

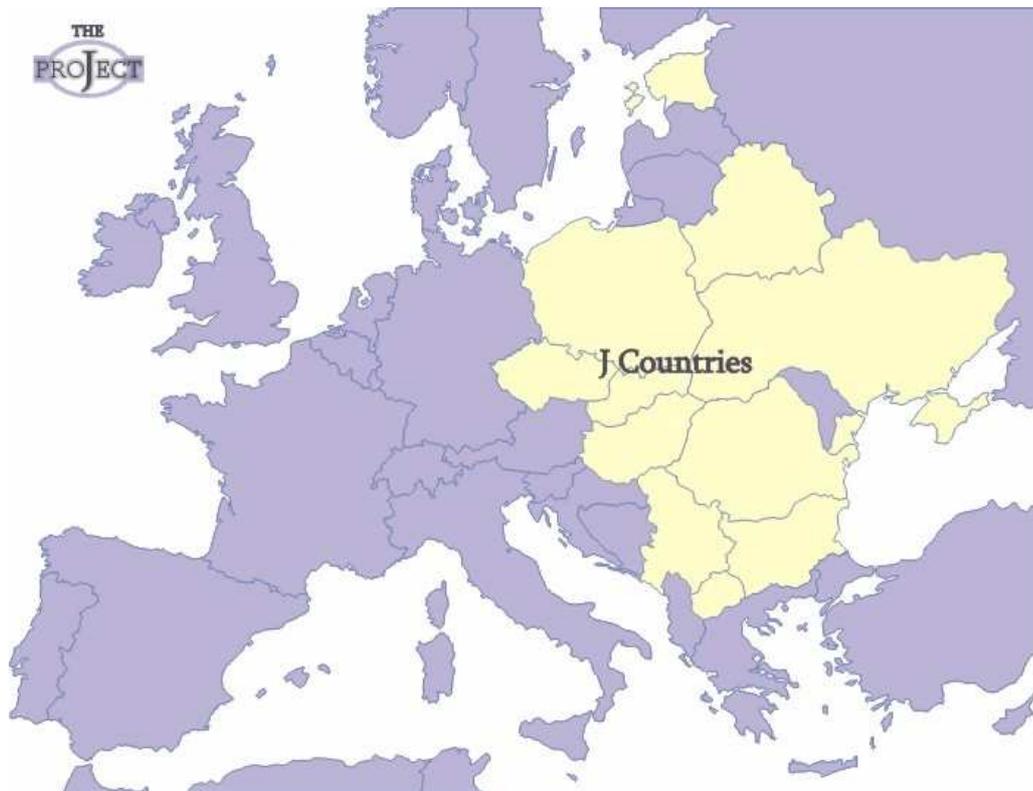
The J Project

2005



**ECE IPI CTR
University of Debrecen
Medical and Health Science Center**

The J Project



Editor: László Maródi

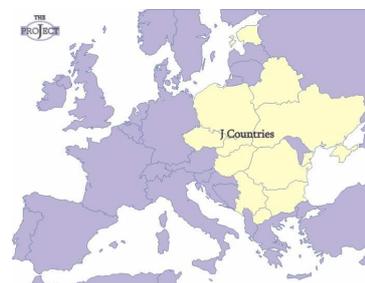
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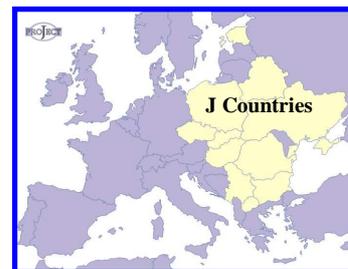
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Foreword



Primary immunodeficiencies (PIDs) are rare diseases that require special medical and psychosocial approaches from doctors and families, and the society. Patients with PIDs are in a special need for particular attention to their problems that are often challenging, especially when it comes to proper diagnosis and early treatment. In contrast, the common experience is that rare diseases, in general, and PIDs, in particular are often pushed to the periphery of medical care. This may be particularly true in countries with lower socio-economic conditions. However, continuing education and awareness campaigns can make the difference. There has always seemed to me to be a need for a concerted action in physician education for a better understanding of the need of PID patients in Eastern Europe. To this end, in March 2004, we launched the **J Project**, a postgraduate meeting series in the region to initiate increased awareness and to stimulate a better PID patient care. These meetings are being organized to attract pediatric immunologists, infectious diseases specialists, hematologists, general practitioners, and others interested in the field. This initiative has become one of the driving forces that may improve PID patient care, and increase awareness on medical, social and psychological aspects of managing PID patients. After successful meetings held in TARGU MURES (Romania), PRAGUE (Czech Republic), BELGRADE (Serbia and Montenegro), SKOPJE (Macedonia), KIEV (Ukraine), and ZAKOPANE (Poland) in 2004, the **J Project** program was continued in SOFIA (Bulgaria), PRAGUE (Czech Republic), GDANSK (Poland), DEBRECEN (Hungary), LVIV (Ukraine), and ORADEA (Romania) in 2005. In addition, we have six meetings scheduled for 2006. During these meetings we have realized that one major difficulty doctors and their patients are experiencing is the lack of a reliable and financially manageable molecular genetic service. Thanks to the University of Debrecen, the Jeffrey Modell Foundation and Biotest Hungary Ltd. we were able to establish the Debrecen Jeffrey Modell Diagnostic Center to provide help in mutational analysis of PID genes in the East Europe. The professional devotion and enthusiasm of our East-European colleagues and the team of the Genetic Laboratory in Debrecen made significant progress over the past year. I hope all of them will continue to find the **J Project** inspiring as it represents the outcomes of steps forward to date since we first met on 12 March, 2004, in Targu Mures, Romania.

Debrecen, March 15, 2006

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.

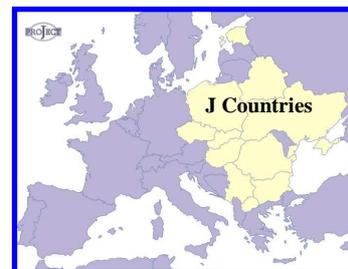
From the conception aspects, ECE IPI CTR is tightly connected with the **Interregional Association of the Carpathian Euroregion**, one of the main aims and functions of which is the formulation of educational and scientific co-operation within the region of East-Central-Europe.



The J Project

PID Awareness Meeting Series in 2005

East Europe



	CITY, Country	Date
1.	SOFIA, Bulgaria	15-16 April, 2005
2.	PRAGUE, Czech Republic	9-10 May, 2005
3.	GDANSK, Poland	23-24 September, 2005
4.	DEBRECEN, Hungary	4-5 November, 2005
5.	LVIV, Ukraine	25 November, 2005
6.	ORADEA, Romania	18 December, 2005

Aims

- 1) To organize professional meetings on PID and related diseases in 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 the night 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.

SOFIA, BULGARIA
15-16 APRIL, 2005



HOST: ELISSAVETA NAUMOVA
(immun@medun.acad.bg)

ORGANIZING COMMITTEE: Ilija Klev
Daniela Baltadjieva
Kalina Penkova
Snejina Mihaylova
Gergana Petrova
László Maródi

VENUE: Medical University of Sofia

MAIN TOPIC:
Recent advances in diagnosis and therapy of PID



PROGRAMME: 16 April

◆ Presentations

An extended J Project initiative
László Maródi (Debrecen)

Primary Immunodeficiency Diseases in Bulgaria
Ilia Kalev (Sofia)

Diagnostic algorithm for immunodeficiency diseases
Elissaveta Naumova (Sofia)

*ESID-registry and upgrade of diagnostic and therapeutic guidelines of
PIDs EURO-POLICY-PID EU grants*
Beata Wolska-Kusnierz (Warsaw)

Immunoglobulin substitution in EC Europe
Alla Volkha (Kiev)

National Registries
Country Representatives

Epidemiology of PID in Romanian children
Margit Serban, Maria Cucuruz, Mihaela Bataneant (Timisoara)

Wiskott-Aldrich Syndrome – Case report
Dora Popova (Sofia)

Temporary IgA deficiency with atypical clinical course
Emil Slavov (Stara Zagora)

Clinical and immunological characteristics of hyper-IgE syndrome
Melinda Erdős, László Maródi (Debrecen)

Late diagnosis of immunodeficiency with thymoma – case report
Krassimira Halacheva (Stara Zagora)

Ataxia telangiectasia – a case report
V. Cvetkova, Svetla Gecheva, Petrunka Petrova, Emiliana Konova (Pleven)

Combined immunodeficiency with defective MHC classII expression
Daniela Baltadjieva, Milena Ivanova, Kalina Penkova, Snežina Mihaylova, A. Perenovska,
Elissaveta Naumova (Sofia)

Hereditary angioedema- clinical picture, diagnostic problems and Bulgarian experience
Petar Krastev, P. Angelova, Maria Simeonova, Ivan Yovchev, Maria Stefanova, Henriette Farkas,
Mariana Murdjeva (Plovdiv)

Clinical manifestation of autoimmunity in children with PID
Mihaela Bataneant, Maria Cucuruz, Margit Serban (Timisoara)

East-Central-European Network for molecular genetics diagnosis of PID

SUMMARY AND CONCLUSIONS:

The Extended J project meeting was held in Sofia, Bulgaria on 16th of April 2005 in the Central Hall of Forum Hotel, situated nearby Medical University Sofia. It has been organized by the Central Laboratory for Clinical Immunology together with the Department of Pediatrics, Medical University Hospital “Alexandrovska”, Sofia. The meeting was attended by more than 50 immunologist and pediatricians from all over the country. Doctors from East-Central Europe attending the meeting were: Prof. László Maródi and Melinda Erdős from Debrecen, Hungary; Beata Wolska-Kusnierz from Warsaw, Poland; Alla Volokha from Kiev, Ukraine; Mihaela Bataneant from Timisoara, Romania; and Prof. Sonja Peova, Katarina Stavric and Lidija Kareva from Skopje, Macedonia. The meeting was organized at the Medical University of Sofia

The main task of the Extended J project meeting in Sofia was to exchange knowledge and experience in PID diagnosis and treatment. Report on the activities of the national registries and a discussion on the establishment of a network for molecular genetic diagnosis of PID was intended.

The first presentation was given by Prof. L. Maródi. He talked on the purpose of the Extended J project and summarized the future tasks of this initiative. An overview on the clinical criteria of PID, differential diagnostic approach in



Bulgaria and their frequencies was presented by Prof. Ilia Kalev. Prof. Elissaveta Naumova presented the diagnostic algorithm used routinely for PID suspected patients in Central Laboratory for Clinical Immunology. ESID – registry, grants, diagnostic and therapeutic guidelines were explained in details by Dr. Beata Wolska. Extensive information on IVIG treatment in EC Europe was given by Alla Volkha. The problems regarding this type of treatment in Bulgaria have been discussed and a necessity of a national policy on IVIG treatment arose.

Topics of the next session were targeted mainly at the PID registries in the countries of the participants. Dr A. Volkha, Dr. B. Wolska, Prof. S. Peova and Dr M. Bataneant gave very detailed presentations proving that the J project initiative works and gives good results

Dr. G. Petrova in a very attractive way presented the first steps of Bulgarian registry and current problems in PID patient registry development in Bulgaria. With enthusiasm she assured the participants that Bulgarian pediatricians will work hard in the near future to achieve the results of their colleagues. And they relay, certainly, on a collaboration with colleagues from the other East- Central European countries as well as on a grant from Jeffrey Modell Foundation.

In the afternoon session there were several very nicely presented case reports with WAS (Dr. D. Popova), AT (Dr. V.



Cvetkova), hyper IgM syndrome (Dr. M. Erdős), IgA deficiency (Dr. E. Savov), MHC class II deficiency (Dr. D. Baltadjieva) and hereditary angioedema (Dr. P. Krastev). Along with, the diagnostic problems and participants' experiences were discussed. Dr M. Bataneant from Romania talked on the clinical manifestation of autoimmunity in children with PID.

At the end of the meeting a concept for the establishment of a network for molecular genetic diagnosis of PID in the region was presented by Dr. D. Baltadjieva. This was supported also by the idea of Prof. L. Maródi who suggested that a meeting should be organized in the near future between representatives of the centers which have already developed molecular-genetic testing for PID: Debrecen is able to detect mutations of 6 PIDs – XLP, HIGM1/2, WAS, SDS, XLA and CLCM, Sofia – of 4 genes involved in MHC class II PID, Zakopanje – of AT and NBS genes. This initiative has been accepted as a good idea for sharing the burden of genetic analysis of PID's between the different countries.

The meeting was closed by Prof. E. Naumova and Prof. L. Maródi, and the guests visited the Central laboratory for Clinical Immunology at the Medical University "Alexandrovska"-Sofia. The Laboratory was founded in 1971. Since 1998 the laboratory has been accredited by the European Federation for Immunogenetics and since 2000 has been chosen for National Reference Laboratory for tissue typing and immunological monitoring by the Bulgarian Ministry of Health. The main directions of diagnostic and research

work are in the field of immunogenetics, transplantation, autoimmunity, allergy, immunodeficiencies and malignancies. The most recent technology in molecular biology and flowcytometry is used in any of the directions. The laboratory is participating in several international and national research projects.



I do believe that the Extended J project meeting in Sofia was a very successful one not only because it contributes to achieve the purpose of the project. As a final step of the meeting a decision for organizing a molecular genetic diagnosis network for PID was taken which will develop in the future under the coordination of Prof. Maródi.

Elissaveta Naumova

SPONSORS: ECE IPI CTR, The Jeffrey Modell Foundation, Biotest Pharma GmbH, Baxter

PRAGUE, CZECH REPUBLIC
9-10 MAY, 2005



HOST: ANNA SEDIVA
(AnnaSediva@fnmotol.cz)

VENUE: University Hospital Motol

MAIN TOPIC:
Prague Spring Meeting of PIDs



PROGRAMME: 9 May

◆ Presentations

Welcome speech

Anna Šedivá, Jiřina Bartůňková

Short film – Second Medical Faculty, Charles University, Czech Republic and Prague

Mycobacterial infections:

From idiopathic infectious diseases to novel primary immunodeficiencies

Jean Laurent Casanova (France)

BCG vaccination in SCID

Petr Sedláček (Prague)

National registries and patients' organizations:

ESID Register of PID

Bodo Grimbacher (Germany)

Upgrade of diagnostic and therapeutic guidelines of PID's - EURO - POLICY PID EU

Malgorzata Pac (Poland)

Patient organisation

IPOPI – the International Patient Organisation for Primary Immunodeficiencies

Eva Soergel (Germany)

Patients in the Czech Republic

Radana Zachová (Prague)

Immunodeficiency and autoimmunity

APECED: monogenic autoimmune disease" or just "APECED and AIRE

Part Peterson (Finland)

APECED – endocrinology

Jan Lebl (Prague)

APECED – immunology

Anna Šedivá (Prague)

Raffaele Badolato (Italy)

PID - case reports

Hematopoietic stem cell transplantation in children with severe combined immunodeficiency

Tuba Turul (Turkey)

Donor lymphocyte infusion as a treatment of a complete DiGeorge syndrome

Kateřina Zdráhalová, Ester Mejstříková (Prague)

Diagnosis and the treatment of FHL – first patient with complete perforin deficiency diagnosed in the Czech Republic

Renata Formánková, Ester Mejstříková (Prague)

Tour through the Institute of Immunology, Department of Pediatric Hematology and Oncology and Institute of Hematology and Blood Transfusion

10 May

Laboratory Methods

EXBIO – company presentation, news

Miloslav Suchánek (Prague)

Is there any genotype-phenotype relationship in patients with hereditary angioedema?

Tomáš Freiburger (Brno)

Applications for a rapid method of diagnosis using DHPLC system

Gigliola Di Matteo (Italy)

Detection of four herpesviruses in HLH patient after haploidentical HSCT

Petr Hubáček (Prague)

The Czech National Database of PID

Vojtěch Thon (Brno)

Round table: availability of laboratory tests of PID in participating countries/centres

Co-ordination: Andrea Poloučková (Prague)

Antibody deficiencies

Genetic cause of hypogammaglobulinemia

Bodo Grimbacher (Germany)

Potential classification of CVID patients based on B cell phenotypes

Berne Ferry (UK)

Illustrative cases of patients with CVID

Aleš Janda (Prague)

CVID and ITP – case reports

Marta Sobotková (Prague)

Asymptomatic hypogammaglobulinemic patients – do we need immunoglobulins?

Jiří Litzmann, Marcela Vlková, Mária Šárfyová (Brno)

News, case reports

XLA – contiguous deletion syndrome

Anna Šedivá (Prague)

Dendritic cells in BTK

Klára Sochorová (Prague)

Mucocutaneous candidosis

Pavel Rozsival, Eva Pařízková, D. Vokurková (Hradec Králové)

Agammaglobulinemia. Case report

Rasa Duobiene (Lithuania)

Immunological alterations in a cohort of patients with 22q11 syndrome

Caterina Cancrini (Italy)

Closing speech ESID Prague Spring Meeting 2005, May the 9th – 10th

Anna Šedivá

SUMMARY AND CONCLUSIONS:

On May 9 and 10, 2005, the fourth ESID Prague Spring meeting was held in the University Hospital Motol, Prague, Czech Republic. This year 32 participants from 10 countries, namely Czech Republic, Estonia, France, Germany, Italy, Lithuania, Poland, Slovakia, Turkey and UK, attended and actively participated in the event.

The main task of Prague ESID meeting is traditionally devoted to the exchange of information on PID between Western and Eastern Europe. This task was accomplished this year by an excellent attendance from both parts of the European continent. Invited participating speakers for 2005 were Jean Laurent Casanova, Bodo Grimbacher, Rafaele Badolato and Part Peterson.

The first day was devoted to introductory invited lectures. Exceptionally interesting topics on the interaction between infection and immunity were discussed. Complexity of such interactions underline the importance of research targeted at pathogenesis of PID. Traditionally the next panel was devoted to registries of PID. Participation of representatives from 10 European countries clearly demonstrated the need for joined activities including registry and was beneficial for further development of such concerted actions. Lectures in the afternoon panel covered the area between immunodeficiency and autoimmunity with an effort of multidisciplinary approach to autoimmune polyglandular syndrom. This entity that opens new aspects of the establishment of immune tolerance was presented from the point of view of molecular biologists,

endocrinologists, pediatricians and immunologists. Laboratory methods and antibody deficiencies were on program on the second day of the meeting. Substantial part of the program was devoted to the case reports. Unusual and interesting cases were the most valued among all participants.

Complementary activities form traditionally important and interesting part of Prague Spring ESID meeting: Concert in Prague Castle and walks through City of Prague, located in the heart of Europe, reminded participants of one unified European area, particularly in 2005, the year of 60th anniversary of the end of Second World War.

The meeting was supported by Charles University, Second Medical Faculty, Prague, further by University hospital in Motol, Prague, by Research project 0021620812 of Ministry of Education, Czech Republic, EU 6th Framework Program Allostem LSHB-CT-2004-503319 and by companies Grifols, Exbio and Immunotech.

The meeting was also organized on the occasion of the Day of Immunology, declared by EFIS on 29th April, 2005 and it was a part of the J Project Programme.

This year's meeting confirmed still growing scientific quality of the conference and the usefulness of these educational activities. The tradition will continue on May 8 and 9, 2006.

Anna Sediva

G DANSK, POLAND
23-24 SEPTEMBER, 2005



HOST: EWA BERNATOWSKA (Immuno@czd.waw.pl)

MAIN TOPIC:
Progress in Education in Clinical Immunology

SUMMARY AND CONCLUSIONS:

The third Autumn – Winter School of Clinical Immunology was organized in Gdansk- the town lying on the Bay of Gdansk and the southern coast of the Baltic Sea. Gdansk was first mentioned as a Polish city in 997. Under the rule of Polish Crown the city became a wealthy member of the Hanseatic League. The meeting took place close to the Old Town, which lies on the banks of the Motława River.

Now, about the meeting, which was the first one organized as a part of a new EU project: ‘Policy oriented and harmonizing research activities in the field of primary immunodeficiency diseases’- EURO-POLICY-PID. The initiator and coordinator of this project is C. I. Edward Smith, Carolinska Institute, the leader of the former EU project EURO-PID-NAS. The objectives of the new project are to build up the necessary research capacity at the European level in order to: increase the understanding of PIDs etiology and identify novel mutations, to carry out epidemiological studies in order to determine the prevalence of PIDs in Europe, to establish harmonized guidelines for diagnosis and treatment of PIDs and to improve the awareness of PID among the European population.

The first session of the meeting included the state of the art of the project.

Anna Berglöf - the project manager from Carolinska Institute, presented the objectives, deliverables, milestones and expected results of that project and the description of work to accomplish by individual partners. Her presentation was followed by remarks of other partners involved in that project; they presented the updated results of their activities. Next, Bodo Grimbacher from Freiburg, gave a lecture on PanEuropean PID patients registry, which is successfully growing up and has already gathered 1 084 patients from the whole Europe. The biggest number comes from Italy, Turkey and Poland. Mauno Vihinen, from Helsinki, presented PID mutation databases existing on <http://bioinf.uta.fi> and now connected with www.esid-registry.org. Helen Chapel, from Oxford, shared with UK experiences concerning the increase of awareness about PID in national public health groups, government and non-government organizations. Esther de Vries presented diagnostic protocol for suspected immunodeficiency as the statement from the Clinical Working Party of ESID. This manuscript will be published soon. The national Polish project with the aim to improve patients’ care and to raise the awareness on PID across the country was

presented by Ewa Bernatowska, from Warsaw.

An excellent review on advances in molecular diagnosis and clinical correlation of PID was the topic of the next lecturers. C. I. Edward Smith presented the genotype-phenotype correlation in X-linked agammaglobulinaemia. Recent advances in diagnosis and classification of CVID were presented by Bodo Grimbacher. Christine Kinnon spoke about updated genetic diagnostics in PIDs. Krystyna Chrzanowska gave a talk on Nijmegen breakage syndrome (NBS) linking DNA damage with immunodeficiency and cancer predisposition, basing on 90 NBS patients already registered in Poland. Wide spectrum of HIGM syndromes was shown by Anna Durandy. Allison Jones shared her experience on long-term follow-up of different PIDs after bone marrow transplantation. Amos Etzioni gave an excellent and practical review on etiology and clinical aspects of agranulocytosis.

After a long and busy day, altogether - speakers and participants enjoyed themselves listening to classical music and eating Polish food. Later in the evening, the international group consisted of some participants spontaneously initiated singing songs in a tavern located in the Old Town of Gdansk.

The next day of the meeting was

created as a case report session, with Teresa Español and Amos Etzioni as chairmen. Some interesting PID cases and patients with unusual features suspected to PIDs were presented by young immunologists from Central Easter Europe.

More than sixty participants attended this meeting. The list of young immunologists is still growing. For the first time, young colleagues from new EU countries, such as Lithuania, Estonia and Latvia came to the meeting. The group of 29 young Polish physicians specializing in clinical immunology was also present.

The 4th School of Clinical Immunology was sponsored by two EU grants: EURO-POLICY-PID SP23-CT-2005-006411 and PERFECT QLG1-CT-2002-90358. The next meeting will be organized in Ruciane Nida – a little village situated in the very heart of the Mazurian Lake District, among the forests, in the middle of the Old Continent, in the Northern - Eastern part of Poland. The symposium will be held from 9 to 10 of June, 2006. For more information please contact us at immuno@czd.waw.pl.

Ewa Bernatowska



DEBRECEN, HUNGARY
4-5 NOVEMBER, 2005



HOST: LÁSZLÓ MARÓDI (lmarodi@jaguar.dote.hu)

ORGANIZING COMMITTEE: Melinda Erdős
Gabriella Kürti
Melinda Szabó

VENUE: Civis Grand Hotel Aranybika

MAIN TOPIC:

Genetic diagnostics and management of primary immunodeficiency disorders in East-Central-Europe



L-R: Zoltán Ellenes, Maria Cucuruz,
László Maródi



L-R: Henriette Farkas, Beáta
Dérfalvy



L-R: László Karmazsin, Dezső
Schuler, founders of the
HWGPI

PROGRAMME: 4 November

◆ Presentations

ORPHANET: An European database for rare diseases
György Kosztolányi (Pécs)

Rules and exceptions in molecular PID diagnostics
Klaus Schwarz (Ulm)

Molecular genetic diagnostics of PID in the Czech Republic
Thomas Freiberger (Brno)

New disease-causing mutations in Shwachman-Diamond syndrome
Melinda Erdős (Debrecen)

Prenatal diagnostic options in PID
Olga Török (Debrecen)

The HAENETWORK project
Henriette Farkas (Budapest)

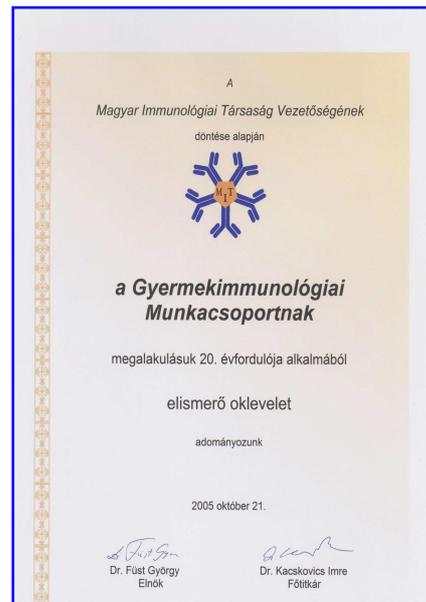
Genetic investigation - decision making factors for diagnosis of PID
Mihaela Bataneant (Timisoara)

WASP mutations in five Hungarian families with WAS
Vera Gulácsy (Debrecen)

SH2D1A mutations in three Hungarian families with XLP
Andrea Lakatos (Debrecen)

Pulmonary diseases in primary antibody deficiencies
Alla Volokha (Kiev)





Certificates of Merit



Gergely Kriván, Olga Török

5 November

Kóros apoptosis és immunhiány betegségek
Réka Tóth (Debrecen)

A szelektív IgA hiány immunpatológiája
Zoltán Ellenés (Oradea)

Össejt immunrekonstitúció
Kálmán Nagy (Miskolc)

A LAD klinikuma és terápiája
Gergely Kriván (Budapest)

A biológiai terápia perspektívái gyermekkori krónikus arthritisben
Zoltán Szekanez (Debrecen)

IVIG szubsztitúció – 2005
Gabriella Vértesi (Miskolc)

Makrofág aktivációs szindróma juvenilis idiopathiás arthritisben
Ildikó Csürke (Nyíregyháza)

Elkülönítő diagnosztikai nehézségek gyermekkori coxitisben
Katalin Fazekas (Debrecen)

Abscessus hepatis krónikus granulomás betegségben
Éva Rákóczi (Debrecen)

Mycobacterium fertőzés Hyper-IgE szindrómában
Beáta Dérfalvi (Budapest)

X-kromoszómához kötött súlyos kombinált immundefektus
Ágnes Vojcek (Pécs)



Alla Volokha



Michaela Baetenant



Melinda Erdős



Thomas Freiburger



László Szabó



Imre Kacs Kovics



Pál Gergely

SPEAKERS
(above)
and
GUESTS
(left)

SUMMARY AND CONCLUSIONS:

The J Project meeting in Debrecen was jointly organized by the Department of Infectious and Pediatric Immunology of the University of Debrecen Medical and Health Science Center and the Hungarian Working Group for Pediatric Immunology (HWGPI) established in 1995. This Meeting provided the possibility to celebrate the 20th anniversary of the establishment of the HWGPI. In addition, the Meeting was part of a series of scientific symposia organized all over Hungary under the heading of Hungarian Science Day. The venue was the Civis Grand Hotel Aranybika, the most traditional hotel in Debrecen.

The Meeting was opened by Dr. László Szabó, secretary of the Hungarian Society of Pediatricians, who honored the HWGPI with a Certificate of Merit on behalf of the Society. Similarly, Dr. Imre Kacsokovics presented a Certificate of Merit to the HWGPI on behalf of the Board of the Hungarian Society for Immunology. Dr. Pál Gergely, science director of the University of Debrecen Medical and Health Science Center expressed his appreciation to the successful activity and achievements of the HWGPI over the past twenty years.



L-R: Klaus Schwarz, György Kosztolányi

A special talk was delivered by Dr. György Kosztolányi, member of the Hungarian Academy of Sciences, on a European database for rare diseases. He invited experts from J Project countries to join this program also referred to as

ORPHANET. Another network project developed for the benefit for patients with C1-esterase inhibitor deficiency was presented by Dr. Henriette Farkas. Dr. Maria Bataneant and Dr. Alla Volokha from Romania and Ukraine, respectively, presented data on autoimmune lymphoproliferative diseases and pulmonary manifestations in patients with PIDs. Two presentations were focused on molecular genetic aspects of PID diagnostics, one by Dr. Karl Schwartz and another by Dr. Thomas Freiburger. Case reports were presented by Dr. Melinda Erdős, Vera Gulácsy, and Andrea Lakatos, all from Debrecen, in which molecular genetic analysis of Shwachman-Diamond syndrome, Wiskott-Aldrich syndrome, and X-linked lymphoproliferative syndrome were shown. Dr. Olga Török gave a very nice talk on methods used in fetal sampling for prenatal diagnosis in PID. At the end of the first day of the program a panel discussion moderated Dr. László Maródi was devoted to discuss technical details of how the Debrecen Jeffrey Modell diagnostic Center can be of help to provide molecular genetic diagnosis for patients in East-Central Europe.

The main subjects of the second day of



L-R: Gyula Szegedi, Kálmán Nagy

the Meeting included talks on apoptosis in PIDs, advances in understanding immunopathology of selective IgA deficiency, and updates of immune-reconstitution and IVIG substitution. A clinical summary of LAD deficiency was

given by Dr. Gergely Kriván. Dr. Zoltán Szekanecz, and adult rheumatologists gave a comprehensive talk on perspectives of biologic therapy in rheumatic disease.

The final session was devoted to case reports on macrophage activation

syndrome, recurrent hepatic abscess as the sole manifestation of CGD, mycobacterial infections in HIGE, and X-linked SCID.

László Maródi



← Gusztáv Magócs,
Éva Rákóczi,
↓ Vera Gulácsy



L-R: Gabi Lakos, Szilvi Taskó, Gabi Kovács



L-R: Andrea Lakatos, Krisztina Alapi



Bernadett Mosdósi, Zoltán Nyul

SPONSORS: Biotest Hungary, Octapharma Hungary, Human RT, Civis Grand Hotel Aranybika, PharmaCenter Hungary Kft, Genzyme CEE GmbH, Baxter Hungary Kft, Hungarian Foundation for Children with Immunodeficiencies, East-Central-European Infectology and Pediatric Immunology Center, Jeffrey Modell PID Center in Debrecen, University of Debrecen, Medical and Health Science Center.

L VIV, UKRAINE
25 NOVEMBER, 2005



HOST: LARYSA KOSTYUCHENKO (lkostyuchenko@ukr.net)

ORGANIZING COMMITTEE: Jarema Volznyiysia (Lviv)
Ludmyla Chernyshova (Kiev)
László Maródi (Debrecen)
Oleksandr Myndyuk (Lviv)
Valentyna Chopyak (Lviv)

VENUE: Hotel “Suputnyk”, Lviv

MAIN TOPIC:

Diagnosis and Treatment of PID



The group from Kiev



L-R: László Maródi, Malgorzata Pac,
Fedir Lapij



Ludmyla Chernyshova

PROGRAMME: 25 November

◆ Presentations

Primary immunodeficiency patient care and treatment in East-Central Europe
László Maródi (Hungary)

IVIG substitution in patients with primary immune deficiencies in Ukraine
Lyudmila Chernyshova (Kiev)

Treatment of primary immune deficiencies in adults in conditions of Ukraine
Valentyna Chopjak (Lviv)

Clinical and diagnostic aspects of Di-George syndrome
Malgorzata Pac (Warsaw)

Clinical manifestations and molecular genetics of hyper-IgM syndrome
László Maródi (Hungary)

Immunodeficiencies in patients with syndromes of chromosomal instability
Larysa Kostyuchenko (Lviv)

Current situation and future trends in diagnostics of syndromes of chromosomal instability
Gajane Akopjan (Lviv)

WASP mutations in Hungarian patients with the Wiskott-Aldrich syndrome
Vera Gulácsy (Hungary)

SH2D1A mutations in three Hungarian families with XLP
Andrea Lakatos (Hungary)

Chronic muco-cutaneous candidiasis
Oleksandr Pryluckyj (Donetsk)

IL-12 receptor β 1-defect in patient with BCG-infection and chronic Salmonella infection
Yaryna Boyko (Lviv)

Nezelof syndrome
Dmitriy Samarin (Kiev)

Severe combined immunodeficiency- way to the diagnosis
Yaryna Romanyshyn (Lviv)

Immunochemiluminescence analysis in evaluation of patients with PID
Andrij Petruh (Lviv)

SUMMARY AND CONCLUSIONS:

The 11th event of the PID Awareness Meeting Series in East Europe "Diagnosis and Treatment of Primary Immunodeficiency Diseases" took place in Lviv, Ukraine, on November 25th, 2005. It was organized in the form of school-seminar with international participation. This J Project meeting in Lviv attracted 137 participants, including 4 international guest-speakers: 46 participants from different regions of Ukraine and abroad and 88 participants from Lviv (among them representatives of Danylo Halyt'skyj Lviv National Medical University and 2 Lviv Scientific-Research Institutes).

This meeting was not only a chance for the Ukrainian doctors and scientists to get a profound and up to date information about the PID, their diagnostic and treatment procedures, but also to learn about the work of Western Ukrainian Center of Pediatric Immunology that already functions for 10 years on the base of Lviv Regional Specialized Children's Clinic. The PID registry of the Center includes about 90 children with PID. It was the first and only institution in Ukraine that started to use using cytogenetic and molecular-genetic methods for diagnosis of PIDs, in particular in NBS and Ataxia-Teleangiectasia (AT).

The opening speech was presented by Prof. László Maródi, in which he has informed the audience about the concept and the work of "J-project".

Prof. Lyudmila Chernyshova, the chief children's immunologist of Ukraine pointed out the current problems of substitutional therapy, especially the fact that not all the children are receiving the needed IVIG in time.

The report on the treatment of adults with PID was presented by chief immunologist of Ukraine V. Chopjak.

The greatest interest of the audience was drawn to the presentations of the guest-speakers from abroad: M. Pac, L. Maródi, V. Gulácsy and A. Lakatos. They fully and precisely described the problems of diagnosis and treatment of different PIDs including hyper-IgM syndrome (L. Maródi), Di-George syndrome (M. Pac), WAS (V. Gulácsy), and XLP (A. Lakatos).

Larysa Kostyuchenko and Gayane Akopyan shared their experience of diagnosis and treatment of the syndromes of chromosomal instability in the large group of patients with NBS and AT.

The last part of the Meeting included presentations of clinical cases of PIDs from different regions of Ukraine which were difficult to diagnose or treat.

The resolution of the meeting was that Ukraine needs to join the European PID Programmes "J Project" and "ESID Online Registry" in order to cooperate in the field of clinical and molecular-genetic investigations. Also, more effort should be given to the organization of PID Parents Group in Ukraine.

The day before and after the meeting, the guests from the other regions of Ukraine and the foreign guests had a chance to see one of the most famous Ukrainian operas "Natalka-Poltavka" and to explore beauties of the historical part of Lviv on the sightseeing tour.

Larysa Kostyuchenko

SPONSORS: BIOPHARMA, Pfizer, Nestle, GlaxoSmithKline, Parmasco, Yamanouchi, DrFalk, Solvay, Novartis, Sanofi Aventis, OMB, charity foundation "Svit Dytyny".

ORADEA, ROMANIA
18 DECEMBER, 2005



HOST: ZOLTÁN ELLENES (e_zoltan@yahoo.com)

VENUE: Oradea Children's Hospital, Oradea

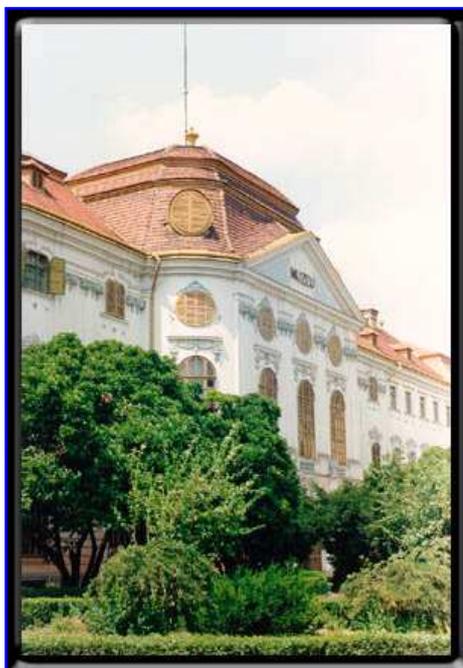
MAIN TOPIC:
PID genetic diagnostics for Romanian patients



L-R: Zoltán Ellenés, László Ritli, Maria Cucuruz, László Maródi

SUMMARY AND CONCLUSIONS:

On December 18, 2005, a meeting was organized to coordinate molecular genetic analysis of Romanian primary immunodeficiency patients from different regions of the country (Transylvania, South- and South-East Romania, and East-Romania). László Maródi from the Debrecen Jeffrey Modell Center was invited to this meeting. Romanian participants were Prof. Spineanu Radu, head of the Pediatric Department, University of Oradea, Prof. László Ritli, head of the Oradea Children's Hospital, Dr. Carp Gheorghe, general manager of the Oradea Children's Hospital, Dr. Maria Cucuruz, head of the Division of Immunology at the Timisoara Children's Hospital, and Dr. Zoltán Ellenés, pediatric immunologist at the Oradea Children's Hospital. The meeting was held in the Children's Hospital of Oradea



that is one of the most important centers of PID patient care in Romania.

Prof. László Maródi informed the Romanian colleagues on the development of the Debrecen Jeffrey Modell Center and the spectrum of PID diseases in



which genetic analysis has recently become available in Debrecen. These diseases include XLP, XLA, WAS, SDS, CD40L deficiency, AID deficiency, WHIM syndrome, and IL-2RG deficiency. However, the center is able to elaborate on the diagnostic spectrum in case in special request and need. In particular, it was offered that prenatal diagnosis may be available in addition to analysis of the disease-causing gene in the proband. To this end, a special team of geneticist, gynecologist, pediatrician, immunologist, and medical ethics expert was organized in Debrecen. Prof. Maródi emphasized that genetic tests are free of charge up to a certain limit due to the generous help provided by the Jeffrey Modell Foundation and Biotest Hungary. Dr. Cucuruz and Dr. Ellenés presented a list of patients whose clinical and immunological evaluation have been completed, but genetic analysis could not be done. It was agreed that Romania is a large enough country to develop additional PID centers, one in East-Romania (IASI, Moldova), and another in South-East Romania (Bucharest).

The one day meeting was very inspiring and the conclusion was made that time has arrived for the J Project countries to enter into joint research on epidemiology and mutational spectrum of PIDs in the East-European region.

Zoltán Ellenés

SPONSOR: ECE IPI CTR

J Project Meetings in 2006

January 16, 2006

Perspectives of Gene Therapy in the Wiskott-Aldrich Syndrome

Debrecen, Hungary

Organizer: **László Maródi** (Imarodi@jaguar.dote.hu)

May 8-9, 2006

ESID Prague Spring Meeting

Prague, Czech Republic

Organizer: **Anna Sediva** (AnnaSediva@fnmotol.cz)

June 9-10, 2006

Primary Immunodeficiency Update

Bucharest, Romania

Organizer: **Nicolae Iagăru** (iagarun@yahoo.com)

June 9-10, 2006

Progress in Education in Clinical Immunology

Ruciane Nida, Poland

Organizer: **Ewa Bernatowska** (immuno@czd.waw.pl)

November 9-10, 2006

Clinical, Immunological and Genetic Aspects of Primary Immunodeficiencies

Minsk, Belarus

Organizer: **Michael Belevtsev** (belevtsev_m@mail.ru)

December 8-9, 2006

East-European Primary Immunodeficiency Mutation Database

Debrecen, Hungary

Organizer: **László Maródi** (Imarodi@jaguar.dote.hu)

ESID/IPOPI/INGID Meeting 2006

October 4-7, Budapest

www.ESID2006.com

Dear Colleague,

On behalf of the ESID Board and the Scientific Committee I invite you to attend the XIIth Meeting of the European Society for Immunodeficiencies which will be held in the renovated Novotel Budapest Congress Centre of the beautiful city of Budapest, on 4-7 October, 2006. The ESID Meeting will be jointly organized with the International Parent Organization for Primary Immunodeficiencies, and the International Nursing Group for Immunodeficiencies. The preliminary scientific program has been set and it includes keynote talks and invited presentations on, among others, the clinical and genetic aspects of innate immunodeficiencies, defects of T and B cell development, bone marrow failure syndromes, and gene therapy. Interactive workshop sessions will be organized to discuss infections in PID patients, immunoglobulin replacement therapy, and long-term outcome of stem cell transplantations. To facilitate active participation of the broad membership of ESID as well as non-ESID members from various fields of clinical immunology, poster sessions will be integral part of the scientific program. According to the tradition, an Educational Day will precede the two and a half day Meeting. This time the subject of the Educational Day will be the development and defects of B cells.

All contributions will be evaluated by the Scientific Committee and recommended for either an oral or poster presentation during the main program. Because of the busy schedule, the Meeting will also include lunch symposia. During these symposia you will have the opportunity to exchange views on topics such as current status of the ESID Registry and update on the ESID Website. I hope that the preliminary program of the Meeting that you can see on the web is inspiring and you will join us to make another successful ESID Meeting together.

Your participation is critical for the success of this Meeting and I encourage you to call the attention of your colleagues and collaborators to this important scientific and professional event.

I look forward to seeing you in Budapest!

László Maródi
Congress President

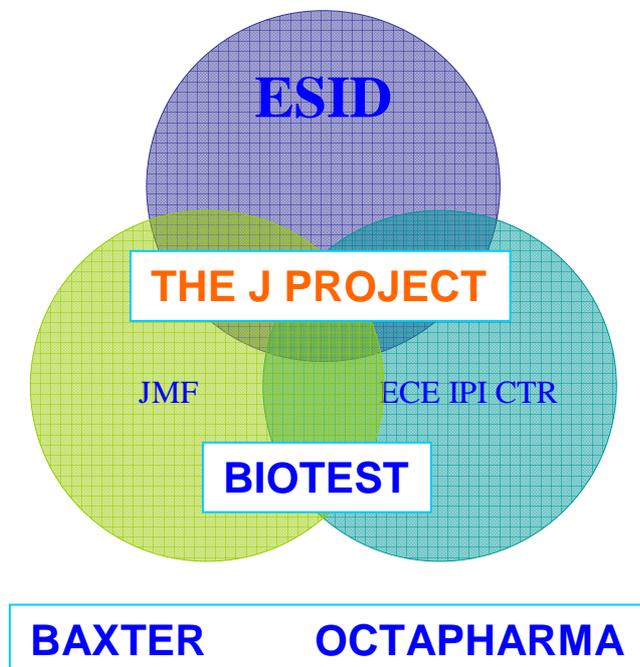
THE J PROJECT

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2. Elissaveta Naumova	Bulgaria
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4. Svetlana Aleshkevich	Belarus
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7. Ale Janda	Czech Republic
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10. Urve Putnik	Estonia
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18. Malgorzata Pac	Poland
19. Mihaela Bataneant	Romania
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21. Nicolae Iagaru	Romania
22. Zoltán Ellenés	Romania
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24. Olga Paschenko	Russia
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26. Aleksandra Minic	Serbia/Montenegro
27. Peter Ciznar	Slovakia
28. Lyudmila Chernishova	Ukraine
29. Larysa Kostyuchenko	Ukraine
30. Ala Volokha	Ukraine

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AN EAST-EUROPEAN PERSPECTIVE



AIM: To increase awareness on PID