THE BULGARIAN NATIONAL REGISTRY FOR PRIMARY IMMUNE DEFICIENCIES

Elisaveta Naumova[1,2,3], Nevena Gesheva[1,2], Spaska Lesichkova[1,2,3], Petya Yankova[1,2,3], Gergana Nikolova[1], Veneta Milenova[1,2], Nedelcho Ivanov[1,2], Snezhina Michailova[1,2,3]

[1]*Clinic of Clinical Immunology with Stem Cell Bank, University Hospital "Alexandrovska", Sofia, Bulgaria*

[2] *Expert Center for Rare Diseases - PID, University Hospital "Alexandrovska", Sofia, Bulgaria*

[3]*Department of Clinical Immunology, Faculty of Medicine, Medical University, Sofia, Bulgaria*

The Expert Center (EC) for Rare Diseases - Primary Immune Deficiencies (PID) at University Hospital "Alexandrovska" has been officially established since 05.04.2016. In May 2017 The National Register of Patients with Rare Diseases, established and administrated by the National Center for Public Health and Analysis, also became operational. The registry contains clinical and genetic information for PID patients. In 2020 PID patients' data started to be entered in the existing European Primary Immunodeficiency Database, managed by the European Society for Primary Immunodeficiency (ESID). The total number of patients entered in the PID register is 184 (97 men and 87 women), with more than half being children (100; 56.8%). Regular updating of the information in the register shows that 8 (4.3%) of those registered have died and 176 (95.7%) are subject to continuous monitoring, which is similar to the registers in other European countries. Before the establishment of the EC in Bulgaria, in the period 2005-2015, 87 patients with PID were diagnosed. From 2016 to September 2020, 97 patients with various forms of PID have been diagnosed and included in the national registry, which is also due to the increased number of centers supplying information for newly diagnosed or suspected PID patients. The predominant diagnosis (84 patients; 45.6%) was antibody deficiency. Seventy (38.4%) of the patients underwent genetic testing, and 52 (28.2%) had a proven genetic diagnosis, which corresponds to the data of the European registry. Confirmation of the phenotypic diagnosis is present in 61% of cases, change in the diagnosis in 16% and lack of genetic diagnosis in 23%. Fifty-five patients received fully covered replacement therapy or other specific therapy, and the rest were receiving symptomatic treatment.

The delay between the onset of symptoms, diagnosis and initiation of treatment has decreased significantly for most PID the last 15 years, thanks to increased public awareness, significantly improved diagnostic capabilities, experience in PID and excellent collaboration between physicians in various medical specialties at national and international level.