medicinska revija
medical review

UDK : 572.522(571.1/.5) ;
616.5-003.829(571.1/.5)
ID : 189221644

Komarovskikh N. E. et al. MD-Medical Data 2012;4(1): 017-021 *MEDICAL DATA / Vol. 4. No 1 /* III 2012.

Originalni članci/
Original articles

OCULO-CUTANEUS ALBINISM IN SIBERIA

OKULO-KUTANI ALBINIZAM U SIBIRU

Elena N. Komarovskikh, Ailana H. Delger

Krasnoyarsk state medical university

Correspondence to:

Prof. Dr Elena Komarovskikh,
Faculty ophthalmology with a rate after the diploma of education,
Krasnoyarsk state medical university,
1, Part. Jeleznyaka str.,
Krasnoyarsk, 660022,
Russia

Tel.: 8 9607607904,
E-mail: Komarovskih_e.n@mail.ru

Key words
oculo-cutaneus albinism, races, Tuva, Siberia

Ključne reči
okulo-kutani albinizam, rase , Tuva, Sibir

Abstract

The aim of this research is mapping of the incidence and prevalence of albinism among the inhabitants of the Republic of Tuva in Siberia. Frequency and clinical manifestation are investigated in the frame of races and entices considerations. High prevalence of coetaneous albinism or oculo-cutaneus albinism was found among inhabitants of the Republic of Tuva. The proportion of albinism from the indigenous population of Tannu Tuva was 0.02% that's 5 times more than 0.004% of the population outside. The preservation of genetic isolation, low frequency inter-ethnic marriage and migration, territorial exclusion and fragmentation of mountain ranges and distance, according to the authors, could be the reason for the random accumulation of oculo-cutaneus albinism indigenous inhabitants – oriental sectors.

INTRODUCTION

The name «albinism» comes from the Latin word "albus" with the meaning "white". Albinism is hereditary disease, that is characterized by reduction or total lack of pigmentation of the skin, hair, eyes (oculo-cutaneus albinism) or only the eye (ocular albinism). Normal process of developing the melanin consists of turning the amino acid tyrosinase residue in the enzyme involved in melanin enzyme tyrosinase. Albinism refers to altered metabolism of amino acids based on hereditary effects or resulting from melanin metabolism defect mutations [1-4] The importance of melanin for organism is significant. The normal process of melanin formation takes place in the presence of the enzyme tyrosinase. Melanin is derived from this transformation.[12, 13] . The disease is characterized by a significant reduction in Visual acuity and result in a violation of the quality of life, and disability. Patients need a special care provided by well trained persons, additional social protection and adaptation in the modern world (Fig 1.).

Figure 1. A Europe child - albino.

As the albinism is a genetic disease, deterministic precautionary measures should be taken for risky groups. These measures include: the surveys on patients and their relatives; the medical-genetic consultation; proper and timely informing about the nature and possible complications of the disease and also the ways

of its prevention and treatment. Almost all forms of albinism inherited by autosomal - recessiv type [3, 4].The disease leads to low vision and to violation of the quality of life and disability, therefore the patients need special training and subsequent job placement, improved social protection and the best possible inclusion in the normal life. Albinisms is hereditary disease or can be result of melanin metabolism defect mutations that lead to a reduction or totally absence of pigment in the skin, mucous, hair and eyes. The role of melanin in the body is essential. Melanin gives color to skin, hair and eyes. Skin and its function related to the production of melanin reflects the response of organism to the environment in general. [2 - 4].

It is proved that the presence of melanin is needed during gestational period. Melanin plays a significant role in the formation and development of the nervous system that is indirectly confirmed by the presence of persons with albinism pathological crossing in hiazme of Visual nerves, the malformed macular area of the retina and the frequent combination of albinism with the deaf and other anatomical disorders [1 - 4, 13]. The functions of the melanin in the eye are very diverse and complex and yet to be explored. Meanwhile, according to available knowledge [4, 13] the melanosomes and pigment in the yellow spot retinal provides optical and protection from light of Visual cells and pigment epithelium, and also perform biochemical (antioxidant) protection of the retina. In the case of albinism, Visual ways in hiazme irregularly cross the axons ganglion cells: overlapping the fibers reduced from 45° (standard) up to 20°. Afferent fibers with a central 20 degrees both temporal areas of the retina instead of having to stay on the same side, overlap with fibers from the nasal part of the retina and brain boom come in opposing kernel [3, 4, 13]. As a consequence of this the violation of the normal retinal Anatomy of lateral crankshaft kernel. In the case of albinism it is less correctly focused and has irregular structure. Various violations inherent in albinisms, apparently due to changes in fiber optic afferent path. At the outset, the formation of these neurological defects attributed the loss of function enzyme tyrosinase. Recently, the need for the presence of melanin in gestational period for formation of normal Visual path is proved. [13].

Data on the prevalence of albinisms available in worldwide literature are contradictory. In the works of C.J. Witkop [12] prediction prevalence is 1: 17000, while G.M. Wilson gives 1: 15000 [13]. Even the albinism is considered as a rare disease, certain racial groups have unusually high incidence. Hermansky-Pudlak syndrome is the most frequent among Puerto Ricans [10] common P-gene-dependent albinism is the most common among the population of the United States but is also dominant in the African and Asian regions. "Pockets" of albinism are identifies in Northern Ireland and hundreds of albinos are found in the tribe Kuna in southern Panama, due to the tendency of increased occurrence of hereditary diseases in small ethnic groups resulted from a great frequency of intermarriage [7, 8, 14]. «Brown», which is a variant of oculocutaneus albinism type 2 oculo-cutaneus albinism, and TRP-dependent oculo-cutaneus albinism (type 3) are found in dark-skinned populations. These types described in [7 - 9] patients from Nigeria, South Africa and New Guinea, where the albinism has be seen as a relatively common genetic condition, despite a dramatic contrast with the normal dark pigmentation. Oculocutaneus form albinisms from the indigenous population here has spread 1: 4182 (fig. 2). Epidemiology of hereditary diseases is an important tool to describe the level of genetic health of populations. Studies evaluating the «cargo» hereditary diseases clarified the structure of the medical-genetic assistance [5, 11].

Figure 2. A Negro child - albino.

Epidemiological studies of various ethnic groups, peoples and nationalities in the vast Siberian territory are becoming more and more interest. Available literature provides information on the genetics of albinos appeared in Japan, Korea and China [6]. Studies of racial aspects of albinisms in oriental sectors of the Russian Federation were launched. The clinical aspects of albinisms in the Republic of Tuva, living in southern Siberia, stay unknown.

The aim of the research: mapping the incidence and prevalence of albinisms in Orientals living in southern Siberia to the indigenous inhabitants of the Republic of Tuva.

Materials and methods: A clinical study of patients with albinism has research and clinical bases in Republican Consultative-Diagnostic Clinic, urban clinics, Kyzyl and Central District hospitals Republic of Tuva. Expeditionary survey were conducted in Todjinskom and Mongun - Tajginskom areas of midwifery. Totally 51 persons with albinism were registered at the medical-genetic consultation of the Ministry of health of the Republic of Tuva, and the Main Bureau of Medical and social assessment for the

Republic of Tuva. Among the people with albinism Asians constituted an absolute majority of 94 % (48 people), while registered Europeans represented only 6 % (3 patients). According to Helsinki Declaration all ethical principles for medical research involving human were respected.. Thus, 48 patients with albinism tuvinians (96 eyes) were comprehensively surveyed. Patients age varied from 4 up to 53 years, the median age was 20.9 ± 1.7 years. Patients in age up 29 years were dominated (79.1 %). Children under 17 years represented 45.8 % of the total surveyed. The average age of men and women was approximately the same: 20.2 ± 2.7 years for men and 21.4 ± 2.3 years for women. The gender distribution was not quite symmetrical, total number of women included in the study was 56.2%, opposite to 43.8%. of man Traditional ophthalmic research methods were used for testing of Visual acuity, Visual field inspection of slit lamp, examination of fundus oculi, refractive available definition, including the disabled known as accommodation, against the backdrop of the electrophysiological studies, General examination of patients.

The results: All surveyed Tuvinians-albinos have oculo-cutaneus albinism I and II types. We had a genealogical survey of the closely related marriages. The 40 patients experiencing oculo-cutaneus albinism, inherited by autosomal - recessiv type. The total population of the Republic of Tuva is 311619 inhabitants (2008), and 75 % (235313)of them are tuvinians. According to number of inhabitants it can be concluded that the prevalence of albinism is 1 patient on 4902. The indigenous albinism met more than 5 times less - 1 patient on 25435. According to our studies, the prevalence of albinism among tuvinians was 2.0 per 10000 of population, indigenous - 0.4. The proportion of albinism from the indigenous population of Tannu Tuva was 0.02% that's 5 times more than 0.004% of the population outside. In General, tuvinians frequency of albinism is 3.5 times higher that it was reported in available domestic and foreign sources. An especially common albinism is reported in Bai-Tajginskom area of the Western Group, 1 patient per 645 inhabitants. A particularly high number of patients with albinism in this area can be explained, in our view, «the effect of the founder», which is local accumulation of subpopulation in albinism, living in the same ecological niche. This contributes to the preservation of genetic and territorial exclusion, resulting in accidental patchy drug accumulation diseases (figs. 3. and 4.).

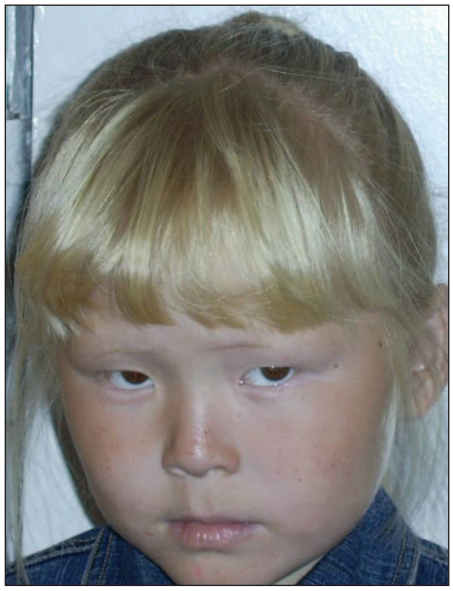

Figure 3. A Asian (tuvinka) child - albino.

Figure 4. A Asian (tuvinka) child - albino.

For the first time it was found that all the patients-Tuvinias suffered oculo-cutaneus form albinism I-B-tyrosinase-positive type, which are more favourable than I-A-tyrosinase - negative type, characterized by the complete absence of pigment. Surveyed patients oculo-cutaneus albinism Tuvin's were typical of the albinism reduced pigmentation of the skin and hair, skin was especially pale «monotonic» with the exceptional sensibility to the Sun and the frequent occurrences of sunburns even when short insolation. Level of pigmentation of hair, eyebrows and eyelashes albinos-therefore vary from very light golden to dark chestnut. Photophobia, the information available to all countries surveyed, impeded the holding of ophthalmologic survey and had varying degrees of gravity. Drawn attention was paid to the coloring of the Iris with albino-oriental sectors. Unlike the inherent Tuvin cam of dark brown and black colors, Iris varied from light brown to grey with green hues. Stroma of Iris

atrophy has been tamed, not similar to bovine spongiform structure healthy tuvinians, its smoothness and color reminiscent of the felt. Decrease of pigmentation on eye grounds, head of the optic nerve hypoplasia, absence of reflexes in the macular area and increased pigmentation in the area of Central holes netted casing were reported in all cases. All surveyed attended horizontal fine-sprawling nystagmus. Component rotation in the two cases. In 87.5% of nystagmus was permanent, 12.5% – a non-permanent. Explicit squint was a 20.8% of patients. In any case, the patients had not been commensurate refraction albinism. The degree of myopia, had averaged 5.6 ± 1.2 Dptr. Myopia refraction combined with different types and degrees of astigmatism. Anisometropia in our surveys. Hypermetropy in all cases combined with astigmatism. The data obtained are different from known where peak refractive curve albinos moved towards hypermetropy accompanied by astigmatism [4, 5].

Astigmatism in Asian - patients with oculo-cutaneus albinism is 2.7 times higher than for European patients (83.3 % : 31 %). Visual acuity below 0.1 was identified at 42 eyes (43.8%). Visual acuity below 0.3 was found in 87 cases (90.7%) above the Visual acuity was 9 (9.3%). No statistically reliable differences of Visual acuity without correction on two eyes in patients ($p = 0.65$). Optimal optical correction does not change the acuity 33.3% of patients. The increase of Visual acuity at 0.01-0.3 with optimal correction received from 52.1% of patients. 12.5% of patients have Visual acuity has increased by 0.3 -0.6 and 2.1% to 0.6 - 0.9. Acuity patients on average increased by 0.16 ± 0.02, i.e. 2.2 times. At 35.4% surveyed had abnormal color vision: deiteranomalya - 35.4%, protanomalya - 14.6%, which as a slight violation of the perception of color violation (type C). P. E. Eskina disorder patients color vision was A and B types (sharp and average reduction) [4]. We believe that less defeat of organ of vision, higher functional performance, positive effects of optical correction on Visual acuity in patients oculo-cutaneus albinism-oriental sectors due to favorable I-B - tyrosinase-positive type and relatively higher content of melanin by ethnicity.

General attitude of population towards the people with albinism in different countries vary considerably. In most African countries they are ostracized from the community, their killing and injury are not considered as a crime and they are not investigated. Serious disadvantages of Africans with albinism are not only the matter of human rights but more the matter of unfavorable environment and negative influence of Sun.

In contrast to this negative attitude and environmental circumstances, in the Republic of Tuva, albinos are not considered to be ill, despite the bright phenotype is significantly different from the phenotype of the mainstream population. People with albinism are perceived as happy people who managed to keep the signs of those present were bright tuvinians, with blue eyes until the Mongol «Golden Horde». Of particular relevance to children fathered by white, is evident in the names by which they are called. They are called names and surnames, respectively its Phenotype: name of AK - «white», names: Aldyn - «Golden», Aldyn-Sai and Aldynaj – the «Golden Pebble» and «Golden Moon», Angyrak - «light», «pale yellow», Ajyran – «colorful», Aydýn-ool - «Moon boy», Mengilen - «snow snowdrift».

CONCLUSION

It was found that among Tuvians there is mainly oculo-cutaneus albinism autosomal recessive inheritance type. According to the literature, virtually all forms of oculo-cutaneus albinism are inherited by autosomal recessiv type, however, there is a dominant form of inheritance, and albinism, which disregards the popularizations factor. We believe that particular demographic and genetic processes in ethno-territorial groups (maintaining genetic isolation, low frequency inter-ethnic marriage), low rates of migration and territorial exclusion and fragmentation of mountain ranges and distance can cause accidental localized accumulation of disease, in other words, a manifestation of the effect of the founder, when created by an ancestor, the ancestor of albinism is transmitted to his descendants, remaining in the land of their ancestors.

Apstrakt

Cilj ovih istraživanja je mapiranje pojave i preovlađivanje albinizma među stanovništvom Republike Tuva u Sibiru. Istraživani su učestanost i kliničke manifestacije u okviru rasnih i etničkih razmatranja. Medju stanovništvom Republike Tuva preovladjuju dva oblika albinizma coetaneous albinism i oculo-cutaneus albinism. Pojava albinizma kod domorodačke populacije Tannu Tuva je bila 0.02% što je 5 puta više nego kod stanovništva van ove etničke grupe. (0.004%) Razlozi za slučajnu akumulaciju slučaja oculo-cutaneus albinizma mogu biti zatvorenost etničkih grupa, teritorijalna izolacija i odvajanje, mali broj međuetničkih brakova, genetička izolacija i ograničena migracija.

REFERENCES

1. Bogdanova V. Crips membership and questions the origin of tuvinians. Problems of anthropology of the ancient and modern populations of Soviet Asia. Novosibirsk, Nauka, 1986; 203-206.

2. Kalachev I., Mojerenkov B. Albinism in the practice of Ophthalmology (overview). Journal of Ophthalmology, Moscow, 1989; 2, 71-74.

3. Shamshinova A. Hereditary and congenital diseases of the retina and optic nerve. M.: Medicine; 2001, 528 p.

4. Eskina E., Smolyaninova J., Shamshinova A. Diagnosis and assist patients with albinism. Journal of Ophthalmology, 1994; 3, 35-38.

5. Hedrick P. Hopi Indians, «cultural» selection, and albinism. Am. J. Phys. Anthropol., 2003; 121, 2, 151-156.

6. Inagaki K., Suzuki T., Ito S. et al. OCA4: evidence for a founder effect for the p.D157N mutation of the MATP gene in Japanese and Korean. Pigment. Cell. Res., 2005; 18, 5, 385-388.

7. Lee S., Nicholls R., Schnur R. Diverse mutations of the P gene among African-Americans with type II (tyrosinase-positive) oculo-cutaneous albinism (OCA2). Hum. Mol. Genet., 1994; 3, 2047–2051.

8. Lund P. Oculo-cutaneous albinism in southern Africa: population structure, health and genetic care. Ann. Hum. Biol., 2005; 32, 2, 168-173.

9. Oetting W., Fryer J., Shriram S. et al. Oculo-cutaneous albinism type 1: the last 100 years. Pigment. Cell. Res. 2003; 16, 3, 307-311.

10. Santiago-Borrero P., Rodriguez-Perez Y., Renta J. Genetic testing for oculo-cutaneous albinism type 1 and 2 and Hermansky-Pudlak syndrome type 1 and 3 mutations in Puerto Rico. J. Invest. Dermatol., 2006; 126, 1, 85-90.

11. Schroeder H., Orth U., Meyer-Konig E. Familiare foveahypoplasie – klinische Einordnung. Klin. Monatsbl. Augenheilkd, 2003; 220, 8, 559-562.

12. Witkop C., Quevedo W., Fitzpatrick T., King R. Albinism. The metabolic basis of inherited disease, New York: McGraw-Hill, 1989, 2905-2947.

13. Wilson G., Maumenee I. Albinism. Retinal dystrophies and degenerations New York, 1998, 305-317.

14. Woolf C. Albinism (OCA2) in Amerindians. Am. J. Phys. Anthropol., 2005; 41, 118-140.

■ The paper was received on 19.01.2012.
Revised and accepted on 30.01.2012.