

OVERCOMING PSYCHOLOGICAL BARRIERS IN PATIENTS WITH RARE GENETIC DISORDERS

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Nowadays there are lots of rare (orphan) genetic disorders caused by different types of mutations. Obviously, defect genes encode dysfunctional proteins which modify general biochemical pathways, but also it's worth mentioning about a great number of patients with rare diseases who have psychological problems, such as high levels of stress, depressions and problems with social adaptation. Moreover physicians face difficulties in sharing experience of treatment of these diseases.

The research aims to assemble all types of knowledge concerning treatment, diagnosis of orphan genetic disorders, prevention of mental issues and to make statistical assessment of Ukrainian progress in this field of medicine. Several tasks were set:

- detailed review of online blogs, groups, sites, where patients and doctors from different parts of the world can fluently communicate with each other;
- searching for Ukrainian patient communities;
- analysis of the statistics of diagnostics and treatment of rare genetic disorders in Ukraine;
- assessment of the level of psychological assistance to people with rare genetic syndromes.

To complete the tasks of the research online clinical bioinformatics and genomic resources (Undiagnosed Diseases Network), medical databases (OMIM, Genetics Home References, NCBI, PHG Foundation), medical organizations' sites (Genetic Alliance), patients' personal blogs (written by parents of the child with NGLY1 deficiency, by family with rare mitochondrial disease) and patient communities (RareConnect) have been used. In order to find out real information about diagnostics and treatment of unique illnesses in Ukraine, some sites of patient organizations were reviewed (Debra Ukraine, Orphan Ukraine).

Surprisingly many Ukrainian patients with orphan genetic disorders have depression. These people always live in stress, because actually they are not treated or even consulted by specialists. The main cause of mental problems is the lack of adequate financial government support. According to the statistics given by Ukrainian healthcare committee, financing has increased by 4.45 times (2011-2015 y.), but that is not enough for providing a good healthcare. It was estimated that this financial support is only 10-30% of the required sum. We also have people with undiagnosed pathologies. Only 5% of Ukrainian patients with rare ailments know their true diagnosis. Surely, some international organizations help our patients. In fact, people with such disorders should find assistant communities by themselves. In foreign countries (UK as an example), scientists use next generation sequencing that provides better diagnostics of rare diseases and helps in treatment, also physicians collaborate with patients by using blogs, healthcare sites.

There is an extremely critical situation with financial support for patients with orphan genetic syndromes in Ukraine, but international organizations are ready to help. People don't know about their possibilities, and that is the key problem. Ukrainian physicians should integrate their knowledge with colleagues from other countries, pay attention to the patients' psychological problems. International cooperation will help patients with undiagnosed pathologies, which are mostly caused by mutations in genes, and have polysymptomatic presentation pattern.