THE J PROJECT

EDITORS:
László Maródi & Zsuzsa Horváth
with the contribution of
Tadej Avcin, Ewa Bernatowska, Peter Ciznar, Liudmyla Chernyshova,
Ismail Reisli, Nima Rezaei, Anna Sediva, Miklós Szolnoky, Irina Tuzankina

SPONSOR:
ECE IPI CTR

December, 2013
Contents

Foreword ................................................................. 3
ECE IPI CTR ............................................................... 4
The J Project – Meetings in 2013 .................................. 5
  Meeting in Mukachevo .............................................. 6
  Meeting in Tehran .................................................... 9
  Meeting in Bratislava ................................................. 10
  Meeting in Debrecen ................................................. 12
  Meeting in Ljubljana .................................................. 13
  Meeting in Prague ..................................................... 15
  Meeting in Klimkovics Spa ......................................... 18
  Meeting in Budapest ................................................ 19
  Meeting in Cracow .................................................... 23
  Meeting in Szolnok .................................................... 24
  Meeting in Tehran .................................................... 26
  Meeting in Ptuj ......................................................... 28
  Meeting in Krasnoyarsk ............................................. 30
  Meeting in Malatya .................................................... 31
  Meeting in Budapest ................................................ 32
J Project Meetings in Central and Eastern-Europe (2004–2013) ................ 33
J Project Steering Committee Members ............................ 36
Genetic testing in the Debrecen PID Reference Center .................... 38

Attila Kondor: Numen I (2012)
Foreword

This J Project booklet is dedicated to the late Pál Megyeri, MD, PhD, who was one of the outstanding academic pediatrician. Dr. Megyeri not only understood the importance and future perspectives of molecular medicine but he made remarkable progress to implement this in everyday clinical practice.

Pál Megyeri began his carrier in Szeged where he trained and specialized in pediatrics at the Pediatric Department of Albert Szent-Györgyi University, and he was a research fellow in immunology. After he established the pediatric immunology center at the Pediatric Department he worked alongside Andrew Issekutz for years in Halifax, Canada and studied inflammatory cytokines. After he completed his post-doctoral fellowship, he was eager to further develop his Center in Szeged in order to increase competence in primary immunodeficiency disease (PID) patient care. His endeavor was not without local difficulties and obstacles. He was endowed with a good sense of humor assuring an immediate pleasant atmosphere in his company. He started to laugh in his unique way before he told us one of his many jokes and stories. Similarly to most PID experts he never received remarkable professional honor or award from authorities. He inspired countless medical students, pediatric residents and immunology and infectious disease fellows with his passion for clinical knowledge both in the lecture room and the bedside and compassion for ill children. Even after he developed a rapidly progressive neurological disease and became restricted to wheelchair he still continued to teach students and see patients. During his last months he withdraws and Miklós Szolnoky was the only colleague and friend who was privileged to see and meet him until his last days when he deleted all his files from his computer. He has left a huge mark in the field of Hungarian Pediatric Immunology and we thank him for all he did on behalf of the pediatric immunology community as well as his patients whose life have been better through his talent, professional responsibility and devotion. Surely, Pali would have been a fan and helpful supporter of the J Project we have started in 2004.

László Maródi
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. 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 is 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 2013**

<table>
<thead>
<tr>
<th>City (Country)</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Mukachevo (Ukraine)</td>
<td>April 18</td>
</tr>
<tr>
<td>2. Tehran (Iran)</td>
<td>April 23</td>
</tr>
<tr>
<td>3. Bratislava (Slovakia)</td>
<td>April 24</td>
</tr>
<tr>
<td>4. Debrecen (Budapest)</td>
<td>April 26</td>
</tr>
<tr>
<td>5. Ljubljana (Slovenia)</td>
<td>May 9</td>
</tr>
<tr>
<td>6. Prague (Czech Republic)</td>
<td>May 13–14</td>
</tr>
<tr>
<td>7. Klimkovice Spa (Czech Republic)</td>
<td>May 31</td>
</tr>
<tr>
<td>8. Budapest (Hungary)</td>
<td>May 31–June 1</td>
</tr>
<tr>
<td>9. Cracow (Poland)</td>
<td>September 20–21</td>
</tr>
<tr>
<td>10. Szolnok (Hungary)</td>
<td>October 5</td>
</tr>
<tr>
<td>11. Tehran (Iran)</td>
<td>October 12–13</td>
</tr>
<tr>
<td>12. Ptuj (Slovenia)</td>
<td>October 18–19</td>
</tr>
<tr>
<td>13. Krasnoyarsk (Siberia, Russia)</td>
<td>October 22</td>
</tr>
<tr>
<td>14. Malatya (Turkey)</td>
<td>November 16</td>
</tr>
<tr>
<td>15. Budapest (Hungary)</td>
<td>November 22</td>
</tr>
</tbody>
</table>

**Aims**

1. To organize professional meetings on PID and related diseases in regions of Central and Eastern 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.
MUKACHEVO, UKRAINE

Host: Liudmyla Chernyshova (luchernyshova@yandex.ua)
Venue: Hotel “Chervona Hora”, Mukachevo
Main Topic: Recent advancement in diagnosis and treatment of complex immunodeficiency disorders

PROGRAM

April 18, 2013

Symposium “Recent advancement in diagnosis and treatment of complex immunodeficiency disorders” in ranks of Central and Eastern European Project – J Project

Diagnostics of primary (innate) immunodeficiencies, medical management of children with PID on different stages of medical care – interactive communication

Diagnostics of primary (innate) immunodeficiencies of antibody production – interactive communication
(Alla Volokha, Kiev)

Markers of combined immune deficiency – interactive communication
(Larisa Kostyuchenko, Lviv)

Peculiarities of Intravenous immunoglobulins production
(Skrynnik Maksim, Kiev)

Extending of clinical usage of intravenous immunoglobulins with their new generations production
(Rybakov Andrey, Kiev)

Attestation and testing of attendees of School-Seminar

First session of scientific-practical symposium with international attendance “Recent advancement in diagnosis and treatment of complex immunodeficiency disorders in children” in ranks of Central and Eastern European Project – J Project

Completion of PID registry in Ukraine in 2012, analyses of achievements in treatment of children with CID
(Liudmyla Chernyshova, Kiev)

IL-17-mediated immunity in patients with CMC
(László Maródi, Debrecen)

Differential role of anti-IL-17 and anti-IL-22 antibodies in CMC pathology in APS-1 patients
(Adrien Katalin Sarkadi, Debrecen)

Large deletion mutations in patients with WAS
(Vera Gulácsy, Debrecen, Hungary)

Organization and functioning of HOPI
(Zsuzsa Horváth, Debrecen)
SUMMARY AND CONCLUSION

One hundred physicians from the most western region of Ukraine, 50 pediatric immunologists from the majority of regions of Ukraine and invited speakers from Hungary, Belarus and Latvia took part at the symposium. In the beginning of the symposium, opinion leader of the Ministry of Health of Ukraine and Head of Ukrainian Association “Pediatric Immunology” professor Liudmyla Chernyshova highlighted main achievements in diagnostics and treatment of primary immunodeficiency in Ukraine during last ten year. The Ukrainian text-book on pediatric immunology with authorship by professors and faculty of department of Pediatric Infectious Diseases and Clinical Immunology of National Medical Academy for post-graduate education (Kiev, Ukraine) was presented. The first part was addressed to general practitioners and pediatricians with the aim to raise awareness concerning primary immunodeficiency for early referring of patients to pediatric immunologists by teaching main clinical features and markers of the most common pathologic forms of primary immune deficiencies. Lectures of chief Ukrainian pediatric immunologists on main topics of deficiency of antibody-production and combined immunodeficiency were presented by professors Alla Volokha (Kiev) and Larysa Kostyuchenko (Lviv). After that, there was an interactive communication focusing on following up whether the knowledge was transferred to the participants properly. At the second part of the Meeting, profound topics of laboratory diagnostics, possibilities of molecular genetics search of primary immunodeficiency were discussed. Well-known scientists from Ukraine, Hungary, Belarus and Latvia presented their lectures on this issue. By the help of interactive communication, discussion focused on problems and questions of primary immune deficiency diagnostics and treatment was organized among pediatric immunologists. In the ranks of symposium, sessions of members of Ukrainian Association “Pediatric Immunology” and discussions on changes in the Status of Association were
held. At the end of symposium, excursion for participants to Mukachevo “Palanok” Castle was organized. Informal atmosphere and hospitality of “Zakarpatty”, blossoming Sakura, allowed us to give the perfect impression of the organized Symposium.

Liudmyla Chernishova
Program and Summary

The 85th J Meeting was organized in Tehran, Iran during the World PI Week (April 22–29, 2013). The Research Center for Immunodeficiencies (RCID) and the Children’s Medical Center (the Pediatrics Center of Excellence in Iran) affiliated to Tehran University of Medical Sciences, organized an immunodeficiency symposium. Pediatricians as well as pediatric fellows, residents and medical students attended the meeting to become aware on recent classification of PID and approach to patients with recurrent infections. The symposium was started with case discussions, and ended with an expert PID panel. The media were also invited for interviews. Medical ethics session showing the movie on “Bubble Baby” was an additive in this program.

Nima Rezaei, Asghar Aghamohammadi
BRATISLAVA, SLOVAKIA  
April 24, 2013

Host: Peter Ciznar (ciznar@dfisp.sk)
Venue: Children’s University Hospital, Bratislava
Main Topic: 3rd Symposium on primary immunodeficiencies

PROGRAM

Innate immunity deficiencies
Basics of antifungal immunity
(Paulovicová Ema, Bratislava)
STAT mutations in patients with CMC
(László Maródi, Debrecen)
DNA reparation defects
(Milos Jesenák, Martin)
BMT in patients with CGD – case presentations
(Júlia Horáková, Bratislava)
Di George syndrome patient group in DFNsP
(Linda Libai Véghová, Bratislava)

Defects in specific immunity
Common variable immunodeficiency
Univ. (Hermann Wolf, Vienna)
Hyper-IgM syndrome, know-unknown
(Ales Janda, Freiburg)
Agammaglobulinemia – case presentation
(Denisa Ilencíková, Bratislava)
Case presentation – CVID in adults
(Jana Strakova, Martin)
Neonatal screening for SCID – how far is the issue?
(Peter Ciznar, Bratislava)

SUMMARY AND CONCLUSION

The 3rd nationwide conference on primary immunodeficiency (PID) with international participation was held on April 24, 2013 in the setting of the Children’s University (Faculty) Hospital in Bratislava. It was attended by 92 registered participants of whom 11 were active lecturer. Four of the speakers were from Germany, Austria and Hungary. The conference was organized around two thematic sections – primary immunodeficiencies of innate immunity and primary immunodeficiencies of adaptive immunity. Both groups represent a heterogeneous group of hereditary diseases that are classified as rare diseases with an overall prevalence estimated to be 1:20,000. The event was held during the week of global primary immunodeficiency week (www.worldpiweek.org), organized by professional societies and non-profit organizations from around the world (e.g. ESID, INGID, IPOPI, JMF, EFIS, CIS, IDF, LASID, PPTA and others). Public awareness activities, including TV media information preceded the conference. First Pediatric Department of the Children University Hospital a.k.a. the Center for Primary Immunodeficiencies organized this symposium already for the third time as a part of Central and East Europe Initiative, a J Project, under the auspice of the CEE-PID-Center in Debrecen, Hungary and the Jeffrey Modell Foundation Networking (JMCN). Both Slovak immunology societies, the Slovak Society of Clinical Immunology and Allergology (SSAKI) and Slovak Immunology Society (SIMS) actively supported the whole event. Symposium was intended for general doc-
tors and medical specialists in internal disciplines (pediatrics, hematology, dermatology, ENT etc.) as well for the laboratory medicine specialists. During the symposium nurses had the opportunity to participate in a site training program for home immunoglobulin treatment and lectures to become familiar with the topic of primary immunodeficiencies. Symposium initial lectures were focused on antifungal immunity with respect to the inherited genetic defects. This was followed by lectures addressing clinical experience in transplantation of complex disorders of neutrophil function, chronic granulomatous diseases with the presentation of first experiences of successful HSCT in Bratislava Center. Subsequently an analysis of patients with DNA repair defects and Di George syndrome were presented. The second part of symposium covered the topic of common variable immunodeficiency and hyper-IgM syndrome by an experienced specialist in the field from Vienna and Freiburg. Finally the topic of PI screening in newborns was discussed. All the presentations were accompanied by live discussions. Conference language was parallel Slovakian and English, interpreted continuously by a moderator. The event was generally assigned as a high professional level meeting. Evaluation forms showed that lectures highlighted new theoretical and practical information and allowed improving clinical practice of participants in the sake of early diagnosis and treatment of patients with PI. Organizers were recommended to continue in the series of annual nationwide meetings by the representatives of the local professional community.

Peter Ciznar
PROGRAM

The current financial background of IVIG/SCIG treatment in Hungary
(Tamás Szamosi, Budapest, Margit Zeher, Debrecen)

Problems of financing of PID gene sequencing
(László Maródi, János Kappelmayer, Debrecen)

Hungarian PID registry
(Melinda Erdős, Beáta Tóth, Debrecen, Beáta Dérfalvi, Budapest, Miklós Szolnoky, János Kórház, Budapest)

Collaborations and activities of the Hungarian PID centers
(Kálmán Nagy, Miskolc, Bernadett Mosdósi, Zoltán Nyul, Pécs)

SUMMARY AND CONCLUSION

This J Project meeting organized during the World Primary Immunodeficiency Week was entirely devoted to the management of Hungarian patients with immunoglobulin deficiencies. Various reimbursement policies used previously in Hungary was discussed. Immunoglobulin treatment in this country was started in 1981 by using Sandozglobulin and continued with the novel, safer and more tolerable preparations later on. The reimbursement of treatment by the insurance company (OEP) has never been completely managed before. At this J Project meeting we decided to discuss this issue with an authentic representative of OEP and clinical immunology leadership of the country. We came to an agreement by the end of the meeting that all patients in Hungary who are diagnosed with antibody deficiency or other PIDs that should be treated with immunoglobulin replacement should have access to the currently available preparations free of charge. This milestone meeting proved to be the first success of the mission of the recently established Hungarian Society for Immunodeficiencies (HUSID). We also overviewed the PID national registry and discussed current state and future challenges of collaboration between the Hungarian PID Centers.

László Maródi
LJUBLJANA, SLOVENIA

May 9, 11, 2013

Host: Tadej Avcin (tadej.avcin@gmail.com)
Venue: University Children’s Hospital, Ljubljana
Main Topic: J Project spring meeting

PROGRAM

May 9

Primary immunodeficiencies in adults and national PID registry
(Tadej Avcin, Ljubljana)
Tests of immune system in adults with PID
(Alojz Ihan, Ljubljana)
Genetic tests in adults with PID
(Marusa Debeljak, Ljubljana)

Presentation of center for adults with PID,
Department of Infectious Diseases and Febrile Illnesses, UMC Ljubljana
Presentation of center for adults with PID, University Clinic of Pulmonary and Allergic Diseases Golnik
Presentation of other centers for adults with PID
Case presentations and transition of care
May 11

Report on activities of our Society in 2012 and goals for future activities
*(Tadej Avcin, Ljubljana)*

Recurrent fever syndromes in children: etiology, clinical picture and treatment
*(Natasa Toplak, Ljubljana)*

Presentation of poster about human immune system function
*(Stefan Blazina, Ljubljana)*

Coping with chronic illness in family
*(Mateja Gorenc, Ljubljana)*

Protection from infections for children suffering from PID
*(Maja Camernik, Ljubljana)*

Living with PID – personal experience and report on PID meeting in Vienna
*(Sandra Sparas, Maribor)*

### SUMMARY AND CONCLUSION

Two J Project meetings were organized by the Slovenian Society for Children with Immune Diseases in 2013. The spring meeting (May 9, 2013) was organized at the University Children’s Hospital Ljubljana and was dedicated to establish national multidisciplinary team for management of adult patients with PID. Primary immunodeficiencies that are more common among adults and updated data from the Slovenian PID registry were presented, followed by descriptions of specific tests of immune system and genetic tests in adult PID patients. These patients are treated in two centers, both of which were represented on the meeting. After the presentations, specific clinical cases of PID patients that reached adulthood were presented and discussed including transition of care in one of the centers for adults. As part of the spring J Project meeting and World PI week campaign, a scientific program for parents of

*Participants of the J Project spring meeting*
children with PID was held near capital city Ljubljana on May 11, 2013. Topics for patients and parents were activities of the patients’ society, autoinflammatory diseases, presentation of translated poster about human immune system (from JMF), psychological aspects of PID burden for families and protection from infections. One of our patients presented her personal experiences about living with PID. She also reported in a PID meeting held in Vienna. Social program was organized for younger children during the presentations. They enjoyed in magician’s show and making their own paper kites.

Stefan Blazina, Tadej Avcin

**PRAGUE, CZECH REPUBLIC**

May 13–14, 2013

**Host:** Anna Sediva (anna.sediva@fnmotol.cz)

**Venue:** Department of Immunology, 2nd Medical School, Charles University, Motol University Hospital

**Main Topic:** 12th ESID Prague Spring Meeting

**PROGRAM**

**May 13**

**B cells**

B cell development

*(Menno van Zelm, Amsterdam)*

Diagnostic vaccination in hypogammaglobulinemic patients

*(Vojtech Thon, Brno)*

Subgroup of Common Variable Immunodeficiency patients with distinct clinical and biological features revealed by B-cell immunophenotyping

*(Tomás Kalina, Prague)*

Impaired B cell development in a patient with Xq24 deletion

*(Anne-Kathrin Kienzler, London)*

Management of patients with CVID in South Bohemia

*(Tomás Milota, Prague)*

Soluble CD14 but not Endotoxin is Increased in CVID

*(Jiri Litzman, Brno)*

From autoimmunity to immunodeficiency – regression of systemic lupus after development of common variable immunodeficiency syndrome

*(Pavlina Králícková, Eva Malá, Sárka Imrichová, Hradec Hrálové)*

Practical experiences with s.c. immunoglobulin replacement therapy in Motol Hospital

*(Radana Zachová, Prague)*

**Rare immunodeficiencies and syndromes**

Hermansky Pudlak and related disorders of organelle formation

*(Raffaele Badolato, Brescia)*

Deficit of GATA-2 transcription factor:
new immunodeficiency syndrome with broad phenotype. First patients diagnosed in the Czech Republic  
(Ales Janda, Freiburg Ester Mejstríková, Prague)

HLH in developing countries challenges  
(Mohamed Abdelaziz Almalky, Zagazig)

Hematopoietic stem cell transplantation in cartilage-hair hypoplasia – case report  
(Renata Formánková, Prague)

FMF in Czech Republic  
(Anna Sedivá, Prague)

Is this PID? – Case report  
(Andrea Poloucková, Radana Zachová, Prague)

May 14

Other well defined PID

Mucocutaneous candidiasis  
(Anne Puel, Paris)

Chronic intestinal inflammation in the context of PIDs  
(Cecilia Domínguez Conde, Vienna)

The first experience of genetic diagnosis of ataxia-telangiectasia in Ukraine  
(Ihor Savchak, Lviv)

NBS – fear of neoplasia, not easy to be proved/ Fever of unknown origin – what should be the next step  
(Ewa Wiesik-Szewczyk, Warsaw)

Family case of Nijmegen breakage syndrome, complicated by EBV-lymphoproliferative disease and lymphoma  
(Larysa Kostyuchenko, Lviv)

Clinical and genetic characteristics of nine Ukrainian patients with Wiskott–Aldrich syndrome  
(Yaryna Romanyshyn, Lviv)

Humoral immunity in patients with Di George syndrome  
(Adam Klocperk, Prague)

Laboratory diagnostics and novel technologies

Autoimmunity and autoinflammation in PIDs  
(Kaan Boztug, Vienna)

The immunomodulatory role of endogenous glucocorticoids in ovarian cancer  
(Ahmed Adel Seida, Cairo)

Massively parallel sequencing for rapid and accurate newborn screening of patients with severe combined immunodeficiencies (SCIDs)  
(Nina Pergemann, Vienna)

How to handle intronic mutations?  
(Tomás Freiberger, Brno)

SCID newborn screening study in Czech Republic  
(Michal Svaton, Adam Klocperk, Anna Sediva, Prague)

Establishment of an optimized data analysis pipeline for next generation sequencing data to identify monogenetic defects in patients with primary immunodeficiencies  
(Christine Jandrasits, Vienna)

SUMMARY AND CONCLUSION

The 12th ESID Prague Spring Meeting was organized by the Department of Immunology and the Clinic of Pediatric Hematooncology of the 2nd Medical faculty of the Charles University and the Motol University Hospital, Prague, Czech Republic, on May 13 and 14, 2013. The meeting took place in its traditional settings in the Olympus facility in Prague. It was attended by 32 participants from 9 countries, majority of
them were from the Czech Republic, more arrived from Austria, Egypt, France, Italy, Poland, Ukraine, United Kingdom and The Netherlands, in alphabetical order. The meeting emphasized the active participation of young immunologists and gave them the opportunity to present their work in oral presentations. Since its launch in 2002 the Prague ESID meeting has been devoted to the exchange of information on primary immunodeficiencies (PIDs) between Western and Central and Eastern Europe. The continuous effort to promote exchange of information and rise of scientific levels in Eastern Europe is partly successful. So far several excellent groups were formed in Eastern European countries. There is, however, still a visible gap between the East and West part of the Old Continent in diagnosing and treating primary immunodeficiency patients and also in related research activities. The program of the meeting was divided into four major blocks supported by contributions and supervision of invited speakers. B cell panel was introduced by Menno van Zelm and later on, it was extensively covered by series of lectures on pathogenesis, diagnosis and treatment of B mediated disorders. Rare immunodeficiencies and well defined syndromes included very interesting talks on novel discoveries in disorders of organelle formation, given by Reffaele Badolato, and showed several other novel genetic causes of immunodeficiencies. Anne Puel gave an excellent review on new findings in pathogenesis of mucocutaneous candidiasis. The last afternoon was devoted to novel technologies and laboratory diagnostics. Sufficient amount of time for discussions and a very active and relaxing atmosphere established a traditionally creative environment, typical for ESID Prague Spring Meeting series. Social programs were also very interesting. The special evening took place in a newly opened University Club, belonging to Charles University. Situated in city center and located in a gothic basement, the club provided unusual and inspiring settings for a cultural program. It included the opening exhibition of paintings of Academy of Fine arts on the theme Prague, was followed by a block of folk songs and yoyo performance. Traditional Czech dinner in a close by restaurant finished the evening. The meeting was traditionally organized as a part of the activities related to the Day of Immunology, declared by EFIS on April 29, 2013 and to Primary Immunodeficiency Week 2013. The meeting is also an event of J projects meetings, promoting awareness about PIDs in Eastern European Countries. ESID Prague Spring Meeting was supported by the Charles University, 2nd Medical School, Prague and by Motol University Hospital, Prague. General partners of this year events were Baxter, CSL Behring, Olympus and Shire, further contributions came from Grifols, Exbio, Octapharma, Schoeller and Synlab. Special support was provided by Czech Immunology Society. The meeting is traditionally supported by Jeffrey Modell Centers Network.

Anna Sediva
PROGRAM

Humoral immunodeficiencies
(Jiri Litzman, Brno)
Pneumopathology of CVID in children and adults
(Pavlína Králícková, Hradec Králové)
Laboratory investigation in humoral immunodeficiencies
(Vojtech Thon, Brno)
Lymphocyte subpopulations in immunodeficient patients
(Jana Nechvátalová, Brno)
Current possibilities of laboratory diagnostics of cellular immunodeficiencies
(Alexandra Lochmanová, Ostrava)
10 warning signs of primary immunodeficiencies
(Andrea Poloucková, Prague)
Immunoglobulin replacement therapy, Practical aspects
(Radana Zachová, Prague)
Cellular immunodeficiencies and Di George syndrome
(Anna Sediva, Prague)
Transplantation of hematopoietic stem cells in primary immunodeficiencies
(Renata Formánková, Prague)
Multifunctional CMV specific T-cells are the sign of successful defense against reactivation of CMV in children after HSCT
(Jan Stuchly, Prague)
Deficiency of transcription factor GATA-2: New immunodeficiency with broad phenotype spectrum
(Ester Mejstríková, Prague)
Molecular genetic testing for primary immunodeficiencies
(Tomáš Freiberger, Brno)

SUMMARY AND CONCLUSION

The main purpose of a meeting is education in PIDs. The program is composed a very practical way, to give an audience the overview of an immune system and primary immunodeficiencies. Part of the program is practical, such as SCIG practical tips and experience (see the program). The meeting was attended by 60 physicians mostly from local area, from North Moravia, a region with less developed care for primary immunodeficiencies, but on the other had a region with many cases of these diseases, including SCIDs. Based on reactions of attending physicians the meeting was successful. We also distributed educational materials, booklet on PIDs for patients and parents and flyers with information on PIDs.  

Anna Sediva
Conclusions and agreements
In his general introduction, László Maródi proposed a consideration of the items of the Minutes of the 2012 SC Meeting, with discussion of the achievements of the last one and a half years and future plans.

1. Summary reports for PID organizations: Working groups/Societies/Patients’ groups/Nursing groups

L Maródi reported on the establishment of a legally registered, independent, professional PID society, the Hungarian Society for Immunodeficiencies (HUSID), and on the Hungarian Organizations for Patients with Immunodeficiencies (HOPI) and the Hungarian Nursing Group for Immunodeficiencies (HUNGID), which are also independent and legally registered. The representatives of all countries then reported on their management of the development of equivalent organizations in their countries. Hungary is currently the only country with a nursing group. Patient groups have been organized, but their activity varies significantly between countries. Professional organizations mostly exist as working groups (WG) under the umbrella of national allergy and clinical immunology societies. In Poland, the PID WG is independent; in Iran, the WG is functioning as a network.

It was agreed that professional organizations and patient groups should be developed further and, if feasible, they should attain legally independent status. Organizations of nursing groups are needed in all J Project countries.

2. PID registry – overlapping registries (ESID, JMF, JMF-Octapharma, J Project)

Several PID registries currently exist, but some are complicated (ESID registry) and others are incomplete (JMF, JMF-Octapharma). A new, relatively simple, but informative questionnaire was proposed by L Maródi and several suggestions were made as to how to improve it, by including data on consanguinity and treatment and omitting dates of birth and death, for ethical and legal reasons, for example.

It was agreed to develop a uniform national registry in each J Project country, with data entry in English, to facilitate the combination of national registries into a J Project registry. Data should be entered online where possible, and there may be disease-specific registries in addition to com-
prehensive national registries. A J Project Registry Committee was established, with the following members: B Tóth, T Avcin, P Ciznar, A Aghamohammadi.

3. Affiliation of the J Project

E Bernatowska reported on the affiliation of the J Project group to the Central European Journal of Immunology (CEJI). The J Project group will be indicated on the front cover and the first page of this journal in the near future. L Maródi informed the SC that the last issue of CEJI in 2012 published an editorial that he had written, together with two papers focusing on PIDs, one from Ljubljana-Trieste and one from Warsaw.

The Committee was informed that new members of the editorial board from the J Project group may be proposed for consideration by the Editor-in-Chief of CEJI. The proposed editorial board members are: E Bernatowska, M Serban, M Erdős, T Avcin, J Litzman, P Ciznar.

L Maródi informed the SC that Vincent Bonagura, Chief Editor of the Journal of Clinical Immunology (JoCI) had asked the J Project group to associate itself with the JoCI. This association would be best achieved by the J Project group members becoming affiliated members of the Clinical Immunology Society (CIS). J Project group members would be offered reduced rates of $50 per year for online subscription to the JoCI and membership of the CIS. An additional $20 would be required to obtain the printed version of JoCI. It is particularly important for J Project members to embrace the JoCI given that its current editors, V. Bonagura and Jean-Laurent Casanova, intend to develop this journal to focus primarily on PIDs in the future.

SC members will communicate this information in their home countries, so that all healthcare professionals and scientists working in the field of PIDs have the opportunity to become CIS members at the reduced rate. SC members were asked to provide feedback about the interest of healthcare professionals from their countries in this possibility.

4. J Project clinical research collaborations

Ongoing clinical research collaborations and future plans were discussed, as follows:

– **CGD project (coordinator: T Avcin)**
  **Achievements:** 59 patients with CGD from ECE countries have been analyzed, to improve the definition of genotype-phenotype relationships and sequence variation in the region. A research paper is currently in preparation.

– **NBS project (coordinator: E Bernatowska)**
  **Achievements:** 149 patients with NBS have been studied and the results of this clinical and genetic evaluation will be submitted to JACI in the near future.

– **IVIG/SCIG therapy in Europe (coordinator: A Sediva)**
  **Achievements:** Comprehensive analyses of the availability of intravenous and subcutaneous immunoglobulin and of new treatments of this type in
Western and Eastern European countries have been performed. The major conclusions will be summarized and published in a medical immunology journal.

- **WHIM syndrome project (coordinator: K Mironska)**

  **Achievements:** Four families from Macedonia, Romania, and Hungary have been diagnosed and followed. All patients had recurrent \textit{CXCR4} sequence variants. This study will continue and additional patients must be included before publication can be considered.

- **CMC project (coordinator: L Maródi)**

  **Achievements:** Nine patients diagnosed with chronic mucocutaneous candidiasis (CMC) and gain-of-function mutations of \textit{STAT1} have been analyzed by molecular genetic and immunological approaches. A paper reporting the clinical, immunological and genetic data for these patients has recently been accepted and published by J Med Genet (May 24, 2013).

5. **J Daughters Project**

The spirit and concept of the J Project have recently been extended to Eastern Europe and Western Asia. The J Daughters Projects include:

- **The J Anatolia Project (coordinator: Ismail Reisli)**
- **The J Persia Project (coordinator: Nima Rezaei)**
- **The J Siberia Project (coordinator: Irina Tuzankina)**
- **The J Central Asia Project (coordinator: Nurzhan Otarbayev)**

Reports about the exciting and ambitious work of the J Persia Project and the J Siberia Project were presented by Asghar Aghamohammadi and Irina Tuzankina, respectively. A research paper by Zs. Horváth \textit{et al.} concerning “The spread of the J Project” was recently published in JoCI (May 19, 2013).

6. **J Project Meetings 2013**

Fourteen J Project Meetings are scheduled for this year, culminating in the 97th J Project Meeting at the end of 2013. Thus, early in 2014, probably in March, we shall celebrate the 100th J Project Meeting in Hungary.

7. **The J2 Project**

The J Project was launched nine years ago, as a physician education program in Central Europe. It subsequently spread to Eastern Europe and, more recently, to Siberia (Russia) and Western Asia, Iran and Eastern Turkey (Anatolia). It has also gradually developed into clinical research collaboration, resulting in the publication of more than 10 clinical research papers in international immunology journals. The J Project received a considerable impetus from the genetic diagnostic service provided by the Debrecen PID Center to all J Project countries lacking genetic diagnostic laboratories for the analysis of genes involved in PIDs. With the continued generous support of Biotest Hungaria Kft., sequence analyses have been performed free of charge at the Debrecen Center. However, one of the major aims
of the Project formulated in recent years was the establishment of genetic diagnostic laboratories in all ECE countries. This program is known as the J2 Project. Genetic diagnostic testing facilities are now available not only at the Debrecen Center, but also in Brno in the Czech Republic, Ljubljana in Slovenia, and Minsk in Belarus. However, the J2 Project remains in its infancy in most countries.

All ECE countries should become self-sufficient in terms of genetic analysis for PIDs in the near future. However, until this objective is achieved, the existing genetic laboratories should continue to provide assistance in studies of PID genes in other countries. The recently launched Central and Eastern European PID Centers Network (CEE-PID-CN) program, which provides financial support to selected PID centers, should contribute to progress in the J2 Project. It was proposed that the CEE-PID-CN project should allow the Centers to use their support more flexibly, increasing the proportions of their budgets spent on genetic testing rather than raising awareness.


One of the suggestions made at the SC Meeting was that a J Project Junior Meeting (JPJM) should be organized, to be attended mostly, but not exclusively, by young research fellows from Central and Eastern Europe. As we will celebrate the 10-year anniversary of the J Project in 2014 and the 100th J Project meeting, which will be organized next March, it would be reasonable to launch the JPJM in Hungary in conjunction with the 100th J Project Meeting. We plan to organize this 100th meeting in Budapest, and experts from all over the world will be invited. The JPJM could be scheduled to take place before the 100th J Project meeting. After this unique event, the location and structure of the JPJM may change.

9. J Project Society

This item was not considered appropriate for discussion.

10. Harmonization of the Central and Eastern European PID Centers Network and the J Project

This item was not considered appropriate for discussion.

11. J Project Bulletin

A Aghamohamaddi suggested developing the J Project website further and creating a J Project Bulletin. This suggestion was accepted.
CRACOW, POLAND

September 20–21, 2013

Host: Ewa Bernatowska (ewa.bernatowska@gmail.com)
Venue: Qubus Hotel, Cracow
Main Topic: Second Conference of the Polish Working Group on primary immunodeficiencies

PROGRAM

September 20

Jeffrey Modell Diagnostic and Research Center – Children’s Memorial Health Institute activity report
(Ewa Bernatowska, Cracow)

Polish registry of PID
(Ewa Bernatowska, Cracow)

Treatment with gammaglobulins in children and adults – data from national Centers for Diagnosis and Treatment of primary immunodeficiencies
(Małgorzata Pac, Warsaw)

HSCT in PID patients – polish experience
(Beata Wolska-Kusnierz, Warsaw)

Diagnostics and treatment of adult patients with PID
(Karina Jahnz-Różyk, Warsaw, Jacek Musial, Cracow, Jacek Rolinski, Lublin)

Therapeutic program for immunoglobulin treatment of PID patients – current problems
(Maciej Siedlar, Cracow)

Newborn screening in PID
(Krzysztof Zeman, Lodz)

Newborn screening in Europe – preliminary data
(Małgorzata Pac, Warsaw)

Discussion on possibilities of introduction of newborn screenings tests in Poland
(Katarzyna Kuśmierska, Cracow, Mariusz Ołtarzewskim, Warsaw)

Consensus on standards of vaccination in children and adults with PID
(Ewa Bernatowska, Cracow)

September 21

Chronic granulomatous disease – current diagnostics and therapeutic approach

CEJI – partner of J Project Group
Janusz Marcinkiewicz, Cracow, Włodzimierz Maśliński, Warsaw

Plenary lecture: From BMT to Gene Therapy: Special challenges and progress in CGD
Reinhard Seger, Genetics of CGD in Central and Eastern European countries
(Marusa Debeljak, Ljubljana)

Flow cytometry in differential diagnostics of primary immune deficiencies with granulomatous lesions
(Barbara Piatosa, Warsaw)

Genetic and clinical picture of CGD
(Edyta Heropolitska-Pliszka, Warsaw)

Egypt – one center experience with CGD
(Aisha Elmarsafy, Cairo)

Liver involvement in X-CGD
(Anna Székely, Péter Mózes, Jr., Melinda Erdős, László Maródi, Debrecen)

HSCT in CGD – polish centers experience
(Beata Wolska-Kusnierz, Warsaw, Krzysztof Kalwak, Wroclaw, Katarzyna Drabko, Lublin; Jolanta Goździk-Spychalska, Cracow, Jacek Wachowia, Poznan)
SUMMARY AND CONCLUSION

Seventy-five clinical immunologists, pediatricians and internists attend the conference, to discuss how to improve PID patient care. It has been presented that the number of the diagnosed patients with PIDs increased from 2,976 in 2012 to 3,440 in 2013. Some improvement concerning replacement therapy for adults has been noticed; patients diagnosed in pediatric centers with agammaglobulinemia/CVID could continue IVIG/ScIg therapy in adult’s center, as part of a National Therapeutic Program, which is fully guaranteed by the National Health Fund. Discussion on possibilities of implementation of newborn screenings in Poland was made between immunologists and representatives from the Center of Newborn Screening, Institute of Mother and Child, Warsaw. The pilot study with TREC screening assay is going to start next year, as a part of the Pomerania ongoing NBS program for the new metabolic errors, in North-West region of Poland, and will cover 18 000 newborns/year. It was announced that new members of the Editorial Board of the Central European Journal of Immunology have been nominated by the J Project Steering Committee: E Bernatowska (Poland), M Erdős (Hungary), T Avcin (Slovenia), J Litzman (Czech Republic), P Ciznar (Slovakia).

Ewa Bernatowska

SZOLNOK, HUNGARY

<table>
<thead>
<tr>
<th>Host:</th>
<th>Zsuzsa Horváth (<a href="mailto:hzsu86@gmail.com">hzsu86@gmail.com</a>)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Venue:</td>
<td>Congress Center, Szolnok</td>
</tr>
<tr>
<td>Main Topic:</td>
<td>Clinical, immunopathological and genetic aspects of chronic mucocutaneous candidiasis</td>
</tr>
</tbody>
</table>

Chairs: László Maródi, Zoltán Péter

PROGRAM

Chronic mucocutaneous candidiasis in patients with DOCK8 deficiency
(Beáta Tóth, Debrecen)  
Anti cytokin antibodies in patient with APECED syndrome
(Adrien Sarkadi, Debrecen)
Ophtalmological manifestation of APECED syndrome  
(Judit Dohán, Budapest)
STAT1 GOF mutation in patients with CMC  
(Beáta Soltész, Debrecen)
Gastrointestinal complications of CMC  
(Zoltán Péter, Budapest)
Immunological studies in patients with genital candidiasis  
(Márta Szegedi, Debrecen)

**SUMMARY AND CONCLUSION**

In concert with the agreements at the 3rd J Project Steering Committee meeting, a new professional society: Hungarian Society for Immunodeficiencies (HUSID) was established. The major aims of the Society are to promote PID patient care, education and research in Hungary. Among several approaches HUSID organizes sessions at meetings of other professional and scientific societies. This time we organized a session on chronic mucocutaneous candidiasis, a cross road of immunology and infectious diseases at the meeting of the Hungarian Society of Infectious Diseases. Various aspects of CMC including genetics, immunopathology, clinical manifestations and management were discussed.

*László Maródi*

Anna Tusnádi (L), organizer of the meeting
SUMMARY AND CONCLUSION
The 6th Meeting on Clinical Immunology and Immunodeficiencies, also considered as the 94th J Project Meeting, was organized in October 12–13, 2013 (Tehran, Iran), along with 25th International Congress of Pediatrics. 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 and provide updates in this field, whereas many scientists and researchers took part in this congress to broaden 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-day meeting was 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, 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. Since 2011, as of establishment of the Research Center for Immunodeficiencies, the Meetings on Clinical Immunology and Immunodeficiencies is organized annually along with International Congress of
Pediatrics in October. This year, the 6th Meeting on Clinical Immunology and Immunodeficiencies, considered as the 94th J Project Meeting, was organized along with the 25th 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 was the main objective of the meeting. We were very pleased to host a total of more than 1,000 delegates in this meeting. The majority of the audience included general practitioners, pediatricians, and pediatric subspecialists; however, medical students, pediatric residents and fellows in the field of clinical immunology and infectious diseases actively participated in the meeting. The meeting was organized in four sessions in four different halls. The main session of the meeting was started by a key-note lecture by Prof. Jean-Laurent Casanova who gave a presentation with the title of “Toward a genetic theory of childhood infectious diseases”. Dr. Asghar Aghamohammadi presented a lecture on evaluation of the immune system on those suspicious to immunodeficiencies, while Dr. Nima Rezaei presented an update on the classification of primary immunodeficiencies. Then a lecture on recent advances on bone marrow transplantation of immunodeficient patients was presented by Dr. Amir Ali Hamidieh, and a report of primary immunodeficiencies from Azerbaijan was presented by Dr. Gulnara Nasrullayeva. After two further talks, an official memorandum of understandings between the centers in Iran and Azerbaijan was signed.

Nima Rezaei

PTUJ, SLOVENIA October 18–19, 2013

Host: Tadej Avcin (tadej.avcin@siol.net), Stefan Blazina (stefanblazina@yahoo.com)
Venue: Old town of Ptuj
Main Topic: Slovenian registry of PID and clinical aspects of IgA deficiency

PROGRAM

Slovenian registry of PID and clinical characteristics of patients with IgA deficiency
(Stefan Blazina, Markelj Gasper, Tadej Avcin, Ljubljana)
Tests of immune system in adult patients with suspected PID
(Ihan Alojz, Ljubljana)
Screening of Swiss blood donors for IgA deficiency: Significance for the investigation and transfusion of patients with anaphylactic transfusion reactions
(Hustinx Hein, Bern)
Screening blood donors for IgA deficiency – new registry
(Hrasovec Vesna, Levicnik Stezinar Snezna, Krafic Ana, Ljubljana)
Guidelines for blood products supply for IgA deficient patients in Slovenia
(Mali Polona, Ljubljana)
SUMMARY AND CONCLUSION

The autumn meeting (18th–19th Oct 2013) was organized together with the Slovenian society for transfusion medicine and the Slovenian Society for Haematology in the old town of Ptuj. The PID program focused on the Slovenian registry of PID and clinical aspects of IgA deficiency. The session included presentations of immunological tests of patients with suspected PID and presentation of novel in house method to determine of the low level of concentration of the IgA antibodies. A case of severe anaphylactic reaction during IVIG application was presented and served for discussion on screening blood donors for severe IgA deficiency which started last year. Guidelines regarding transfusions in IgA deficient patients were presented. Swiss and Slovenian registries of IgA deficient blood donors were compared. There was extensive discussion on IgA deficiency in relation to screening blood donors, transfusion reactions and follow up of healthy IgA deficient blood donors.

Stefan Blazina, Tadej Avcin
**Krasnoyarsk, Siberia, Russia**  
October 22, 2013

**Hosts:** Irina Tuzankina (*ituzan@yandex.ru*)
- Managing Committee of international project for primary immunodeficiency J Project, daughter J Project for Ural and Siberia;
- Institute of Immunology and Physiology of Ural Branch of Russian Academy of Sciences;
- Ministry for Public Health of Krasnoyarsk region
- Medical Scientific Research Institute for Northern Problems of Siberian Division of Russian Academy of Medical Sciences (MSR Institute for Northern Problems SD RAMS);
- V. F. Voino-Yasenetskii Krasnoyarsk State Medical University

**Venue:** Clinics of MSR Institute for Northern Problems SD RAMS, Krasnoyarsk

**Main Topic:** Clinical discussions and a practical seminar

**PROGRAM**

**October 22**

Diagnostics of immunopathology primary immunodeficiency: orphan diseases  
(*Irina Tuzankina, Ekaterinburg*)

Clinical diagnostics of immune system functional disorders  
(*Svetlana Smirnova, Krasnoyarsk*)

Principles of therapy for primary immunodeficiency. Clinical polymorphism PID  
(*Alexandr Pishchalnikov–Chelyabinsk*)

**SUMMARY AND CONCLUSION**

October 22, 2013 in Krasnoyarsk (Russia) in the framework of the Russian school with international participation “Immunology in Clinical Practice” was held a seminar on “Primary immunodeficiencies” with the assistance of international project “J Project”.

*Prof. Sergey Tereschenko, Prof. Svetlana Smirnova, Prof. Irina Tuzankina, Yuriy Klimov (L to R)*

*Krasnoyarsk railway station*
The seminar was organized by Siberian Branch of Russian Academy of Sciences, the International Steering Committee of the project on primary immunodeficiencies “The J Project”, affiliate project of “The J Project” the Urals and Siberia, The Ministry of Health of the Krasnoyarsk region, Institute of Medical Problems of the North (Siberian Division of Russian Academy of Medical Sciences), Krasnoyarsk State Medical University, Institute of Immunology and Physiology (Ural Branch of the Russian Academy of Sciences), Russian Society of Immunology. The seminar was attended by more than 60 doctors and researchers of the Krasnoyarsk Territory. The clinic of Institute of Medical Problems of the North was conducted clinical examination of patients with suspected primary immunodeficiencies. Then professor Tuzankina I. A. (project manager of the subsidiary J project in the Urals and Siberia, head of the Regional Center for Clinical Immunology in the Regional Childrens Hospital of Ekaterinburg) read two lectures. The seminar participants were awarded with certificates of participants.

Irina Tuzankina

MALATYA, TURKEY

November 16, 2013

Host: Ismail Reisli (ireisli@hotmail.com)
Venue: Departments of Pediatric Immunology of Necmettin Erbakan University, Anatolia
Main Topic: Primary immunodeficiency disorders. Clues to diagnoses

PROGRAM

PIIDs in pediatric patients. Clinical and laboratory clues
(Ismail Reisli, Konya)

PIIDs in adult patients (Ahmet Zafer, Konya)

SUMMARY AND CONCLUSION

We had planned to organize educational meetings in Anatolia, especially Eastern Anatolia where has no immunologist in, to increase the awareness of primary immunodeficiency diseases (PID) among the physicians. The third meeting of Eastern Anatolian J Project, also the sixth J Project in Turkey, was held on 16–17 November, 2013 in Malatya. The meeting was organized by the Departments of Pediatric Immunology of Necmettin Erbakan University and İnönü University at Ramada Convention Center. First, I would like to thank Dr. Ferhat Çatal and Dr. Erdem Topal for their contribution to the meeting. The meeting was primarily focused on the awareness, clinical signs and early diagnosis of PIIDs among the medical students and physicians (pediatricians, family physicians, etc). More than 100 participants attended the meeting. The meeting was started with an opening lecture by Prof. Ismail Reisli focused on the awareness of primary immunodeficiency. After this, two lectures were given in the meeting which
was based on diagnostic problems, clinical signs, screening tests of PIDs by Prof Ismail Reisli and Prof Zafer Caliskaner. After the meeting, the shuttle buses transferred the participants to a nice restaurant to have a traditional Malatya dinner. I would like to thank all speakers and participants for their contributions, especially to our sponsor (Gen Ilac) for their financial help which made it possible to organize this educational meeting.

Ismail Reisli

**BUDAPEST, HUNGARY**

November 22, 2013

| Host: | László Maródi (lmarodi@med.unideb.hu) |
| Venue: | Hungarian Academy of Sciences, Budapest |
| Main Topic: | JAKs and STATs in health and disease |

**Chairs:** László Maródi, Zsolt Tulassay

**PROGRAM**

The biology of the JAK/STAT signaling pathway  
*Gabriella Sármay (Budapest, Hungary)*

Deciphering the genetic etiologies of chronic mucocutaneous candidiasis disease (CMCD)  
*Sophie Cypowij (New York, USA)*

Impaired development of IL-17-mediated immunity in patients with STAT1 gain of function mutations  
*Beáta Soltész (Debrecen, Hungary)*

Management of gastrointestinal complications of CMCD  
*Zoltán Péter (Budapest, Hungary)*

**SUMMARY AND CONCLUSION**

Each year the Hungarian Academy of Sciences organizes the „Celebration of Hungarian Science“ (CHS) program series. The Hungarian Society for Immunodeficiencies (HUSID) took the opportunity this year to organize a CHS event in the central building of the Academy with a worldwide known invited speaker, Sophie Cypowij. The event focused on JAKs and STATs in health and disease. The discovery of the JAK/STAT signaling pathway greatly enhanced our understanding of the immunopathology and genetics of human diseases. Sequence variant in STAT1, STAT3 and STAT5 have been described in connection with human diseases. Most recently, STAT1 gain of function mutation was discovered as the most common cause of chronic mucocutaneous candidiasis. This particular subject was discussed by Dr. Cypowij from the group of Prof Jean-Laurent Casanova, Rockefeller University, NYC.

László Maródi
J Project Meetings in Central and Eastern-Europe
2004-2013

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. Kiev, 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 9–10, 2005; Organizer: Larysa Kostyuchenko
12. Oradea, Romania, December 20, 2005; Organizer: Zoltán Ellenes
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: Mikhail Belevtsev
17. Debrecen, Hungary, December 8–9, 2006; Organizer: László Maródi
18. Zaporozhzhye, Ukraine, April 19–20, 2007; Organizer: Liudmila 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: Liudmila Chernyshova
24. Sunny Beach, Bulgaria, May 22–23, 2008; Organizer: Elissaveta Naumova
25. Sarajevo, Bosnia-Herzegovina, October 10–11, 2008; Organizer: Velma Mulaos-manovic
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: Liudmila 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: Natasa Toplak
36. Tehran, Iran, October 11–12, 2009; Organizer: Asghar Aghamohammadi, Nima Rezaei
37. Cairo, Egypt, October 18–19, 2009; Organizer: Nermeen Galal
38. Minsk, Belarus, October 22–23, 2009; Organizer: Mikhail Belevtsev
39. Tirana, Albania, April 8–9, 2010; Organizer: Georgina Kuti-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, Azerbaijan, 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
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, 2011; Organizers: Fazil Orhan, Ismail Reisli
60. Riga, Latvia, November 26, 2011; Organizer: Tatjana Prokofjeva
61. Lillafüred, Hungary, January 20–21, 2012; Organizer: Miklós Szolnoky
63. Rostov-on-Don, Russia, March 16, 2012; Organizer: Ludmila Sizyakina
64. Moscow, Russia, March 29, 2012; Organizer: Irina Kondratenko
65. Chernigov, Ukraine, April 5–6; 2012; Organizer: Liudmyla Chernyshova
66. Tallinn, Estonia, April 19, 2012; Organizer: Krista Ress
67. Tehran, Iran, April 22, 2012; Organizers: Asghar Aghamohammadi, Nima Rezaei
68. Bratislava, Slovakia, April 27, 2012; Organizers: Peter Ciznar
69. Diyarbakir, Turkey; May 12, 2012; Organizer: Ismail Reisli
70. Prague, Czech Republic; May 14–15, 2012; Organizer: Anna Sediva
71. Konya, Turkey; May 26–27, 2012; Organizer: Ismail Reisli
72. Warsaw, Poland; 2012; Organizer: Ewa Bernatowska
73. Sibiu, Romania; June 1–2, 2012; Organizer: Sorin Iurian
74. Tzigov Chark; June 8–9, 2012; Organizer: Elissaveta Naumova
75. Astana City, Kazakhstan, June 12–15, 2012; Organizer: Panteley Popandopulo
76. Tyumen, Russia-Siberia, July 1–2, 2012; Organizer: Irina Tuzankina
77. Ljubljana, Slovenia, September 14–15, 2012; Organizer: Tadej Avcín
78. Tehran, Iran, October 13–14, 2012; Organizer: Nima Rezaei
79. Timisoara, Romania, October 25–26, 2012; Organizer: Mihaela Bataneant
80. Minsk, Belarus, November 17, 2012; Organizer: Mikhail Belevseev
81. Ufa, Russia, November 23, 2012; Organizer: Irina Tuzankina
82. Sanliurfa, Turkey, November 25, 2012; Organizer: Ismail Reisli
83. Krakow, Poland, December 15, 2012; Organizer: Ewa Bernatowska
84. Mukachevo, Ukraine, April 18, 2013; Organizer: Liudmyla Chernyshova
85. Tehran, Iran, April 23, 2013; Organizer: Nima Rezaei
86. Bratislava, Slovakia, April 24, 2013; Organizer: Peter Ciznar
87. Debrecen, Hungary, April 26, 2013; Organizer: László Maródi
88. Ljubljana, Slovenia, May 9, 2013; Organizer: Tadej Avcin
89. Prague, Czech Republic, May 13–14, 2013; Organizer: Anna Sediva
90. Moravia, Czech Republic, May 31, 2013; Organizers: Jiri Litzman, Anna Sediva
91. Budapest, Hungary, May 31–June 1, 2013; Organizer: László Maródi
92. Cracow, Poland, September 20–21, 2013; Organizer: Ewa Bernatowska
93. Szolnok, Hungary, October 5, 2013; Organizer: Zsuzsa Horváth
94. Tehran, Iran, October 12–13, 2013; Organizer: Nima Rezaei
95. Ptuj, Slovenia, October 18–19, 2013; Organizer: Tadej Avcin
96. Krasnoyarsk, Siberia, Russia, October 22, 2013; Organizer: Irina Tuzankina
97. Malatya, Turkey, November 16, 2013; Organizer: Ismail Reisli
98. Budapest, Hungary, November 22, 2013; Organizer: László Maródi

Attila Kondor: Modification of the light (2012)
<table>
<thead>
<tr>
<th>Country</th>
<th>City</th>
<th>Name</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Albania</td>
<td>Tirana</td>
<td>Georgina Kuli-Lito</td>
<td><a href="mailto:gkuli_lito@hotmail.com">gkuli_lito@hotmail.com</a></td>
</tr>
<tr>
<td>Azerbaijan</td>
<td>Baku</td>
<td>Gulnara Nasrullahayeva</td>
<td><a href="mailto:g.nasrullahayeva@hotmail.com">g.nasrullahayeva@hotmail.com</a></td>
</tr>
<tr>
<td>Belarus</td>
<td>Minsk</td>
<td>Mikhail Belevtsev</td>
<td><a href="mailto:belevtsev_m@mail.ru">belevtsev_m@mail.ru</a></td>
</tr>
<tr>
<td>Bosnia-Herzegovina</td>
<td>Sarajevo</td>
<td>Velma Mulaosmanovich</td>
<td><a href="mailto:velmamulaosmanovic@hotmail.com">velmamulaosmanovic@hotmail.com</a></td>
</tr>
<tr>
<td>Bulgaria</td>
<td>Sofia</td>
<td>Elissaveta Naumova</td>
<td><a href="mailto:naumovaej@gmail.com">naumovaej@gmail.com</a></td>
</tr>
<tr>
<td>Croatia</td>
<td>Zagreb</td>
<td>Jadranka Kelečić</td>
<td><a href="mailto:jkelecie17@gmail.com">jkelecie17@gmail.com</a></td>
</tr>
<tr>
<td>Czech Republic</td>
<td>Prague</td>
<td>Anna Sediva</td>
<td><a href="mailto:AnnaSediva@fnmotol.cz">AnnaSediva@fnmotol.cz</a></td>
</tr>
<tr>
<td>Czech Republic</td>
<td>Brno</td>
<td>Jiri Litzman</td>
<td><a href="mailto:jiri.litzman@fnusa.cz">jiri.litzman@fnusa.cz</a></td>
</tr>
<tr>
<td>Egypt</td>
<td>Cairo</td>
<td>Aisha El-Marsafy</td>
<td><a href="mailto:aisha_mars@hotmail.com">aisha_mars@hotmail.com</a></td>
</tr>
<tr>
<td>Estonia</td>
<td>Tallinn</td>
<td>Sirje Velbri</td>
<td><a href="mailto:velbrisir@hotmail.com">velbrisir@hotmail.com</a></td>
</tr>
<tr>
<td>Estonia</td>
<td>Tallinn</td>
<td>Krista Ress</td>
<td><a href="mailto:krista.ress@gmail.com">krista.ress@gmail.com</a></td>
</tr>
<tr>
<td>Hungary</td>
<td>Debrecen</td>
<td>László Maródi</td>
<td><a href="mailto:lmarodi@med.unideb.hu">lmarodi@med.unideb.hu</a></td>
</tr>
<tr>
<td>Hungary</td>
<td>Debrecen</td>
<td>Melinda Erdős</td>
<td><a href="mailto:melinda.erdos@yahoo.com">melinda.erdos@yahoo.com</a></td>
</tr>
<tr>
<td>Hungary</td>
<td>Debrecen</td>
<td>Bea Tóth</td>
<td><a href="mailto:toth.beata.anna@freemail.hu">toth.beata.anna@freemail.hu</a></td>
</tr>
<tr>
<td>Iran</td>
<td>Tehran</td>
<td>Asghar Aghamohammadi</td>
<td><a href="mailto:aghamohammadi@Tums.ac.ir">aghamohammadi@Tums.ac.ir</a></td>
</tr>
<tr>
<td>Iran</td>
<td>Tehran</td>
<td>Nima Rezaei</td>
<td><a href="mailto:rezaei_nima@yahoo.com">rezaei_nima@yahoo.com</a></td>
</tr>
<tr>
<td>Kazakhstan</td>
<td>Astana City</td>
<td>Nurzhan Otarbayev</td>
<td><a href="mailto:otarbaev@rambler.ru">otarbaev@rambler.ru</a></td>
</tr>
<tr>
<td>Latvia</td>
<td>Riga</td>
<td>Tatjana Prokofjeva</td>
<td><a href="mailto:monja@balticom.lv">monja@balticom.lv</a></td>
</tr>
<tr>
<td>Lithuania</td>
<td>Kaunas</td>
<td>Brigita Sitkauskiene</td>
<td><a href="mailto:brigita.sitkauskiene@kaunoklinikos.lt">brigita.sitkauskiene@kaunoklinikos.lt</a></td>
</tr>
<tr>
<td>Poland</td>
<td>Warsaw</td>
<td>Ewa Bernatowska</td>
<td><a href="mailto:bernatowskae@yahoo.com">bernatowskae@yahoo.com</a></td>
</tr>
<tr>
<td>Poland</td>
<td>Warsaw</td>
<td>Malgorzata Pac</td>
<td><a href="mailto:malgorzata.pac@wp.pl">malgorzata.pac@wp.pl</a></td>
</tr>
<tr>
<td>No.</td>
<td>Country</td>
<td>City</td>
<td>Name</td>
</tr>
<tr>
<td>-----</td>
<td>----------------</td>
<td>---------------</td>
<td>-----------------------</td>
</tr>
<tr>
<td>22.</td>
<td>Poland</td>
<td>Warsaw</td>
<td>Malgorzata Skomska</td>
</tr>
<tr>
<td>23.</td>
<td>Poland</td>
<td>Krakow</td>
<td>Anna Szafiarska</td>
</tr>
<tr>
<td>24.</td>
<td>Rep. of Macedonia</td>
<td>Skopje</td>
<td>Kristina Mironska</td>
</tr>
<tr>
<td>25.</td>
<td>Rep. of Moldova</td>
<td>Chisinau</td>
<td>Liudmyla Cerempei</td>
</tr>
<tr>
<td>26.</td>
<td>Romania</td>
<td>Timisoara</td>
<td>Mihaela Bataneant</td>
</tr>
<tr>
<td>27.</td>
<td>Romania</td>
<td>Timisoara</td>
<td>Margit Serban</td>
</tr>
<tr>
<td>28.</td>
<td>Russia</td>
<td>Moscow</td>
<td>Irina Kondratenko</td>
</tr>
<tr>
<td>29.</td>
<td>Russia</td>
<td>St. Petersburg</td>
<td>Marina Guseva</td>
</tr>
<tr>
<td>30.</td>
<td>Russia</td>
<td>Chelyabinsk</td>
<td>Alexandr Pischalnikov</td>
</tr>
<tr>
<td>31.</td>
<td>Russia</td>
<td>Yekaterinburg</td>
<td>Irina Tuzankina</td>
</tr>
<tr>
<td>32.</td>
<td>Serbia</td>
<td>Belgrade</td>
<td>Srdjan Pasic</td>
</tr>
<tr>
<td>33.</td>
<td>Slovakia</td>
<td>Bratislava</td>
<td>Peter Ciznar</td>
</tr>
<tr>
<td>34.</td>
<td>Slovenia</td>
<td>Ljubljana</td>
<td>Tadej Avcin</td>
</tr>
<tr>
<td>35.</td>
<td>Turkey</td>
<td>Konya</td>
<td>Ismail Reisli</td>
</tr>
<tr>
<td>36.</td>
<td>Turkey</td>
<td>Konya</td>
<td>Zafer Caliskaner</td>
</tr>
<tr>
<td>37.</td>
<td>Ukraine</td>
<td>Kyiv</td>
<td>Liudmyla Chernyshova</td>
</tr>
<tr>
<td>38.</td>
<td>Ukraine</td>
<td>Kyiv</td>
<td>Anastasia Bondarenko</td>
</tr>
<tr>
<td>39.</td>
<td>Ukraine</td>
<td>Lviv</td>
<td>Larysa Kostyuchenko</td>
</tr>
<tr>
<td>40.</td>
<td>Ukraine</td>
<td>Kyiv</td>
<td>Alla Volokha</td>
</tr>
</tbody>
</table>
Genetic testing in the
Debrecen PID Reference Center

PRIMARY IMMUNODEFICIENCIES (Genes)
1. X-linked agammaglobulinemia (BTK)
2. Common variable immunodeficiency subtype 1 (ICOS)
3. Common variable immunodeficiency subtype 2 (TACI)
4. X-linked hyper-IgM syndrome (TNSF5=CD40L)
5. Hyper-IgM syndrome (UNG)
6. Activation induced cytidinedeaminase deficiency (AICDA)
7. Recombination-activating gene 1 deficiency (RAG1)
8. Recombination-activating gene 2 deficiency (RAG2)
9. X-linked severe combined immunodeficiency (SCIDX1) (IL-2RG)
10. T-, B+, NK+ severe combined immunodeficiency (IL7RA)
11. X-linked chronic granulomatous disease (CYBB)
12. Autosomal recessive chronic granulomatous disease (NCF1)
13. Wiskott-Aldrich syndrome (WAS)
14. AR-Hyper-IgE syndrome (TYK2)
15. AD-Hyper-IgE syndrome (STAT3)
16. Severe congenital neutropenia type 1 (ELANE)
17. Severe congenital neutropenia type 2 (GFI1)
18. Severe congenital neutropenia type 3 (HAX1)
19. Severe congenital neutropenia type 4 (G6PC3)
20. Hereditary neutrophilia (CSF3R)
21. Shwachman-Diamond syndrome (SBDS)
22. Familial hemophagocytic lymphohistiocytosis syndromes (PRFI)
23. Warts, hypogammaglobulinemia, infections and myelokathexis (WHIM) (CXCR4)
24. Complement component 2 deficiency (C2)
25. X-linked lymphoproliferative syndrome (SH2D1A)
26. X-linked lymphoproliferative syndrome type 2 (XIAP)
27. DOCK8 deficiency (DOCK8)
28. Nijmegen breakage syndrome (NBN)
29. Chronic mucocutaneous candidiasis disease – DC-SIGN (CLEC4L)
30. Chronic mucocutaneous candidiasis disease – dectin-1 (CLEC7A)
31. Chronic mucocutaneous candidiasis disease – dectin-2 (CLEC6A)
32. Chronic mucocutaneous candidiasis disease (MRC1)
33. Chronic mucocutaneous candidiasis disease (CARD9)
34. Chronic mucocutaneous candidiasis disease (STAT1)
35. Chronic mucocutaneous candidiasis disease (IL17RA)
36. Chronic mucocutaneous candidiasis disease (IL17F)
37. Chronic mucocutaneous candidiasis disease (TRAF3IP2)
38. Cartilage hair hypoplasia (RMRP)
39. X-linked Dyskeratosis congenita (DKC1)
40. Autoimmune lymphoproliferative syndrome type 1A (CD95=FASG)
41. Autoimmune lymphoproliferative syndrome type 1B (CD178=FASLG)
42. Autoimmune polyendocrine syndrome (*AIRE*)
43. Hyper-IgD syndrome (*MVK*)
44. IL18R1 deficiency (*IL18R1*)
45. X-linked ectodermal dysplasia, incontinentiapigmenti (*IKBKG*)
46. Familial Mediterranean fever (*MEFV*)
47. Familial Mediterranean fever (*CIASI*)
48. Familial Mediterranean fever (*TNFRSF1A*)
49. Mendelian susceptibility to mycobacterial disease (*IL12RB1*)
50. Mendelian susceptibility to mycobacterial disease (*IL12B*)
51. Mendelian susceptibility to mycobacterial disease (*IFNGR1*)
52. Mendelian susceptibility to mycobacterial disease (*IFNGR2*)
53. MYD88 deficiency (*MYD88*)
54. WIP deficiency (*WIPF1*)
55. Ikaros deficiency (*IKAROS*)
56. X-linked autoimmunity-allergic dysregulation syndrome (IPEX) (*FOXP3*)
57. IRAK4 deficiency (*IRAK4*)

**LYSOSOMAL STORAGE DISEASES (GENES)**
58. Fabry disease (*GLA*)
59. Gaucher disease (*GBA*)
60. Niemann-Pick Disease (*SMPD1*)

**OTHER DISEASES (GENES)**
61. Von Hippel-Lindau Syndrome (*VHL*)
62. X-linked adrenal hypoplasia congenital (*DAX1*)
63. Galetin 3 deficiency (*LGALS3*)
64. Alström syndrome (*ALMS1*)
65. Atopic eczema (*FLG*)
66. Myhre-syndrome (*SMAD4*)