

The J Project

2008



EAST-CENTRAL-EUROPEAN INFECTIOUS AND PEDIATRIC
IMMUNOLOGY CENTRE FOR TRAINING AND RESEARCH

The J Project

2008



Editor: László Maródi

Co-Editors: Lyudmila Cerempei, Lyudmila Chernyshova,
Velma Mulaosmanovic, Elissaveta Naumova, Tatjana Prokofjeva,
Melinda Erdős

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Foreword

The mission of The J Project is to be the forum in East-European countries to increase awareness, and to improve diagnostic facilities and complex management of primary immunodeficiencies.

Since the first J Project meeting in Targu Mures, Romania, on March 11-12, 2004, more and more in the field have been actively working to achieve these goals. 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, conclusions, 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, in June an EU PID consensus conference was organized in the Paul-Erich-Institute, Langen, Germany (pp...). This Meeting was a milestone in the long term awareness campaign on PIDs ESID, IPOPI, INGID, EFIS among the general public. In October, Budapest saw the biannual ESID/IPOPI/INGID meeting, the summary of which was published not only in the ESID newsletter, but also by Nature Immunology (8:323,2007) indicating the unique achievements made by scientists who participated at the Meeting. Further, the first gene therapy trial in patients with Wiskott-Aldrich syndrome was started at the University of Hanover, Germany (pp..) increasing the number of PIDs in which gene therapy was 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 in gene therapy in Wiskott-Aldrich syndrome (pp...). The traditional Prague Spring Meeting was focused in this year on new primary immunodeficiency disorders (pp...). The first J Project meetings in Bucharest and Minsk were organized in June and November, respectively, and attracted a large number of immunologist, infectious disease specialists, hematologists, transplant physicians, and others with an interest in PID from both South-Romania and Belarus. These meetings were primarily physicians' education events, but also 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 Center was decided on. 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 to the Jeffrey Modell Foundation and Biotest Hungary Kft, our major sponsors.

Debrecen, March 15, 2009.

László Maródi



University of Debrecen
Medical and Health Science Center

ECE IPI CTR

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

- Molecular pathology of primary immune deficiency diseases
- Host defence mechanisms against bacteria and fungi
- Mechanisms of action of intravenous immunoglobulins
- 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. 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.



**EAST-CENTRAL-EUROPEAN INFECTIOUS AND PEDIATRIC
IMMUNOLOGY CENTRE FOR TRAINING AND RESEARCH**

The J Project

PID Awareness Meetings in 2008



CITY, Country	DATE
1. ODESSA, Ukraine	10 April, 2008
2. SUNNY BEACH, Bulgaria	22-23 May, 2008
3. SARAJEVO, Bosnia-Herzegovina	10-11 October, 2008
4. CHISINAU, Republic of Moldova	31 October – 1 November, 2008
5. RIGA, Latvia	27-28 November, 2008

Aims

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

Structure of the Meetings

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

Odessa, Ukraine

10 April, 2008



Host:

Liudmyla Chernyshova (chernyshova@ukr.net)

Venue:

Hotel Valentina, Franzuzsky Bulvar, Odessa

Main Topic:

Diagnostics and management of children with primary immunodeficiency diseases



PROGRAM

April 10

Opening ceremony

Welcome from:
Ministry of Health of Ukraine and
Department of Health Care of Odessa

The evolving J Project
Maródi L. (Debrecen)

Medical management of children with
primary immunodeficiency
Chernyshova L. (Kiev)

Characteristics of B-lymphocyte phenotypes
in patients with PIDs
Volokha A. (Kiev)

PIDs with chromosomal instability
Kostyuchenko L. (Lviv)

Coffee break

Combined immunodeficiency
Samarin D. (Kiev)

Diagnostics and treatment of children with
PID in Russia
Prodeus A. (Moscow)

PID patient care in Latvia
Prokofjeva T. (Riga)



Vaccination in immunocompromised
children
Goldstein O. (Moscow)

Hematopoietic stem cell transplantation in
patients with SCID
Balashov D. (Moscow)

PID case presentations
Mirzoyants G. (Odessa)

Dinner

Indications and safety of vaccination in PID
children
Lapiv F. (Kiev)

Management of patients with chronic
granulomatosis disease
Guseva M., Davydova N. (St. Petersburg)

PID diagnostics in Moldova
Cerempei L. (Chishinau)

PID Patients/Parents group session

Closing ceremony



SUMMARY AND CONCLUSIONS

Up to date three J Project seminars were held in Ukraine - in Kiev (2004), in Lvov (2005), and in Zaporozhye (2007). By this way the central, western, and eastern regions of Ukraine were involved into the *J Project* program. The place of 4th *J Project* Meeting in Ukraine was in Odessa.

Odessa – is a large city and seaport on the Black Sea shore, which is situated on the south of Ukraine. The population of this region is about 2 million people. The Odessa region has a low diagnostic rate of PIDs because of the insufficient information level of pediatricians about primary immunodeficiencies.

Connecting to the scientific program the Meeting also included a workshop section where parents of PID children participated. Parents spoke about the problems in PID care through their families' examples. Their speeches have impressed the audience and touched everybody's heart.



The social program included a City Tour and attending of the Odessa Opera Theatre. The sponsors of the event were Sanofi-Pasteur (France) and Biofarma (Ukraine) companies.

Liudmyla Chernyshova



Alla Volokha, ?, Fedir Lapiy, ?, ?



Liudmyla Chernyshova and László Maródi



?, ?, Liudmyla Chernishova, and Alla Volokha

Sunny Beach, Bulgaria

22-23 May, 2008



Host:

Elissaveta Naumova (immun@sun.medun.acad.bg)

Guergana Stoyanova (gal_ps@yahoo.co.uk)

Venue:

Main Topic:



PROGRAM

23 May, Friday

Opening

Naumova E. (Sofia)

The J project – overview

Maródi L. (Debrecen)

ESID Registry

Kindle G. ()

Bulgarian national PID registry – problems needed to be solved

Petrova G. ()

Molecular defects associated with PIDs

Naumova E. (Sofia)

Flowcytometry in the diagnostic evaluation of PID

Baltadjieva D. ()

Molecular and clinical characteristics of patients with Wiskott-Aldrich syndrome in Hungary

Maródi L. (Debrecen)

Case report: IgA deficiency in a 12 year old girl

Stoeva T. ()

Case report: Idiopathic CD4 immunodeficiency – how can we deal with it

Penkova K. ()

Immunoglobulin therapy

Perenovska P. ()

Immunomodulators in immunodeficiency (Ewopharma)

Case report

Baleva M. ()



SUMMARY AND CONCLUSIONS

Elissaveta Naumova

Sarajevo, Bosnia-Herzegovina

October 10-11, 2008



Host:

Velma Mulaosmanovic (velmamulaosmanovic@hotmail.com)

Venue:

Department of Allergology, Rheumatology and Clinical Immunology, Children's Hospital Sarajevo, University Clinical Center/Pediatric Educational Center Sarajevo (PECS)

Main Topic:

Early diagnosis and therapy of primary immunodeficiencies



PROGRAM

10 October, Friday

Early recognition of PID

Opening

Mesihović-Dinarević S. Director of Children's Hospital/Pediatric Educational Center Sarajevo

Introduction

Mulaosmanović V. (Sarajevo)

The J Project: achievements and current concepts

Maródi L. (Debrecen)

Classification and PID research in Europe

Mulaosmanović V. (Sarajevo)

Ten warning signs of PID

Delihanović I. (Sarajevo)

Coffee break

Assesment of PID

Laboratory evaluation of T and B cell deficiencies

Zečević L., Karmehić J. (Sarajevo)

Laboratory evaluation of the phagocyte system

Šarac-Delić M., Drače Z., Karamehić J. (Sarajevo)

Genetic basis and diagnostics of PIDs

Kapur L. (Sarajevo)

Periodic fever syndrome

Toplak N. (Ljubljana)

Risk of malignancy development in patients with PID

Tunić A. (Sarajevo)

Coffee break

Treatment of PID

Bone marrow transplantation and gene therapy for primary immunodeficiency diseases

Seger R. (Zurich)

Use of human immunoglobulin in pediatrics

Dizdarević A. (Sarajevo)

Vaccination of immunodeficient patients

Mulaomerović M. (Sarajevo)

11 October, Saturday

PID syndromes I

Clinical and genetic characteristics of Hemophagocytic Lymphohistiocytosis /Macrophage Activation Syndrome

Avčín T. (Ljubljana)

Clinical and genetic characteristics of Job syndrome

Maródi L. (Debrecen)

The cardiac-immunologic aspect of DiGeorge syndrome

Mesihović-Dinarević S. (Sarajevo)

Severe combined imunodeficiency and RAG deficiency phenotypes

Pašić S. (Belgrade)

Coffee break

PID syndromes II

Immune Dysregulation, Polyendocrinopathy, Enteropathy and X-linked inheritance (IPEX)

Gambineri E. (Firence)

Case report: steps to diagnose autoimmune lymphoproliferative syndrome

Mulaosmanović V. (Sarajevo)

Bruton's agammaglobulinemia – case report

Tanović-Rujanac S., Mulaosmanović V. (Sarajevo)

Chediak-Higashi syndrome: yes or no? – case report

Čengić A., Mulaosmanović V. (Sarajevo)

SUMMARY AND CONCLUSIONS

On October 10-11, 2008, the *J Project* Primary Immunodeficiency Diseases (PID) Awareness Meeting was held in Sarajevo, Bosnia and Herzegovina. The meeting was organized by the recently founded Department of Allergology, Rheumatology and Clinical Immunology of the Children's Hospital, University Clinical Center Sarajevo in collaboration with the East-Central-European Infectious and Paediatric Immunology (ECE-IPI) Center for Training and Research and PID Care in Development Working Party of the European Society for Immunodeficiencies. This was the first time, that we had PID meeting in B&H. The meeting was attended by participants from several countries including Slovenia, Switzerland, Hungary, Serbia and Bosnia and Herzegovina.

The *J Project* Meeting in Sarajevo was entitled like the previous one held in November 2007 in Ljubljana, Slovenia: „Early Diagnosis and Treatment of Primary Immunodeficiency Diseases“. The keynote speaker was Prof. Dr Reinhard Seger from the University Children's Hospital Zürich, Switzerland who presented an excellent overview of current progress in bone marrow transplantation and gene therapy for PID. Prof. L. Maródi (Debrecen, Hungary) gave an interesting overview about the *J Project* achievements and current concepts. In addition, a number of invited and local speakers presented and discussed specific aspects of PID. The official language of the *J Project* Meeting in Sarajevo was English and Bosnian/Croatian/Serbian in order to foster discussions among participants from different countries and local ones.

The scientific program was divided in sessions as follows :1) Early recognition of PID; 2) Assessment of PID, 3) Treatment of PID; 4) PID syndromes I, 5) PID syndromes II. Aims of the meeting were to increase public awareness, to increase and broaden knowledge of PID, to establish professional collaboration with other centers, to facilitate early diagnosis and treatment, to encourage establishment of patient support group and the main aim was

to improve patient care. Desired concept of the meeting was completely fulfilled - collegial discussion in friendly atmosphere about certain and suspected diagnosis of PID patients and best therapy, emphasizing Education, Sociability and Interaction for all. Main problems that we are facing with in B&H, were well and easily defined: professional education (all levels), under diagnosed number of PID patients and low laboratory facilities. In particular, the *J Project* Meeting provided an initiative to establish the B&H multidisciplinary team for PID. Moreover, the *J Project* Meeting in Sarajevo provided an excellent opportunity to promote international collaboration in the field of PID among Central and South-Eastern European countries.



The scientific and social program of the meeting was rated as highly successful and we would like to thank all the participants, especially our distinguished invited speakers, including Prof. L. Maródi (Debrecen), dr. T. Avčin (Ljubljana) and dr. S. Pašić (Serbia) for their contribution. We thank our sponsors (ECE-IPI, Alkaloid, Roche, Unifarm, Medis, Pfizer) for their support. In addition, we would like to thank Prof. S. Mesihović-Dinarević, Director of the Children's Hospital Sarajevo, Prof. L. Maródi, Chair of the ECE-IPI Center for Training and Research for their continuous support and help with the organization of the meeting. Special thanks to dr Tadej Avčin, Children's Hospital Ljubljana for all his support, for making us scientifically impressed with Primary Immunodeficiency Disease world and for sharing knowledge with us.

There is no doubt that *J Project* PID Meeting in Sarajevo was a very important scientific event in the field of the pediatric primary immunodeficiency in B&H in 2008. We sincerely hope that it would contribute to brighter future for patients with PID.

Velma Mulaosmanović

**In front: László Maródi, Velma Mulaosmanovic,
Tadej Avcin, and Reinhard Seger**



Chisinau, **R**ep. of **M**oldova

31 October–1 November, 2008



Host:

Lyudmila Cerempei (lcerempei@rambler.ru)

Venue:

Primary State Medical and Pharmaceutical University Nicolae Testemitanu

Main Topic:

Primary Immunodeficiencies in children: diagnostic and treatment problems



PROGRAM

31 October, Friday

Courses of the Continuous Medical Education

Basics of the immune response

Cerempei L. (Chisinau)

Primary and secondary immunodeficiency

Cerempei L. (Chisinau)

Chromosomal aberration syndromes: ataxia, telangiectasia and the Niimegen-breakage syndrome

Secara V. (Chisinau)

Common variable immunodeficiency

Schitco O. (Chisinau)

Severe combined immunodeficiency

Gorbunov G. (Chisinau)

Actualities in the treatment of children with primary immunodeficiency

Dumitras S. (Chisinau)

Vaccination in special conditions

Cerempei L. (Chisinau)

1 November, Saturday

Welcome speech

Ababii I., Chairman of the SMPPhU “Nicolae Testemitanu”

Tarus M., Head of the Mother and Child Department of the MOH

Possibilities and constrains in the diagnosis of the primary immunodeficiency conditions in Moldova

Stratulat P. (Chisinau)

Modern approach to clinical and immunological manifestations in the hyper-IgE syndrome

Maródi L. (Debrecen)

Case presentation of a patient with hyper-IgE syndrome

Guseva M. (St. Petersburg)

Immune disorders in children with recurrent infections

Cerempei L. (Chisinau)

Long immunological monitoring of the patients with agammaglobulinemia

Kalinina N. (Chisinau)



Coffee break

Clinical cases of children with defective WAS protein expression

Zueva (St. Petersburg)

Immunotherapy in asthma

Sinitchi G. (Iasi)

Secondary immune disorders – background for ecological diseases in children

Vasilos L., Cojocararu A. (Chisinau)

Diagnosis and treatment of children with PID in Lithuania

Procofiieva T. (Riga)

Primary X-linked immunodeficiency

Rugina A. Gotea S., Moraru E., Murgu A., Alexoae M. (Iasi)

Lunch

Bruton's disease – case report

Sciuca S. (Chisinau)

Extrinsic allergic alveolitis

Sinitchi G. (Iasi)

PID cases in Moldova

Sinitin L. (Chisinau)

Cardiovascular disorders in children with primary immunodeficiency

Stamati A. (Chisinau)

Actualization of the PID diagnosis in children in Moldova and National Register for primary immunodeficiency

Cerempei L. (Chisinau)

Discussion, conclusions

SUMMARY AND CONCLUSIONS

The XXVIth Symposium of the international *J Project* program was organized in Chisinau, Republic of Moldova on 31st of October – 1st of November, 2008. The main topic of this symposium was: “Primary Immunodeficiencies in children: diagnostic and treatment problems”.

The meeting was assured by the Ministry of Health of Moldova, and the Chair of Pediatrics and Neonatology of the State Medical and Pharmaceutical University “Nicolae Testemitanu”.

The aim of the symposium was to get family practitioners and pediatricians acknowledged of the problems encountered with the diagnosis and treatment of patients with PIDs in Moldova. The symposium was attended by 345 medical doctors from national and inner city hospitals, and physicians from the primary health care level in rural areas were also participated.

The guest lecturers of this meeting were: Prof. László Maródi, Head of the “J Project”, and Head of the Department of Infectious and

Pediatric Immunology, University of Debrecen (Debrecen, Hungary); Prof. Natalia Kalinina, Head of the Immunology Center of the Ministry for Emergency Situations (St. Petersburg, Russia); Prof. Georgeta Sinitchi, Head of the Department of Immunology and Allergy, and Head of the Department of Family Medicine, Medical and Pharmaceutical University “Gr.T. Popa” (Iasi, Romania); dr. Marina Guseva, Coordinator of the “J Project” (St. Petersburg, Russia); and dr. Tatiana Procofieva, Coordinator of the “J Project” (Riga, Latvia)

Prof. Maródi visited the National Hospital for Children “Emilian Cotaga” where he took part in a discussion, there an interesting case report of a patient with neutropenia was discussed. After the presentation the general recommendations were to examine the bone marrow and to find the possibilities for molecular diagnostic procedure.

Lyudmila Cerempei



Tatjana Prokofjeva, László Maródi, and Lyudmila Cerempei

Latvia, Riga

27-28 November, 2008



Host:

Tatjana Prokofjeva (monja@balticom.lv)

Venue:

VEF Cultural Palace, Riga, Latvia

Main Topic:

Update on Primary Immunodeficiencies



PROGRAM

November 28

The J Project

Maródi L. (Debrecen)

Report on the XIIIth ESID Meeting (16-19 October 2008, 's-Hercogenbosh, the Netherlands).

Prokofieva T. (Riga)

Immunological and genetic diagnostics of primary immunodeficiencies in Belarus

Belevtsev M. (Minsk)

Clinical presentation and immunopathology of hyper-IgE syndrome

Maródi L. (Debrecen)

New therapy technologies in infected and post hypoxic heart pathologies in child

Degtjarova J. (Moscow)

Case report: a patient with hyper-IgE syndrome

Guseva M. (St. Peterburg)

PID: Major clinical manifestations and therapy principles.

Kondratenko I. (Moscow)

Primary antibody deficiencies in Estonia.

Sirje V. (Tallinn)

Overview of PID patient care in Latvia.

Prokofieva T. (Riga)

T-B+NK+ SCID case report

Eglite I. (Riga)

Case report: Generalized BCG-infection in SCID

Skangale A. (Riga)

Diagnostic problems in patients with T-cell deficiency, elevated IgE, reduced IgG, parathyroid deficiency, and cardiomyopathy

Prokofjeva T. (Riga)

Chronic mucocutaneous candidiasis

Shabashova (St. Petersburg)



Inherited heart diseases: frequency, treatment achievements and registration
Ilyin V. (Moscow)

Case report: hyper-IgM1 syndrome.
Lagzdinya D. (Riga)

Common variable immunodeficiency in a girl with exsudative pleuritis and recurrent maxillary sinusitis

Cirule I. (Riga)

Common variable immunodeficiency in a girl with diabetes mellitus

Kirilova I. (Riga)

Case report: chronic granulomatous disease
Prokofieva T. (Riga)

Clinical approach to selective IgA deficiency in children

Purinya S. (Riga)

Case report: Ataxia-telangiectasia
Lindenberga S. (Riga)

T-cell lymphoma in a child with Nijmegen-breakage syndrome

Voitovich I. (Riga)

First registered case of Wiskott-Aldrich syndrome in Latvia

Medne G. (Riga)

Transient hypogammaglobulinemia in infancy

Puchuka Z. (Riga)

General discussion

SUMMARY AND CONCLUSIONS

In November 2008 the first *J Project* meeting was held in Riga, Latvia. The meeting was organized by the Latvian Immunology Society and the Latvian Pediatrician Society.

Latvia is a small country near the Baltic Sea with a population of less than 2.2 million people. Among the 33 members of the Latvian Immunology Society (whom 15 are biologists) only one treats patients with PIDs. Because of the limited capabilities of the laboratories diagnosing PIDs in Latvia, to establish the correct diagnosis is sometimes very difficult. Latvia does not have national PID register and PID professional working groups.

The main aims of the meeting were to improve awareness of PID among the pediatricians and other specialists, and to discuss the diagnostic aspects, clinical symptoms and treatment of PIDs. The meeting was attended by 470 participants including pediatricians, immunologists, neonatologists and infectologists from all regions of Latvia. The guest lecturers of the meeting were prof. L. Maródi (Debrecen), M. Belevtsev (Minsk), prof. N. Shabashova and M. Guseva (St. Petersburg), prof. V. Ilyin and J. Degtjarova (Moscow), prof. L. Cerempei (Chisinau), and V. Sirje (Tallin).

The first session of meeting was focused on the newest informations about the *J Project* program. In the first part an overview of the XIIth ESID Meeting in 's-Hercogenbosh, the Netherlands was also presented. M. Belevtsev from the Belarus Research Center for Pediatric Oncology and Hematology (Minsk) reported on immunological and genetics diagnostic possibilities of PIDs in Belarus. Prof. L. Cerempei (Rep. of Moldova), S. Velbri (Estonia), and T. Prokofjeva also presented the organization of diagnostics and treatment of patients with PID in their countries. Prof. L. Maródi (Debrecen) gave a summary about the clinical presentation and immunopathology of the hyper-IgE syndrome. M. Guseva (St. Petersburg)

presented an interesting case report about a patient with hyper-IgE syndrome.

The second part of the meeting contains a series of presentations from the local speakers about SCID and CVID. Prof. N. Shabashova from the Institute of Medical Mycology, Medical Academy of Postgraduate education (St. Petersburg) gave an interesting lecture about chronic mucocutaneous candidiasis. A case report about a family with hyper-IgM syndrome was also presented. The genetic diagnosis in this family was made in a collaboration with the Debrecen Jeffrey Modell PID Reference Center.

In the third part of the meeting the clinical presentations of a patient with chronic granulomatosis disease, a patient with Nijmegen-breakage syndrome associated with T-cell lymphoma, and presentation of the first registered patient with Wiskott-Aldrich syndrome in Latvia demonstrated our own experiences in this field.

The social events included a visit to the Old Town of Riga and the Lido Recreation Centre. We would like to thank all the participants who contributed to the succes of the first *J Project* meeting in Latvia. We hope that the experiences of this meeting will undoubtedly improve the situation of diagnostics and management of patients with primary immunodeficiency in Latvia.

In summary we think that *J Project* is an essential and important program especially for small countries with limited financial resources. The collaborations with the East-Central-European Infectious and Pediatric Immunology Centre for Training and Research, Debrecen, Hungary and others give us possibilities to improve the level of the diagnostics of primary immunodeficiencies in Latvia. We hope we are looking forward a successful cooperation in the next years.

Tatjana Prokofjeva



?, Marina Guseva, ?, Michael Belevtsev, Tatjana Prokofieva



The Old Town of Riga



J Project Meetings in 2009 N^o 28-40

City (country)	Organizer	Date
28. Timisoara (Romania)	Margit Serban mserban@rdstm.ro	March 19-20
29. Sevastopol (Ukraine)	Liudmyla Chernyshova chernyshova@ukr.net	April 09-10
30. Prague (Czech Republic)	Anna Sediva Anna.Sediva@fnmotol.cz	May 11-12
31. Ekaterinburg (Russia)	Irina Tuzankina I.Tuzankina@iip.uran.ru	May 13-14
32. St. Petersburg (Russia)	Marina Guseva gusevamarina@mail.ru	June 9-10
33. Konya (Turkey)	Ismail Reisli ireisli@hotmail.com	June 25-26
34. Cracow (Poland)	Danuta Kowalczyk dkowalcz@cm-uj.krakow.pl	September 24-25
35. Tallin (Estonia)	Sirje Velbri velbrisir@hotmail.com	October 7-8
36. Ljubljana (Slovenia)	Nataša Toplak natasa.toplak@ardi.si	October 9-10
37. Tehran (Iran)	Asghar Aghamohammadi aghamohammadi@sina.tums.ac.ir and Nima Rezaei rezaei_nima@yahoo.com	October 11-12
38. Cairo (Egypt)	Nermeen Galal nermeengalal@gmail.com	October 18-19
39. Minsk (Belarus)	Michael Belevtsev belevtsev_m@mail.ru	October 22-23
40. Tirana (Albania)	Georgina Kuli-Lito gkuli_lito@hotmail.com	November 19-20

J Project Meetings in East-Europe 2004-2008

1. Targu Mures, Romania, March 11-12, 2004; Organizer: Csilla Todea
 2. Prague, Czech Republic, May 10-11, 2004; Organizer: Anna Sediva
 3. Belgrade, Serbia/ Montenegro, June 11-12, 2004; Organizer: Srdjan Pasic
 4. Skopje, Macedonia, September 17-18, 2004; Organizer: Katarina Stavrik
 5. Kyiv, Ukraine, November 18-19, 2004; Organizer: Alla Volokha
 6. Zakopane, Poland, December 16-17, 2004; Organizer: Ewa Bernatowska
 7. Sofia, Bulgaria, April 15-16, 2005; Organizer: Elissaveta Naumova
 8. Prague, Czech Republic, May 9-10, 2005; Organizer: Anna Sediva
 9. Gdansk, Poland, September 23-24, 2005; Organizer: Ewa Bernatowska
 10. Debrecen, Hungary, November 4-5, 2005; Organizer: László Maródi
 11. Lviv, Ukraine, November 25, 2005; Organizer: Larysa Kostyuchenko
 12. Oradea, Romania, December 20, 2005; Organizer: Zoltan Ellenés
 13. Debrecen, Hungary, January 16, 2006; Organizer: László Maródi
 14. Prague, Czech Republic, May 8-9, 2006; Organizer: Anna Sediva
 15. Bucharest, Romania, June 9-10, 2006; Organizer: Nicolae Iagaru
 16. Minsk, Belarus, November 9-10, 2006; Organizer: Michael Belevtsev
 17. Debrecen, Hungary, December 8-9, 2006; Organizer: László Maródi
 18. Zaporozhzhye, Ukraine, Apr 19-20, 2007; Organizer: Lyudmila Chernishova
 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: Lyudmila Chernishova
 24. Sunny Beach, Bulgaria, May 22-23, 2008; Organizer: Elissaveta Naumova
 25. Sarajevo, Bosnia-Herzegovina, October 10-11, 2008; Organizer: Velma Mulaosmanovic
 26. Chisinau, Rep. of Moldova, Oct. 31-Nov. 1, 2008; Organizer: Lyudmila Cerempei
 27. Riga, Latvia, November 27-28, 2008; Organizer: Tatjana Prokofjeva
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Steering Committee Members

1.	Gene Sulcebe	Albania
2.	Georgina Kuli-Lito	Albania
3.	Velma Mulaosmanovic	Bosnia-Herzegovina
4.	Michael Belevtsev	Belarus
5.	Elissaveta Naumova	Bulgaria
6.	Daniela Baltadijeva	Bulgaria
7.	Guergana Stoyanova	Bulgaria
8.	Darko Richter	Croatia
9.	Anna Sediva	Czech Republic
10.	Jiri Litzman	Czech Republic
11.	Nermeen Galal	Egypt
12.	Sirje Velbri	Estonia
13.	László Maródi	Hungary
14.	Melinda Erdős	Hungary
15.	Miklós Szolnok	Hungary
16.	Asghar Aghamohammadi	Iran
17.	Nima Rezaei	Iran
18.	Sonja Peova	Macedonia
19.	Kristina Mironska	Macedonia
20.	Katarina Stavrik	Macedonia
21.	Lyudmila Cerempei	Rep. of Moldova
22.	Ewa Bernatowska	Poland
23.	Malgorzata Pac	Poland
24.	Anna Szaflarska	Poland
25.	Danuta Kowalczyk	Poland
26.	Margit Serban	Romania
27.	Mihaela Bataneant	Romania
28.	Aurica Rugina	Romania
29.	Ion Ghergina	Romania
30.	Nicolae Iagaru	Romania
31.	Zoltán Ellenés	Romania
32.	Irina Kondratenko	Russia
33.	Marina Guseva	Russia
34.	Andrei Prodeus	Russia
35.	Irina Tuzankina	Russia
36.	Srdjan Pasic	Serbia/Montenegro
37.	Peter Ciznar	Slovakia
38.	Nataša Toplak	Slovenia
39.	Ismail Reisli	Turkey
40.	Lyudmila Chernishova	Ukraine
41.	Larysa Kostyuchenko	Ukraine
42.	Ala Volokha	Ukraine

Debrecen Jeffrey Modell PID Reference Center

Genetic testing of PID genes in 2004-2008

Primary immunodeficiency disorders													
Gene	HU	UK	RO	PO	RU	LT	SER	BU	MAC	SK	SLO	CRO	SUM
1. <i>AICDA</i>	15	2											17
2. <i>AIRE</i>	24												24
3. <i>BTK</i>	31	43	29	13	3		1	1	1		2		124
4. <i>C2G</i>	8												8
5. <i>CYBB</i>	3		3	4		4							14
6. <i>CSF3R</i>	9												9
7. <i>CD40LG</i>	26	2										1	29
8. <i>CXCR4</i>	6		1										7
9. <i>DKC1</i>	1												1
10. <i>ELA2</i>	13												13
11. <i>FASG</i>	1												1
12. <i>FASLG</i>	1												1
13. <i>GFII</i>	9												9
14. <i>HAX1</i>	9												9
15. <i>ICOS</i>	6												6
16. <i>IL2RG</i>	4	6											10
17. <i>MVK</i>	3												3
18. <i>NBS1</i>	1		1			4							6
19. <i>NCF1</i>	-		1										1
20. <i>PRF1</i>	3										1		4
21. <i>RAG1</i>	1					1							2
22. <i>RAG2</i>	1					1							2
23. <i>RMRP</i>	3												3
24. <i>SBDS</i>	7	4					1						12
25. <i>SH2DIA</i>	52	1	3							2			57
26. <i>STAT3</i>	16	1		3	2								22
27. <i>TACI</i>	8												8
28. <i>WASG</i>	41	12	2		12	2		1	2				72
Summary :	302	71	40	20	17	12	2	2	3	3	2	1	475

Genes available for genetic testing in the Debrecen *Jeffrey Modell* PID Reference Center

PRIMER IMMUNODEFICIENCIES (*Genes*)

1. X-linked agammaglobulinemia (*BTK*)
2. X-linked hyper-IgM syndrome (*TNSF5=CD40L*)
3. Activation induced citidine deaminase deficiency (*AICDA*)
4. Common variable immunodeficiency (*TACI*)
5. Common variable immunodeficiency (*ICOS*)
6. Hyper-IgD syndrome (*MVK*)
7. Hyper-IgE syndrome (*STAT3*)
8. X-linked severe combined immunodeficiency (*IL-2RG*)
9. IL7R deficiency (*IL7R*)
10. Rekombinase activator gene 1 deficiency (*RAG1*)
11. Rekombinase activator gene 2 deficiency (*RAG2*)
12. Familiar hemophagocitic lymphohistiocytic syndrome (*PRF1*)
13. X-linked lymphoproliferative syndrome (*SH2D1A*)
14. Autoimmune lymphoproliferative syndrome (*FAS*)
15. Autoimmune lymphoproliferative syndrome (*FASLG*)
16. Autoimmune polyendocrinopathy I syndrome (*AIRE*)
17. Dyskeratosis congenita (*DKC1*)
18. Cartilag-hair hypoplasia (*RMRP*)
19. Nijmegen-breakage syndrome (*NBS1*)
20. Shwachman-Diamond syndrome (*SBDS*)
21. Warts-Hypogammaglobulinemia-Infection-Myelokathexis syndrome /WHIM syndrome (*CXCR4*)
22. Wiskott-Aldrich syndrome (*WAS*)
23. Kostmann syndrome (*HAX1*)
24. Ciclic neutropenia (*ELA2*)
25. Severe congenital neutropenia (*GF11*)
26. Severe congenital neutropenia (*CSF3R*)
27. X-linked chronic granulomatous disease (*CYBB*)
28. Autosomal recessive chronic granulomatous disease (*NCF1=p47phox*)
29. C2 deficiency (*C2*)
30. Hereditary angioedema (*SERPING1*)

LYSOSOMAL STORAGE DISORDERS (*Genes*)

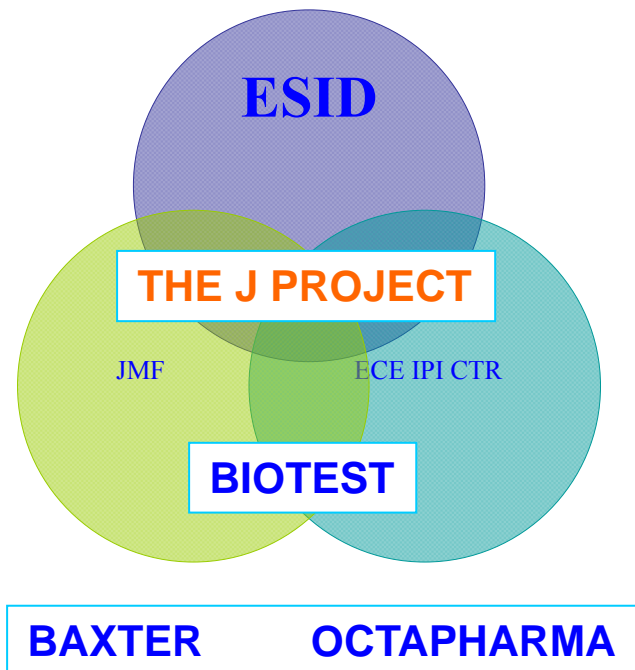
31. Fabry disease (*GLA*)
32. Gaucher disease (*GBA*)
33. Niemann-Pick disease (*SMPD1*)

OTHERS (*Genes*)

34. Von Hippel-Lindau syndrome (*VHL*)
35. X-linked congenital adrenal hypoplasia (*DAX1*)
36. Galectin 3 deficiency (*LGALS3*)

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AN EAST-EUROPEAN PERSPECTIVE



AIM: To increase awareness on PID

