

# PROJECT

**2004-2014**

First announcement of the

**100<sup>th</sup> J Project Meeting**  
**12-15 March 2014**



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*Rixos Lares Hotel, Lara, Antalya, Turkey*  
*Contact: Zsuzsa Horváth ([hzsus86@gmail.com](mailto:hzsus86@gmail.com))*

# Preliminary PROGRAM

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## 12 March 2014

10:00-18:45 **Registration**

18:45-19:15 **Welcome addresses**

*Amos Etzioni, President, ESID*

*Jose Drabwell, Chair, IPOPI Board*

*Dilşad Mungan, Turkish Society of Allergy and Clinical Immunology*

*Günnur Deniz, President, Turkish National Society of Immunology*

*Ismail Reisli, Leader, J Daughter Anatolia Project*

*László Maródi, Leader, J Project*

19:15-20:15 **SESSION 1: KEYNOTE LECTURE**

Chairs: *Ismail Reisli, Konya*  
*László Maródi, Debrecen*

**Life-threatening infectious diseases of childhood: single-gene inborn errors of immunity**

*Jean-Laurent Casanova, Paris-New York, France-USA*

20:15-21:45 **CHEESE AND VINE WELCOME RECEPTION and J PROJECT POSTER SESSION I.**

## POSTERS SESSION I.

### PID registries and databases

Chairs: *Anna Sediva, Prague*  
*Elissaveta Naumova, Sofia*  
*Marina Guseva, St. Petersburg*

1. **Epidemiological and genetic features of PID in children at Belarus**  
*Mikhail Belevtsev, Minsk, Belarus*
2. **Update of PID activities in Iran**  
*Nima Rezaei, Asghar Aghamohammadi, Tehran, Iran*
3. **Polish registry of Primary Immunodeficiency**  
*Malgorzata Pac and the Polish Working Group for Primary Immunodeficiencies*
4. **Epidemiological and genetic features of PID patients in Russian Children's Clinical Hospital (20 years experience)**

*Olga Paschenko, Irina Kondratenko, Andrey Bologov, Moscow, Russia*

**5. Primary immunodeficiencies in childhood and adults: The Siberian PID registry**

*Irina Tuzankina, Yelena Vlasova, Marina Karakina, Evgeniya Bass, Ekaterinburg, Russia*

**6. National PID registry in Ukraine**

*Liudmyla Chernyshova, Anastasia Bondarenko, Alla Volokha, Kiev, Ukraine*

**7. Registered PID patients in Zonguldak, Turkey**

*Mutlu Yüksek, Zonguldak, Turkey*

**8. Adult PID patients in a newly established immunology/allergy division in Konya, Turkey**

*Caliskaner Zafer, Arslan Sevket, Konya, Turkey*

**9. Registered PIDs patients between 2006 and 2013 in Gaziantep, Turkey**

*Özkars Mehmet Y. G. Antep, Turkey*

**10. The spectrum of primary immunodeficiency disorders: 3 year experience in a single center**

*Fazıl Orhan, Zekiye İlke Kılıç Topçu, Mehtap Haktanır Abul, Trabzon, Turkey*

**11. Education and PID awareness among medical students**

*Nesrin Reisli, İsmail Reisli, Konya, Turkey*

**12. Frequency and characteristics of patients with CVID over 18 years of age**

*Fevzi Demirel, Uğur Muşabak, Sait Yeşillik, Abdullah Baysan, Özgür Kartal, Mustafa Güleç, Osman Şener, Ankara, Turkey*

**13. PID - Common manifestations of rare diseases**

*M Karakina, Irina Tuzankina, Ekaterinburg, Russia*

**14. Adult PID screening program in out-patient settings**

*Arslan Sevket, Caliskaner Zafer, Konya, Turkey*

## 13 March, 2014

### SESSION 1 Adaptive immune deficiencies I.

Chairs: *Irina Tuzankina, Ekaterinburg*  
*Jiri Litzman, Brno*  
*Necil Kütükçüler, Izmir*

- 9:00-9:20 **Novel agammaglobulinemia genotypes**  
*Mary Ellen Conley, Memphis, TN*
- 9:20-9:35 **Immunodeficiency due to CD19 mutations**  
*Ismail Reisli, Konya, Turkey*
- 9:35-9:50 **Update on CVID and IgA deficiency**  
*Lennart Hammarström, Stockholm, Sweden*
- 9:50-10:00 **Chronic immune activation in CVID is associated with elevated serum levels of soluble CD14 and CD25**  
*Jiri Litzman, Brno, Czech Republic*
- 10:00-10:10 **Clinical and molecular characteristics of 200 Iranian CVID patients**  
*Asghar Aghamohammadi, Tehran, Iran*
- 10:10-10:20 **Progressive degenerative damage of the central nervous system in X-linked agammaglobulinemia patients**  
*Irina Tuzankina, Ekaterinburg, Russia*
- 10:20-10:35 **Recent advances in understanding immunoglobulin class switch deficiencies**  
*Anne Durandy, Paris, France*
- 10:35-10:50 **Novel genetic etiologies of Hyper IgM syndrome**  
*Sergey Nejentsev, Cambridge, UK*
- 10:50-11:00 **Class switch recombination defects and unique cases of CD40 and CD40L deficiencies**  
*Necil Kütükçüler, Izmir, Turkey*

**11:00-11:30 COFFEE**

### SESSION 2 Immunoglobulin replacement therapy

Chairs: *Yıldız Camcıoğlu, Istanbul*  
*Peter Ciznar, Bratislava*  
*Tatiana Latysheva, Moscow*

- 11:30-11:45 **IVIG/SCIG replacement: the golden standard of treatment**  
*Vincent R Bonagura, New York, NY*
- 11:45-12:00 **IVIG treatment of patients with CVID and agammaglobulinemia**  
*Tatiana Latysheva, Moscow, Russia*
- 12:00-12:15 **Chances and limitations of Ig replacement in the care of patients with CVID**  
*Klaus Warnatz, Freiburg, Germany*
- 12:15-12:25 **Clinical and immunological efficacy of SCIG treatment of children and adults**  
*Anna Szaflarszka, Cracow, Poland*

- 12:25-12:40 **Clinical study of a novel 10% intravenous immunoglobulin preparation**  
*Gergely Kriván, Budapest, Hungary*
- 12:40-12:50 **Specific antibody deficiencies with normal immunoglobulins**  
*Ricardo Sorensen, New Orleans, LA*
- 12:50-13:00 **Hospital screening for antibody deficiencies**  
*Krista Ress, Tallinn, Estonia*

**13:00-14:00 LUNCH**

**SESSION 3**  
**Adaptive immune deficiencies II.**

Chairs: *Ewa Bernatowska, Warsaw*  
*Aydan İkinciogulları, Ankara*

- 14:00-14:15 **Novel combined immunodeficiency genotypes**  
*Stuart Tangye, Sydney, Australia*
- 14:15-14:30 **Di George syndrome: where do we stand now?**  
*Malgorzata Pac, Warsaw, Poland*
- 14:30-14:40 **T and B cell development in DiGeorge syndrome**  
*Adam Klocperk, Prague, Czech Republic*
- 14:40-14:50 **SCID before and after neonatal BCG vaccination in Slovakia**  
*Peter Ciznar, Bratislava, Slovakia*
- 14:50-15:00 **Functional characterization of the novel 137+5G>A splice donor site mutation in the SH2D1A gene**  
*Beáta Tóth, Debrecen, Hungary*
- 15:00-15:10 **Nijmegen Breakage syndrome: long-term outcome and treatment**  
*Beata Wolska-Kuśnierz, Warsaw, Poland*
- 15:10-15:20 **Nijmegen breakage syndrome in Ukraine: frequency, medical support and follow-up**  
*Ihor Savchak, Lviv, Ukraine*
- 15:20-15:40 **BMT/HSCT: a success story of treating PID patients**  
*Andrew Cant, Newcastle upon Tyne, UK*
- 15:40-15:50 **How should we treat patients with TAP deficiency**  
*Figen Dogu, Ankara, Turkey*
- 15:50-16:00 **CD25 deficiency: Clinical and laboratory features**  
*Aydan İkinciogulları, Ankara, Turkey*

**16:00-16:30 TEA**

**SESSION 4:**  
**PID CARE IN THE SOUTH HEMISPHERE AND SOUTH-ASIA**  
**16:30-17:40**

16:30-17:30 Chairs: *Aisha El-Marsafi, Cairo*  
*Olcay Yeğın, Antalya*  
*Hamoud Al-Mousa, Riyadh*

16:30-16:40 **Latin-America**

- 16:40-16:50 *Ricardo Sorensen, New Orleans, LA*  
**Africa**  
*Ahmed Aziz Bousfiha, Casablanca, Marocco*
- 16:50-17:00 **India**  
*Surjit Singh, Chandigarh, India*
- 17:10-17:20 **PID care and research in Saudi-Arabia**  
*Hamoud Al-Mousa, Riyadh, Saudi Arabia*
- 17:20-17:30 **Australia/New Zealand**  
*Jane Peake, Brisbane, Australia*
- 17:30-17:40 **Exome sequencing in the Asian PID Network**  
*Yu-Lung Lau, Hong Kong, China*

**20:00-21:30 SOCIAL PROGRAM and J PROJECT POSTER SESSION II.**

## POSTERS SESSION II.

### B CELLS

*Chairs: Margorzata Pac, Warsaw*  
*Krista Ress, Tallinn*  
*Adrien Sarkadi, Debrecen*

1. **Recurrent wheezing and hypogammaglobulinemia during infancy**  
*Ozdemir Oner, Bozdogan S. Sakarya, Turkey*
2. **A clinical syndrome of elevated serum IgM, massive splenomegaly and recurrent lower respiratory tract infection in an adult patient**  
*Şengül Aksakal, Nihal Mete, Aytül Sin, Okan Gülbahar, Ali Kokuludağ, Ömür Ardeniz, İzmir, Turkey*
3. **Rheumatoid arthritis, sclerosing cholangitis and recurrent thymoma in an adult CVID patient**  
*Rabia Bilge Özgül Özdemir, Nihal Mete, Aytül Sin, Okan Gülbahar, Ali Kokuludağ, Ömür Ardeniz, İzmir, Turkey*
4. **B cell subsets in isolated IgM deficiency**  
*Deniz Güloğlu, Funda Erol Çipe, Senem Koçak, Şule Haskoloğlu, Caner Aytekin, Figen Doğu, Aydan İkinciogulları Ankara, Turkey*
5. **Steroid resistant idiopathic thrombocytopenic purpura in a patient with CVID**  
*Sait Yeşillik, Uğur Muşabak, Abdullah Baysan, Fevzi Demirel, Özgür Kartal, Mustafa Güleç, Osman Şener, Ankara, Turkey*
6. **Evaluation of bronchiectasis in PID patients**

*İbrahim Cemal Maslak, Ayşen Bingöl, Tuba Turul Özgür, Olcay Yeğın, Akdeniz Üniversitesi, Antalya, Turkey*

7. **A case of common variable immune deficiency associated with hemolytic anemia**  
*Mehtap Haktanır Abul, Zekiye İlke Kılıç Topçu, Fazıl Orhan, Trabzon, Turkey*
8. **Selective immunoglobulin M deficiency associated with angioedema**  
*Zekiye İlke Kılıç Topçu, Mehtap Haktanır Abul, Fazıl Orhan, Trabzon, Turkey*
9. **Autoimmune hepatitis and scleroderma in a patient with X - linked agammaglobulinemia**  
*Mustafa Yılmaz, Gülbin B Karakoç, Derya Ufuk Altıntaş, Aylin Kotil, Adana, Turkey*
10. **B cell lymphopenia and lymphadenitis in a newborn infant**  
*Şükriü Nail Güner, Esra Hazar Sayar, Mukaddes Ağirtıcı, İsmail Reisli, Konya, Turkey*
11. **The effect of IVIG therapy on specific antibody responses in children with transient hypogammaglobulinemia of infancy**  
*Elif Azarsız, Neslihan Edeer Karaca, Güzide Aksu, Necil Kütükçüler, İzmir, Turkey*
12. **Chronic inflammatory disease in patients with sIgAD and CVID**  
*Zoltán Ellenés, István Kovács, Michaela Bataneant, Maria Cucuruz, Margit Serban, László Maródi, Oradea-Timisoara, Romania; Debrecen, Hungary*
13. **Selective IgA deficiency: analysis of 100 Latvian patients**  
*Tatjana Prokofjeva, Riga, Latvia*

## T CELLS

*Chairs: Andrey Prodeus, Moscow  
Alla Volokha, Kiev  
Ludmilla Cerempei, Chisinau*

1. **Clinical and laboratory features and outcome of MHC class II deficiencies**  
*Gökalp BolKent, Şule Haskolođlu, Funda Erol Çipe, Zülfikar Akelma, Sevda Çam, Deniz Gülođlu Ankara, Turkey*
2. **Late-onset purine nucleoside phosphorylase deficiency with spastic paraplegia**  
*Fatih Celmeli, Antalya, Turkey*
3. **Screening Di George Syndrome among patients with congenital cardiopathies**  
*Esra Toprak Kanık, Neslihan Edeer Karaca, Ertürk Levent, Özge Altun Körođlu, Emin Karaca, Ferda Özkınay, Güzide Aksu, İzmir, Turkey*
4. **A novel STK4 deficiency in two siblings**  
*Sevil Oskay Halacli, Deniz Cagdas Ayvaz, Cagman Tan, Elif Uz, İlhan Tezcan, Nurten Akarsu, Ozden Sanal, Ankara, Turkey*

## 5. SCID: analysis of 17 Latvian patients

*Tatjana Prokofjeva, Latvia, Riga*

## HSCT

*Chairs: Gergely Kriván, Budapest  
Anna Szaflarszka, Cracow  
Tatjana Prokofjeva, Riga*

### 1. HSCT for patients with SCID: A single center experience

*Deniz Çağdaş Ayvaz, Tuba Turul Özgür, Gülten Türkkani Asal, Çağman Tan, Özden Sanal, İlhan Tezcan, Ankara, Turkey*

### 2. Immuno-reconstitution in the first three years after HSCT

*Alişan Yıldırım, Samsun, Türkiye, Stephan Borte, Leipzig, Germany*

### 3. Food and respiratory allergy in patients after solid organ transplantation

*Ozdemir Oner, Bozdogan S. Sakarya, Turkey*

### 4. A single center experience of HSCT in Ankara.

*Zülfikar Akelma, Figen Doğu, Şule Haskoloğlu, Funda Çipe, Caner Aytekin, İsmail Reisli, Mutlu Yüksek, Alişan Yıldırım, Sevda Çam, Metin Aydoğan, Deniz Güloğlu, Tanıl Kendirli, Aydan İkinciogulları Ankara, Turkey*

### 5. The use of mesenchymal stem cell infusions for the treatment of GvHD in PID patients

*Aydan İkinciogulları, Şule Haskoloğlu, Figen Doğu, Birsal Küçükersan, Ercüment Ovalı Ankara, Turkey*

### 6. Hematopoietic stem cell transplantation in primary immunodeficiencies: a single center experience in Hungary, 1992-2014

*Gergely Kriván, Budapest, Hungary*



**14 March, 2014**

**SESSION 5  
INNATE IMMUNE DEFECTS I.**

Chairs: *Georgina Kuli-Lito, Tirana*  
*Ricardo Sorensen, New Orleans, LA*

- 09:00-09:15 **Aicardi-Goutières syndrome**  
*Yanick J Crow, Manchester, UK*
- 09:15-09:30 **Hyperimmunoglobulinemia D syndrome in two children with periodic fever**  
*Georgina Kuli-Lito, Tirana, Albania*
- 09:30-09:45 **Genetic basis of Behçet's disease**  
*Daniel L Kastner, Bethesda, MD*
- 09:45-10:00 **Autoimmune manifestations of immune deficiency**  
*Tadej Avcin, Ljubljana, Slovenia*
- 10:00-10:15 **Mitochondrial pathology in autoinflammatory disorders**  
*Anna Sediva, Prague, Czech Republic*
- 10:15-10:30 **Primary OX40 deficiency and genetic etiologies of Kaposi sarcoma**  
*Minji Byun, New York, NY*
- 10:30-10:40 **Anti-IL-17A, but not anti-IL-17F and anti-IL-22 antibodies, may be correlated with the severity of mucocutaneous candidiasis in APECED patients**  
*Adrien Sarkadi, Debreceni, Hungary*
- 10:40-10:50 **Autosomal recessive HIES**  
*Ozden Sanal, Ankara, Turkey*
- 10:50-11:00 **Different severity of phenotypes in patients with STAT1 gain-of-function mutations**  
*Alla Volokha, Kiev, Ukraine*

**11:00-11:30 COFFEE**

**SESSION 6  
INNATE IMMUNE DEFECTS II.**

Chairs: *Ozden Sanal, Ankara, Turkey*  
*Nima Rezaei, Tehran, Iran*

- 11:30-11:45 **PID patients with neutropenia: differential diagnostic algorithms**  
*Nima Rezaei, Tehran, Iran*
- 11:45-11:55 **GATA2 deficiency in children with hematological abnormalities**  
*Svetlana Sharapova, Minsk, Belarus*
- 11:55-12:05 **Chronic mild neutropenia revealing MonoMac syndrome**  
*Mihaela Bataneant, Timisoara, Romania*
- 12:05-12:20 **STAT2 deficiency**  
*Sophie Hambleton, Newcastle upon Tyne, UK*
- 12:20-12:35 **Immunodeficiency phenotype of MAGT1 mutation**  
*Michael J Lenardo, Bethesda, MD*

- 12:35-12:50 **Challenges of HSCT to treat patients with phagocytic cell defects**  
*Reinhard Seger, Zurich, Switzerland*
- 12:50-13:00 Lung and brain aspergillosis in a patient with AR CGD  
*Anastasia Bondarenko, Kiev, Ukraine*

**13:00-14:00 LUNCH**

**SESSION 7  
MISCELLANEOUS  
14:00-15:00**

Chairs: *Nadejda Sabashova, St. Petersburg*  
*Beáta Tóth, Debrecen*  
*Anna Shcherbina, Moscow*

- 14:00-14:15 **Novel adhesion protein deficiencies**  
*Amos Etzioni, Haifa, Izrael*
- 14:15-14:30 **Novel and recurrent WASP mutations in Eastern and Central European patients with WAS**  
*Vera Gulácsy, Debrecen, Hungary*
- 14:30-14:45 **Novel STAT1 GOF mutations in ECE patients with CMCD**  
*Beáta Soltész, Debrecen, Hungary*
- 14:45-15:00 **Characterizations of CGD patients in Turkey**  
*Yavuz Köker, Kayseri, Turkey*

**POSTERS SESSION III.**

**PID SYNDROMES**

Chairs: *Irina Kondratenko, Moscow*  
*Margit Serban, Timisoara*  
*Anastasia Bondarenko, Kiev*

1. **Histological findings in ALPS patients**  
*Irina Kondratenko, Dmitry Rogozhin, Olga Paschenko, Andrey Bologov, Moscow, Russia*
2. **Kimura disease in a patient with the Wiscott-Aldrich syndrome**  
*Irina Tuzankina, Yelena Vlasova, Anna Scherbina, Ekaterinburg-Moscow, Russia*
3. **Unusual presentations of PID**  
*Marina Karakina, Ekaterinburg, Russia*
4. **Wiskott-Aldrich syndrome: report on a unique case**  
*Gulnara Nasrullayeva, Baku, Azerbaijan*

**5. Lymphocyte subsets in patients with ataxia-telangiectasia***Halil Çeliksoy, Samsun, Erdem Topal, Malatya, Alişan Yıldırım, Samsun, Turkey***6. Unusual clinical presentation of ataxia-telangiectasia in two siblings***Alişan Yıldırım, Samsun, Türkiye, Stephan Borte, Leipzig, Germany***7. The lack of WASP gene and multiple food allergy in a Wiskott-Aldrich syndrome patient***Nurşen Ciğerci Gunaydın, Esen Demir, Vera Gulácsy, László Maródi, Remziye Tanac, Necil Kutukculer, İzmir, Turkey***8. Unique ocular findings in a patient with 22q11.2 deletion syndrome***Gokturk Bahar, Bozkurt Bozkurt, Reisli Ismail, Konya, Turkey***9. Delayed diagnosis of NBS in an adult patient presenting with recurrent infections from childhood***Zeynep Peker Koç, Emin Karaca, Nihal Mete, Aytül Sin, Okan Gülbahar, Ali Kokuludağ, Ömür Ardeniz, İzmir, Turkey***10. STK-4 deficiency among patients with autosomal recessive HIES phenotype***Sevil Oskay Halacli, Deniz Cagdas Ayvaz, Cagman Tan, Elif Uz, İlhan Tezcan, Nurten Akarsu, Ozden Sanal, Ankara, Turkey***11. IVS10+1 G>A and IVS9+1>C splicing mutations in WASP gene***Ayşenur Öztürk, Figen Doğu, Seda Tasır Yılmaz, Hilal Özdağ, Aydan İkinciogulları, Ankara, Turkey***12. WHIM syndrome without hypoglobulinaemia and without warts***Kristina Mironska, Skopje, Rep. of Macedonia***13. The value of mean platelet volume/platelet count ratio to predict 22q11.2 deletion syndrome***Bahar Gokturk, Sukru Nail Guner, Reyhan Kara, Mine Kırac, Sevgi Keles, Hasibe Artac, Ismail Reisli, Konya, Turkey***INNATE IMMUNITY**

*Chairs: Beáta Soltész, Debrecen  
Mihaela Bataneant, Timisoara  
Alexander Pischalnikov, Chelyabinsk*

**1. CGD patients treated with interferon- $\gamma$  in Turkey***Serkan Filiz, Dilara Fatma Kocacık Uygun, Antalya, Turkey***2. Early diagnoses leukocyte adhesion molecule deficiency type-1***Şükür Nail Güner, Esra Hazar Sayar, Melike Emiroğlu, İsmail Reisli, Konya, Turkey***3. Eye infections as presenting manifestations of chronic granulomatous disease**

*Esra Hazar Sayar, Şükrü Nail Güner, Mine Kırac, Melike Emirođlu, İsmail Reisli*

**4. Presentation of interleukin-12/-23 receptor beta1 deficiency with varying clinical symptoms**

*Deniz Çađdaş Ayvaz, Çađman Tan, Ayşe Metin, Özlem Keskin, Mehmet Yaşar Özkars, Özden Sanal, İlhan Tezcan, Ankara, Turkey*

**5. Salmonella enteritidis infection associated with cutaneous leukocytoclastic vasculitis in a patient with IL-12R  $\beta$ 1 deficiency**

*Dilara Fatma Kocacık Uygun, Serkan Filiz, Antalya, Turkey*

**6. Rare and remarkable manifestations of a case with complete interferon-gamma receptor-1 deficiency**

*Neslihan Edeer Karaca, Stephanie Boisson Dupuis, Guzide Aksu, Jackinta Bustamante Jean-Laurent Casanova, Necil Kütükçüler, İzmir, Turkey*

**7. C8 deficiency with or without recurrent meningitis in a Turkish family**

*Nesrin Gulez, İzmir, Turkey*

**SESSION 8**  
**THE ANTALYA DECLARATION**  
**16:00-16:30**

Chairs: *László Maródi, J PROJECT*  
*Ismail Reisli, J DAUGHTER ANTALIA PROJECT*  
*Amos Etzioni, ESID*  
*Jose Drabwell, IPOPI*

16:00-16:05 **THE ANTALYA DECLARATION FOR PID PATIENTS**

Reader: *Melinda Erdős, Debrecen, Hungary*

16:05-16:10 Government representative of Hungary

16:10-16:15 Sema Özbaş, Government representative of Turkey

16:15-16:20 Government representative of Germany

16:20-16:25 **Signature of the Declaration**

16:25-16:30 Closing the Meeting

17:00-19:00 Sport and relaxation program

19:30 Cultural program