

The J Project

2007



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

University of Debrecen Medical and Health Science Centre

The J Project

2007



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Foreword

Long-term awareness campaigns exemplified by the *J Project* are most beneficial for patients with rare diseases in terms of improving diagnostics, medical treatment, and public attention. The year 2007 witnessed five additional primary immunodeficiency awareness meetings organized in different East-European countries. We first had J Meetings in East-Ukraine (Zaporizhzhya), South-Macedonia (Ohrid), East-Romania (Iasi), and Slovenia (Ljubljana). An important advance of the project was the involvement of Russia in the *J Project* initiative by organizing a meeting jointly by our pediatric immunologist colleagues and friends from St. Petersburg and Moscow.

Over the past four years the *J Project* has grown into a widely appreciated and acknowledged postgraduate physician education program with a clear concept to improve PID patient care in a complex way. A growing number of professionals including pediatric immunologists, infectious diseases experts, and geneticists have become part of the project by active contribution to the success of the meetings, and patient care. Based on activities of previously existing PID centers we have been able to make measurable progress including the emerging molecular genetic laboratories for PID mutational testing in Belarus, Bulgaria, Macedonia, and Romania. With ongoing support by the Jeffrey Modell Foundation and the Biotest Hungary Kft. we have been able to provide molecular genetic testing in Debrecen and help to define the genetic diagnosis of more than 150 patients (page 28).

The J Project has created a network between 25 PID Centers in 18 East-European countries. Analysis of disease causing genes in increasing number of patients has led to establishment of mutational databases which will be used in the near future for joined publication adding new flavor and quality to the professional achievements of the *J Project*.

With all these advances, however, the treatment options of many PID patients in East-Europe are still extremely limited. Availability of high quality intravenous immunoglobulin preparations and bone marrow transplantation centers are desperately needed among other, more specifically targeted immunotherapies. In this regard, one way to step forward is a joined meeting scheduled together with the ambassadors of the *J Project* countries in Hungary, the members of the J Project Steering Committee, and leaders of the Jeffrey Modell Foundation on May 13, 2008, when the officinal dedication of the Debrecen Jeffrey Modell Diagnostic Center for Primary Immunodeficiencies will take place.

The dynamics of the J Project is indicated by the fact that we already have three scheduled meetings for 2009, in addition to the five meetings (N^o 23-28) set for 2008. I am grateful to all who have joined the *J Project* since March 12, 2004, the first meeting in Romania, and have become committed to its aims and goals for the benefit of PID patients.

Debrecen, March 15, 2008.

László Maródi



University of Debrecen Medical and Health Science Centre

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 2007



CITY, Country	DATE
1. ZAPORIZHZHAYA, Ukraine	19-20 April, 2007
2. ST. PETERSBURG, Russia	30 May, 2007
3. OHRID, Macedonia	20-21 September, 2007
4. IASI, Romania	12-13 October, 2007
5. LJUBLJANA, Slovenia	16-17 November, 2007

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.

Zaporizhzhya, Ukraine

19-20 April, 2007



Host:

Lyudmila Chernyshova (chernyshova@ukr.net)

Venue:

National Medical Academy of Postgraduate Education
Department of Pediatric Infectious Diseases and Clinical
Immunology

Main Topic:

Primary immunodeficiencies in children



The J Project Meeting venue

PROGRAM

April 19, Thursday

About the J Project
Maródi L. (Debrecen)

Medical management of children with primary immunodeficiencies (lecture)
Chernyshova LI. (Kyiv)

Organization of medical help to children with PID in Hungary
Maródi L. (Debrecen)

Medical help to children with PID in Latvia
Prokofieva T. (Riga)

Characterization of primary hypogammaglobulinaemias in Ukraine
Volokha À. (Kyiv)

Diagnostics and treatment of children with PID in Belarus
Belevtsev M. (Minsk)

Difficulties in diagnostics of ataxia-telangiectasia
Kostuchenko L. (Lviv)

Finish of plenary session.



Larysa Kostuchenko (Lviv) presenting her talk,
Chairing, Lyudmila Chernyshova, Kyiv

April 20, Friday

The case of X-linked agammaglobulinemia in twins
Ovcharenko L. (Zaporizhzhya)

Experience of regional center for revelation of PID
Pushkarenko L. (Kharkiv)

Wiskott-Aldrich syndrome in children from the same family
Samarin D. (Kyiv)

The consequences of afterward diagnostics of hereditary hypogammaglobulinemia (cases from the same family)
Benko G. (Iv.-Frankivsk)

The case of hyper-IgE syndrome
Sychova ². (Dnipropetrovsk)

Severe combined immunodeficiency (case report)
Bondarenko À., Michaylova Ò. (Kyiv, Kharkiv)

Discussion
Prodeus À. (Moscow)

Coffee break

Flaccid paralysis in patient with antibody deficiency
Lapiy F. (Kyiv)

Management of patients with PID in Kherson region
Vasilenko N. (Kherson)

Vaccinations of patients with PID
Goldshtein A. (Moscow)

Nijmegen-breakage syndrome (case report)
Bobkova Î. (Zaporizhzhya)

Differential diagnostics of the Wiskott-Aldrich syndrome
Boyko Y. (Lviv)

Experience of additional vaccination in patients with PID
Yakimovich S. (Kyiv)

Management of patients with chromosomal instability in Donetsk region
Nikonets L. (Donetsk)

Shwachmann syndrome (case report)
Savchak ². (Lviv)

Discussion

Dinner

Session of parents of children with PID

The reasons of afterward diagnostics of PID in a child
Strelnikova Î . (Kyiv)

Evolution of parent's opinion about treatment of children with PID
Yarmilko N. (Vinnitsa)

Difficulties in substitution therapy in children with adverse reactions to IVIG
Schuklina G. (Zaporizhzhya)

The problems of providing the treatment of child with Nijmegen syndrome in Zaporizhzhya region
Griban (Melitopol)



Speaker, Andrei Prodeus (Moscow);
Chair, Lyudmila Chernyshova (Kyiv)



Discussion of treatment difficulties. Two brothers with XLA. R, the mother of the twins



Leonid Ovcharenko, presenting historical highlights of the Pediatric Department



Leonid Ovcharenko (L) with colleagues from Zaporizhzhya (M), Kyiv (F), and Debrecen (R)

SUMMARY AND CONCLUSIONS

Up to date two *J Project* meetings have been held in Ukraine - in 2004 in Kyiv, and in 2005 in Lviv. By this way, central and western regions of Ukraine were involved. In 2008 the meeting was organized in Zaporizhzhya, in Hotel Khortitsa on the Khortitsa island. The number of participants were about two hundred. For the first time a section for parents of children with primary immunodeficiencies was also organized. Parents talked about their problems through their families' examples. Their speeches were very impressive and touched everybody's heart.

Zaporizhzhya is situated on the east part of Ukraine in a big industrial region. The region's population is 2 million. There are several big cities in this region, like Melitopol, Krivoy Rog, and Dnipropetrovsk. The diagnostic rate of PID in Zaporizhzhya region is low, because of the insufficient information level of pediatricians about primary immunodeficiencies.

Prof. L. Ovcharenko, the head of Department of Pediatrics of Zaporizhzhya Medical Academy leads the pediatric immunology care in Zaporizhzhya region. Under Prof. L. Ovcharenko's initiative several organizations of the Pediatric Immunology Center in Zaporizhzhya were held.



Hosts (L and F) and guests (M and R) at the meeting reception in Hotel Khortitsa

The events of the *J Project* meeting in Zaporizhzhya were the follows. Prof. L. Maródi has consulted two patients with X-linked agammaglobulinemia in Prof. L. Ovcharenko's clinic and then visited the immunologic laboratory of the Pediatric Immunology Center. Prof. L. Maródi, Prof. L. Chernyshova, and representatives of the city's administration acquainted with opportunities of the immunologic laboratory. Unfortunately we had to conclude that the laboratory is poorly equipped by apparatuses and test-systems. The social program include an excursion on the Khortitsa island and the visiting of the Kazak's Glory museum – “Zaporiz'ka sich”.

Alla Volokha



The Kozaks of Zaporizhzhya

St. Petersburg, Russia

20-21 September, 2007



Host:

Marina Guseva (gusevamarina@mail.ru)

Venue:

Anichkov Palace, St. Petersburg

Main Topic:

Diagnostics of primary immunodeficiencies in St. Petersburg and North-West of Russia



The J Project Meeting venue at
Anichkov Palace, St. Petersburg

PROGRAM

30 May, Friday

First Symposium

Congenital disorders of complement system
Prodeus AP. (Moscow)

Disease of the respiratory tract among patients with primary immunodeficiency
Kalinina NM. (St. Petersburg)

Prenatal infections and primary immunodeficiency
Shabalov NP. (St. Petersburg)

Defects of IL-12 and IFN- γ dependent immunity
Galkina EV. (Moscow)

Disorders of IL-12 and IFN- γ dependent immunity among patients with generalized mycobacterium infection
Mushkin AN. (St. Petersburg)

Prenatal diagnostics and medico-genetic consultation of patients with primary immunodeficiency
Pashchenko OE. (Moscow)

Structure of primary immunodeficiency care in Latvia
Prokofieva TÅ. (Riga)

Primary immunodeficiencies with predominant disorders of the antibody synthesis
Guseva MN. (St. Petersburg)

Plenary session

Filatov's disease and the genetics of XLP
Maródi L. (Debrecen)

The principles of therapy of primary immunodeficiencies
Kondratenko IV. (Moscow)

Primary chemokine receptor defect
Erdős M. (Debrecen)

Molecular-genetic inspection of patients with primary immunodeficiencies
Polyakov AV. (Moscow)

Second Symposium

Immunodeficient and immunocompromised child
Jartsev MN. (Moscow)

Complexities in diagnostics and conducting of primary immunodeficiency among adults
Latysheva TV. (Moscow)

Vaccination of children with primary immunodeficiencies
Harit SM. (St. Petersburg)

Questions of the organization of the medical aid giving patients with immunodeficiencies
Sidorenko IV. (Moscow)

Possibilities in diagnostics and treatment of patients with primary immunodeficiencies in Ukraine
Chernyshova LI. (Kyiv)

15 years experience in clinical immunology in versatile children's hospital
Bologov AA. (Moscow)

Congenital immunodeficiencies in practice of versatile children's hospital in St. Petersburg
Hiseva ÅS. (St. Petersburg)

Experiences in allogenic hematopoietic stem cells transplantation in Wiskott-Aldrich syndrome
Staicheva NV. (St. Petersburg)



Andrei Prodeus (Moscow) is presenting

SUMMARY AND CONCLUSIONS

The *J Project* meeting was held for the first time in Russia on the 30th of May 2007. The meeting was organized as part of the XI Russian scientific forum called “Days of immunology” with international participation in St. Petersburg. The “Days of immunology” are annually held in our city, demonstrating the rapid development of fundamental and applicative immunology.

The meeting was very vital for practical diagnostics and management of patients with PID in our region. It was the first pediatric forum where a lot of specialists of our city exchanged experience in management of patients with PID. Our colleagues from Moscow have shared their great experience with the participants. The Forum contributed to establish new professional international relations, open new possibilities of diagnostics and exchange practical experiences. Presentations of clinical cases about the management of patients with PIDs are the best school in this area.

Reports of our colleagues, Professor László Maródi and Melinda Erdős from the University of Debrecen (Hungary) showed latest scientific achievements in molecular diagnostics of XLP and primary chemokine receptor defects. Report of Professor A.V. Polyakov showed possibilities of molecular biology diagnostics of primary immunodeficiencies in Moscow. Reports of our colleagues from RDKB of Moscow were very actual and important for pediatricians and clinical immunologists. Report of Professor I.V. Kondratenko was dedicated to the principles of the therapy of primary immunodeficiencies. The report of our colleagues from Moscow about the 15 year experience at the Department of Clinical Immunology, functioning as a general pediatric hospital was very interesting and useful for the participants. Results of the collaboration between the Department of Clinical Immunology RDKB from Moscow and the Department of Osseous-Articular Tuberculosis of Institute of Phthisiopulmonology from St. Petersburg were

represented in the reports of E.V. Galkina and A.N. Mushkin.

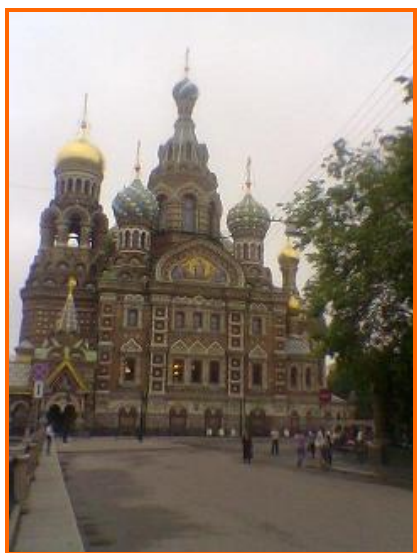
The diagnostic difficulties of primary immunodeficiencies were emphasized in the report of Professor N.P. Shabalov. Diseases of respiratory system among patients with primary immunodeficiency were analyzed in the report of Professor N.M. Kalinina. New directions of practical use of immunologic research methods were represented in report of Professor A.P. Prodeus. Our colleagues from Institute of Immunology (Moscow) represented interesting case reports: Professor M.N. Yartsev talked about Immunodeficient and immunocompromised child, and Professor N.X Setdikova talked about “Primary immunodeficiency in adults: the issues of diagnostics and management”.

The diagnostics and management of patients with primary immunodeficiency in the practice of a general pediatric hospital was represented by Professor E.S. Nisheva. The experiences in the management of patients with primary immunodeficiencies with predominantly antibody defects was showed in the report of M.N. Guseva. Violent discussion concerning children vaccination developed after the report from the Institute of Pediatric Infections (St. Petersburg). The first experiences in allogenic hematopoietic stem cells transplantation in Wiskott-Aldrich syndrome in our city was presented by N.V. Staicheva. Our colleague from Riga shared experience in diagnostics and management of patients with primary immunodeficiencies in Latvia.

Professional experience of colleagues from other regions and countries was investigated during the whole meeting and discussions. The experiences of this meeting will undoubtedly improve the situation of diagnostics and management of patients with primary immunodeficiency in our region.

Marina Guseva

Pictures of St. Petersburg and the dinner



Tatjana Prokofieva (L)
and Marina Guseva



Irina Kondratenko (L),
László Maródi, and
Melinda Erdős



Natalia Kalimina, Areg Totolyan, Melinda Erdős, László Maródi,
Marina Guseva, Elena Galkina, and Irina Kondratenko

Ohrid, Macedonia

20-21 September, 2007



Host:

Kristina Mironska (mironska2002@yahoo.com)

Venue:

Hotel Metropol, Ohrid

Main Topic:

Genetic testing and complex management of patients with primary immunodeficiencies in East-Europe



PROGRAM

20 September, Thursday

Welcome reception

21 September, Friday

Introduction

Peova S., Maródi L

Diagnostic and therapeutic guidelines for PID

Bernatowska E.

Molecular genetic analysis of Hungarian patients with the hyper-IgM syndrome

Maródi L. (Debrecen)

Diagnosis of primary immunodeficiencies in Albania

Sulcebe G. (Tirana)

Laboratory diagnostics of primary immunodeficiencies in Macedonia

Spirovski M. (Skopje)

Flowcytometry assays in the diagnostics of PID

Baltadjieva D.

Early diagnosis of Di George syndrome

Sukarova-Angelovska E.

Macedonian report of primary immunodeficiencies in children

Peova S. (Skopje)

Discussion

Coffee break

Severe combined immunodeficiency in Serbia: clinical presentation, diagnosis and outcome

Pasic S. (Belgrade)

The clinical features of patients with X-linked agammaglobulinemia

Pac M. (Warsaw)

Joint disease in children with X-linked agammaglobulinemia

Kareva L.

Job syndrome

Petrova G. (Skopje)

Wiskott-Aldrich syndrome in a female patient

Mironska K. (Skopje)

Clinical and genetical aspects of chronic granulomatous disease in patients of the Department of Immunology, CMHI, Warsaw
Kurenko-Deptuch M. (Warsaw)

Autoinflammatory disorders

Stavric K. (Skopje)

PNP deficiency

Bataneant M. (Timisoara)



László Maródi is giving his talk



Speakers and Participants



Chairs: Genc Sulcebe (Tirana), László Maródi (Debrecen) and Kristina Mironska (Skopje)

SUMMARY AND CONCLUSIONS

In September 2007, for the second time in Macedonia, a meeting of the *J Project* for PID was held. The meeting was organized as the part of the Fourth Congress of the Pediatricians in Macedonia with international participation to achieve an increase in the number of correctly diagnosed PID patients and obtain greater success in raising the awareness of the existence of primary immunodeficiencies among the general practitioners and specialists. Besides the guests members of the *J Project* from Hungary (L. Maródi), Poland (E. Bernatowska, M. Pac, M. Kurenko-Deptuch), Romania (M. Bataneant), Bulgaria (D. Baltadjieva, G. Petrova), and Serbia (S. Pasic), for the first time, and to our great pleasure, our colleague G. Sulcebe from the University of Tirana, Albania, attended and participated in the event. The auditorium of the Paediatricians' Congress was attended by about 50 participants who enriched the importance of this meeting. The major topic of the meeting was: "Genetic diagnosis and complex management of patients suffering from primary immunodeficiencies in East Europe"

The meeting was organized as a symposium, consisting of two sessions, in which the data of the PID registries, the opportunities of the diagnostic laboratories in the appropriate centers, the therapeutic aspects and problems in the treatment of the patients were reported. Some cases of PID patients with diagnostic problems, needing complex management were also presented. The discussions during the lectures, and the social programs, gave a good opportunity to exchange experiences and informations about the latest diagnostic and therapeutic achievements in the East European region.

At the beginning of the meeting, in front of the entrance of the conference room, different materials, both in textual and in pictorial form, which included pamphlets titled "PID- what you should know and what you can do", posters showing the 10 warning signs of PID were distributed to all



Ewa Bernatowska and László Maródi

the participants. These materials were translated, printed and distributed in all the medical centers and institutions throughout the country) by the Macedonian working team for Primary Immunodeficiency (Peova, Mironska, Kareva, Stavric), due to the support of the Jeffrey Modell Foundation JMF, USA (www.info4pi.org). We have also the possibility to send blood samples from our PID patients to the JM Reference Center in Debrecen for molecular genetic analysis.

The symposium was held in friendly and professional atmosphere, and it was rated as successful and useful. We want to express our deepest appreciation to all the participants of this meeting as well as to our sponsors Alkaloid, Makedonija Lek and Replek Farm.

Kristina Mironska



Coffee break at old town Ohrid

After the Meeting at Lake Ohrid



Iasi, Romania

12-13 October, 2007



Medical Faculty of the Univ. of Iasi

Host:

Aurica Rugina (rugvictor@yahoo.com)

Venue:

The Hall Of The "Sf. Maria" Children Emergency Hospital, Iasi

Main Topic:

Limitations in care of patients with primary imunodeficiencies in East-Romania and Moldavia



The „Sf. Maria” Children Hospital

PROGRAM

12 October, Friday

Welcome reception

Primary and secondary immunodeficiencies

Rugina A. (Iasi)

Intravenous immunoglobulin therapy in pediatric patients

Rugina A. (Iasi)

The role of laser biostimulation in the therapy of an immunodeficient child with chronic arthritis

Ailioaie C. (Iasi)

Update on therapy with monoclonal antibodies

Rugina A. (Iasi)

Immunotherapy in autoimmune diseases

Gotia S. (Iasi)

Vaccination in special situations

Moraru E. (Iasi)

13 October, Saturday

Review reports

The regulatory T cells: the implications of deficiency in the pathogenesis of allergic and autoimmune diseases

Gotia S. (Iasi)

Update on primary immunodeficiencies

Gherghina I. (Bucurest)

Primary immunodeficiency in velocardiofacial syndrome Di George – one of the most common genetic disease

Panzaru M. (Iasi)

The cytokine profile in digestive diseases

Cerempei L. (Chi^oin^ău)

Sarcoidosis – immunological disorders

Sinitchi G. (Iasi)

Hereditary angioedema – a relatively well-defined primary immunodeficiency

Moldovan D. (Targu Mures)

Primary immunodeficiencies and digestive diseases

Moraru E. (Iasi)

Adverse reactions to intravenous immunoglobulins

Iagaru N. (Bucurest)

Clinical and molecular forms of X-linked lymphoproliferative disease

Maródi L. (Debrecen)

Immune disorders in common pediatric diseases

Cerempei L. (Chisinau)

Immunodeficiencies in premature newborns: major infectious risk factors

Bivoleanu A. (Iasi)

Schimke immunoosseous spondyloepiphyseal dysplasia

Brumariu O. (Iasi)

Transient IgA deficiency – possibility of error in celiac disease diagnose in children (a case study)

Cirdei E. (Iasi)

Severe neurodegenerative disease in Wiskott-Aldrich syndrome

Iagaru N. (Bucurest)

Humoral immunity perturbations in malnutrition

Moraru D. (Iasi)

EUROPIDNAS (European Primary Immunodeficiency Program for Newly Associated States) report

Update of the National Primary Immunodeficiency Registry in Romania

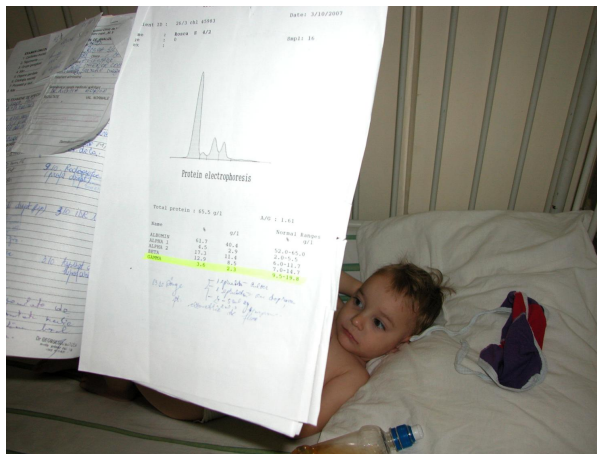
The importance of complete immunologic investigation in tuberculosis in children

Brinza N. (Iasi)

Discussion and conclusions

CASE REPORT FROM IASI

The case of a 2-year-old boy (below) was discussed. The team of the Immunology-Allergy group had treated him for recurrent respiratory tract infections including pneumonias. Serum electrophoresis performed in another hospital showed decreased gamma globulin fraction (see below). On physical examination, the lymph nodes and tonsils were hardly palpable and visible. Blood sample was taken for flow cytometry to be performed in Timisoara (Romania) by Dr. M. Bataneant, and for mutational analysis to be performed in Debrecen (Hungary).



The results showed negligible number of B cells and genetic analysis revealed mutation in the *BTK* gene.

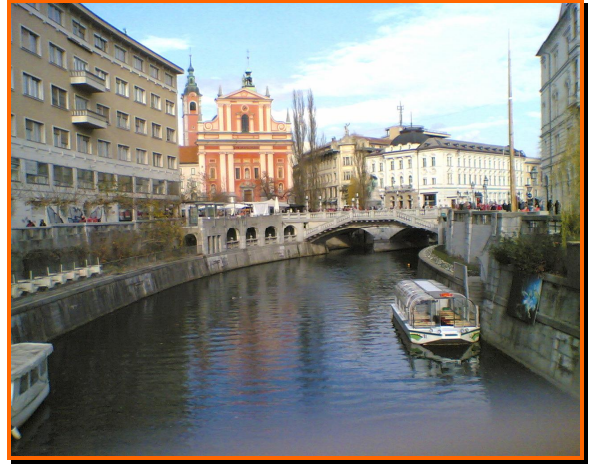
One outcome of the *J Project* meeting in Iasi definitely was the molecular diagnosis of Bruton agammaglobulinemia in this boy in a week after the meeting. This case report is an example of functioning of the *J Project* network in East-Europe. It also indicates the need for molecular genetic testing of PIDs in East-Romania.

Aurica Rugina



Ljubljana, Slovenia

16-17 November, 2007



Host:

Tadej Avcin (tadej.avcin@siol.net)

Venue:

Medical Faculty, University of Ljubljana

Main Topic:

Early diagnosis and treatment of primary immunodeficiency diseases



The University of Ljubljana



Medical Faculty of the UL
(the Meeting venue)

PROGRAM

16 November, Friday

Early recognition of PID

Introduction

Avcin T., Maródi L.

Classification, epidemiology and current perspectives on PID

Avcin T. (Slovenia)

Warning signs of PID in neonates and infants

Nosan G. (Slovenia)

Warning signs of PID in children and young adults: Could it be an immune deficiency?

Vesel T. (Slovenia)

Recurrent fever as a differential diagnostic challenge

Toplak N. (Slovenia)

Oncologic risks in children with PID

Faganel-Kotnik B. (Slovenia)

Primary immunodeficiency diseases in adults

Tomazic J. (Slovenia)

Assessment of primary immune disorders

Laboratory evaluation of T and B cell deficiencies

Ihan A. (Slovenia)

Laboratory evaluation of deficiencies of the phagocytic system

Simcic S. (Slovenia)

Genetically determined deficiencies of the complement system

Kotnik V. (Slovenia)

Molecular genetic diagnostics of PID

Debeljak M. (Slovenia)

New aspects in the pathogenesis of X-linked agammaglobulinemia

Sediva A. (Czech Republic)

Treatment of PID

Antimicrobial therapy and prophylaxis in immunodeficient patients

Mrvic T. (Slovenia)

Vaccination of immunodeficient patient
Zakotnik B. (Slovenia)

Bone marrow transplantation in ADA deficiency: case report and review of the literature

Ciznar P. (Slovakia)

Bone marrow transplantation and gene therapy for PID

Seger R. (Switzerland)

Welcome reception

17 November, Saturday

PID Syndromes I

Severe combined immunodeficiency

Avcin T. (Slovenia)

SCID: a case of a child with Omenn syndrome

Kitanovski L. (Slovenia)

Diagnosis of CVID and recent pathogenetic insights

Wolf H. (Austria)

Common variable immunodeficiency – case presentation

Mulaosmanovic V. (Bosnia and Herzegovina)

Clinical and genetic features of patients with the Wiskott-Aldrich syndrome in Hungary
Maródi L. (Hungary)

Rituximab treatment of autoimmune hemolytic anemia in a child with suspected T cell immunodeficiency

Gagro A. (Croatia)

Ataxia telangiectasia and other immunodeficiencies associated with defects of DNA repair

Praprotnik M. (Slovenia)

Diagnosis and follow-up of pulmonary disease in adult patients with PID

Skrgat-Kristan S. (Slovenia)

PID Syndromes II

X-linked lymphoproliferative disease

Jazbec J. (Slovenia)

Autoimmune lymphoproliferative syndrome

Glavnik V. (Slovenia)

IPEX syndrome – experience from an Italian network

Tommasini A. (Italy)

Clinical and genetic features of patients with APECED

Avbelj M. (Slovenia)

Chronic granulomatous disease – clinical and molecular genetic aspects

Markelj G. (Slovenia)

Long-term follow-up and prognosis of CGD in Serbia

Pasic S. (Serbia)

Disorders of the innate immune system

Zitnik SE. (Slovenia)

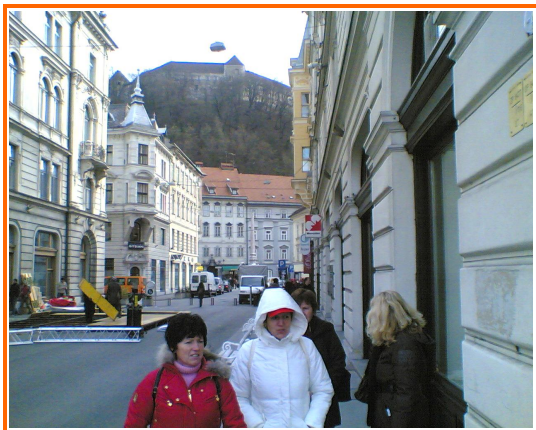
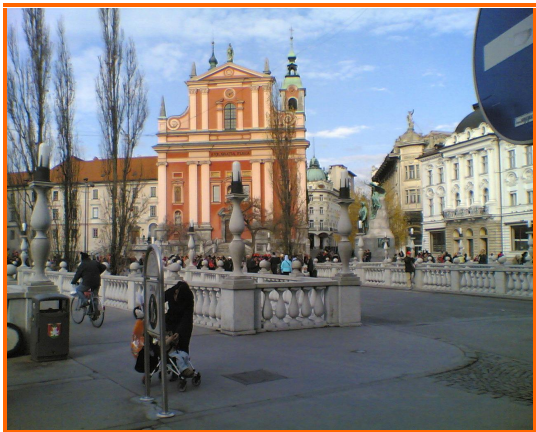
National registry of PID in Slovenia

Koren A. (Slovenia)

The Auditorium of the Medical Faculty



Downtown Ljubljana



People at stike in downtown Ljubljana on November 16, 2007

SUMMARY AND CONCLUSIONS

On November 16-17, 2007, the *J Project* Primary Immunodeficiency Diseases (PID) awareness meeting was held in Ljubljana, Slovenia. The meeting was organized by the Department of Allergology, Rheumatology and Clinical Immunology of the University Children's Hospital, University Medical Center Ljubljana and the Medical Faculty Ljubljana in collaboration with the East-Central-European Infectious and Pediatric Immunology Centre for Training and Research (ECE IPI CTR) and PID Care in Development Working Party of the European Society for Immunodeficiencies. The meeting was attended by more than 100 participants from 10 countries, including Austria, Bosnia and Herzegovina, Croatia, Czech Republic, Hungary, Italy, Serbia, Slovakia, Slovenia and Switzerland.

The *J Project* meeting in Ljubljana was entitled "Early diagnosis and treatment of primary immunodeficiency diseases" with the aim to discuss current diagnostic and therapeutic practices and problems in patients with PID. The keynote speaker was 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. In addition, a number of invited and local speakers presented and discussed specific aspects of PID. We would like to specifically thank our distinguished invited speakers including H. Wolf (Austria), A. Sediva (Czech Republic), L. Maródi (Hungary), A. Tommasini (Italy), S. Pasic (Serbia), and P. Ciznar (Slovakia). The official language of the *J Project* Meeting in Ljubljana was English in order to foster discussions among participants from different countries. Proceedings of the meeting will be published as a special issue of the Slovenian Journal of Pediatrics.

Specific care for patients with PID has been organized at the University Children's Hospital Ljubljana for more than two decades and the *J Project* Meeting in Ljubljana provided an opportunity to further

improve recent protocols for diagnostics and management of patients with PID. In particular, the *J Project* Meeting provided an initiative to establish the Slovenian multidisciplinary team for PID (including physicians of different subspecialties, basic immunologists and geneticists), to improve genetic diagnostics and to update the Slovenian National Registry for PID. Moreover, the *J Project* Meeting in Ljubljana provided an excellent opportunity to promote international collaboration in the field of PID among Central and South-Eastern European countries and to discuss future clinical and research projects.

The scientific and social program of the meeting was rated as highly successful and we would like to thank all the participants for their contributions as well as our sponsors Octapharma, Biotest/Mediline and Altea svetovanje d.o.o. for their generous support. In addition, we would like to thank Prof. Ciril Krzisinik, Director of the University Children's Hospital Ljubljana, Prof. Dusan Suput, Dean of the Medical Faculty Ljubljana, and Prof. László Maródi, Chair of the ECE-IPI CTR for their continuous support and help with the organization of the meeting.

Tadej Avèin



J Project Meetings in 2008

Ukraine
(Odessa)

April 9-10

Lyudmila Chernyshova
chernyshova@ukr.net

Bulgaria
(Sunny Beach)

May 22-23

Elissaveta Naumova
immun@sun.medun.acad.bg
Guergana Stoyanova
gal_ps@yahoo.co.uk

Bosnia-Herzegovina
(Sarajevo)

October 10-11

Velma Mulaosmanovic
velmamulaosmanovic@hotmail.com

Republic of Moldova
(Chisinau)

October 31-
November 1

Lyudmila Cerempei
lcerempei@rambler.ru

Latvia
(Riga)

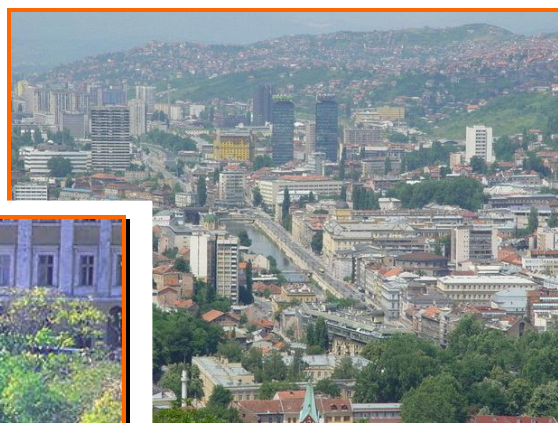
November 27-28

Tatjana Prokofjeva
monja@balticom.lv



Sunny Beach

Odessa



Sarajevo



Chisinau

Riga



J Project Meetings in East-Europe 2004-2007

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
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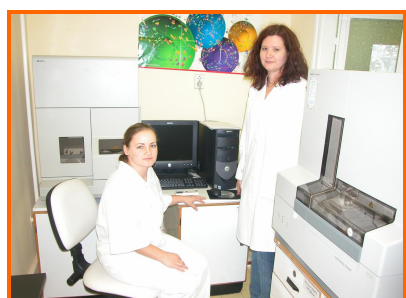
THE J PROJECT

Steering Committee Members

1.	Genc Sulcebe	Albania
2.	Velma Mulaosmanovic	Bosnia-Herzegovina
3.	Michael Belevtsev	Belarus
4.	Elissaveta Naumova	Bulgaria
5.	Daniela Baltadjieva	Bulgaria
6.	Guergana Stoyanova	Bulgaria
7.	Darko Richter	Croatia
8.	Anna Sediva	Czech Republic
9.	Jiri Litzman	Czech Republic
10.	Sirje Velbri	Estonia
11.	László Maródi	Hungary
12.	Melinda Erdős	Hungary
13.	Miklós Szolnoky	Hungary
14.	Kristina Mironska	Macedonia
15.	Katarina Stavrik	Macedonia
16.	Sonja Peova	Macedonia
17.	Lyudmila Cerempei	Moldova
18.	Ewa Bernatowska	Poland
19.	Malgorzata Pac	Poland
20.	Anna Szaflarska	Poland
21.	Margit Serban	Romania
22.	Mihaela Bataneant	Romania
23.	Aurica Rugina	Romania
24.	Ion Ghergina	Romania
25.	Nicolae Iagaru	Romania
26.	Zoltán Ellenés	Romania
27.	Irina Kondratenko	Russia
28.	Marina Guseva	Russia
29.	Andrei Prodeus	Russia
30.	Srdjan Pasic	Serbia/Montenegro
31.	Peter Ciznar	Slovakia
32.	Lyudmila Chernyshova	Ukraine
33.	Larysa Kostyuchenko	Ukraine
34.	Ala Volokha	Ukraine

Mutational analysis of genes at the Debrecen JM Diagnostic Center for PID 2004-2007

Genes	H	UA	RO	PL	RUS	LV	SRG	BG	MK	SK	SLO	HR	SUM
1. <i>AICDA</i>	15	5											20
2. <i>AIRE</i>	11												11
3. <i>BTK</i>	28	43	28	6	2			1	1		2		111
4. <i>C2G</i>	7												7
5. <i>CYBB</i>	4		3	8		4							19
6. <i>CD40LG</i>	23	2										1	26
7. <i>CXCR4</i>	6		1										7
8. <i>DKC1</i>	1												1
9. <i>ELA2</i>	9												9
10. <i>FASG</i>	1												1
11. <i>FASLG</i>	1												1
12. <i>HAX1</i>	9												9
13. <i>ICOS</i>	6												6
14. <i>IL2RG</i>	4	8											12
15. <i>LGALS3</i>	1												1
16. <i>MVK</i>	4												4
17. <i>NBS1</i>	1		1			4							6
18. <i>NCF1</i>			1										1
19. <i>PRF1</i>	3									3			6
20. <i>RAG1</i>	1					1							2
21. <i>RAG2</i>	1					1							2
22. <i>RMRP</i>	3												3
23. <i>SBDS</i>	7	8		3			4						22
24. <i>SH2D1A</i>	48	1	3										52
25. <i>SMPD1</i>	8												8
26. <i>STAT3</i>	11			3	1								15
27. <i>TACI</i>	7												7
28. <i>WAS</i>	29	10	2		12	2		2	2				59
SUM:	249	77	39	20	15	12	4	3	3	3	2	1	428



Molecular Genetic Laboratory,
Deaprt of Infectious and Pediatric
Immunology and JM Diagnostic
Center for PID, Debrecen

Jeffrey Modell Diagnostic Center for PID in Debrecen

PRIMARY IMMUNODEFICIENCY DISEASES (*Genes*)

1. XLA (*BTK*)
2. WAS (*WAS*)
3. XLP (*SH2D1A*)
4. X-CGD (*CYBB*)
5. X-SCID (*IL-2RG*)
6. X-HIGM (*TNSF5*)
7. ALPS (*FAS*)
8. HIGD (*MVK*)
9. HIES (*STAT3*)
10. ALPS (*FASLG*)
11. AR-CGD (*NCF1*)
12. APECED (*AIRE*)
13. C2 deficiency (*C2*)
14. IL7R deficiency (*IL7R*)
15. AID deficiency (*AICDA*)
16. RAG-1 deficiency (*RAG1*)
17. RAG-2 deficiency (*RAG2*)
18. Cyclic neutropenia (*ELA2*)
19. WHIM syndrome (*CXCR4*)
20. Kostmann syndrome (*HAX1*)
21. Dyskeratosis congenita (*DKC1*)
22. Cartilage-hair hypoplasia (*RMRP*)
23. Hereditary angioedema (*SERPINC1*)
24. Nijmegen breakage syndrome (*NBS1*)
25. Shwachman-Diamond syndrome (*SBDS*)
26. Common variable immunodeficiency subtype (*TAC1*)
27. Common variable immunodeficiency subtype (*ICOS*)
28. Familial hemophagocytic lymphohistiocytosis (*PRF1*)

OTHER DISEASES (*Genes*)

1. (*LGALS3*)
2. X-AHC (*DAX1*)
3. Fabry disease (*GLA*)
4. Gaucher disease (*GBA*)
5. Niemann-Pick disease (*SMPD1*)

ESID/IPOPI/INGID Meeting 2008

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St. John's Cathedral









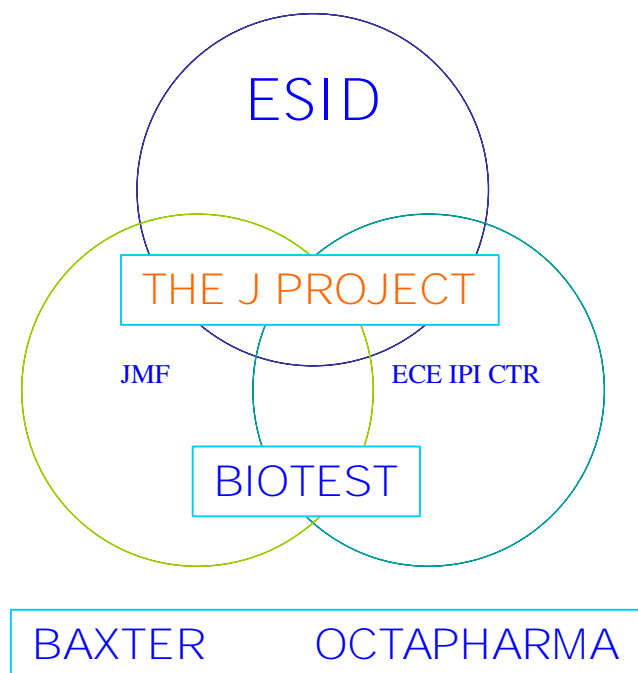
Theater aan de Parade

's-Hertogenbosch
The Netherlands

October 16-19, 2008

THE J PROJECT

AN EAST-EUROPEAN PERSPECTIVE



AIM: To increase awareness on PID