The J Project

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Co-editors: Michael Belevtsev, Melinda Erdős, Nicolae Iagaru, Ales Janda, and Anna Sediva,

Sponsor: ECE IPI CTR
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Foreword

The mission of The J Project is to be the forum in East-European countries to increase awareness on primary immunodeficiency disorders (PIDs). Since the first J Project meeting in Targu Mures, Romania, on March 11-12, 2004, more and more of us in the field have been actively working to achieve this goal. It is amazing that in the beginning of 2007 we have not only scheduled six J Project meetings for the year, but have already defined the venue of one of the 2008 events (Bulgaria). Detailed information on programs, achievements, and the atmosphere of these meetings are described in the annual J Project booklets as well as on the web (www.ece.dote.hu).

The year of 2006 was highlighted by several events to remember by the primary immunodeficiency community. Just to mention a few, an EU PID consensus conference was organized in the Paul-Erlich-Institute, Langen, Germany, in June. This Meeting was a milestone in the long-term awareness campaigns among the general public. Budapest saw the biannual ESID/IPOI/INGID meeting in October, the summary of which was published in the ESID newsletter and this booklet. Further, the first gene therapy trial in patients with Wiskott-Aldrich syndrome was started at the Department of Pediatric Hematology/Oncology, Hannover Medical School increasing the number of PIDs in which gene therapy has been initiated.

The J Project events in 2006 covered a wide range of diagnostic and therapeutical aspects of PID patient care. We started in Debrecen with a discussion on current status, and future challenges of gene therapy in Wiskott-Aldrich syndrome. The traditional Prague Spring Meeting was focused this year on new primary immunodeficiency disorders. The first J Project meetings in Bucharest and Minsk were organized in June and November, respectively, and attracted a large number of immunologists, infectious disease specialists, hematologists, transplant physicians, and others with an interest in PID from both South-Romania and Belarus. These meetings were physicians’ education events that provided opportunity to discuss recent diagnostic and treatment achievements in these large areas of East-Europe. The perspective of PID gene sequencing in Minsk, Belarus was discussed and collaboration to this end with the Debrecen Jeffrey Modell PID Reference Center was started. The meeting series in 2006 ended up in Debrecen with a summary of PID mutation database in East-Europe. I am grateful to all who have joined this Project, and take the opportunity to thank our major sponsors, the Jeffrey Modell Foundation and Biotest Hungary Kft. for their generous support and encouragement.


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

- Molecular pathology of primary immunodeficiency diseases
- Host defence mechanisms against bacteria and fungi
- Mechanisms of action of intravenous immunoglobulins
- Developmental biology of macrophages

The Center 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 Center. Its 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 Euroregion, 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 2006

CITY, Country                      DATE
1. DEBRECEN, Hungary                16 January, 2006
2. PRAGUE, Czech Republic           8-9 May, 2006
3. BUCHAREST, Romania               9-10 June, 2006
4. MINSK, Belarus                    9-10 November, 2006
5. DEBRECEN, Hungary                8-9 December, 2006

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.
Debrecen, Hungary
16 January, 2006

Hosts:
László Maródi and Melinda Erdős

Venue:
Department of Infectious and Pediatric Immunology,
University of Debrecen Medical and Health Science Center

Main Topic:
Perspectives of gene therapy in the Wiskott-Aldrich syndrome

Zoltán Gyeszli, Melinda Erdős, Lyudmila Chernishova,
László Maródi, Christoph Klein, Kaan Boztug
Genetically analyzed patients with the Wiskott-Aldrich syndrome in Hungary
L. Maródi, V. Gulácsy

c.58C>T nonsense mutation in two siblings with the Wiskott-Aldrich syndrome
L. Chernishova, V. Gulácsy, L. Maródi

Gene therapy for Wiskott-Aldrich syndrome: rescue of T-cell signaling and amelioration of colitis upon transplantation of retrovirally transduced hematopoietic stem cells
C. Klein

Development of hematopoietic stem cell gene therapy for Wiskott-Aldrich syndrome
K. Boztug, C. Klein

Discussion

Break

An approved protocol for human gene therapy trial in the Wiskott-Aldrich syndrome
C. Klein

Discussion with parents

Joint visit to the Clinical Center for Cell Therapy, University of Debrecen, Medical and Health Science Center
Host: J. Hunyadi
SUMMARY AND CONCLUSIONS

This unique J Project Meeting was aimed at discussing the perspectives of gene therapy in patients with severe Wiskott-Aldrich syndrome, who have no appropriate family or foreign donor for bone marrow transplantation. Participants in the Meeting included professionals from Germany, Hungary and Ukraine, and the parents of two Hungarian patients with severe Wiskott-Aldrich syndrome. In both families hemizigosity in the WAS gene was diagnosed in the affected boys at the Debrecen JMF PID Reference Center. None of the patients had HLA-matched donor for bone marrow transplantation at the time of the discussion.

We invited Prof. C. Klein and Dr. K. Boztug, and requested them to participate in a face-to-face discussion with the parents, and to answer all kind of question that was raised in particular, potential risks of GT. First, professional presentations on clinical, immunological, and genetic data of the two patients were given by Hungarian doctors. Then, Prof. Klein and Dr. Boztug presented in vitro and animal data concerning gene therapy in WAS. Prof. Klein also described the gene therapy protocol currently approved by German authorities. Finally, the parents were questioning the doctors and asked intriguing, sometimes astonishingly smart questions that were focused mostly on possible risks of this therapy. Of course, no decision was requested or made by the parents at the Meeting. It was made clear that gene therapy today is experimental and may be a therapeutical option only if BMT is not available, the disease is severe and progressing, and the family decided for this medication.

László Maródi

Department of Infectious and Pediatric Immunology,
University of Debrecen Medical and Health Science Center
SUMMARY AND CONCLUSIONS

On May 8 and 9, 2006, the fifth ESID Prague Spring Meeting was held at the University Hospital Motol, Prague, Czech Republic. This year 27 participants from 10 countries, namely Czech Republic, Estonia, Germany, Hungary, Lithuania, Poland, Slovakia, Romania, Russia and the Great Britain, attended and actively participated in the event. The main task of the Prague ESID meeting is traditionally devoted to the exchange of information on PIDs between Western and Eastern Europe. This task was accomplished this year through an excellent attendance from both parts of the European continent. The invited speakers were Christine Kinnon from Molecular Immunology Unit, UCL Institute of Child Health in London and
Benjamin Gathmann from Department of Rheumatology and Clinical Immunology in Freiburg. Helen Chapel from Oxford University has provided great support in organizing the Meeting and especially in its educational dimension.

The introductory lecture of the Meeting served as an overview of the current state of the gene therapy. Christine Kinnon covered the history of this pioneering treatment option for severe forms of PIDs and outlined its potential prospects as well. One section of the program of the first day focused on the issues with pan-European impact, as for example the new ESID on-line registry of PIDs and the disparities in the diagnosis, care and treatment of PIDs among European countries. All the participants, representing 8 EU member states, and Romania and Russia, took active part in the discussions. The outcome from the fruitful discussions would serve as a base for the negotiations during the forthcoming EU consensus meeting in June 2006 in Langen, Germany.

The program of the second day was concentrated on humoral deficiencies, the crucial topic of this year’s Meeting. The latest advancements and discoveries in the research of molecular background of antibody deficiencies were discussed as well as laboratory tools used for the classification of these entities. Case reports as for example the recently described Cernunnos deficiency formed substantial part of the program. Newly discovered immunodeficiencies and unusual and interesting cases were highly appreciated.

The social program was quite an important part of the Prague Spring ESID Meeting as it enabled further fostering of close cooperation between the Eastern and Western European countries. The relaxing stroll through the Prague Castle, dinner in a restaurant situated in the historical quarter of Prague and a concert in a jazz club were all enjoyed by the participants.

The Meeting was supported by the Charles University, 2nd Medical School, Prague, and by University Hospital in Motol, Prague, and substantial contribution from pharmaceutical companies Baxter, Grifols, Exbio and Immunotech.

Meeting was organized as a part of the activities related to the Day of Immunology, declared by EFIS on 29th April, 2006.

This year Meeting has confirmed the still growing scientific quality of the conference and the usefulness of this educational activity.

The next ESID Prague Spring Meeting will be directed mainly on ESID juniors and will be held on May 14 and 15, 2007.

Anna Sediva and Ales Janda

University Hospital Motol, Prague
Bucharest, Romania
9-10 June, 2006

Hosts: Nicolae Iagaru and Ioan Gherghina

Venue: Amfiteatrul IOMC

Main topic:
Diagnostic facilities and therapeutical practices in the management of PID patients in South-Romania

Margit Serban (middle) and her colleagues
PROGRAM

Primary immunodeficiencies update
I. Gherghina, A. Cochino

The J Project - an East-European perspective
L. Maródi

Primary immunodeficiency epidemiology in Romania
M. Serban

Oncologic risks in children with primary immunodeficiencies
M. Bataneanț, M. Cucuruz, C. Petrescu, E. Boeriu, S. Arghirescu, L. Pop, M. Serban

Genetic basis of immunoglobulin synthesis
C. Bara

Primary immunodeficiencies: from theory to clinical practice
N. Iagaru, M. Iusan, A. Cochino, I. Stan

IGIV in primary immunodeficiencies: current practice and perspectives
N. Iagaru, C. Dragomir, M. Iusan, E. Ionita, M. Ritivoiu

Rebuilding of the immune system after bone marrow transplantation
C. Arion, A. Colita, L. Dumitrache, A. Dumitrescu, G. Nitu

Chronic neutropenia associated with scleroderma
Z. Ellenes

Recurrent fever as a diagnostic challenge
M. Stefan

Chronic granulomatous disease
I. Stan, M. Ritivoiu

Louis-Bar syndrome
A. Cochino, M. Iusan, N. Iagaru

Cyclic neutropenias
A. Cochino

CATCH22 syndrome
V. Bica, M. Iordachescu

Selective IgA deficiency
I. Stan, N. Iagaru

General discussion
SUMMARY AND CONCLUSIONS:

The Institute for Mother and Child Care organized the first J Project Meeting in Bucharest, on 9th-10th June 2006, with the help of the Debrecen Jeffrey Modell PID Reference Center. The goal of this Meeting was to improve recognition and treatment of patients with PID in South Romania. In order to meet this task, our Institute invited the regional representatives of all the counties of South Romania and Bucharest, and also the heads of the Pediatric Departments in this area (Craiova, Constanta, Galati, Bucharest).

The program included 16 PID-related presentations by remarkable doctors and scientists from Romania and Hungary (see attached program). The main areas of discussion were as follows:

- Current practices and management of IVIG substitutions in Romania
- Diagnosis and treatment of patients with IgG deficiencies
- Sequencing PID genes, and collaboration at this point with the Debrecen JMF PID Reference Center
- Availability of flow cytometry for analysis of patients with cellular ID

It was concluded that the Institute for Mother and Child Care should remain and further develop its activity as the center of physician education and PID screening in Southern Romania and should further develop collaboration with other centers in Romania, as well as with the Debrecen JMF PID Reference Center.

Nicolae Iagaru
Minsk, Belarus
9-10 November, 2006

Hosts: Michael Belevtsev and Olga V. Aleinikova

Venue: Belarusian Research Center for Pediatric Oncology & Hematology

Main Topics:
National Registry for PID in Belarus
Molecular genetic diagnostics of PID
Informatics technologies in management of PID

Olga V. Aleinikova (middle), her colleagues, and Hungarian guests
9th November, 2006

Welcome addresses
O. V. Aleinikova and L. Maródi

Care of children with PID at the Research Center for Pediatric Oncology/Hematology
O. V. Aleinikova

Clinical detection of patients with primary immunodeficiency disease
I. E. Gurmanchuk, L. P. Titov

Shwachman-Diamond syndrome
M. Erdős

Classification and principles of diagnostics of immunodeficiency diseases
D. K. Novikov

Organization of diagnostics and treatment of PID in the Ukraine
L. I. Chernishova

PID registries and databases
E. V. Galkina

Primary immunodeficiency patient care in Latvia
T. E. Prokofyeva

Clinical aspects, diagnostics and treatment of children with CVID
V. I. Novikova

PIDs in adults
L. R. Vichristenko
10th November, 2006

Clinical aspects and principles of treatment of PIDs
A. S. Yurasova

Antibody deficiencies
A. P. Voloha, L. I. Chernishova

IgA deficiency
I. I. Savanovich

Wiskott-Aldrich syndrome: Approach to diagnostics and treatment
D. V. Samarin, L. I. Chernishova, L. Maródi, C. Klein, K. Boztug

Complement defects: clinical manifestation and diagnostics
L. P. Titov

Molecular diagnostics of PID
A. A. Mihas, A. M. Kustanovich

Application of IVIG for children with PID
T. A. Uglova, S. N. Aleskevich

National registry of PID in the Republic of Belarus
N. N. Savva, O. I. Bydanov

Informational and analytical program application in the complex management of patients with PID
O. V. Krasko, N. Novoselova, I. E. Tom

Genetic engineering technologies
N. Meleshko

Lyudmila I. Chernishova (left) visiting the Lab of Michael Belevtsev (right)

Alla Volokha and Michael Belevtsev
SUMMARY AND CONCLUSIONS

On November 9-10, 2006 the 1st International Conference on Primary Immunodeficiencies was carried out in Minsk, Belarus.

The Conference was organized by the Ministry of Health of the Republic of Belarus, the Belarusian Research Center for Pediatric Oncology and Hematology, and the East-Central European Infectious and Pediatric Immunology Center, Debrecen, Hungary.

The following topics were scheduled in the program:

- Epidemiology and registration of PIDs.
- Clinical aspects of PIDs.
- Immunological and molecular genetic diagnostics of PIDs.
- Modern approach to treatment of patients with PIDs.
- Informatics technologies in the diagnostics of PIDs.

More then 150 specialists (pediatricians, immunologists, hematologists, researchers) from Belarus, Hungary, Latvia, Ukraine, and Russia had participated at this conference. Opening addresses were made by the deputy director of the Ministry of health; Prof. Olga V. Aleinikova, the head of BRCPOH, and Prof. László Maródi, the head of the J Project. The Meeting was aimed at discussing the followings:

- Medical care of children with PID in Belarus
- Diagnostic possibilities of PIDs
- Establishment of the educational center for PID in Belarus

First, Prof. Aleinikova gave an intriguing summary on medical care of children with PID in Belarus, and then Prof. Gurmanchuk and D. K. Novikov talked about clinical detection and diagnostics of PIDs, respectively.

Reports of our colleagues from Ukraine were also of considerable interest. Prof. Chernishova presented the organization of diagnostics and treatment of patients with PID in Ukraine, and A. Volokha made an interesting talk about antibody deficiencies. Experience exchange with our colleague from Latvia (T. E. Prokofyeva) was also of great interest. The first patient in Hungary diagnosed with Shwachman-Diamond syndrome was presented by M. Erdös. Our colleagues from Russia E. V. Galkina, and A. S. Yurasova talked about registries, databases, and clinical aspects and treatment of PIDs.

Reports on patients with CVID (V. I. Novikova), IgA deficiency (I. I. Savanovich), Wiskott-Aldrich syndrome (D. V. Samarin), and complement defects (L. P. Titov) were also presented. The molecular diagnostics of PIDs and the application of flow cytometry were summarized by A. A. Mihas and M. Belevtsev, respectively. The engineering technologies were discussed by A. N. Meleshko. The final part was devoted to therapy (T. A. Iglova), and the national registries (N. N. Savva). O. V. Krasko also talked about the informational and analytical program application in the complex management of patients with PID.

Michael Belevtsev
Debrecen, Hungary
7-8 December, 2006

Host: László Maródi

Venue: Debrecen Centre of the Hungarian Academy of Sciences

Main Topics:
Pediatric infectious diseases and immunology training in Europe.
Immunodeficiency databases in East-Europe

University of Debrecen
PROGRAM

7th December, 2006

EDUCATION AND TRAINING

Training in pediatric infectious diseases and immunology in Europe
A. Cant

Pediatric antibiotic prescribing in Europe
M. Sharland

The J PROJECT: From physician education to genetic databases
L. Maródi

8th December, 2006

MOLECULAR DIAGNOSTICS AND DATABASES

Recent advances in genetic analysis of PID patients in Romania
M. Bataneant
László Maródi and Mike Sharland

Application of fluorescence in situ hybridization technique in patients with CATCH 22
V. Bica

Clinical and molecular diagnostics in patients with SCID
A. Cochino

Identification of Btk mutations in Hungarian families with agammaglobulinemia
B. Tóth

Recurrent CXCR4 sequence variation in WHIM syndrome
M. Erdős

Mutational spectrum of Btk in Ukrainian patients with XLA
A. Volokha

WAS mutation profile in patients from East-European countries
V. Gulácsy

Molecular approach to selective immunoglobulin A deficiency
I. Stan

Molecular pathology of XLP
Á. Lányi

Clinical and genetic diagnostics in Ukrainian patients with Nijmegen breakage syndrome
I. Savchak

Combined immunodeficiency with defective MHC classII expression
D. Baltadjieva

PID case reports
G. Petrova
We can make the difference together.
SUMMARY AND CONCLUSIONS

At this J Project Meeting we took advantage of the visitation by Dr. Mike Sharland (London), Prof. Andrew Cant (New Castle), members of the Educational and Training Committee (ETC) of the Confederation of European Specialists in Pediatrics (CESP), to the Department of Infectious and Pediatric Immunology of the University of Debrecen. Dr. Sharland (Chair of the ETC) gave an intriguing and comprehensive talk on antibiotic prescribing practice in some countries of the European Union and emphasized the risk of overusing antibiotics in pediatric practice. Prof. Cant was talking about the CESP-approved pediatric infectious diseases tertiary program and the pediatric immunology add-on program. Both presentations precipitated hot discussions with doctors from East-Europe.

An overview on mutational analysis during the past years at the Debrecen JMF PID Reference Center of more than seventy patients from East-European countries was presented. The papers from Romania (presented by doctors M. Bataneant, V. Bica, I. Stan, and A. Cochino) gave insights into the survey of management practices in the major Romanian PID centers, Timisoara and Bucharest. A. Volokha from Kiev summarized the mutational spectrum in more than 20 Ukrainian patients with X-linked agammaglobulinemia and emphasized that these data represent one of the outcomes of collaboration between East-European PID Centers and the Debrecen JMF PID Reference Center. B. Tóth presented data on Hungarian patients and family members with BTK mutation. The first patient in Hungary diagnosed with the WHIM syndrome was presented by M. Erdős. V. Gulácsy, one of the medical students working in the lab of the Department of Infectious ad Pediatric Immunology in Debrecen, gave a summary on mutations in the WAS gene of patients from Hungary and five East-European countries. One of the highlights of the Meeting was a great talk by Á. Lányi on molecular pathology of XLP. I. Savchak from the group of Prof. Kostuchenko presented genetic data of Nijmegen-breakage syndrome patients diagnosed and cared in Lviv. It was a pleasure to have a nice group of immunologists from Sofia, who contributed to the success of the Meeting by intriguing case presentations.

László Maródi
The ESID-IPOPI-INGID Meeting on October 4-7, 2006 was organized at the Budapest Novotel Congress Center. We made every effort with colleagues at my Department and at Convention Budapest Kft. to ensure that the Meeting would be of the highest quality from both scientific and organization points of view. The ESID Board and the Scientific Committee provided great help throughout the organization process. The regular sponsors (Baxter, Octapharma, ZLB, Biotest, just to mention a few) continued to be as generous as they were at our previous meetings and helped to reach a balanced budget and to be able to invite the most outstanding speakers in the field.

When I first proposed in Weimar 4 years ago that Budapest might be a nice venue for a future ESID meeting, I was considering a meeting with about 500 participants. However, with the background described above, we were able to attract a record number of 982 participants from 54 countries. We had 94 East-European participants and this may have been related to The J Project activity in the region over the past years.

There were several reasons that might explain the high level of interest in this Meeting. First, we had a great program and I want to appreciate the help of the ESID Board in the formulation of the scientific program. The program was actually quite busy and the table below shows statistics of different presentations.

### Presentations at the 2006 ESID Meeting

<table>
<thead>
<tr>
<th>Type</th>
<th>Number</th>
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<tbody>
<tr>
<td>Keynote lectures</td>
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<tr>
<td>Introductory lectures</td>
<td>14</td>
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<td>Educational Day presentations</td>
<td>8</td>
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<td>Plenary oral presentations</td>
<td>29</td>
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<tr>
<td>Workshop presentations</td>
<td>12</td>
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<tr>
<td>Posters</td>
<td>358</td>
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<tr>
<td>Lunch Symposium presentations</td>
<td>10</td>
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</table>

**Sum:** 433

It was clear in Budapest that the PID community in Europe is not only dynamic but it is getting more and more heterogeneous. ESID, IPOPI, INGID, the Jeffrey Modell Foundation, Prague Spring Meetings, Winter Meetings in Poland, The J Project, and recently the EU-PID initiative are active components of the
professional, scientific, and social work for the benefit of immunodeficient children. Although it is not just ESID any more, I think it is important that ESID remains the driving force and the core organization of all these activities. Our understanding of PIDs today is different from what we thought about it 8-10 years ago. The scientific level of the Meeting was indicated by the presentation of 9 new PIDs and disease-causing genes (table below).

**Novel immunodeficiency diseases**

<table>
<thead>
<tr>
<th>Immunodeficiency disease</th>
<th>Disease-causing gene</th>
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<tbody>
<tr>
<td>Autosomal recessive UNC-93B deficiency</td>
<td>UNC-93B1</td>
</tr>
<tr>
<td>Autosomal recessive Tyk2 deficiency</td>
<td>TYK2</td>
</tr>
<tr>
<td>X-linked recessive MSMD</td>
<td>IKBKG</td>
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<tr>
<td>HAX-1 deficiency</td>
<td>HAX1</td>
</tr>
<tr>
<td>p14 deficiency</td>
<td>MAPBPIP</td>
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<tr>
<td>Ig-β deficiency</td>
<td>CD79B</td>
</tr>
<tr>
<td>X-linked inhibitor of apoptosis deficiency</td>
<td>BIRC4</td>
</tr>
<tr>
<td>Hepatic veno-occlusive disease with immunodeficiency Sp110</td>
<td></td>
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<tr>
<td>Autosomal recessive IPEX syndrome</td>
<td>IL2RA</td>
</tr>
</tbody>
</table>

MSMD, mendelian susceptibility to mycobacterial disease; IPEX, immune dysregulation, polyendocrinopathy, enteropathy, X-linked inheritance.

ESID INGID IPOPI Meeting

Aquincum Quartet

József Balog

St. Stephen Cathedral
Budapest Hungary 2006

Karoly Bagdi

Julia Kukely

Horse show and banquet dinner in Domonyvölgy

25
### J Project Meetings in 2007 (No. 18 to 23)

<table>
<thead>
<tr>
<th>Country (city)</th>
<th>Organizer</th>
<th>Date</th>
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<tbody>
<tr>
<td>18. Ukraine (Zaporozhye)</td>
<td>Lyudmila Chernishova</td>
<td>Apr 19-20</td>
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<td>(<a href="mailto:chernyshova@ukr.net">chernyshova@ukr.net</a>)</td>
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<tr>
<td>19. Russia (St. Petersburg)</td>
<td>Marina Guseva</td>
<td>May 29-30</td>
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<td>(<a href="mailto:gusevamarina@mail.ru">gusevamarina@mail.ru</a>)</td>
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<td>20. Hungary (Pécs)</td>
<td>Bernadett Mosdósi</td>
<td>June 22-23</td>
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<td>(<a href="mailto:bernadett.mosdosi@aok.pte.hu">bernadett.mosdosi@aok.pte.hu</a>)</td>
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<td>21. Romania (Iasi)</td>
<td>Aurica Rugina</td>
<td>Sep 10-11</td>
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<td>22. Macedonia (Ohrid)</td>
<td>Kristina Mironska</td>
<td>Sep 21-22</td>
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<td>23. Slovenia (Ljubljana)</td>
<td>Tadej Avcin</td>
<td>Nov 16-17</td>
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**ZAPOROZHYE**  
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**PÉCS**

**IASI**  
**OHRID**  
**LJUBLJANA**

26
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<th>No.</th>
<th>Name</th>
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<td>1.</td>
<td>Michael Belevtsev</td>
<td>Belarus</td>
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<td>Olga A. Aleinikova</td>
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Molecular genetic testing available at the Debrecen Jeffrey Modell PID Reference Center

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PRIMARY IMMUNODEFICIENCY DISEASES (Genes)

- XLA (BTK)
- WAS (WAS)
- XLP (SH2D1A)
- X-CGD (CYBB)
- X-SCID (IL-2RG)
- X-HIGM (TNFSF5)
- ALPS (TNFSF6)
- AR-CGD (NCF1)
- C2 deficiency (C2)
- IL7R deficiency (IL7R)
- AID deficiency (AICDA)
- RAG-1 deficiency (RAG1)
- RAG-2 deficiency (RAG2)
- Cyclic neutropenia (ELA2)
- WHIM syndrome (CXCR4)
- Kostmann syndrome (HAX1)
- Dyskeratosis congenita (DKC1)
- Cartilage-hair hypoplasia (RMRP)
- Hereditary angioedema (SERPING1)
- Nijmegen breakage syndrome (NBS1)
- Shwachman-Diamond syndrome (SBDS)

LYSOSOMAL STORAGE DISEASES (Genes)

- Fabry disease (GAL)
- Gaucher disease (GBA)

Information:
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