

The J Project



2011

Editors:

László Maródi and Zsuzsa Horváth

with the contribution of

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Foreword

Awareness about primary immunodeficiency disorders and the availability of diagnosis and treatment need to be permanently increased, particularly in countries with poor socioeconomic conditions. The Jeffrey Modell Foundation, the Immunodeficiency Foundation, the International Patient Organization for Primary Immunodeficiencies, International Nursing Group for Immunodeficiencies and national PID foundations and national patients' groups like HOPI (Hungarian Organization for Patients with Immunodeficiency) have been the pioneer organizations to meet the challenges. Professional and scientific societies including the societies for immunodeficiencies in Europe (European Society for Immunodeficiencies, South America (Latin American Society for Immunodeficiencies), Africa (African Society for Immunodeficiencies) and Asia provide invaluable support and encouragement to promote awareness and physician education campaigns all over the world. Countries in Eastern- and Central Europe belong to those that desperately need increased awareness because of the relatively low number of registered and diagnosed patients and insufficient resources available for diagnosis and treatment of PIDs. The Eastern- and Central European Infectious and Pediatric Immunology Center for Training and Research (ECE IPI CTR) was established in 1999 in Debrecen, Hungary. From educational and scientific points of view, the Center was closely associated with the Department of Infectious and Pediatric Immunology at the University of Debrecen. ECE IPI CTR functions as an institutional and regional Center. Its essential role is the initiation of regular postgraduate courses for specialists in infectious diseases and immunology in countries of ECE. One of the most successful programs of the Center has been the J Project. The J Project is an ECE perspective which was formulated and has been run by clinician scientists in a network. Regular meetings organized by local professionals to disseminate knowledge among general practitioners are important components of the Project. Research collaboration is an integral part of the program. Activities of the Project are coordinated by the J Project Steering Committee and a J Project Booklet is issued each year since 2004 and also available online at <http://jproject.dote.hu>.

Debrecen, December, 2011

László Maródi



University of Debrecen
Medical and Health Science Center

ECE IPI CTR

The **East-Central-European Infectious and Pediatric Immunology Centre for Training and Research (ECE IPI CTR)** was established on December 22, 1999. From educational and scientific points of view, the Centre is closely associated with the **Department of Infectious and Pediatric Immunology** at the **University of Debrecen Medical and Health Science Centre**. The Department has a strong profile of immunology including research into antimicrobial host defense mechanisms. The main fields of research activity of the Department and Centre are as follows:

- Molecular pathology of primary immune deficiency diseases
- Host defense mechanisms against bacteria and fungi
- Mechanisms of action of intravenous immunoglobulin
- Developmental biology of macrophages

The Centre provides modern educational utilities, demonstrational materials, and rooms that are suitable for practical seminars as well as lecture rooms. ECE IPI CTR functions as an institutional and regional Infectious and Pediatric Immunology Centre. It's essential role is the initiation of regular postgraduate courses for specialists in Infectious Diseases and Immunology in countries of East Europe.

From the conception aspects, ECE IPI CTR is tightly connected with the **Interregional Association of the Carpathian Euro-region**, one of the main aims and functions of which is the formulation of educational and scientific co-operation within the region of East-Central-Europe.



The J Project

PID Awareness Meetings in 2011

City (Country)	Date
1. Pushkin Hills (Russia)	February 03-04
2. Zakopane (Poland)	February 24-26
3. Oradea (Romania)	March 01
4. Debrecen (Hungary)	March 10-11
5. Moscow (Russia)	March 31-April 01
6. Baku (Azerbaijan)	April 22-23
7. Bratislava (Slovakia)	April 28-29
8. Lutsk (Ukraine)	May 11-12
9. Sibiu (Romania)	June 10-11
10. Chelyabinsk (Russia)	June 26-27
11. Ohrid (Rep. of Macedonia)	October 08
12. Tehran (Iran)	October 15
13. Trabzon (Turkey)	November 19-20
14. Riga (Latvia)	November 26

Beijing (China)*

Sept 23

**J Project session at BIT's 4th Congress and Exposition of Molecular Diagnostics*

Aims

1. To organize professional meetings on PID and related diseases in regions of East-European countries with low number of registered PID patients.
2. To discuss diagnostic and therapeutic practices and problems, and to define specific areas to be improved and to be supported by other European groups, institutions, companies, and foundations.
3. Updating national PID registries.
4. Establishing PID professional working groups.
5. Establishing PID patients, groups.

Structure of the Meetings

1. Informal discussion on the day before the meeting.
2. Introductory lectures by invited and local speakers.
3. Case reports mostly by local speakers.
4. PID WG and patients' group activity in the country.
5. National registry update.

Pushkin Hills, Russia

03-04 February, 2011

Host: Marina Guseva (gusevamarina@mail.ru)
Areg Totolian (totolian@spbraaci.ru)

Venue: Pushkin Hills, Region of Pskov

Main Topic: International educational symposium on primary immunodeficiency disorders



Participants of the meeting

Program

2 February

Molecular mechanisms of primary immunodeficiencies

Poliakov A. (Moscow)

Macrophage activating syndromes

Kondratenko I. (Moscow)

Screening program for immunodeficiencies

Prodeus A. (Moscow)

DNA-diagnostics of PIDs

Poliakov A. (Moscow)

Autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy and chronic mucocutaneous candidiasis

Kondratenko I. (Moscow)

February 3

X-linked lymphoproliferative disease

Maródi L. (Debrecen)

Pneumococcal genetic susceptibility (IRAK-4 and MyD88 deficiencies)

Capucine P. (Paris)

Clinical, immunological and molecular features of patients with primary antibody deficiencies

Plebani A. (Brescia)

Methods of intravenous immunoglobulins safety testing

Prodeus A. (Moscow)

Immunobiologics treatment safety issues

Hadzhidis A. (St. Petersburg)

Lunch

Afternoon session

Biotest Farma GmbH symposium

Chairman: *Reznikov Y. (Moscow)*

Primary and secondary immunodeficiencies: Classification, diagnostics and therapy

Reznikov Y. (Moscow)

Algorithm of therapy of PID patients

Kondratenko I. (Moscow)

Bone marrow transplantation in PID: The role of intravenous immunoglobulin treatment

Skorobogatova E. (Moscow)

Discussion

4 February

Morning session

Hyper IgE syndromes

Maródi L. (Debrecen)

Mendelian susceptibility to mycobacterial infections

Capucine P. (Paris)

Improving awareness on PIDs among physicians: The IPINET (Italian Primary Immunodeficiency Network) model

Plebani A. (Brescia)

Morphological analysis in PID diagnostics

Tuzankina I. (Ekaterinburg)

The ESID diagnostic protocol for PID

Guseva M. (St. Petersburg)

Discussion and conclusion

Summary and conclusion

The second All-Russian School of Clinical Immunology was held in Pushkin Mountains from January 29 to February 5, 2011. Participants and speakers from 23 regions of Russia, as well as Latvia, Belarus, Hungary, France, Italy got together to attend the J Project symposium held on February 2-4. Introductory lectures on topics of fundamental immunology relevant to primary immunodeficiencies were presented. The first report by László Maródi, head of the J Project and ECE-IPI-CTR, University of Debrecen Medical and Health Science Center, Hungary, was devoted to the history and prospects for the J Project in Central and Eastern Europe, including Russia, and its march to different cities of our country. Ekaterinburg and Chelyabinsk also joined the J Project in addition to St. Petersburg in 2007. It is also scheduled to hold J Project meeting in Moscow in Russian State Children Hospital. All this activity is expected to increase awareness on PID among physicians of various specialties, and the public. Thematic reports by professor Maródi were dedicated to hyper-IgE syndrome and XLP (X-linked lymphoproliferative disease). Clinical, immunological, and molecular genetic aspects of these diseases were covered in these reports. An update by professor Alessandro Plebani (Brescia, Italy) on clinical, immunological and molecular characterization of patients with defects of antibody synthesis was provided. He also gave a presentation on "Perfection of knowledge about primary immunodeficiencies among practicing physicians: The Italian model of networking". Professor Plebani shared the experience of detection of patients with primary immunodeficiency in Italy. There is computer program for the detection of these patients (PID registry) and co-operation between immunological centers in Italy by regular meetings. Reports by professor Kondratenko I. (Moscow, RSChH) were focused on clinical experience demonstrating knowledge and practical experience in managing patients with PID. Report by professor Prodeus A. P. (Moscow) demonstrated screening program for the diagnosis of immunodeficiency in the U.S., and modern methods of testing immunoglobulin safety. Report of professor Tuzankina I. A. (Ekaterinburg, Center of Clinical Immunology) was devoted to the pathologic interpretation of PID, and demonstrated experience in diagnosing and managing patients with PID in the Ural region. Report from St. Petersburg presented the work of PID working group. Guseva M. N. talked about "Protocols for diagnosis of primary immunodeficiency diseases and management of patients with PID in Consulting Center of St. Petersburg State Pediatric Clinic. Excursion allowed the visitors to develop an understanding of the history of the famous Russian poet Alexander Pushkin, whose family estate is located here, where he wrote his wonderful poems.

Marina Guseva and Areg Totolian



**L-R: Guseva M., Kalinina N.,
Tuzankina I.**



**Totolian A. and Maródi L.
co-chairing the PID session**



Areg Totolian and his colleagues



Andrei Prodeus



**At the tomb of Alexander Pushkin.
R-L: Plebani A. and Maródi L.**



Visiting Pushkin Hills

Zakopane, Poland

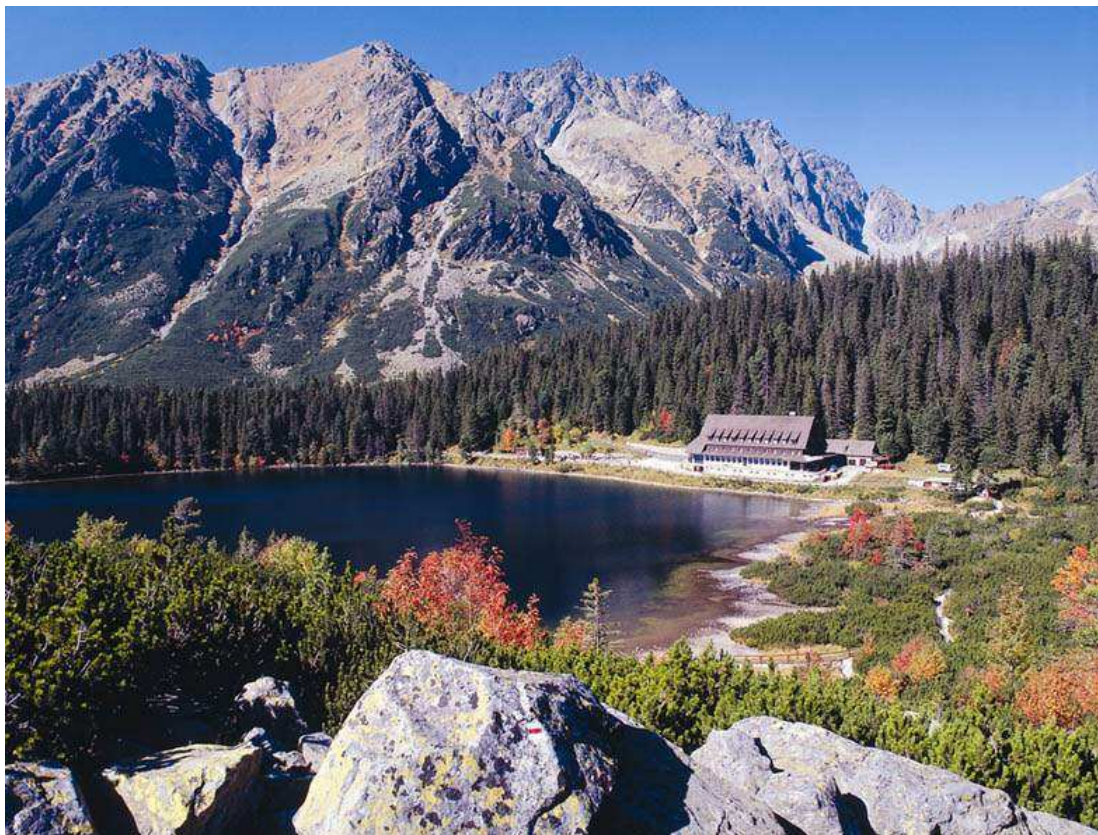
24-26 February, 2011



Host: Ewa Bernatowska (bernatowskae@yahoo.com)

Venue: Grand Hotel in Nosalowy Dwor

Main Topic: Standardization of diagnostic and therapeutic guidelines in primary immunodeficiencies and autoinflammatory diseases.



Zakopane

Program

25 February

Opening and introduction

Marcinkiewicz J. (Warsaw)

Bernatowska E. (Warsaw)

Euro-Gene – Scan Project

Berglof A. (Stockholm)

The J Project

Maródi L. (Debrecen)

ESID Subregistry on-line of Nijmegen

Breakage Syndrome

Bernatowska E. (Warsaw)

Nijmegen Breakage Syndrome – a cancer prone disease in clinical practice

Chrzanowska K. (Warsaw)

Peripheral T-cell subsets in NBS patients

Langerak T. (Rotterdam)

The precursor B-cell analyses in bone marrow of NBS patients

van der Burg M. (Rotterdam)

Peripheral B-cell abnormalities in children with NBS

Piatosa B. (Warsaw)

Discussion

Warsaw syndrome

Chrzanowska K. (Warsaw)

Primary defects of immune regulation

Maródi L. (Debrecen)

Mutational spectrum of WASP in Eastern and Central Europe

Gulácsy V. (Debrecen)

HOPI (Hungarian Organization for Primary Immunodeficiency)

Tóth-Apáti Nagy M. (Debrecen)

Regulatory T cells in transient hypogammaglobulinemia of infancy

Siedlar M. (Krakow)

Studies of TACI gene mutations in children with different types of hypogammaglobulinemia

Kowalczyk D. (Krakow)

Stomatological problems in immunodeficient patients

Olczak D.- Kowalczyk D. (Krakow)

IFNGR/IL12R – deficiencies

Pac M. (Warszawa)

26 February

Immune reconstitution after treatment of childhood leukemia

Szczepanski T. (Warsaw)

Allogenic haematopoietic stem cell transplantation as high effective and safe therapy for chronic granulomatous disease – single centre experience

Pituch-Noworolska A. (Krakow)

Autoinflammatory diseases – overview

Gathorno M. (Genoa)

Increasing awareness of periodic fever syndromes in Poland

Wolska-Kusnierz B. (Warsaw)

Family case of Netherton Syndrome

Kostyuchenko L. (Lviv)

HAE

Volokha A. (Kiev)

The antibodies against IBD in primary immune deficiency patients

Pituch-Noworolska A. (Krakow)

Subsets of B cells with THI and CVID
Bukowska-Strakova K., Kowalczyk D.
(Krakow)

Haemophagocytic lymphohistiocytosis-
patients at the Dept of Immunol Children's
Memorial Health Institute in Warsaw

Klaudel-Dreszler M. (Warsaw)

Autosomal Dominant Hyper-IgE
Syndrome - clinical and genetic analysis of
polish patient
Heropolitańska-Pliszka E. (Warsaw)

Summary and conclusion

On February 24-26, 2011 the 6th School of Clinical Immunology took place in Zakopane in Grand Hotel in Nosalowy Dwór. It was jointly organised by the Department of Immunology, Children's Memorial Health Institute, Warsaw, the JM Foundation, New York, and the ECE-IPI-CTR (J Project program). The introductory remarks were made by professor Ewa Bernatowska and professor Janusz Marcinkiewicz – the President of the Polish Society of Experimental and Clinical Immunology. There were about 50 participants including young immunologists from Central- and Eastern-European countries. The main topic of the meeting was standardization of diagnostic

and therapeutic guidelines in primary immunodeficiencies and autoinflammatory diseases. We were also proud to have well-known lecturers such as Mirjam van der Burg and Ton Langerak from the Netherlands, professor László Maródi from Debrecen, and excellent clinicians and scientists from Poland including professor Krystyna Chrzanowska from Warsaw and professor Anna Pituch-Noworolska from Krakow. One session was fully dedicated to the problems of Nijmegen breakage syndrome. There was also time for presentation of patients with unusual features of primary immunodeficiencies such as XLP, Netherton syndrome and periodic fever syndromes.

Ewa Bernatowska



THE AUDIENCE



László Maródi



Krystyna Chrzanowska

Oradea, Romania

01 March, 2011

Host: Ladislau Ritli (ritli_ladislau@yahoo.com)

Venue: "Dr Gavril Curteanu" Clinic Municipal Hospital, Oradea

Main Topic: Future strategies of management of PIDs



Oradea

Program

The J Project: Recent achievements and future challenges

Maródi L. (Debrecen)

The situation of the PID patient care in Romania

Șerban M. (Timisoara), Bataneanț M. (Timisoara)

The PID registry in Oradea: 2007- 2010

Ellenes J. Z. (Oradea), Miculschi G. (Oradea), Ritli L. (Oradea)

Gene expression studies in PID patients with novel missense mutations

Tóth B. (Debrecen)

ELA2 gene mutation in neutropenic patients

Székely A. (Debrecen)

Refractory cytopenias in siblings with CVID

Miculschi G. (Oradea)

Round table discussion

Summary and conclusion

The J Project meeting was organized in order to overview recent advances of the J Project in general and to discuss the possibilities of closer collaboration between the two neighboring PID centers, Oradea and Debrecen in Romania and Hungary, respectively. Professor Maródi L. gave a summary on the collaboration between the two centers as well as the remarkable activity of the Timisoara group, the leading Romanian center for immunodeficiencies patient care in the country. Professor Șerban M. and Dr. Bataneanț reported on the PID registry in Romania, and Dr. Ellenes informed us on the activity of the Oradea PID team. Beáta Tóth gave a very important and instructive talk on the diagnostic challenges when patients with novel missense mutation are defined. She presented the establishment of new cell laboratory in Debrecen which provides all the facilities to perform gene expression studies. Dr. Székely presented Hungarian patients with defined *ELA2* mutation, and Dr. Miculschi closed the session with the presentation on cytopenias as complications of CVID.

Ladislau Ritli

Debrecen, Hungary

10-11 March, 2011

Host: László Maródi (lmarodi@dote.hu)

Venue: Hotel Aquaticum

Main Topic:

Interdisciplinary Immunodeficiency



The participants of the 50th J Project Meeting in Debrecen

Program

10 March

J PROJECT ROUND TABLE

PID care in Slovakia: achievements and perspectives

Ciznar P. (Bratislava)

The emerging PID Center in St. Petersburg Region

Guseva M. (St. Petersburg)

Management of PID patients in Bulgaria: diagnostics and treatment

Naumova E. (Sofia)

Lithuanian primary immunodeficiencies

Duobiene R. (Lithuania)

The success of the J Project in Ukraine

Kostyuchenko L. (Lviv), Chernishova L. (Kiev)

Current state of PID care in Azerbaijan

Nasrullayeva G. (Baku)

Juniors PID School: a new perspective of the J PROJECT

Mironska K. (Skopje), Maródi L. (Debrecen)

The Primary Immunodeficiency Centers Network Project

Lebedowicz J. (Stockholm)

Current concepts and future challenges of the J PROJECT

Maródi L. (Debrecen)

KEYNOTE LECTURE – 1

Autoinflammatory diseases: a new category of primary immunodeficiencies

Aksentijevich I. (Bethesda)

11 March

WELCOME

Szöllősi J. (Budapest)

INNATE IMMUNE DEFICIENCIES AND PID SYNDROMES

Genetic defects of TLR-mediated immunity

Picard C. (Paris)

IL-12 receptor deficiency

Savchak I. (Lviv)

Comel-Netherton syndrome

Kostyuchenko L. (Lviv)

Hyper-IgE syndromes

Maródi L. (Debrecen)

Atypical hemolytic uremic syndrome as a primary immunodeficiency

Prohászka Z. (Budapest)

Primary MBL deficiency

Dérfalvi B. (Debrecen)

Ocular manifestations in APS type I.

Dohán J. (Budapest)

COMBINED IMMUNODEFICIENCIES AND SCT

Genetic defects that may cause Omenn syndrome

Bondarenko A. (Kiev)

Immunological archeology

Richter D. (Zagreb)

Post-vaccinal BCG infection in SCID patients

Prokofjeva T. (Riga)

Stem cell transplantation in WHIM syndrome patient

Kriván G. (Budapest)

B CELL DEFICIENCIES

X-linked agammaglobulinemia: The Italian experience

Plebani A. (Brescia)

Atypical findings in X-linked agammaglobulinemia

Sediva A. (Prague)

Follow-up of children with mild forms of antibody deficiency

Velbri S. (Tallinn)

J PROJECT COHORT STUDIES

Periodic fever syndromes in Central- and Eastern Europe

Toplak N. (Ljubljana)

Genetic characteristics of patients with the Wiskott-Aldrich syndrome in Central- and Eastern Europe

Gulácsy V. (Debrecen)

Nijmegen Breakage Syndrome – experience of Children's Memorial Health Institute

Pac M. (Warsaw)

KEYNOTE LECTURE – 2

Transposon technologies to manipulate the stem cell genome

Ivics Z. (Berlin)

Hearing impairment in patients with BTK mutation

Volokha A. (Kiev)





László Maródi (Hungary)



Peter Ciznar (Slovakia)



Elissaveta Naumova (Bulgaria)



Liudmyla Chernysova (Russia)



**Rasa Duobiene
(Lithuania)**



**Gulnara Nasrullayeva
(Azerbaijan)**



**János Szöllősi
(Hungary)**



Joanna Lebedowicz (Sweden)



Ioan Gherghina (Romania)



Capucine Picard (France)



Igor Savchak (Ukraine)



Zoltán Prohászka (Hungary)



Beáta Dérfalvi (Hungary)



**Nastya Bondarenko
(Ukraine)**



**Tatjana Prokofjeva
(Latvia)**



**Gergely Kriván
(Hungary)**



Darko Richter (Croatia)



Alessandro Plebani (Italy)



Alla Volokha (Ukraine)



Sirje Velbri (Estonia)



Press conference. L-R: Miklós Szolnoky (Hungary), László Maródi (Hungary), Joanna Lebedowicz (Sweden), László Harsányi (Hungary)

Summary and conclusion

The J Project physician education campaign was started in 2004, and since then has established itself as a prototype successful awareness program for primary immunodeficiencies (PIDs). The Project originated from the critical analysis of data in the ESID Registry before 2002, which showed that most Eastern European countries had reported less than ten patients with such disorders. These data suggested that PIDs may have not only been under-reported but under diagnosed in ECE. We launched the J Project meeting series in Targu Mures, Romania, on March 12, 2004 and celebrated the 50th J Project meeting in Debrecen in March, 2011 and by the end of the year, sixty awareness meetings on PIDs will have been organized in Albania, Azerbaijan, Belarus, Bosnia and Herzegovina, Bulgaria, China, Croatia, Czech Republic, Estonia, Hungary, Latvia, Poland, Republic of Macedonia, Republic of Moldova, Romania, Russia, Serbia, Slovakia, Slovenia, and Ukraine. In parallel, the number of diagnosed PID patients has increased to thousands in ECE. In addition, three non-European countries (Egypt, Iran, and Turkey) have also joined the Project by organizing “invited” J Meetings and the first J Project event entitled “*Clinical and Molecular Immunodeficiencies and Inflammatory Diseases: A J Project Session*” took place in Beijing at the BIT's 4th Annual International Conference and Exposition of Molecular Diagnostics. These meetings and the joint publications in international journals provide clear evidence of the success of our J Project. Even if we are pleased to see J Project events outside Europe, it is important to emphasize that the J Project has always been and will remain an ECE PID awareness activity.

László Maródi

Moscow, Russia

31 March- 01 April

Host: Irina Kondratenko (ikondratenko@rambler.ru)

Venue: Russian Children's Clinical Hospital

Main Topic: Educational Meeting “Primary Immunodeficiencies
in Pediatric Practice”



Moscow

Program

Mechanisms of innate and adaptive immunity

Kovalchuk L. (Moscow)

Interactions of innate and adaptive cells in immune response

Ignat`eva G. (Moscow)

Classification, clinical and laboratory presentations of severe combined immunodeficiencies

Kondratenko I., Pashchenko O., Deripapa E. (Moscow)

Principals of BMT and Hematopoietic Stem Cell Transplantation (HSCT)

Skorobogatova E. (Moscow)

Immunological and genetics changes in Common Variable Immunodeficiency (CVID)

Belevtsev M., Sharapova S. (Minsk)

Hereditary angioedema: Clinical and genetical aspects

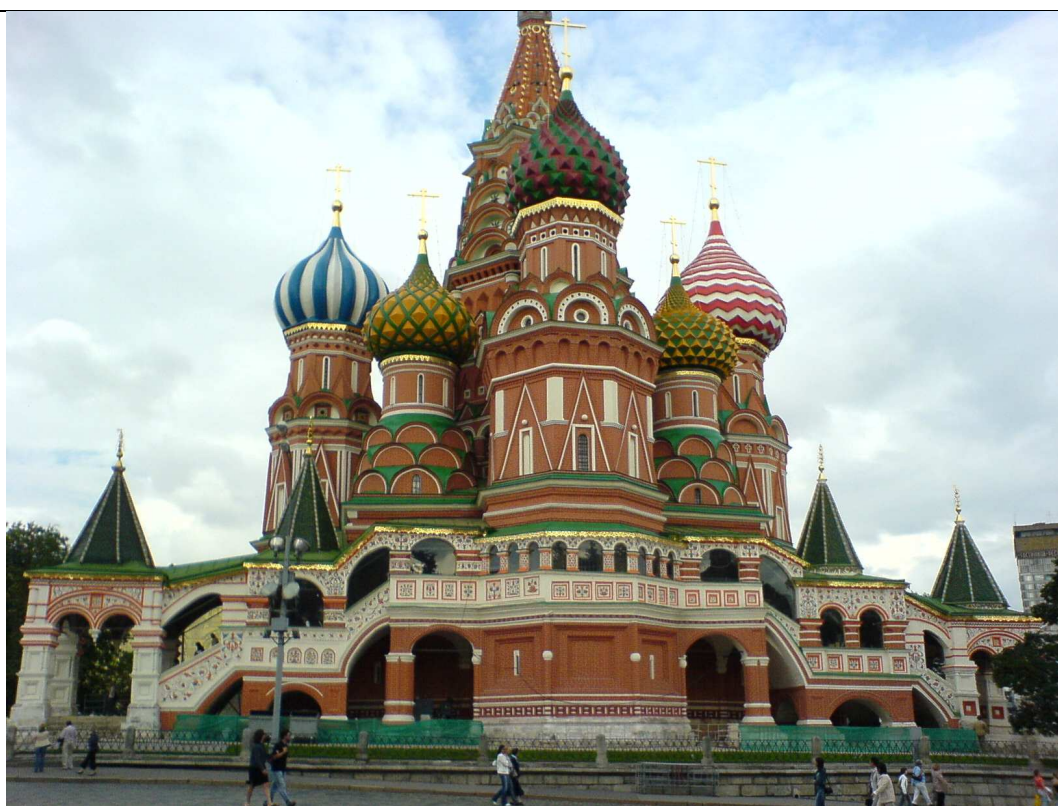
Kapustina A. (Moscow)

Summary and conclusion

On March 31 – April 1, 2011, the J Project Primary Immunodeficiency Diseases (PID) awareness meeting was held in Moscow, Russia. The meeting was organized by the Department of Clinical Immunology of Russian Children's Clinical Hospital, Russian State Medical University. The meeting was attended by 146 pediatricians and allergologists-immunologists from Moscow and Moscow Region outpatient departments and specialists from other regions of Russia, including St. Petersburg, Tula, Voronezh, Vladimir, Orenburg, and Orel from Belorussia. The J Project meeting in Moscow was entitled “Primary immunodeficiencies in pediatric practice” with the aim to discuss current diagnostic and therapeutic practices and problems in patients with PID. The main speakers were professor Leonid Kovalchuk – the chief of Immunology department of Russian State Medical University (Moscow), and professor Irina Kondratenko - the chief of Clinical immunology department of Russian Children's Clinical Hospital (Moscow). Professor Kovalchuk L. presented an overview of mechanisms of innate and adaptive immunity. Than his colleague

professor Ignat`eva G. presented an excellent report about interactions of cells in immune response. Professor Kondratenko I. with her colleagues from Clinical immunology department (assistant professor Pashchenko O. and doctor Deripapa E.) presented detailed overviews of classification, clinical and laboratory presentations of severe combined immunodeficiencies, combined T and B-cell immunodeficiencies, other well defined immunodeficiency syndromes, phagocytic defects and algorithms for diagnostic and therapy of PID. The chief of BMT department Skorobogatova E. talked over the principals of BMT and HSCT. Speakers from Belarus - Belevtsev M. and Sharapova S. presented reports about immunological and genetics changes in CVID. Dr. Kapustina A. made a high quality report about hereditary angioedema. Most lections contented the case reports and raised discussions with participants. The official language of the J Project Meeting in Moscow was Russian. Proceedings of the meeting will be published as a special issue of the Russian Journals of Pediatrics. The aim of our meeting to acquaint the pediatricians and immunologists from outpatient departments of Moscow and several regions of Russia with basic concepts of immunology and immunodeficiencies was fully realized.

Irina Kondratenko



Saint Brasil's Cathedral

Baku, Azerbaijan

22-23 April, 2011



Host: Gulnara Nasrullayeva (g.nasrullayeva@hotmail.com)

Venue: Landmark Business Center

Main Topic: Diagnostic and therapeutic challenges of PIDs



L-R: Ulviya Alekberova, Vafa Mamedova, Gulnara Nasrullayeva, László Maródi, Sevinj Isgandarova, Elnara Gurbanova, Shalala Ibragimova

Program

23 March

J Project Round Table

Welcome address

Nasrullayeva G. (Baku)

Status of Azerbaijan Society of Allergy and Immunology

Ayyubova A. (Baku)

The experience of European Societies of PID

Nasrullayeva G. (Baku)

Primary immunodeficiency syndromes and PID genes

Maródi L. (Debrecen)

Case of Lui-Bar Syndrome

Kabulov H. (Baku)

The influence of intrauterine infections on the forming of immune deficient in children

Mollayeva N. (Baku)

Immune diagnostics of PID: Main Directions

Isqandarova S. (Baku)

Coffee break

Cytokine therapy in immune rehabilitation of frequent diseased children

Sultanova N. (Baku)

Family case of selective IgA deficiency

Panahova T. (Baku)

Features of immune prophylaxes of immune compromising children

Isayeva R. (Baku)



**L-R: Namik Pashayev, László Maródi,
Gulnara Nasrullayeva, Namig Nasrullayeva**

Summary and conclusion

The first J Project meeting in a Caucasian region was organized in Baku, Azerbaijan right after the 50th meeting in Budapest which was attended by professor Gulnara Nasrullayeva, head of the PID team in Azerbaijan State Medical University. The meeting was attended by opinion leaders in the field of allergy and clinical immunology from the different places of the country. László Maródi, the leader of the J Project program was invited to summarize achievements of the Project over the past 7 years. Such a PID awareness program is desperately needed in a country like Azerbaijan with a population of more than 9 million, but only with just a few immunologically diagnosed PID patients and without any possibility of PID genetic testing. The enthusiastic and professionally prepared group of clinical immunologists and laboratory staff from Baku is very keen to start awareness campaign countrywide and to improve the figures in the near future. This activity is now supported by the availability of genetic testing in Debrecen for patients in Azerbaijan under the J Project program. In addition, the Baku PID center based on the immunological and pediatric departments has become part of new network called “Central and Eastern European Primary immunodeficiency Centers Network (CEE-PID-CN)” jointly supported by the Jeffrey Model Foundation and Octapharma Nordic. It is among the plans in the near future to establish PID genetic testing in Baku by making use of this support. Our next step to increase awareness will also include the organization of PID-centered meetings in other university cities and large hospitals in Azerbaijan. The Baku initiative will likely be an example to be followed by other Caucasian and post-Soviet Union countries in the region.

Gulnara Nasrullayeva

Bratislava, Slovakia

28-29 April, 2011

Host: Peter Ciznar (ciznar@dfnsp.sk)

Venue: Pediatric Teaching Hospital Bratislava, Lecture Hall

Main Topic: Symposium on Primary Immunodeficiencies, Early Diagnosis and Treatment



Bratislava at night

Program

Timely diagnosis of PID

Welcome

Ciznar P. (Bratislava)

Primary immunodeficiencies and EU actions

Mikolásik M. (Bratislava)

Classification and manifestation of primary defects of immune regulation

Maródi L. (Debrecen)

Infectious complications of PID patients

Richter D. (Zagreb)

Clinical manifestations of humoral immunodeficiencies

Litzmann J. (Brno)

Coffee break

PID therapy

Replacement therapy with immunoglobulins

Ciznar P. (Bratislava)

Bone marrow transplantation

Horáková J. (Bratislava)

BCG vaccination in immunocompromised child

Thon V. (Brno)

Lunch break

PID syndromes

Hereditary angioedema

Hrubisko M. (Bratislava)

X-linked agammaglobulinaemia

Jesenak M., Banovcín P. (Martin)

Chronic granulomatous disease

Kayserová H. (Bratislava)

Familial hemophagocytic lymphohistiocytosis

Kayserová J. (Bratislava)

Coffee break

Case presentations

Organ complications of CVID in adults

Straková J. (Martin)

Autoimmune complications in twins with CVID

Raffac S. (Kosice)

Complete deficiency of IFNGR1 receptor for interferon gamma

Ostró R. (Kosice)

Severe combined immunodeficiency

Sufliarska S. (Bratislava)

T-ALL bcr/abl positive patient with Nijmegen Breakage syndrome

Mikesková M. (Bratislava)

Syndrome papillon Lefèvre

Staníková D. (Bratislava)

Hyper-IgE syndrome

Véghová L. (Bratislava)

Dinner

Summary and conclusion

A one-day meeting was the first meeting of J Project in Slovakia. It was held at the Detská fakultná nemocnica (Pediatric Teaching Hospital), the largest facility of its kind in Slovakia, seated in the capital city Bratislava, under the auspice of Slovak Immunology Societies. Event was attended by 58 registered participants, mostly pediatricians and clinical immunologists, from all over the Slovakia. The program was designed to introduce the broad topic of Primary immunodeficiencies with the focus on early diagnosis and readily available treatment strategies. At the beginning of the symposium Dr. Miroslav Mikolasik, Slovak member of the European Parliament presented policy of EP regarding rare diseases including Primary immunodeficiencies and the EP support for dissemination of knowledge and information about rare diseases. Subsequently professor Maródi introduced the J Project mission and its activities. Thereafter he continued with the perspectives of PID, focused on the defects of immune regulation. Next among the invited speakers was Dr. Richter from Zagreb, Croatia who had a lecture on the clinical consequences of different PIDs and professor Litzman from Brno, Czech Republic on the clinical phenotype of predominantly B cell immune defects. Presentations of clinical experiences with antibody replacement therapy and bone marrow transplantation at the Pediatric Teaching Hospital in Bratislava followed after a short break. The morning part ended with a hot topic issue for Slovakia, the BCG vaccination and its complication in PID children, presented by Dr. Thon from Brno. Afternoon session was divided into two parts, PID syndromes and case presentations. Colleagues from Prague and all parts of Slovakia had an opportunity to present here their experiences and address various medical issues. The official program ended by a panel discussion. All participants of the meeting obtained credits of Slovak Accreditation Board for Continuing Medical Education (SACCME). The primary objective of the symposium, to increase the awareness of PID in the medical community seems to be achieved and hopefully started a new era of a regular platform for “meeting with experts” on the PID topic in Slovakia.

Peter Ciznar



L-R: Olga Cervenova, Zuzana Lalahova Striezencova, Daniel Zitnan, László Maródi



Participants of the Meeting



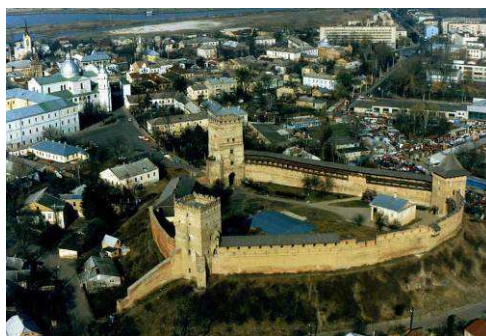
L-R: Darko Richter (Croatia), László Maródi (Hungary)



**L-R: Peter Ciznar (Slovakia),
László Maródi (Hungary),
Miroslav Mikulasek (EP member,
Slovakia)**

Lutsk, Ukraine

11-12 May, 2011



Host: Liudmyla Chernyshova (chernyshova@ukr.net)

Venue: Regional Health Administration Center of Lutsk region

Main Topic: Primary Immunodeficiencies-School seminars



Symposium organizers and speakers

Program

Opening Ceremony

Results of J Project
activity in Eastern
European countries
Maródi L. (Debrecen)

Achievements and
problems of pediatric
immunology in Ukraine
Chernyshova L. (Kiev)

Approaches for creating
the National Register of
PID
Bondarenko A. (Kiev)

Slavic mutation -
Nijmegen Syndrome -
cases from practice
Kostyuchenko L. (Lviv)

BMT: the Brescia
experience
Porta F. (Brescia)

Importance of genetic
investigation for
diagnostics and
prohylactics of primary
immunodeficiency
Volokha A. (Kiev)

Early symtoms of
combined
immundeficiency in
children
Samarin D. (Kiev)

Defect of phagocytosis:
diagnostic problems
Lapyi F. (Kiev)

Section with parents of
patients with primary
immunodeficiency

Possibilities of
immunologic
investigation of children
in Lutsk
Silich P. (Lutsk)

Case of family
Mediterranean fever
Stepanovsky Y. (Kiev)

Discussion

Closing of symposium



L-R: Fulvio Porta, Alla Volokha, and Liudmyla Chernyshova



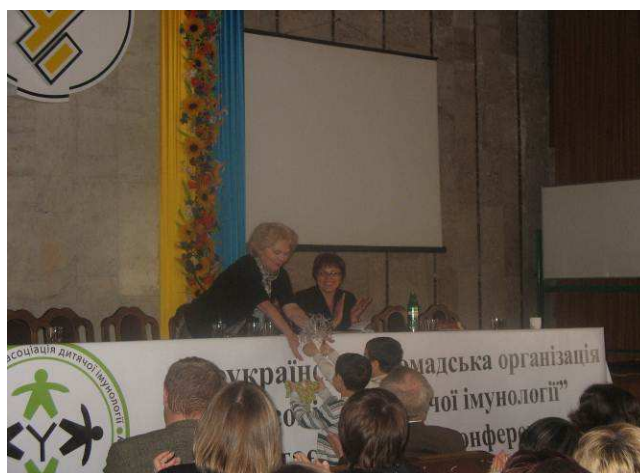
Anastasia Bondarenko



Liudmyla Chernyshova



THE AUDIENCE



Present from the children

Summary and conclusion

The Seventh Ukrainian J Project Meeting had held on May 12, 2011 in Lutsk. Lutsk is a city located by the Styr River in northwestern Ukraine. It is the administrative center of the Volyn Oblast (province). Improving the awareness of primary immunodeficiencies in Ukraine is provided by holding local seminars in different regions for primary care contact doctors and Lutsk became the seventh of this educational program. Over 200 doctors from the Volyn region, about 20 immunologists from other regions of Ukraine as well as guests from Italy had took part in the seminar. At the beginning of the symposium participants heard a statement by professor L. I. Chernyshova, the Head of Ukrainian Association "Pediatric Immunology", which highlighted the main achievements of Pediatric Immunology in Ukraine over the last decade and prospects for development. Reports made at the seminar were devoted to the diagnosis of major groups of PID, presenting cases, genetic testing capabilities, and the formation of a national registry. Besides, the participants became acquainted with the latest developments in the diagnosis of combined immunodeficiency, which were announced by an expert in stem cell transplantation of the European Society of Primary Immunodeficiencies Dr. Fulvio Porta. This was particularly informative due to the fact that stem cell transplantation for PID patients in Ukraine is still unavailable. Dr. Fulvio Porta is a head of the Department of hematology and bone marrow transplantation (Brescia, Italy), where Ukrainian patients can receive treatment due to help of charity organization "Lifeline Italia". In particular, successful stem cell transplantation was done for two patients with severe combined immunodeficiency from the Volyn region. PID patient's parents were traditionally invited to participate in the seminar. Their reports described the path to the diagnosis of their children and the current problems they face during treatment. Speeches of representatives of the patients had a great impression on the audience of doctors. Also, using this opportunity an international consultation was held for two brothers with WAS. Conducting School in an atmosphere of elevated mood, lively discussions and the target themselves to international standards allows hoping for more effective detection of primary immunodeficiencies in the Volyn region and effective cooperation in the future.

Liudmyla Chernyshova

Sibiu, Romania

10-11 June, 2011

Host: Iurian Sorin Ioan (iurian_sorsab@hotmail.com) and Neamtu Mihai

Venue: Aula "Avram Iancu" of "Lucian Blaga", University of Sibiu

Main Topic: Crossroads of medicine: hematology and primary immunodeficiencies



Sibiu – main square

Program

10 June

SATELLITE COURSE WITH TOPIC „BASICS IN IMMUNOLOGY. PRIMARY IMMUNODEFICIENCIES”

11 June

„J PROJECT MEETING” LECTURES

Crossroads of hematology and immunology: X-linked lymphoproliferative disease
Maródi L. (Debrecen)

Clinical and biological aspects in a combined immunodeficiency
Gherghina I. (Bucharest)

Neutropenia associated with hypogammaglobulinemia in infancy. Case report
Bataneant M. (Timisoara)

Romanian Association for Patients with Primary Immunodeficiencies, "ARPID"
From a dream to reality
Baldea A. (Hateg)

Benign Tumor Pathology Associated With Common Variable Immunodeficiency
Rugina A. (Iasi)

What was first: immunodeficiency or malabsorption?
Capilna B. (Sibiu)

Primary immunodeficiencies in Romania
Past, present and future
Bataneant M. (Timisoara)

Severe neutropenia. Case report
Iurian, S. I. (Sibiu)

Malignant hemopathies One side of the primary immunodeficiencies
Bataneant M. (Timisoara)

CVID, a complex disorder of the immune system
Cristea V., Miron N. (Cluj-Napoca)



Sabina Iurian and Sorin Iurian



Brindusa Capilna

Summary and conclusion

The 55th J Project Meeting dedicated to PIDs was held in Sibiu under the auspices of “Lucian Blaga” University from Sibiu, Romanian Medical Council and Romanian Working Group for Primary Immunodeficiencies. This conference was organized by professor Neamtu Mihai and Dr. Iurian Sorin and represented the 7th J Project organized in Romania since 2004. The meeting was attended by about 30 immunologists from other medical universities from Romania (Targu-Mures, Timisoara, Cluj-Napoca, Bucharest, Iasi), general practitioners, residents in pediatrics and pediatricians and represented a huge opportunity to share their experience and to improve the PIDs’ diagnosis. The clinical cases raised a lot of interest among the participants and many of the presenting authors stressed that PIDs diagnosis improved in Romania due to genetic sequencing made in the PID Center in Debrecen, Hungary. The conference was preceded by a satellite course entitled „Basics in immunology. Primary immunodeficiencies” attended by more than 100 general practitioners. In context of reduced number of PIDs cases diagnosed in Sibiu county, the course was focused on pathogenesis and clinical features of primary immunodeficiencies in order to improve the early diagnosis and treatment. We noticed the participation at the meeting not only of patients and parents diagnosed with PIDs but also of board members from Romanian Association for Patients with Primary Immunodeficiencies (ARPID) that recently became a part of International Patient Organization for PIDs. The aims of ARPID consist in supporting children and adults with PIDs and their families to provide and protect their fundamental rights concerning medical, social and educational assistance, establishing collaboration with physicians working with PIDs patients, sustaining their interests, collecting funds from programs and establishing contacts with scientific and charitable organizations from medical field in Romania and other countries. Even if there is no immunology center in Sibiu yet, we remarked a growing interest of pediatricians to increase diagnosis and treatment of children with PIDs. In this context, we’ll start making efforts to initiate a Diagnosis and Treatment Center for PIDs in Sibiu County using genetic diagnostic methods with the support of Professor László Maródi and his colleagues from Debrecen, Hungary. Such a center should provide diagnostic help nationwide in Romania and would be part of the *J2 Project program* aimed at establishing targeted gene sequencing in all Eastern and Central European countries.

Iurian Ioan Sorin



L-R: Profs. Gherghina I. and Capilna B.



Members of ARPID



Sibiu: City of Europe 2007



ASTRA National Museum Complex

Chelyabinsk, Russia

26-27 June, 2011



Kurchatov

Hosts: Irina Tuzankina (ituzan@yandex.ru)
Alexander Pishchalnikov (moiseevatn@yandex.ru)

Venue: Chelyabinsk State Medical Academy

Main Topic: Diagnostics of PIDs in the Urals region



“No one forgets and nothing is forgotten”

Program

June 21

SYMPOSIUM FOR PRACTITIONERS “IMMUNOPATHOLOGY IN CLINICAL PRACTICE” PROJECT

What can we do more for a patient with asthma?

Ignatova G. L. (Chelyabinsk)

New approaches to the treatment of mild bronchial asthma

Ryabova L.V. (Chelyabinsk)

Features of hormonal status in pregnant women with asthma

Studneva N. A. (Chelyabinsk)

Bronchial asthma and chronic obstructive pulmonary disease: similarities and differences

Emelianov A.V. (St. Petersburg)

Modern understanding of the pathogenesis and principles of therapy of atopic dermatitis

Monahov K. N. (St. Petersburg)

Skin Dysbiosis

Pisklakova T. P. (Russia)

Intestinal dysbiosis - imbalance of immune system

Zhuravleva T. V. (Chelyabinsk)

Immunopathology and genetics of mucocutaneous candidal disease

Maródi L. (Debrecen)

Clinical Immunology portraits

Tuzankina I. (Ekaterinburg)

Rare forms of PID in the register of Sverdlovsk region

Vlasova E. V. (Ekaterinburg)

Immunodeficiencies. View of Clinical immunologist

Pishchalnikov A. Y. (Chelyabinsk)

PID registry of Chelyabinsk region

Moiseeva T. N. (Chelyabinsk)

Cytokines in the practice of a physician today and tomorrow

Simbirtsev A. S. (St. Petersburg)

Some aspects of PID laboratory diagnostics

Pashnina I. (Ekaterinburg)

Thymus abnormalities at children with multiple malformations”.

Bass E. A., Abolina T., Kobeleva Y. (Ekaterinburg)

Respiratory infections and immunity: new approaches to the diagnosis of disorders and their correction

Karaulov A. V. (Moscow)

Immunomodulators - Pros and Cons

Ketova G. G. (Chelyabinsk)

Problems of adult allergology-immunology service in Chelyabinsk region.

Abramova N. N. (Chelyabinsk)

Contemporary problems in allergologist's and immunologist's practice.

Zherebtsova I. A. (Chelyabinsk)

Children's allergology-immunology service in Chelyabinsk region.

Malakhova S. I. (Chelyabinsk)

Summary and conclusion

In June, 2011 International PID school for physicians within the framework of “J Project” was held in Chelyabinsk. The school was entitled “Immunopathology in clinical practice.” It was organized by Russian Society for immunology (Tuzankina I., Ekaterinburg; Pishchalnikov A., Chelyabinsk) and professor Maródi L., Debrecen, Hungary. This school attracted physicians, pediatricians, allergists, immunologists and other doctors not only from Chelyabinsk, but from other cities as well (Ekaterinburg, Perm, Tyumen, Orenburg, Kurgan, Ufa etc.). More than 150 specialists of Ural region as well as immunologists from Moscow and St. Petersburg took part in this conference. This school gave an opportunity to exchange experience between doctors that work with PID patients and doctors of other specialties. Presentation of PID registries data in Ekaterinburg and Chelyabinsk regions was very interesting

for the audience. Also presentation of rare and difficult cases in differential diagnostics at children with primary immunodeficiency diseases was made. Most lectors noted that PID diagnostics in Chelyabinsk region became better. Professor Maródi L. (Debrecen, Hungary) got acquainted with Chelyabinsk Regional Clinical Children’s Hospital on the basis of which the regional PID register is conducted by Pishchalnikov A. for 15 years. Professor Maródi also observed PID patients and noted a rather high diagnostic level in Hospital. During the conference was also noted that molecular genetic testing is a very important part of diagnostic process. That is why Urals doctors came to agreement with prof. Maródi to conduct some research together. Also interactive consilium “Diagnostics of rare PID forms in Ekaterinburg registry” was held with participation of all specialists.

Irina Tuzankina



**Center: Tatjana Moyseeva, Irina Tuzankina,
László Maródi, Alexander Pishchalnikov**

Ohrid, Republic of Macedonia

08 October, 2011

Hosts: Kristina Mironska (mironska2002@yahoo.com),
Katarina Stavric, and Lidija Kareva

Venue: Metropol Lake Resort, Ohrid

Main Topic: Novel primary immunodeficiencies



Lake Ohrid in twilight

Program

J Project Session

Novel Primary Immunodeficiency disease

Maródi L. (Debrecen)

Neonatal screening for severe combined immunodeficiency

Kareva L. (Skopje)

Epidemiological, clinical and molecular characteristics of 38 East-Central European patients with chronic granulomatous disease

Avcin T. (Ljubljana)

Hemophagocytic lymphohistiocytosis and macrophage activating syndrome in a case with EBV infection. Case report.

Kuli-Lito G. (Tirana)

Severe congenital neutropenia

Mironska K. (Skopje)

Vlproic ACID as a trigger of HAE attacks case report

Stavric K. (Skopje)

Severe pseudallescheria boydi infection in a boy with X-linked CGD-case report

Pasic S. (Belgrad)

Periodic Fever Syndromes

Toplak N. (Ljubljana)



Famous pianist from Macedonia **Simon Trpchevski** gave an unforgettable recital after the J Meeting.

Summary and conclusion

Macedonia was one of the first countries to join the J Project program with a meeting in Skopje, September 18, 2004 organized by the team of Macedonian pediatric immunologists Peova Sonja, Mironska Kristina, Stavrik Katarina and Kareva Lidija. The next meeting in Macedonia, was organized in Ohrid, September 20-21, 2007 by the same team. Now, the meeting of the 58th J Project for PID was held, for the third time in Macedonia, under the auspices of the Department of Immunology, University Clinic for Children`s Diseases, and Medical Faculty in Skopje and the Pediatric Association of Macedonia. The meeting also took place in Ohrid, at Metropol Lake Resort, along the beautiful Ohrid Lake. The program put together by Kristina Mironska was integrated to the 5th Congress of Pediatric Association of Macedonia with international participation, from October 05-09, 2011. The aim, the same as the last time, was to achieve a bigger turnout, thus obtaining greater success in rising the awareness of the existence of Primary Immunodeficiency Diseases among the general practice doctors, specialists and even wider. The auditorium was represented by about 100 participants of the Pediatricians' Congress which enriched the importance of this meeting. László Maródi gave an introductory talk on novel primary immunodeficiencies with an emphasis on recent discoveries of chronic mucocutaneous candidiasis disease. Lidija Kareva summarized the immunological and genetical approaches and the availability of screening severe combined immunodeficiency disorders. Tadej Avcin discussed the clinical, epidemiological and genetic characteristics of patients diagnosed with CGD in ECE countries. Georgina Kuli-Lito presented an intriguing case with HLH and macrophage activating syndrome which developed in a girl after EBV infection. Srdjan Pasic presented an interesting case of severe,

disseminated Pseudall-escheria boydi infection in 12 year old boy with x-linked CGD, with conclusion that in spite of the new generation of antifungal drugs, SCT represents the only hope for definite cure of CGD in setting of deep-seated fungal infection. Katarina Stavric presented the first case of patient with hereditary angioedema and epilepsy, where the valproic acid is a suspected trigger of the HAE attacks. Kristina Mironska presented case with severe congenital neutropenia and the discussion this case raised the possibility of WHIM syndrome, rare form of this PID. Genetic analysis of the patient in Debrecen was later performed and unveiled disease-causing mutation in the CXCR4 gene. This case nicely demonstrated the positive outcome of the J Project meetings not only as general awareness activities but also add the individual patient level. Natasha Toplak, who was attending the Congress, enriched the J Project meeting with her presentation of the periodic fever syndromes, pointed that the possibility for free genetic testing for the countries in central and south-eastern part of Europe, should be offered by the countries which has the means to do so. During the social events there were enough spaces for discussion of any topics related to PID in quite informal way. The social part was enriched with attendance a Concert of the famous pianist in Macedonia and abroad, Simon Trpchevski in a very acoustic hall of the church “St.Sofia”. This meeting was rated as successful and it completely justified the purpose of its taking place. We are using the opportunity to thank all the participants for their contributions, as well as our platinum sponsor Alkaloid Skopje, and Prof. László Maródi, Chair of the ECE-IPI CTR for his continuous support and help with the organization of the meeting.

Kristina Mironska



László Maródi (Hungary)



Lidija Kareva (Macedonia)



Georgina Kuli-Lito (Albania)



Tadej Avcin (Slovenia)



Kristina Mironska (Macedonia)



Katarina Stavric (Macedonia)



Srdjan Pasic (Serbia)



Natasha Toplak (Slovenia)

Tehran, Iran

15 October, 2011

Host: Nima Rezaei (rezaei_nima@yahoo.com)

Venue: Imam Khomeini Hospital

Main Topic: Clinical Immunology and Immunodeficiencies

Meeting



Speakers and organizers. Back center: Hans Ochs and Nima Rezaei

Program

October 15

Session One: Asthma and Allergy: Diagnosis and Treatment

Approach to a Child with Asthma
Moin M. (Iran)

Approach to a Child with Allergic Rhinitis
Gharagozlou M. (Iran)

Approach to a Child with Atopic
Dermatitis
Movahedi M. (Iran)

Panel Discussion

Session Two: Clinical Immunology: Approach to Immunodeficiencies

Approach to a Child with Recurrent
Infections
Aghamohammadi A. (Iran)

Approach to a Child with Congenital
Neutropenia
Rezaei N. (Iran)

Autoimmunity in Primary
Immunodeficiency Disorders
Al-Herz W. (Kuwait)

Panel Discussion

Session Three: Advances in Primary Immunodeficiencies: Diagnosis and Treatment

Advances in Primary Immune Deficiency
Diseases
Ochs H. (Seattle)

World-Wide Review of Progress in Gene
Therapy for Primary Immune Deficiencies:
Current Studies and Future Technologies
Malech H. (Bethesda)

Transplantation for Primary
Immunodeficiencies with a focus on
Chronic Granulomatous Disease: Who,
When and How?
Kang E. (Bethesda)

Panel Discussion

Session Four: Poster Presentation (Lunch time)

Session Five: Selected Oral Presentation

Session Six: Case Discussion



**L-R: Elisabeth Kang, Nima Rezaei, Asghar Aghamohammadi,
Hans Ochs, Asghar Ramyar, Harry Malech**

Summary and conclusions

The 4th Meeting on Clinical Immunology and Immunodeficiencies, which was also considered as 59th J Project Meeting was organized in October 15th, 2011 (Tehran, Iran). In 2005, first International Congress on Immunodeficiency Disorders was organized in Tehran. A number of PID experts from different countries (USA, UK, Germany, France, Italy, Sweden, Spain, Japan, and Turkey) attended the congress to present an update in this field, whereas many scientists and researchers took part in this congress to increase their knowledge. This congress was a great event to further develop bilateral scientific exchange of Iranian scientists with other researchers of the world. In October 2009, alongside with the 21st International Congress of Pediatrics, a joint meeting on Immunodeficiency Diseases was established. That two days meeting was also supported by the Jeffrey Modell Foundation (JMF). The great success of these conferences brought up the idea of organizing the Clinical Immunology, Allergy and Immunodeficiency Diseases Meeting this year, just before the 22nd International Congress of Pediatrics. This meeting, which was also considered as The 46th J Project Meeting, is organized by the

Research Group for Immunodeficiencies in collaboration with the Department of Pediatrics, Children's Medical Center Hospital, Tehran University of Medical Sciences. As of establishment of the Research Center for Immunodeficiencies, the 4th Meeting on Clinical Immunology and Immunodeficiencies was organized by this center in October 15, 2011 (Tehran, Iran) along with the 23rd International Congress of Pediatrics to focus on the linkages of fundamental sciences and patient-oriented research under the main theme of clinical immunology and immunodeficiency diseases. Improving the awareness of physicians, pediatricians and other specialists would be the main objective of the meeting. The major part of audiences includes general practitioners, pediatricians, and pediatric subspecialists; however, medical students, pediatric residents and fellows in the field of clinical immunology and infectious diseases are expected to actively participate in the meeting. In addition to local speakers, some international well-known persons were also attended this meeting and presented interesting lectures for the audiences. Professor Hans Ochs (Seattle Children's Hospital, University of

Washington, Seattle, WA), Professor Harry Malech (National Institute of Allergy and Infectious Diseases, NIH, Bethesda, MD) and Elisabeth Kang (National Institute of Allergy and Infectious Diseases, NIH, Bethesda, MD) travelled from the US, while Dr. Waleed Al-Herz came from Kuwait which all presented excellent talks on recent advances in diagnosis and treatment of patients with primary

immunodeficiencies. The meeting which was supported by the JMF, as before, consisted of 6 sessions, including Asthma and Allergy: Diagnosis and Treatment, Clinical Immunology: Approach to Immunodeficiencies; Advances in Primary Immunodeficiencies: Diagnosis and Treatment; Poster Presentations; Selected Oral Presentations; and Case Discussion.

Nima Rezaei
Asghar Aghamohammadi



Trabzon, Turkey

19-20 November, 2011

Host: Fazil Orhan (fazilorhan@yahoo.com)
Ismail Reisli (ireisli@hotmail.com)

Venue: Grand Zorlu Hotel, Trabzon

Main Topic: Primer immune deficiencies in diagnosis and treatment



Program

19 November



AVRUPA İMMÜN YETMEZLİKLER DERNEĞİ
EUROPEAN SOCIETY FOR IMMUNODEFICIENCIES

KARADENİZ TEKNİK ÜNİ. TIP FAKÜLTESİ
ÇOCUK İMMÜNOLOJİSİ ve ALLERJİ BİLİM DALI

PRİMER İMMÜN YETMEZLİKLERİN TANI VE TEDAVİSİ

GRAND ZORLU HOTEL TRABZON
19 - 20 KASIM 2011



PROGRAM

19.11.2011

1430	AÇILIŞ
	PRIMARY IMMUNE DEFECTS OF TH17-MEDIATED IMMUNITY Prof. Dr. Laszlo Marodi - University of Debrecen, Hungary
1500	İMMÜN YETMEZLİK SINIFLAMASI VE TARAMA TESTLERİ Prof. Dr. Alihan Yıldırım - Ondokuz Mayıs Üniversitesi Tıp Fakültesi
1520	ÇOCUKLARDA PRİMER İMMÜN YETMEZLİK DÜŞÜNDÜREN BULGULAR Prof. Dr. Fazıl Örnek - Karadeniz Teknik Üniversitesi Tıp Fakültesi
1540	KAHVE ARASI
1600	YETİŞKİN ÇAĞDA PRİMER İMMÜN YETMEZLİKLER Prof. Dr. Zafer Çalıklar - Selçuk Üniversitesi Meram Tıp Fakültesi
1620	PRİMER İMMÜN YETMEZLİKLERİN TEDAVİSİ Prof. Dr. İsmail Reisi - Selçuk Üniversitesi Meram Tıp Fakültesi
1645	KAPANIŞ

20.11.2011

SOSYAL PROGRAM



Summary and Conclusions

I believe that primary immunodeficiency (PID) is an important public health problem in Turkey because of high rate of consanguineous marriage. The prevalence of severe combined immunodeficiency is also very high in Anatolia (*one in 10,000 live births in Konya*) due to high ratio of consanguinity than in literature. We have to organize educational meetings in Anatolia to increase the awareness of primary immunodeficiency diseases. For this purpose, the second J Project Meeting in Turkey was held on 19-20 November, 2011, in Trabzon. The meeting was organized by Department of Pediatric Immunology and Allergy in Medical School of Karadeniz Technical University. The meeting was focused on the awareness of PID among the medical students, physicians and pediatricians, also to discuss PID diagnostic problems, clinical symptoms, treatments and to clarify importance of early diagnose in PID. More than 70 participants were attended the meeting. The meeting was started with an opening lecture by Prof. Dr. László Maródi from Hungary. He had given a lecture about primary immune defects of TH17-mediated immunity. After, four lectures were given in the meeting which was based on diagnostic problems, clinical signs, screening and treatment of PID. The first part of meeting was started with a lecture about the classification and screening tests of PID by Prof. Dr. Alişan Yıldırım from Ondokuz Mayıs University (Samsun) and continued with the warning signs in children with PID by Prof. Dr. Fazıl Orhan from Karadeniz Technical University (Trabzon). The second part of the meeting were dedicated to the warning signs of PID in adults by Prof. Dr. Zafer Çalışkaner from Selcuk University (Konya) and the treatment of PID by Prof. Dr. İsmail Reisli from Selcuk University (Konya). On the evening, the shuttle buses transferred the participants to a nice restaurant where was situated at near the sea side for a traditional Trabzon dinner. Among social events, there were visits to Ataturk's house, Trabzon's Museum, Ayasofya (Hagi Sofia) Museum and sightseeing of Trabzon. We would like to thank all speakers and participants for their contributions, especially Prof. Dr. László Maródi and the local sponsors (Eczacıbaşı-Baxter) for their financial help which made it possible to organize this event.

Ismail Reisli



**L-R: László Maródi, Zafer Caliskaner, Fazil Orhan, and Alisan Yildiran
in front of Ayasofya Museum.**



**L-R: Fazil Orhan, Zafer Caliskaner, László Maródi, and Ismail Reisli
in front of Ataturk's House and Museum in Trabzon**

Riga, Latvia

26 November, 2011



Host: Tatjana Prokofjeva (monja@balticom.lv)

Venue: Maritim Park Hotel, Riga

Main Topic: “PIDs in children and adults”



Program

Registration

Welcome and Introduction

Overview of PID patients care in Latvia
Prokofjeva T. (Riga)

International protocols for the PID diagnosis
Guseva M. (St. Petersburg)

Possibilities for the laboratory PID diagnostics in Lithuania
Malickaite R. (Vilnius)

Immunological and genetic variability of SCID
Sarapova S. (Minsk)

Clinical manifestations, immunology dysfunctions and genetic disorders in patients with ALPS
Belevtsev M. (Minsk)

Two cases of Good syndrome. Diagnostic difficulties.
Kalinina N. (St. Petersburg)

Coffee Break

Common variable immunodeficiency in adults
Jaunalksne I. (Riga)

Biotest
Becker M. (Dreieich)

Hyper IgE Syndrome: difficulties in diagnostics, case report
Kurjane N. (Riga)

Clinical presentation and immunologic aspects in patients with genetically confirmed 22q 11del
Grinfelde I. (Riga)

WAS syndrome. Clinical case.
Medne G. (Riga)

Coffee Break

Autoinflammatory syndromes: Lecture. Periodic Fever syndromes
Toplak N. (Ljubljana)

Discussin. Closing remark



Summary and conclusion

The J Project meeting was held for the second time in Riga, Latvia, on the 26th of November, 2011. The meeting was organized by the Latvian Association of Immunologists. Latvia is a small country near the Baltic Sea with a population of less than 2 million people. Capabilities of the laboratory of PID diagnostics (especially molecular-genetic diagnostics) in Latvia still are limited. The main targets of the conference were to improve awareness of PID not only among the pediatricians but also among general practitioners and other specialists, and provide information about diagnostics and therapeutic practices and problems in patients with PID. The meeting was attended by more than 100 participants (pediatricians, immunologists, infectologists, geneticists, general practitioners as well as students and residents and laboratory specialists). Among the guests there were participants from neighboring countries such as Lithuania, Estonia, Republic of Belarus, St. Petersburg (Russia), as well as from Slovenia and Germany. The first part of the meeting began with the overview of PID patients' care in Latvia. M. Belevtsev from the Republic of Belarus Research Center for Pediatric Oncology and Hematology center (Minsk) reported clinical and genetic diagnostics of 5 cases of ALPS in children in Belarus. Prof. Natalia Kalinina (St. Petersburg, Russia) had presentation of 2 Good syndromes. Sirje Velbri (Tallinn, Estonia) and Inta Jaunalksne (Riga, Latvia) had presentations about CVID, and demonstrated PID cases in adults. The Hyper-IgE diagnostics, clinical aspects and immunopathology were presented by Natalija Kurjane (Latvia). The third part of the meeting was dedicated to autoinflammatory syndromes. Natasa Toplac from the University Children's Hospital, Slovenia, gave a keynote lecture about different forms of periodic fever syndromes. Among public events, there was an organ music concert in the Cathedral of old Riga. The meeting was recognized by the local Medical Collegium and accredited with 8 CME credits. We would like to thank all the invited and local speakers for their excellent lectures, as well as our sponsors Biotest Germany and MEDA for their generous support and help with the organization of the meeting. The collaborations with the East-Central European Infectious and Pediatric Immunology center, Debrecen, Hungary and others give us the possibility to improve the level of the primary immunodeficiencies diagnostics in Latvia.

Tatjana Prokofjeva



Natalja Kurjane (Riga)



Nataša Toplac (Slovenia)



Natalia Kalinina (Russia)



Tatjana Prokofjeva (Riga)



**Michail Belevtsev (Belarus)
and Guseva Marina (Russia)**

J Project Meetings in Central and Eastern-Europe

2004-2011

1. Targu Mures, Romania, March 11-12, 2004; Organizer: Csilla Todea
2. Prague, Czech Republic, May 10-11, 2004; Organizer: Anna Sediva
3. Belgrade, Serbia/ Montenegro, June 11-12, 2004; Organizer: Srdjan Pasic
4. Skopje, Macedonia, September 17-18, 2004; Organizer: Katarina Stavrik
5. Kyiv, Ukraine, November 18-19, 2004; Organizer: Alla Volokha
6. Zakopane, Poland, December 16-17, 2004; Organizer: Ewa Bernatowska
7. Sofia, Bulgaria, April 15-16, 2005; Organizer: Elissaveta Naumova
8. Prague, Czech Republic, May 9-10, 2005; Organizer: Anna Sediva
9. Gdansk, Poland, September 23-24, 2005; Organizer: Ewa Bernatowska
10. Debrecen, Hungary, November 4-5, 2005; Organizer: László Maródi
11. Lviv, Ukraine, November 25, 2005; Organizer: Larysa Kostyuchenko
12. Oradea, Romania, December 20, 2005; Organizer: Zoltan Ellenés
13. Debrecen, Hungary, January 16, 2006; Organizer: László Maródi
14. Prague, Czech Republic, May 8-9, 2006; Organizer: Anna Sediva
15. Bucharest, Romania, June 9-10, 2006; Organizer: Nicolae Iagaru
16. Minsk, Belarus, November 9-10, 2006; Organizer: Michail Belevtsev
17. Debrecen, Hungary, December 8-9, 2006; Organizer: László Maródi
18. Zaporozhzhie, Ukraine, Apr 19-20, 2007; Organizer: Liudmyla Chernyshova
19. St. Petersburg, Russia, May 29-30, 2007; Organizer: Marina Guseva
20. Ohrid, Macedonia, September 20-21, 2007; Organizer: Kristina Mironska
21. Iasi, Romania, October 12-13, 2007; Organizer: Aurica Rugina
22. Ljubljana, Slovenia, November 16-17, 2007; Organizer: Tadej Avcin
23. Odessa, Ukraine, April 10, 2008; Organizer: Liudmyla Chernyshova
24. Sunny Beach, Bulgaria, May 22-23, 2008; Organizer: Elissaveta Naumova
25. Sarajevo, Bosnia-Herzegovina, October 10-11, 2008; Organizer: Velma Mulaosmanovic
26. Chisinau, Rep. of Moldova, Oct. 31-Nov. 1, 2008; Organizer: Lyudmila Cerempei
27. Riga, Latvia, November 27-28, 2008; Organizer: Tatjana Prokofjeva
28. Timisoara, Romania, March 19-20, 2009; Organizer: Margit Serban
29. Sevastopol, Ukraine, April 9-10, 2009; Organizer: Liudmyla Chernyshova
30. Ekaterinburg, Russia, May 13-14, 2009; Organizer: Irina Tuzankina

31. St. Petersburg, Russia, June 9-10, 2009; Organizer: Marina Guseva
32. Konya, Turkey, June 25-26, 2009; Organizer: Ismail Reisli
33. Krakow, Poland, September 24-25, 2009; Organizer: Danuta Kowalczyk
34. Tallinn, Estonia, October 7-8, 2009; Organizer: Sirje Velbri
35. Ljubljana, Slovenia, October 9-10, 2009; Organizer: Nataša Toplak
36. Tehran, Iran, October 11-12, 2009; Organizer: Asghar Aghamohammadi and Nima Rezaei
37. Cairo, Egypt, October 18-19, 2009; Organizer: Nermeen Galal
38. Minsk, Belarus, October 22-23, 2009; Organizer: Michail Belevtsev
39. Tirana, Albania, April 8-9, 2010; Organizer: Georgina Kuli-Lito
40. Porto Marina, Egypt, April 16-17, 2010; Organizer: Aisha El-Marsafy
41. Kharkov, Ukraine, May 19-20, 2010; Organizer: Liudmyla Chernyshova
42. Zlatibor, Serbia, June 1-2, 2010; Organizer: Srdjan Pasic
43. Zagreb, Croatia, July 2-3, 2010; Organizer: Darko Richter
44. Chelyabinsk, Russia, August 30-31, 2010; Organizer: Irina Tuzankina
45. Budapest, Hungary, September 9-10, 2010; Organizers László Maródi & Miklós Szolnoky
46. Tehran, Iran, October 11-12, 2010; Organizers: Asghar Aghamohammadi & Nima Rezaei
47. Pushkin Hills, Russia, February 3-4, 2011; Organizers: Marina Guseva; Areg Totolian
48. Zakopane, Poland, February 24-26, 2011; Organizer: Ewa Bernatowska
49. Oradea, Romania, March 1, 2011; Organizer: Ladislau Ritli
50. Debrecen, Hungary, March 10-11, 2011; Organizer: László Maródi
51. Moscow, Russia, March 31- April 1, 2011; Organizer: Irina Kondratenko
52. Baku, Azerbaydzan, April 22-23, 2011; Organizer: Gulnara Nasrullayeva
53. Bratislava, Slovakia, April 28-29, 2011; Organizer: Peter Ciznar
54. Lutsk, Ukraine, May 11-12, 2011; Organizer: Liudmyla Chernyshova
55. Sibiu, Romania, June 10-11, 2011; Organizer: Sorin Iurian
56. Chelyabinsk, Russia, June 26-27, 2011; Organizer: Irina Tuzankina
- J Project session Beijing, China, September 23, 2011; Organizer: Tina Lee*
57. Ohrid, Rep. Macedonia, October 8, 2011; Organizer: Kristina Mironska
58. Tehran, Iran, October 15, 2011; Organizer: Nima Rezaei
59. Trabzon, Turkey, November 19-20; Organizer: Fazil Orhan & Ismail Reisli
60. Riga, Latvia, November 26, Organizer: Tatjana Prokofjeva

Scheduled J Project Meetings in 2012 N⁰ 61-

<u>City (country)</u>	<u>Organizer</u>	<u>Date</u>
61. Lillafüred (Hungary)	Miklós Szolnoky biotest@biotest.hu	January 20-21
62. Budapest (Hungary)	Miklós Szolnoky Judit Szabó biotest@biotest.hu	January 27-28
63. Rostov-on-Don (Russia)	Liudmila Sizyakina sizyakina_L@rostgmu.ru	March 15-16
64. Moscow (Russia)	Irina Kondratenko ikondratenko@rambler.ru	March 29
65. Bratislava (Slovakia)	Peter Ciznar ciznar@dfnsp.sk	April 28-29
66. Prague (Czech Republic)	Anna Sediva Anna.Sediva@fnmotol.cz	May 14-15
67. Sibiu (Romania)	Sorin Iurian iurian_sorsab@hotmail.com	June 01-02
68. Timisoara (Romania)	Mihaela Bataneant mihabataneant@yahoo.com	June
69. Tyumen (Russia-Siberia)	Irina Tuzankina ituzan@yandex.ru	July 01-02
70. Budva (Montenegro)	Srdjan Pasic pasics@ikomline.net	September
71. Chernigov (Ukraine)	Liudmyla Chernyshova chernyshova@ukr.net	September
72. Sofia (Bulgaria)	Elissaveta Naumova naumovaej@gmail.com	TBA
73. Konya (Turkey)	Ismail Reisli ireisli@hotmail.com	TBA
74. Tehran (Iran)	Nima Rezaei rezaei_nima@yahoo.com	October
75. (Belarus)	Michail Belevtsev belevtsev_m@mail.ru	November
76. (Poland)	Anna Szaflarska miszafla6@wp.pl	November
77. Ufa (Russia)	Irina Tuzankina ituzan@yandex.ru	November 23

THE J PROJECT

Steering Committee Members

1) Albania	Tirana	Georgina KULI-LITO
2) Azerbaijan	Baku	Gulnara Nasrullayeva
3) Belarus	Minsk	Michail BELEVTSSEV
4) Bosnia-Herzegovina	Sarajevo	Velma MULAOSMANOVICH
5) Bulgaria	Sofia	Elissaveta NAUMOVA
6) Croatia	Zagreb	Darko RICHTER
7) Czech Republic	Prague	Anna SEDIVA
8) Czech Republic	Brno	Jiri LITZMAN
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13) Hungary	Debrecen	Beáta TÓTH
14) Hungary	Budapest	Miklós SZOLNOKY
15) Iran	Tehran	Nima REZAEI
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18) Poland	Warsaw	Malgorzata PAC
19) Poland	Krakow	Anna SZAFLARSKA
20) Rep. of Macedonia	Skopje	Kristina MIRONSKA
21) Rep. of Moldova	Chisinau	Liudmyla CEREMPEI
22) Romania	Bucharest	Ioan GHERGHINA
23) Romania	Timisoara	Mihaela BATANEANT
24) Romania	Timisoara	Margit SERBAN
25) Romania	Sibiu	Sorin IURIAN
26) Russia	Moscow	Irina KONDRATENKO
27) Russia	St. Petersburg	Marina GUSEVA
28) Russia	Ekaterinburg	Irina TUZANKINA
29) Russia	Moscow	Anna SHCHERBINA
30) Serbia	Belgrad	Srdjan PASIC
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34) Ukraine	Kiev	Liudmyla CHERNYSHOVA
35) Ukraine	Lviv	Larysa KOSTYUCHENKO
36) Ukraine	Kiev	Alla Volokha

Debrecen Jeffrey Modell PID Reference Center

Genetic testing of PID genes in 2004-2011

Genes	HUN	UKR	ROU	POL	RUS	LAT	SRB	BUL	MAC	SVK	SLO	CRO	TUR	EGY	SUM
1. AICDA	16	2	4		1					1					24
2. AIRE	46	4	1												51
3. BTK	42	50	24	13	9		1	1	1		2	5			148
4. CARD9	2	2	1												5
5. C2G	8														8
6. CLEC4L	3														3
7. CLEC6A	4	1													5
8. CLEC7A	8	2													10
9. CYBB	3		6	6	2	4						1			22
10. CSF3R	10														10
11. CD40LG	30	4	3			5						1		1	44
12. CXCR4	27		4						1						32
13. DKC1	3														3
14. DOCK8	7														7
15. ELA2	22	1							1	2		6			32
16. FASG	1														1
17. FASLG	1														1
18. FLG	10														10
19. GFI1	13														13
20. HAX1	10														10
21. ICOS	9														9
22. IL2RG	4	6										1			11
23. IL18R1	1														1
24. LIG4	1														1
25. MVK	3		1												4
26. MRC1	1														1
27. NBS1	9		3			5									17
28. NCF1			1												1
29. NEMO			3												3
30. PRF1	3									1					4
31. RAG1	2				1	2						1			6
32. RAG2	2				1	2									5
33. RMRP	3														3
34. SBDS	11	5	1				1								18
35. SH2D1A	57	1	3							2					63
36. STAT1	14	7			2										23
37. STAT3	23	7		3	5										38
38. TACI	9														9
39. UNG	1														1
40. WAS	52	14	4	3	13	2		1	2				5		96
41. XIAP	1														1

Genetic testing in the Debrecen Jeffrey Modell PID Reference Center

PRIMER IMMUNODEFICIENCIES (*Genes*)

1. X-linked agammaglobulinemia (*BTK*)
2. X-linked hyper-IgM syndrome (*TNSF5=CD40L*)
3. Activation induced citidine deaminase deficiency (*AICDA*)
4. Common variable immunodeficiency (*TACI*)
5. Common variable immunodeficiency (*ICOS*)
6. Hyper-IgD syndrome (*MVK*)
7. Hyper-IgE syndrome (*STAT3*)
8. X-linked severe combined immunodeficiency (*IL-2RG*)
9. Recombinase activator gene 1 deficiency (*RAG1*)
10. Recombinase activator gene 2 deficiency (*RAG2*)
11. Familiar hemophagocytic lymphohistiocytic syndrome (*PRF1*)
12. X-linked lymphoproliferative syndrome (*SH2D1A*)
13. Autoimmune lymphoproliferative syndrome (*FASG*)
14. Autoimmune lymphoproliferative syndrome (*FASLG*)
15. Autoimmune polyendocrinopathy I syndrome (*AIRE*)
16. Dyskeratosis congenita (*DKC1*)
17. Cartilage-hair hypoplasia (*RMRP*)
18. Nijmegen breakage syndrome (*NBS1*)
19. Shwachman-Diamond syndrome (*SBDS*)
20. Warts-Hypogammaglobulinemia-Infection-Myelokathexis syndrome (*CXCR4*)
21. Wiskott-Aldrich syndrome (*WAS*)
22. Kostmann syndrome (*HAX1*)
23. Cyclic neutropenia (*ELA2*)

24. Severe congenital neutropenia (*GFI1*)
25. Severe congenital neutropenia (*CSF3R*)
26. Severe congenital neutropenia (*G6PC3*)
27. X-linked chronic granulomatous disease (*CYBB*)
28. Autosomal recessive chronic granulomatous disease (*NCF1=p47phox*)
29. C2 deficiency (*C2*)
30. IL18R deficiency (*IL18RG*)
31. Chronic mucocutaneous candidiasis (*CLEC4L*)
32. Chronic mucocutaneous candidiasis (*CLEC6A*)
33. Chronic mucocutaneous candidiasis (*CLEC7A*)
34. Chronic mucocutaneous candidiasis (*MRC1*)
35. Mucocutaneous candidiasis (*CARD9*)
36. Mucocutaneous candidiasis (*STAT1*)
37. Hyper-IgM syndrome (*UNG*)
38. 2-type X-linked lymphoproliferative syndrome (*XIAP*)
39. DOCK8 deficiency (*DOCK8*)
40. DNA ligase 4 (*LIG4*)

LYSOSOMAL STORAGE DISORDERS (*Genes*)

41. Fabry disease (*GLA*)
42. Gaucher disease (*GBA*)
43. Niemann-Pick disease (*SMPD1*)

OTHERS (*Genes*)

44. Von Hippel-Lindau syndrome (*VHL*)
45. X-linked congenital adrenal hypoplasia (*DAX1*)
46. Galectin 3 deficiency (*LGALS3*)
47. Alström syndrome (*ALMS1*)
48. Atopic eczema (*FLG*)

HUNGID - Hungarian Nursing Group for Primary Immunodeficiencies



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Founding Members of HUNGID

**L-R: Ács M, Pongor T, Tóth É,
Garuder M, Veres M, Baranya-Kiss R,
Révész M, Nagy M**

HUNGID was established on March 10, 2011 in Debrecen; the major aims are as follows:

- 1. Dissemination of general and nursing information and knowledge about PIDs in Hungary*
- 2. Representation of Hungary in the International Nursing Group for PIDs*
- 3. Collaboration with and help the work of the Hungarian Organization for Patients with Immune Deficiencies (HOPI), one of the national member organizations of IPOPI*
- 4. To help the activity of the Hungarian Working Group for Primary Immunodeficiencies, the professional organization of doctors and scientists in the country.*

The constitution of HUNGID was approved by the Hungarian authorities and it is now an independent organization. Elected leaders of the Group are Mónika Révész (president) and Róza Kiss –Baranya (vice president). One of the first responsibilities of HUNGID is to promote a wider use of subcutaneous immunoglobulin (SCIG) preparations among PID patients with antibody deficiencies in Hungary.



SCIG therapy: efficient, safe, well tolerated, and improves quality of life

Patients' Groups Pages



Hungarian Organization for Patients with **Immunodeficiencies**

Nagyerdei krt. 98., 4032 Debrecen, Hungary

Phone: **+36 52 255-613**;

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Sport Kids Day

25th of June, 2011



***The Sport Kids Day and the Christmas Day were supported by WIN Grants
from the Jeffrey Model Foundation.***



Christmas Day

10th of December, 2011



Publications

related to the J Project program

- 1) Maródi L. The discovery and taking wing of the J Project in Eastern and Central Europe. *ANN NY ACAD SCI* **1238**:65-73 (2011)
- 2) Tuzankina I, Kobeleva Y, Kiseleva N, Bolkov M, Reuter G, Maródi L. Cytotoxic T lymphocytes mediate neuronal injury in patients with X-linked agammaglobulinemia and progressive neurodegenerative disease. *ALLERGY* **66**:1617-1618 (2011)
- 3) Liu L, Okada S, Kong XF, Kreins AY, Cypowyj S, Abhyankar A, Toubiana J, Itan Y, Audry M, Nitschke P, Masson C, Toth B, Flatot J, Migaud M, Chrabieh M, Kochetkov T, Bolze A, Borghesi A, Toulon A, Hiller J, Eyerich S, Eyerich K, Gulácsy V, Chernyshova L, Chernyshov V, Bondarenko A, María Cortés Grimaldo R, Blancas-Galicia L, Madrigal Beas IM, Roesler J, Magdorf K, Engelhard D, Thumerelle C, Burgel PR, Hoernes M, Drexel B, Seger R, Kusuma T, Jansson AF, Sawalle-Belohradsky J, Belohradsky B, Jouanguy E, Bustamante J, Bué M, Karin N, Wildbaum G, Bodemer C, Lortholary O, Fischer A, Blanche S, Al-Muhsen S, Reichenbach J, Kobayashi M, Rosales FE, Lozano CT, Kilic SS, Oleastro M, Etzioni A, Traidl-Hoffmann C, Renner D, Abel L, Picard C, Maródi L, Boisson-Dupuis S, Puel A, Casanova JL. Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. *J EXP MED* **208**: 1635-1648 (2011)
- 4) Gulácsy V, Freiburger T, Shcherbina A, Pac M, Chernyshova L, Avcin T, Kondratenko I, Kostyuchenko L, Prokofjeva T, Pasic S, Bernatowska E, Kutukculer N, Rascon J, Iagaru N, Mazza C, Tóth B, Erdős M, van der Burg M, Maródi L. Genetic characteristics of eighty-seven patients with the Wiskott-Aldrich syndrome. *MOL IMMUNOL* **48**: 788-792 (2011)
- 5) Tóth B, Wolff A, Halász Z, Tar A, Szüts P, Ilyés I, Erdős M, Szegedi G, Husebye ES, Zeher M, Maródi L. Novel sequence variation of AIRE and detection of interferon- ω antibodies in early infancy. *CLIN ENDOCRINOL* **72**: 641-647 (2010)
- 6) Boztug K, Schmidt M, Schwarzer A, Banerjee PP, Díez IA, Dewey R, Böhm M, Naundorf S, Köhlcke K, Blasczyk R, Maródi L, Kondratenko I, Orange J, von Kalle C, Klein C. Correction of Wiskott-Aldrich syndrome by hematopoietic stem cell gene therapy. *N ENGL J MED* **363**: 1918-1927 (2010)
- 7) Tóth B, Volokha A, Mihás A, Pac M, Bernatowska E, Kondratenko I, Polyakov A, Erdős M, Pasic S, Bataneant M, Szaflarska A, Mironska K, Richter D, Stavrik K, Avcin T, Márton G, Nagy K, Dérfalvi B, Szolnoky M, Kalmár Á, Belevtsev M, Guseva M, Kriván G, Timár T, Nyúl Z, Mosdósi B, Kareva L, Peova S, Chernyshova L, Gherghina J, Serban M, Conley ME, Notarangelo LD, Smith CIE, van Dongen J, van der Burg M, Maródi L. Genetic and demographic features of X-linked agammaglobulinemia in Eastern and Central Europe: a cohort study *MOL IMMUNOL* **46**: 2140-2146 (2009)
- 8) Maródi L, Casanova JL. Primary immunodeficiency diseases: The J Project. *THE LANCET* **373**:2179-2181 (2009)
- 9) Jiao H, Tóth B, Erdős M, Fransson I, Rákóczi É, Balogh I, Magyarics Z, Dérfalvi B, Csorba G, Szaflarska A, Megarbane A, Akatcharian C, Dbaiho G, Rajnavölgyi É, Hammarström L, Kere J, Lefranc G, Maródi L. Novel and recurrent STAT3 mutations in hyper-IgE syndrome patients from different ethnic groups. *MOL IMMUNOL* **46**: 202-206 (2008)
- 10) Botztug K, Germeshausen M, Avedillo Díez I, Gulácsy V, Diestelhorst J, Ballmaier M, Welte K, Maródi L, Chernyshova L, Klein C. Multiple independent second-site mutations in two siblings with somatic mosaicism for Wiskott-Aldrich syndrome. *CLIN GENET* **74**: 68-74 (2008)
- 11) Maródi L, Notarangelo LD. Immunological and genetic bases of new primary immunodeficiencies. *NAT REV IMMUNOL* **7**: 851-861 (2007)
- 12) Maródi L, Notarangelo LD. Education and worldwide collaboration pays off. *NAT IMMUNOL* **8**:323-324 (2007)