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Possibilities of the vegetative resonance test in the screening of orphan diseases NS. Kirgizova

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Patient S., 30 years old. She complained of acne for a long time, not responding to the proposed therapy.

During the initial examination, attention is drawn to the pale skin of the face and upper half of the chest with a pronounced greasy sheen. On the skin in the area of the cheeks, chin, forehead - single acne and cicatricial changes, on the cheekbones pigmented spots. The face and upper eyelids are edematous. The patient has low nutrition, the turgor of the skin of the trunk and extremities is reduced.

History of frequent sore throats, erysipelas of the skin of the lower extremities in adolescence, acne over the past 15 years.

Physical and instrumental examination revealed a pronounced depletion of the immune system, long-term existence of a bacterial streptococcal infection (spores), maximum DNA damage, chromosome XI is tested.

The patient consistently solved the problems of correcting immunity and draining the infection.

Repeated testing confirmed violations associated with the XI chromosome, namely, lipid accumulation diseases (sphingolipids) caused by congenital deficiency of enzymes, such as P, A, B - galactosidases, in this group, in accordance with the List of rare (orphan) diseases of the Ministry of Health Of the Russian Federation, includes lipid storage diseases: Fabry's disease (-Anderson), Gaucher's disease (Gaucher's disease), Krabbe's disease, Niemann-Pick disease (Niemann-Pick disease type C, Niemann-Pick disease type A / B), Faber syndrome (Farber's disease), metachromatic leukodystrophy, sulfatase deficiency (multiple sulfatase deficiency).

In patient S., the study of biochemical blood markers revealed a decrease in the atherogenic coefficient, which has no independent diagnostic value, but in this case, confirming lipid disorders. The patient was sent for examination to clarify the orphan disease.

It is known from clinical practice that early histological and biochemical markers of pathology are rarely detected in women, mainly with heterozygous inheritance. A study using the method of vegetative resonance test allows us to identify the main pathogenetic nuclei, narrow the range of differential diagnostics, offer complex treatment based on the ex juvantibus principle.

Early detection with the help of hardware technologies of markers indicating a possible orphan disease allows for timely specific diagnostics and initiation of substitution therapy. Taking into account the increase in the number of genetically determined pathologies, this will allow avoiding disability and prolonging active longevity. NS. Kirgizova Possibilities of vegetative resonance test in the screening of orphan diseases // "- M .:" IMEDIS ", 2014, v.2 - P.181-182

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