

# The patients' organizations of children with primary immunodeficiency in Poland

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Introduction: In the European Union any disease suffered by fewer than 1 in 2000 people is considered a rare disease. The lack of information about rare diseases often means that sufferers, who together represent more than 7% of the total EU population, do not always benefit from the health services they need. Yet it is often these frequently life-threatening or chronically debilitating diseases that require combined efforts to prevent citizens suffering from a reduced quality of life or dying early. Primary Immunodeficiencies are genetic defects in the immune system that cause increased susceptibility to a wide range of infections, affecting the skin, the ears, the lungs, the intestines and other parts of the body. These infections are often chronic, persistent, recurring, debilitating, and in some cases, fatal. With levels of diagnosis currently estimated at 1 in 10 000, Primary Immunodeficiencies (PIDs) are classified as rare diseases. This is why the PID Community calls upon EU Member States to take heed of these devastating and under-diagnosed diseases by developing national rare disease plans that include PID and ensuring that both new and existing national rare disease plans cover PIDs. See Tab 1

Tab 1  
Components of Primary Immunodeficiencies (PID) Call to Action:

1. Raising Awareness on Primary Immunodeficiencies
2. Education of Health Professionals and Expertise Exchange
3. Early Diagnosis and Screening
4. Gathering Knowledge and Data
5. Comprehensive and Adequate Treatment

Dynamic developments in molecular and genetic techniques led to great progress, in knowledge the pathogenesis of PID, and recognition of new PID types, as well as improvement in treatment protocols. The national programme of substitutional replacement therapy was initiated and conducted by the Department of Immunology, CMHI in 1993. The programme and distribution of intravenous gammaglobulins to immunology centres in Poland was financed to early the 2000s by the Ministry of Health, and now by the National Health Insurance system. Subcutaneous replacement immunoglobulin therapy was introduced in our Department of Immunology in 2001 for the first time in Poland, and has been continued successfully. Increasing experience in hematopoietic stem cell transplantation gives a great chance for correction in 41 of the most severe forms of PID.

Photo 2  
A patient afflicted with AT



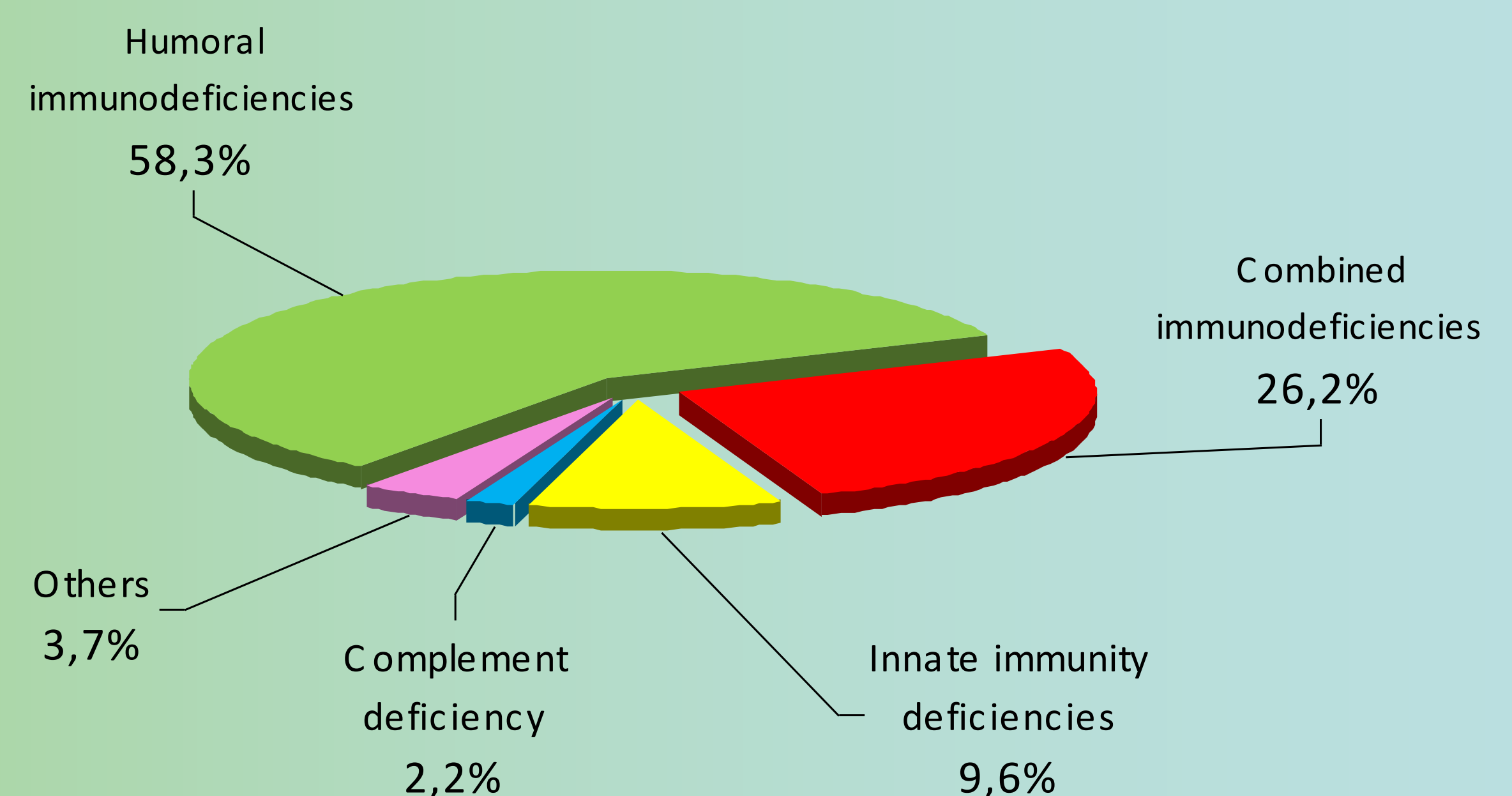
Thanks to the initiative of the Department of Immunology the first patients' organization named the Association of Friends for Children with Immunological System Deficiencies was founded in 1987. The members originate from different walks of life: patients and their families and friends, doctors, nurses and other medical staff. The Association co-operated with similar organizations such as ESID, IPOPI, INGID up to 1999, when finished there activity. The other patients' organizations have been founded, which focused on specific immune diseases (f.e. for DiGeorge syndrome, Nijmegen Breakage Syndrome etc.). These organizations are small and not very active, besides Ataxia-Telangiectasia Foundation which collected large group of patients with AT. The AT patients and their families from the whole Poland meet once a year in May to share their doubts and experience and to widen their knowledge of the disease. See Photo 2,3

Photo 3  
The AT patients and their families from the whole Poland.



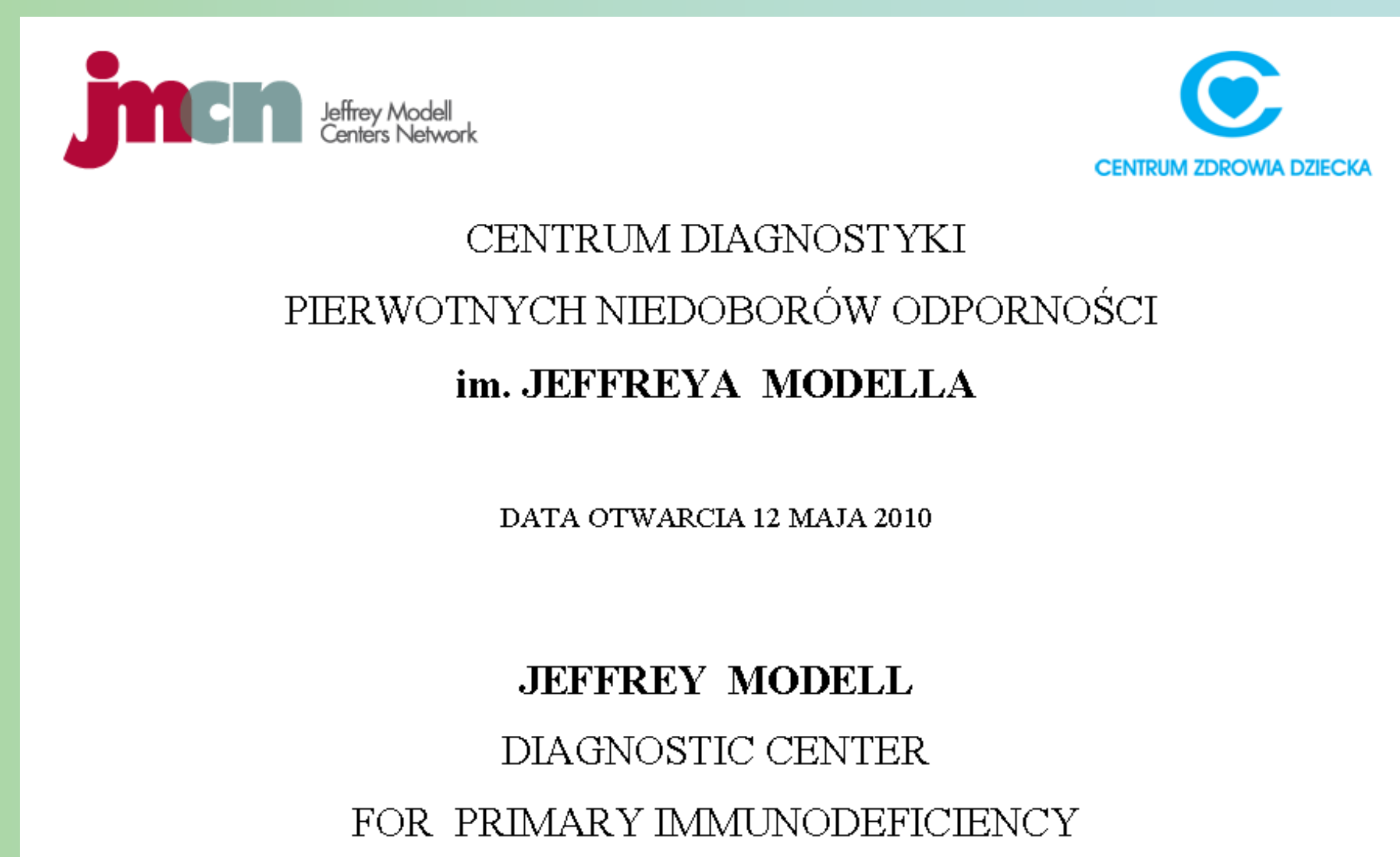
The European Society for Immunodeficiencies (ESID) has developed an internet-based database for clinical and research data on patients with PID. Poland currently showing a prevalence of incident 1,5 patient per 100 000 only, what emphasize that under-diagnosis and under-reporting in the results. Since 1980 the Department of Immunology Children's Memorial Health Institute (CMHI) has been the reference center for diagnosis and therapy of children with primary immunodeficiencies (PID) from the whole of Poland. One thousand one hundred ninety – seven cases of primary immunodeficiency diseases were diagnosed in the Department of Immunology of the Children's Memorial Health Institute in Warsaw during 1980 – 2010. The highest frequency was that of antibody immunodeficiencies - 547 (58.3 %), next by frequency and predominantly T cell deficiencies in 80 ( 8.5 %) children. See Tab 2

Tab 2  
Primary Immunodeficiencies in Polish Registry 1980 – 2009 n = 1179



On May 12, 2010, the first Jeffrey Modell Primary Immunodeficiency Diagnostic Centre was opened at Immunology Unit of the Children's Memorial Health Institute in Warsaw. The Foundation is dedicated to early and precise diagnosis, meaningful treatments, and ultimately new cures that help fighting the Primary Immunodeficiency — a fight which, as there is more and more known, becomes more and more efficacious. The Foundation' aims are met inter alia by means of the international network of 70 research and diagnostic centers. See Photo 1

Photo 1  
Memorial board located in Immunology Department, CMHI in Warsaw



In 2007 a new patients' organization named IMMUNOPROTECT was set up which gathered mainly patients' parents with primary antibody deficiencies. The main aim of setting up this unit has encompassed the support of children and adults with primary immunodeficiencies. Basic activity comprises development of an educational programme for patients and families with primary immunodeficiencies and information initiatives for doctors and parents concerning primary immunodeficiency disorders. IMMUNOPROTECT started to put together other small groups of patients as a reasonable way to build up one powerful, effective organization. This organizations is aimed to provide better patients' care and access to sufficient treatment.



<http://www.immunoprotect.pl>

Summary: Long-lasting polish experience of PIDs patients' parents activity has shown that only one big organization can represent patients' interests successfully.