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Clinical and molecular characteristics of ichthyosis in Yakutia

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Introduction: Hereditary ichthyosis is common name for many dry and scaly skin disorders ranging in frequency from common to very rare [1]. Clinical genealogical analysis of hereditary forms of ichthyosis (from 1989 to 2018) are conducted in the Republic of Sakha (Yakutia), Russia.

Materials and Methods: Data was collected from the medical cards of patients of the "Republican Genetic Register of Hereditary and Congenital Pathology of the Republic of Sakha (Yakutia)" of the Medical Genetics Center of the Republican hospital No1 "National center of medicine". Blood samples were drawn from 10 patients for clinical exome sequencing.

Results: The number of patients with ichthyosis is 126 patients (104 families) according to the genetic register of Yakutia according to date of 2018. The frequency of ichthyosis was about 1/7675, prevalence is 13,0 to 100 000. Gender: 94 men (74.6 %), 32 women (25.4 %). Ethnicity: Yakut – 109 (86.5 %), Russian – 17 (13.5 %). In four families affected patients of both genders and in ten cases only males. All patients had dryness, peeling of the skin. In some cases, more severe symptoms were noted in the form of pityriasis peeling, follicular keratosis. The manifestation varied from birth to adolescence. Results of clinical exome sequencing showed variants in *ALOXE3* c.1604A > G (4/10), *KRT10* c.458T > C (1/10), *STS* c.1334A > G (1/10), suspected deletion in X chromosome (2/10). Two patients were without found variants. Patients with the same c.1604A > G variant in *ALOXE3* demonstrated mild phenotype: mild/moderate erythema with fine white scale, palmoplantar hyperkeratosis as described in literature [2–6]. Patient with c.458T > C variant in *KRT10* demonstrated phenotype of collodion baby.

Conclusions: It is difficult to establish the type of inheritance and further medical and genetic counseling due to genetic heterogeneity and the similarity of the clinical features of ichthyosis. Thus, there is a strong need in a highly specific diagnostic method and in-depth research study for precise diagnosis, treatment and prevention of these diseases.

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