0.24

Distribution of the number and mean number of Q-HRs per individual in neonates and infants died

Number of Q-HRs	Kyrgyz		Russians	
	Neonates	Infants died	Neonates	Infants died
	I	II	III	IV
	(n=145)	(n=17)	(n=37)	(n=5)
0	4			
1	19		3	
2	23		7	
3	38		5	
4	37		12	
5	16	9	7	2
6	5	5	3	2
7	3	3		1
Total	458	79	133	24
Mean number of Q-HRs	3.16±0.13	4.58±0.23	3.59±0.23	4.8±0.37

Цель работы –

определить количественное содержание Q-гетерохроматиновых районов в геноме популяций человека, проживающих постоянно в Республике Казахстан.

Распределение чисел и среднего числа Q-ГР хромосом у индивидов казахской, русской и уйгурской национальностей в возрасте 18 -30 лет г. Алматы

Число Q-ГР хромосом	Популяции		
	Казахи (n = 162) I	Русские (n = 33) II	Уйгуры (n = 40) III
1	15 (10.7)	2 (6.0)	1 (2.5)
2	22 (13.7)	6 (18.2)	5 (12.5)
3	43 (24.4)	11 (33.3)	7 (17.5)
4	38 (25.2)	5 (15.2)	12 (30.0)
5	30 (18.3)	6 (18.2)	8 (20.0)
6	10 (6.1)	2 (6.0)	4 (10.0)
7	4 (1.5)	1 (3.)	2 (5.0)
8	-	-	1 (2.5)
Всего Q-ГР хромосом	578 (99.9)	116 (99.9)	166 (100.0)
Среднее число Q-ГР хромосом	3.57 ± 0.168	3.51 ± 0.254	4.15 ± 0.244
Статистика	$t_{I, II} = 0.080$; df = 153; P = 0.936; $t_{I, III} = 2.410$; df = 160; P = 0.017; $t_{II, III} = 1.791$; df = 71; P = 0.078;		

Распределение чисел и среднего числа Q-ГР хромосом у казахов с. Курдай и г. Алматы.

Число Q-ГР хромосом	Казахи – студенты (Алматы - 2013 г) (n = 162)	Казахи – школьники (с.Курдай – 1982 г) (n = 101)
1	15 (11.5)	4 (4.5)
2	22 (13.9)	13 (14.7)
3	43 (25.4)	15(17.0)
4	38 (22.1)	13 (14.7)
5	30 (18.8)	26 (29.35)
6	10 (6.6)	11 (12.5)
7	4 (1.6)	6 (6.8)
Всего Q-ГР хромосом	578 (99.9)	365 (99.9)
Среднее число Q-ГР хромосом	3.57 ± 0.114	3.61 ± 0.134
Статистика	t = 2.232; df = 536; P = 0.026	

Распределение чисел и среднего числа Q-ГР хромосом у казахов и русских, проживающих в г. Алматы.

Число Q-ГР хромосом в геноме	Популяции					
	Казахи-новорожд. (n = 376) I	Казахи (18– 30 лет) (n = 162) II	Казахи пожилые (n = 20) III	Русские – новорожд-е (n = 83) IV	Русские(18-30 лет) (n = 50) V	Русские пожилые (n=80) VI
0	4		2	1	-	4
1	9	15	2	4	3	6
2	60	22	4	9	6	28
3	82	43	7	14	13	23
4	95	38	4	30	12	14
5	71	30	1	16	10	5
6	36	10		6	4	
7	19	4		3	2	
Всего Q-ГР хромосом	1459	578	52	321	190	212
Среднее значение Q-ГР хромосом	3.88±0.077	3.57±0.114	2.60±0.302	3.87±0.141	3.80 ± 0.208	2.65±0.133
Статистика	$t_{I, II} = 2.232$; df = 536; P = 0.026; $t_{IV, V} = 0.261$; df = 131; P = 0.795; $t_{I, III} = 3.728$; df = 394; P = < 0.001; $t_{IV, VI} = 5.895$; df = 161; P = < 0.001; $t_{II, III} = 2.824$; df = 180; P = 0.005; $t_{V, VI} = 4.886$; df = 128; P = <0.001;					

Выводы:

На основе проведенных исследований были сделаны следующие выводы:

1) **Количество Q-ГР хромосом в геноме казахов на индивида в популяции, составляет ~ 3.51. По этому показателю русские, проживающие в настоящее время на территории Южного Казахстана, статистически достоверно не отличаются от коренного населения РК. Однако, в геноме уйгуров количество Q-ГР хромосом статистически достоверно выше, чем у казахов и русских РК (4.15; 3.56 и 3.51, соответственно).**

2) **Количество Q-гетерохроматиновых районов хромосом в геноме новорожденных детей статистически достоверно выше, чем у молодых и пожилых индивидов, независимо от их расово-этнического происхождения.**

В рамках данного Проекта опубликованы две статьи:

1. Ibraimov A.I., Akanov A.A., Meymanaliev T.S., Karakushukova A.S., Kudrina N.O., Sharipov K.O., Smailova R.D. 2013. *Chromosomal Q-heterochromatin polymorphisms in 3 ethnic groups (Kazakhs, Russians and Uygurs) of Kazakhstan*. Int. J.Genet., 5(1), 121-124. [Импакт фактор 4.56]

2. Ibraimov A.I. 2013. *Is it Possible Artificial Sex Regulation in Mammals?* GJSFR 6(2), 1-7. [Импакт фактор 18]

и три статьи подготовлены для печати.

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CHROMOSOMAL Q-HETEROCHROMATIN POLYMORPHISMS IN 3 ETHNIC GROUPS (KAZAKHS, RUSSIANS AND UYGHURS) OF KAZAKHSTAN

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Abstract- A comparative study of polymorphic variants of chromosomal Q-heterochromatin regions (Q-HRs) was performed in three groups (Kazakhs, Russians, and Uyghurs) of Kazakhstan. The number of chromosomal Q-HRs in the genome of studied individuals r from 0 to 7, with the mean 3.51, 3.51 and 4.15 in Kazakhs, Russians, and Uyghurs, respectively. The studied Kazakhs and Russians sh statistically significant homogeneity in the distribution of the number and mean number of Q-HRs, while the highest amount of chromoso HRs revealed in the Uyghur group. Differences and homogeneity between these three groups in the amount of Q-HRs in their genom discussed as evidence in favor of the hypothesis of the possible selective value of chromosomal Q-heterochromatin material in human a tion to various climate-geographic conditions.

Keywords- chromosomal Q-heterochromatin, Q-heterochromatin polymorphism, human adaptation, human migrations

Citation: Ibraimov A.I., et al. (2013) Chromosomal Q-Heterochromatin Polymorphisms in 3 Ethnic Groups (Kazakhs, Russians and Uygh

Is it Possible Artificial Sex Regulation in Mammals?

A.I. Ibraimov

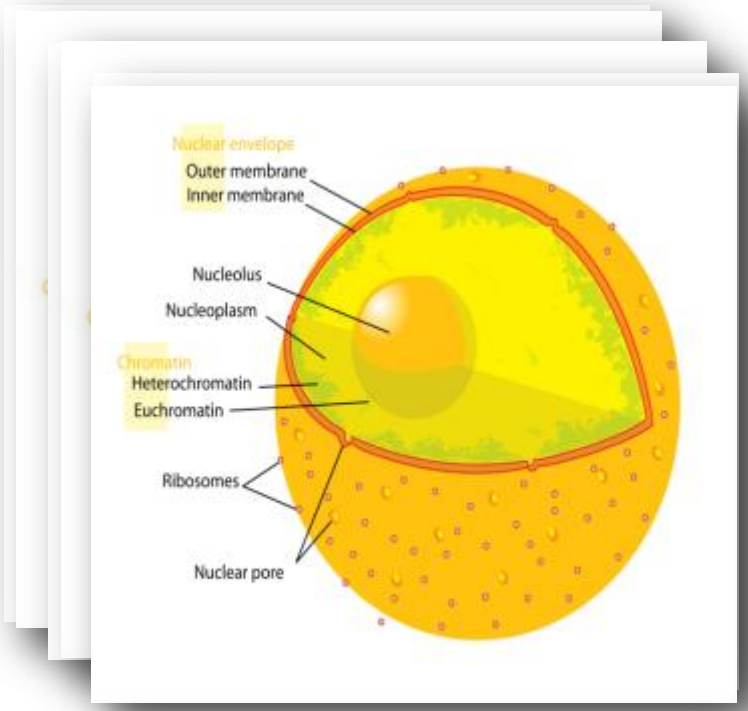
Abstract - In spite of, impressive breakthrough in modern genetics and molecular biology the problem of artificial sex regulation of mammals still remains unsolved. Moreover, the very problem of sex origin of eukaryotes in the process of evolution still has not settled. Existing theories and hypothesizes mainly concern the maintenance and biological reasonability of sexual mode of replication. Their theoretic foundation is based on Darwin's and Mendel's ideas that sex was originated due to natural selection and genes. On the basis of other model of genesis and sex evolution of eukaryote, it was suggested the idea of artificial sex regulation of mammals. Seemingly, the sex differentiation (SD) in animals and human is determined by the amount of constitutive heterochromatin region (cHR) in the Y chromosomes of the undifferentiated embryonic gonads (UEG) via cell thermoregulation. It is assumed the medulla and cortex tissue cells in the UEG differ in vulnerability to the increase of the intracellular temperature because of their anatomical position in genital ridges. If the amount of the cHR on Y chromosome is enough for efficient elimination of redundant metabolic heat from rapidly growing UEG cells the medulla tissue survives. Otherwise it doomed to degeneration and a cortex tissue will remain in the UEG. For

Their theoretic foundation is based on Darwin's Mendel's ideas that sex was originated due to na selection and genes.

However, almost nothing is known a concrete mechanisms and types of g interaction at the SD. The problem also beco complicated especially as: a) the num localization, products, and types of these interactions are not determined; b) the role of the chromosomes in the embryo SD remains not compl clear; c) there are no ideas as regards the possible of a great amount of constitutive heterochromatin re (cHR) of the Y chromosome in the SD.

Indeed it is hard to believe that ha impressive breakthrough in modern genetics molecular biology the reasons and mechanisms o origin are still unknown. This probably has to do wit fact that in the basis of all hypothesizes and theorie sex biology lies idea on all-powered role of na selection and genes in eukaryotic organisms' evolu Although they help to explain reasonably and i

Благодарю за внимание.


Nuclear envelope
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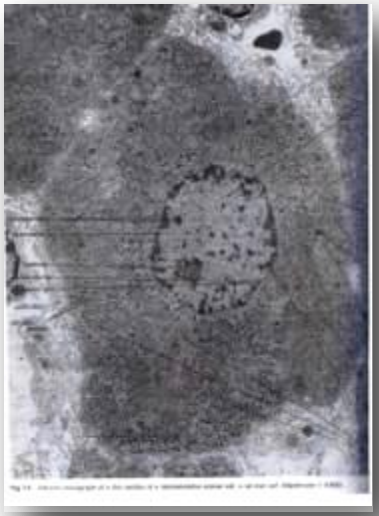
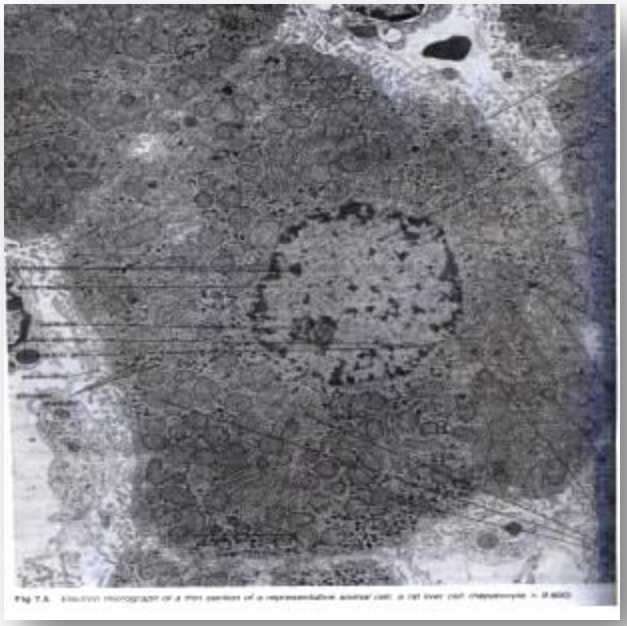
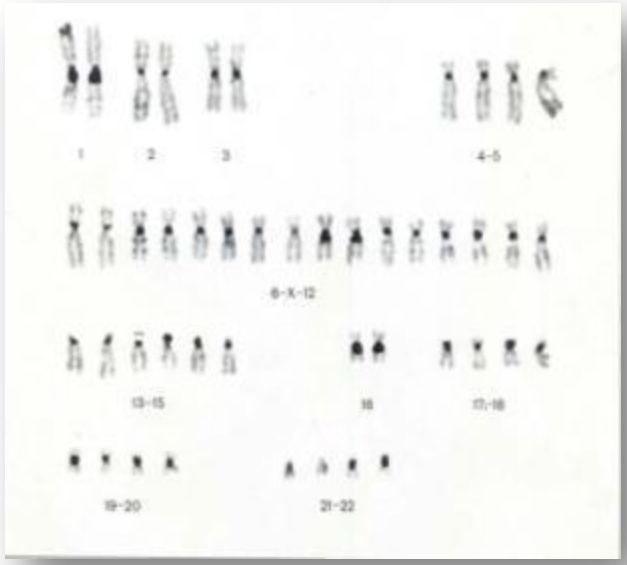
Nuclear pore



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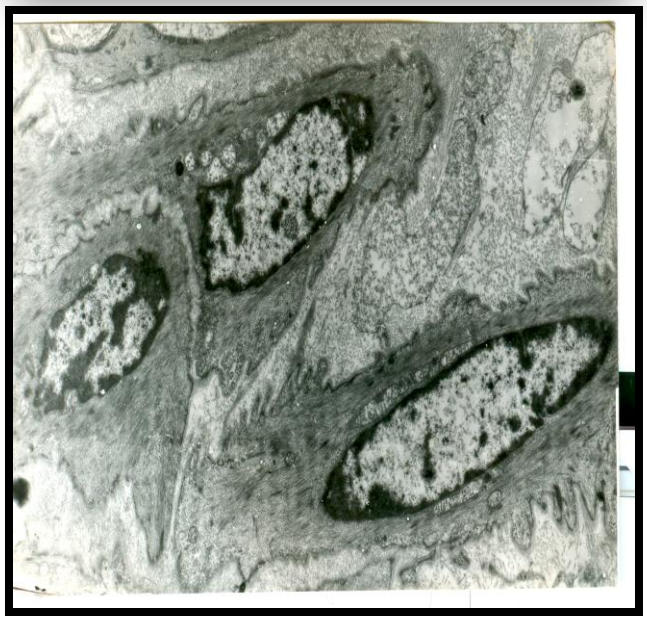

***Вопрос*: отличаются ли индивиды в популяции из разных возрастных групп, постоянно проживающих в РК по количеству Q-ГР хромосом в геноме?**

***Ответ*: в геноме новорожденных Q-ГР хромосом статистически достоверно больше, чем у молодых и пожилых индивидов, независимо от их расово-этнического происхождения.**

Конечная цель исследования *выяснить*: отчего и почему умерли младенцы в первые дни, недели и месяцы жизни, и действительно ли связана ранняя детская смертность с количеством Q-ГР хромосом в геноме?

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Впервые о существовании значительных межрасовых различий

Показал совместное исследование ученых из 5-ти американских университетов, которое было опубликовано на страницах журнала Nature. В частности, у «черных» американцев количество Q-ГР хромосом оказалось больше, чем у «белых».

(Reprinted from Nature, Vol. 268, No. 5621, pp. 633–635, August 18, 1977)

Racial differences in the frequency of Q and C chromosomal heteromorphisms

THE biological and clinical implications of human chromosome heteromorphisms are poorly understood. These heteromorphisms are limited to certain groups, and are heritable[1]. Consistent variability between people, in both size and staining properties, is observed. This may reflect either structural or biochemical variations. Before chromosome banding procedures, the length of the Y was known to vary from person to person and from one ethnic group to another[2]. Some autosomal regions were also found to differ in various racial groups[3,4]. Using Q- and C-banding procedures, we have regions scored were (1) on the long arms (*qh*) of chromosomes 1, 9 and 16; (2) on the short arms (*p*) of the acrocentric autosomes (13–15 and 21–22); and (3) on the centromere (*c*) of all other chromosomes. The results indicate significant differences between the samples of black and white children.

Chromosome studies of peripheral blood lymphocytes were originally done on 4,342, 7–8-yr-old, children from the Collaborative Perinatal Study through the collaboration of cytogenetic laboratories in five cities. The culture and harvest methods are described elsewhere[5]. Most of the data presented here, however, were obtained from a subsample of 415 children. This was derived, using random numbers, from the larger study group and designed to give equal numbers of children of each race above and below an IQ of 85 (ref. 6). Slides from these children were sent to the coordinating centre for Q-banding. The same slides from 200 of the 415 children were also successfully C-banded. In all cases, 5–10 cells were photographed and analysed; two karyotypes were prepared. If C-banding was done, two dual karyotypes with Q- and C-banded chromosomes were prepared from the same cells. It is important to note that the C-banding of all chromosomes could be studied because each chromosome was predentified by Q-banding. Without this sequential staining, only 1, 9, 16 and Y can be studied.

Heteromorphisms were scored according to predetermined criteria without knowledge of race, IQ or other parameters. For Q-banding, the five intensity levels established in 1971 at the Paris Conference were used[7]. The levels are: brilliant, intense, medium, pale and negative; for convenience, they are referred to as levels 5–1, respectively. For C-band heteromorphisms, the karyotype was arranged by decreasing size of the C-band region from largest to smallest[4]. This permitted comparison of C-band regions on homologous and non-homologous chromosomes having similar sized heteromorphisms. Classification of both Q- and C-band heteromorphisms was done by two independent observers, whose scores were virtually identical.

There were 190 black and 194 white children: 98 of the black and 52 of the white children had an IQ less than 85 and the remaining children (92 black and 142 white) had an IQ of 85 or above.

Before scoring the heteromorphisms, each set of slides was scored for quality of culture and quality of banding. There were no significant differences in these distributions between races or between IQ groups. In all groups, the number of heteromorphisms detected directly reflected the quality of the culture. On the average 3.3 Q- and 3.9 C-band heteromorphisms per child were found.

Table 1 Frequencies of Q-banding heteromorphisms in white and black ethnic groups

Chromosome region	Level (s)	White ($N = 205$) N	Prop.*	Black ($N = 210$) N	Prop.*	P value
3*c*	4	57	0.28	89	0.42	<0.01
	5	119	0.58	119	0.57	ns†
4*c*	4,5	42	0.20	20	0.09	<0.01
13*p*	4	87	0.42	113	0.54	<0.01
	5	22	0.11	70	0.33	<0.001
13*s*	4,5	9	0.04	20	0.10	<0.01
14*s*	4,5	23	0.11	27	0.13	ns
15*p*	4,5	2	0.01	6	0.03	ns
15*s*	4,5	21	0.10	21	0.10	ns
21*p*	4,5	1	0.005	4	0.02	ns
21*s*	4,5	16	0.08	28	0.13	ns
22*p*	4,5	4	0.02	8	0.04	ns
22*s*	4,5	12	0.06	29	0.14	<0.01

*Proportion of people, either heterozygous or homozygous. Significantly higher frequencies are underlined.
†ns, not significant χ^2 value.

The frequencies of Q-band heteromorphisms in the two racial groups are given in Table 1. An average of seven heteromorphisms was present in black children and an average of five in white. Where there were significant racial Q-band differences, blacks tended to brilliant or intense heteromorphisms on chromosomes 3, 13 and 22. Chromosome 4 was the single exception. Even when the difference was not statistically significant, the frequency of level 4 and 5 heteromorphism tended to be greater in blacks. We conclude that bright Q-band heteromorphisms are generally more common in blacks.

The centromere (C-band) heteromorphisms were studied in 95 white and 97 black children. These were much less frequent than the prominent Q-band heteromorphisms or *qh* heteromorphisms in 1, 9 and 16. Therefore, data for all chromosomes were pooled. In the white children, the number with small ($N = 44$) and large ($N = 44$) C-band regions was equal, whereas 50 black children had small and 78 had large heteromorphic regions. Overall, larger than usual C-band regions were clearly more common in blacks ($P < 0.005$). Most of this difference was due to chromosomes 4, 18 and 19. No individuals in the white sample had prominent, large centromere heteromorphic regions for chromosomes 4, 18 and 19, but 7·9%, of the black children had large centromere regions for these chromosomes. Five white children had a large centromeric C region in an X chromosome in contrast to only one black child.

The prominent C-band regions on chromosomes 1, 9 and 16 were also studied. These heterochromatic (*h*) regions are usually on the long (*q*) arm of these chromosomes just below the centromere and are termed *qh* regions. There were no obvious racial differences in either extremely small or large *h* regions for chromosomes 1, 9 or 16. Because of greater complexity of these data, they were analysed on a per chromosome basis, rather than a per person basis and will be reported separately by us. Occasionally, however, they seem to be inverted and to be partially or wholly on the short arm of these chromosomes. Although there is no information which permits us to determine whether these are true inversions or whether they represent