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IFN-γ and TNF-α polymorphism in patients with primary immunodeficiency

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Primary immunodeficiencies are rare diseases in which the immune abnormalities observed frequently result in the occurrence of persistent infections. Not recognized and the same not treated properly can lead to a number of complications, abnormalities can sometimes finish in premature death. Therefore, where it is stated recurrent infections efforts should be made to confirm or rule out the disease. Studies on the functioning of the immune system in this group of disorders have been carried out for many years and yet there are still many pathophysiological mechanism is unknown. Therefore, it is important to continue to explore these issues. 87 patients were included in the study of the Department of Clinical Immunology and Pediatrics at the age of 6 m to 18 years old, among whom distinguished group of 26 patients with primary immunodeficiency (PID), 43 patients suffering from recurrent respiratory infections and 18 healthy children (without recurrent infections). All patients were evaluated the screening of selected parameters of immunological hemolytic complement activity (CH50) of the concentration immunolglobulin major classes, properties of neutrophils phagocytosis, and a panel of T cells, B cells and NK cells. Additionally the concentration of IFN-γ and TNF-α was examined in serum it was determined polymorphism of the gene encoding TNF-alpha (TNFA, rs1800629, -308 G/A) and IFN-gamma (IFNg, rs2430561, 874 T/A). Genotyping was performed by analysis of melting curves. This method is based on the reaction of the real-time PCR, i.e., PCR in which the detection of amplified DNA may take place during the reaction, without the need for separate detection of the completion of the reaction. As typing kits used are SNiP Light Assay (TIB MOLBIOL Company) and the mixture Probes Master Mix (Roche). Amplifications are performed with the Light Cycler® 480 Multiwell Plate 96 in the camera 480 II Light Cycler (Roche). It has been demonstrated reduced levels of IFN-γ in children with PID and recurrent respiratory infections compared with the controlled group. The observed most frequent polymorphism groups concerned AT base in both the PID and recurrent respiratory infections, as well as in the control group. TNF-α in the PID group were decreased and increased in patients with recurrent respiratory infections compared to the controlled group. Polymorphism is most frequently found in children with PID base pairs of concern GA, in the group of recurrent respiratory tract infections usually observation GG base pairs.

Biography

Aleksandra Lewandowicz-Uszyńska is currently acting as Manager of the 3rd Department and Clinic of Pediatrics, Immunology and Rheumatology of Developmental Age and Head Physician of the Department of Clinical Immunology and Pediatrics Provincial Hospital J. Gromkowski in Wroclaw. She was graduated from the Medical University of Wroclaw in 1988 and she is a Specialist in Pediatrics and Clinical Immunology. She has participated in numerous scientific internships in the leading Immunological Centers. Her research interests are focused on the phenomena of humoral and cellular immunity (with particular emphasis on the role of neutrophils) in primary and secondary immunodeficiencies (PID). She is also a Provincial Consultant in Pediatrics for Lower Silesia, Member of Board of the Polish Society of Experimental and Clinical Immunology, the Committee of Immunology and Etiology of infections Human Polish Academy of Sciences, Polish Working Group, PIDs, Polish Federation of Pediatric, the Scientific Committee of Standards of Medical and OSOZ. She is the President of the Section of Developmental Immunology Since 1997. She is the author of 102 scientific publications, approximately 100 congress reports, 13 textbook chapters in the field of pediatrics and clinical immunology.