

## **PIDexpert - decision support system for immunodeficiencies. Development and modeling of data**

Crina Samarghitean<sup>1</sup>, Kirsi Vapa<sup>2</sup>, Kati Iltanen<sup>2</sup>, Merja Helminen<sup>3</sup>, Mauno Vihinen<sup>1,4</sup>

<sup>1</sup>Institute of Medical Technology, University of Tampere, Finland

<sup>2</sup>Computer Science Department, University of Tampere, Tampere, Finland

<sup>3</sup>Tampere University Hospital, Finland

<sup>4</sup>Research Unit, Tampere University Hospital, Finland

PIDexpert is a decision support system, which tries to assist physicians in diagnosis of primary immunodeficiencies (PIDs). PIDs are rare hereditary diseases of the immune system, which are often difficult to diagnose, due to similar and overlapping symptoms. Patients have an increased number of infections but also autoimmune and cancer related problems are associated.

PIDexpert gives the diagnostic picture based on symptoms, signs, medical history, physical findings and laboratory tests. PIDexpert includes a knowledge base, a query base, a knowledge acquisition system, an inference engine and a user interface. The knowledge base is built using data and facts from e.g. IDR, (Immunodeficiency Resource, <http://bioinf.uta.fi/idr/>), IDdiagnostics (<http://bioinf.uta.fi/IDdiagnostics>), IDbases, clinical guidelines, literature and medical experts. The query base contains instructions to create the user interface. The questions are organized by distinct topics involved in the diagnostic criteria of PIDs, concerning symptoms and signs, medical and family history, clinical and laboratory findings. The inference engine is rule-based and uses scores to compute the best matched disease profiles using a pattern recognition method. The user interface defines the input and output parameters.