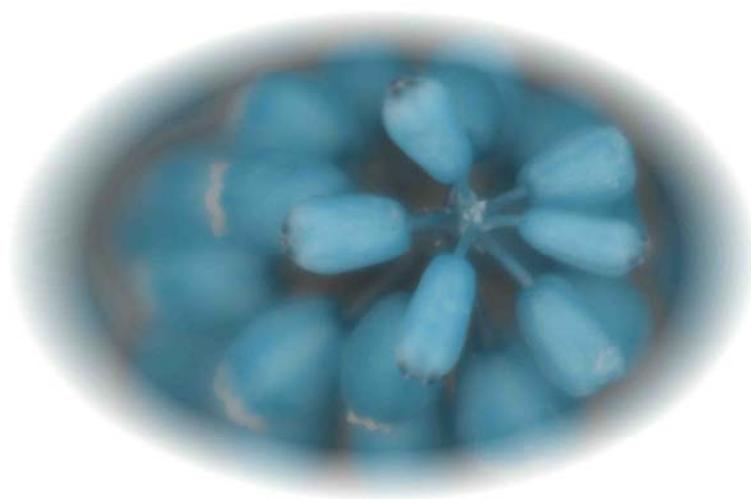


# ESID NEWSLETTER



**2009 - Issue 1**

**April 2009**

**[www.esid.org](http://www.esid.org)**

ESID Editorial Office



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*ESID members are invited to  
publish in this newsletter*



esid@estherdevries.nl

Dear ESID members,

As you will see in this ESID Newsletter, a lot is changing within ESID! We now have Kenes doing a lot of work for us, and this was a good moment to give the ESID Newsletter a 'face-lift'! We hope you like the way it looks now.

You will still find the well-known sections inside, including invitations to the ESID Prague Spring School, ESID Summer School, and upcoming J-Project meeting. Do consider to visit one of these events if they are of interest to you!

We really need some new input in the Editorial Board, so please consider giving us some help, and send me an e-mail at [esid@estherdevries.nl](mailto:esid@estherdevries.nl).

Have a wonderful springtime !

Best wishes,

**Esther de VRIES,**  
*Editor*

**INSIDE THIS ISSUE**

President's letter	4
Secretary's report	4
Treasurer's report	6
News & Views	6
ESID Prague Meeting	10
Working Party Reports	12
Registry WP	12
SCT & GT WP	13
ESID Juniors WP	14
Interesting Papers	14
Information	16



**ESID SPRING SCHOOL**  
May 11-12, 2009  
Prague, Czech Republic



**ESID SUMMER  
SCHOOL**

September 2-6, 2009  
Bled, Slovenia

**NOT YET MEMBER ?**

Become an ESID member and enjoy all the new features of the website :

- Publications in the field of immunodeficiency
- Discussion forums
- Updated working party sections
- Diagnostic criteria
- ESID Juniors Corner
- ESID Members zone
- Ongoing study protocols
- ESID centres

**For more information visit [www.esid.org](http://www.esid.org)**



**Dates changed to  
October 6-9, 2010**

[www.esid2010.org](http://www.esid2010.org)



### A Note from the President



Dear All,

As some of you may know, we have now decided to contract the organization of our ESID meetings with Kenes. You may recall that Kenes had done a superb job, along with Amos Etzioni, in relocating the Jerusalem meeting to Geneva within a very short notice some years ago. The involvement of Kenes in other activities of the ESID was also crucial, as the burden on both our secretary Bodo and treasurer Eleonora was dramatically increasing. We are confident that Kenes will be a great asset to our expanding society and we look forward to working with them. Please join me in welcoming Kenes aboard.

All best wishes,

**Jean-Laurent CASANOVA**

### Secretary's Report



2009 is expected to be a dynamic year for ESID. The Society is not only in the process of redefining some of its internal management but also keeping busy planning new activities!

- The ESID Prague Spring School will be held in May
- The 7th edition of ESID Summer School to be held in Bled, Slovenia in September this year
- The ESID 2010 meeting in Istanbul will also be one of the main highlights of all Board members' efforts during the course of this year

While new projects are in the pipeline, ESID has just recently appointed Kenes International to organise their biennial meeting, following Istanbul, and to manage the Society in its day-to-day business.

### ESID Meetings

Preparations for the Prague Spring School (<http://www.esid.org/home.php?sub=2&id=237>) are being finalized by the local organization committee led by Anna Sediva.

This meeting will run from May 11 to 12, 2009, and ESID has agreed to award 2 to 3 travel grants to successful applicants.

Please go to (<http://www.esid.org/home.php?sub=2&id=230>) for application.

The ESID Summer School will be held in Bled, Slovenia, from September 2 to 6, 2009. For five consecutive days a panel of well known international Faculty will be leading case-based discussions and give lecture presentations on primary immunodeficiencies.

The European Congress of Immunodeficiencies (ECI) happening in Berlin from 13 to 16 September 2009, will include two ESID Symposia, one sponsored by Baxter on Tuesday September 15th, and one sponsored by CSL Behring on Sunday September 13th.

Whilst only occurring in October 2010, the next ESID biennial meeting is one of the top items in our Board members' agenda for 2009.



## Secretary's Report — Continuing

ESID Board members are currently working together with the local organiser to shape the content and format of the education sessions to be the best and trying to optimise interaction amongst participants. The overall aim is to ensure a smooth and successful organisation, raise this meeting's profile and boost attendance. To begin with, dates were changed to Wednesday 6 October to Saturday 9 October 2010.

Membership and registration fees have been considered a very sensitive topic. It was through analysis of former international meetings, that concerns were raised by our Board members in relation to registration fees charged to ESID non-members and to INGID and IPOPI members. It was felt that ESID non-members should pay 125.00 Euros more than members in order to fully cover the two year membership fee of 100 EUR.

Registration for the Istanbul meeting will open in early 2010 and the gala dinner fee will this time be charged separately from the registration fee. ESID Board members are also considering collecting membership fees for the year 2010 and 2011 together with the meeting registration fee.

### Kenes International

ESID has recently appointed Kenes International to be its official congress organiser from 2012. The agency will also provide from 2009 association management services aimed to assist ESID in its day-to-day management. Kenes will pre-finance the congresses and guarantee a minimum revenue to the Society.

Two separate bank accounts have been created under the Kenes umbrella's main account for VAT and administration practicabilities. One for the congress, with all revenues following the event being transferred to the ESID bank account in the Netherlands, and another one for association management. For the latter Kenes will pre-finance all ESID out-of-pocket extra expenses and will invoice the ESID Treasurer, Eleonora Gambineri, every three months.

Kenes will not charge any interest on pre-payments nor on any monies collected.

### Any other business

ESID has decided to apply to become a FOCIS society member. FOCIS is a federation of societies and this will allow us to have a voting representative on their Steering Committee. ESID has currently 347 members and this may help to bring the voice of our Society to a larger international audience thus helping us achieve our goals.

FOCIS will hold its next meeting June 11-14, 2009 in San Francisco. A PID consortium day on June 11th co-organized by the Clinical Immunology Society (CIS) and by the American Academy of Allergy, Asthma & Immunology (AAAAI) will precede the event.

Best wishes,

**Bodo GRIMBACHER**



**Dates changed to  
October 6-9, 2010**

[www.esid2010.org](http://www.esid2010.org)

**ESID to become  
a FOCIS  
society member**



## Treasurer's Report

Dear ESID members,

It is spring already and few months have passed since I was elected new treasurer. Now that I took over all the duties, I must admit that it is very exciting job with lots of opportunities for developments! Thank you again for your support!

Now I would like to provide you few updates:

- For those who haven't noticed, the treasurer's email address has changed. Please refer to [treasurer@esid.org](mailto:treasurer@esid.org) for all your questions or concerns regarding financial issues (i.e. membership payments, travel grants, funding opportunities etc.).
- Some of you have not yet paid the 2008-2009 ESID membership fee. Please do that as soon as possible. You might have noticed that you have been "inactivated" as a member and you are not able to login on the ESID website. So if you wish to be active again, be part of the ESID community and receive all the latest update, please submit your payment by credit card through the ESID website ([www.esid.org](http://www.esid.org) > "Payment of membership/Renewal of membership" section). I would like to remind you that this is the only way to pay the membership fee. We do not accept cash or cheques. Thank you for your collaboration!
- We are supporting many educational activities involving ESID Junior members. We are providing travel grants to participate to the Spring School in Prague and we are organizing the 7th ESID Summer School to be held in Bled, Slovenia on September 2009. Although there is always more to do...so if any of you have contact with possible sponsors please let me know.
- Lately I have received new membership applications from several young trainees. I am very glad to notice that ESID Juniors are interested in being actively part of the Society and I believe that we should provide them all the support needed to contribute to the growth of ESID.

Finally, Kenes International has recently taken over the management of ESID. I would like to welcome all the people that will be working together with us to support practical issues on daily activities, and in particular Rita Louro, the new ESID Administrator, who is already of tremendous help!

Have a super springtime !

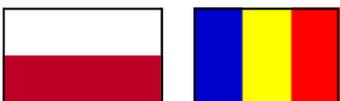
**Eleonora GAMBINERI**

**CALL FOR DUES 2009**  
Support our activities !

## News & Views

### J Project Meetings in 2009

28. Timisoara (Romania)	Margit Serban	March 19-20	<a href="mailto:mserban@rdstm.ro">mserban@rdstm.ro</a>
29. Sevastopol (Ukraine)	Liudmyla Chernyshova	April 09-10	<a href="mailto:chernyshova@ukr.net">chernyshova@ukr.net</a>
30. Prague (Czech Republic)	Anna Sediva	May 11-12	<a href="mailto:Anna.Sediva@fnmotol.cz">Anna.Sediva@fnmotol.cz</a>
31. Ekaterinburg (Russia)	Irina Tuzankina	May 13-14	<a href="mailto:I.Tuzankina@iip.uran.ru">I.Tuzankina@iip.uran.ru</a>
32. St. Petersburg (Russia)	Marina Guseva	June 9-10	<a href="mailto:gusevamarina@mail.ru">gusevamarina@mail.ru</a>
33. Konya (Turkey)	Ismail Reisli	June 25-26	<a href="mailto:ireisli@hotmail.com">ireisli@hotmail.com</a>
34. Cracow (Poland)	Danuta Kowalczyk	September 24-25	<a href="mailto:dkowalcz@cm-uj.krakow.pl">dkowalcz@cm-uj.krakow.pl</a>
35. Tallin (Estonia)	Sirje Velbri	October 7-8	<a href="mailto:velbrisir@hotmail.com">velbrisir@hotmail.com</a>



**News & Views—Continuing**

**J Project Meetings in 2009 –Continuing**

36. Ljubljana (Slovenia)	Nataša Toplak	October 9-10	natasa.toplak@ardi.si
37. Tehran (Iran)	Asghar Aghamohammadi or Nima Rezaei	October 11-12	aghamohammadi@sina.tums.ac.ir rezaei_nima@yahoo.com
38. Cairo (Egypt)	Nermeen Galal	October 18-19	nermeengalal@gmail.com
39. Minsk (Belarus)	Michael Belevtsev	October 22-23	belevtsev_m@mail.ru
40. Tirana (Albania)	Georgina Kuli-Lito	November 19-20	gkuli_lito@hotmail.com



**Invitation to J-Project Meeting in Konya (Turkey)**

One of the following J Project Meeting (No:32) is going to be held in Konya, Turkey between June 25th and June 26th, 2009. We are looking forward to welcoming our colleagues to this meeting that held for the first time in Turkey. The main goal of the meeting is to discuss the diagnostic and therapeutic practices and problems in our region. We hope to get desired results discussing our PID cases.

Konya is situated in the middle Anatolia region and takes place in the huge plain which gives its name.

Konya had been an important Ottoman and Seljuk city in the history. The tomb (now museum) of Mevlana, who is the great philosopher and as the symbol of tolerance and human love, is visited by more than 1.5 million people every year. The region’s population is about 2 million.

In Konya, Department of Pediatric Immunology and Allergy is established in 2001. About 20000 patients are admitted to pediatric clinics in one year and are detected primary immune deficiency (PID) in about

1% of them. Antibody deficiency disorders are the most common PID disease. Severe combined immune deficiency (SCID) is occurs approximately 1:10000 live births in the region because of high consanguineous marriage. The ratio of consanguinity was detected is 37.5% in PID and 84% in the patients with SCID.

We will be proud of being with you in the meeting for two days in Konya, one of the famous Turkish city.

With best regards,

**Ismail REISLI**



**23th J PROJECT Meeting Odessa, Ukraine 10 April, 2008**

**Main Topic:** Diagnostics and management of children with primary immunodeficiency diseases

Three J Project seminars were held in Ukraine - in Kiev, 2004, in Lvov, 2005, in Zaporozhye, 2007. By this way central ,western and eastern regions of Ukraine were involved. The place of 4-th J Project Meeting in Ukraine was chosen as Odessa.

Odessa – is a big city, sea port on the Black Sea shore, and is situated on the south of Ukraine. Region’s population is 2

million. Odessa region has low diagnostic rate of PID, the reason is insufficient information level of pediatricians in aspects of primary immunodeficiency.

To the program of the Meeting was included workshop section where parents of PID children participated. Parents spoke about PID problems on their families’ examples. Their speeches impressed

audience very much and touched everybody’s heart.

Social program included attending Odessa Opera Theatre and City Tour.

Sponsors of the event were Sanofi-Pasteur (France) and Biofarma (Ukraine) companies.

**Liudmyla CHERNYSHOVA**



## News & Views—Continuing

### The 27th J Project Meeting November 28, 2008

VEF cultural palace, 2 Ropažu Str., Riga, Latvia;

**Main Topics :** Update on Primary Immunodeficiencies

**Host:** Tatjana Prokofjeva

The J Project meeting was held for the first time in Riga, Latvia, on the 28th of November, 2008. The meeting was organized by the Latvian Immunology society and the Latvian Pediatrician society.

Latvia is a small country near the Baltic Sea with a population of less than 2.2 million people. In the Immunology society (with 33 members, from which 15 are biologists) only one works with PID of children. Capabilities of the laboratory of PID diagnostics in Latvia are limited. To choose the right diagnosis sometimes becomes very problematic. Latvia does not have neither national PID registers, nor PID professional working groups.

The main targets of the conference were to improve awareness of PID among the pediatricians and other specialists, to discuss PID diagnostic problems, clinical symptoms and treatment aspects.

The meeting attracted much interest. During the day, the conference was attended by 470 participants (pediatricians, immunologists, neonotologists, infectionists etc.) from all regions of Latvia.

Among the guests, there were prof. L. Marodi (Hungary), M. Belevtsev (Minsk, Belarus), prof. N. Shabashova and M. Guseva (St. Petersburg, Russia), prof. V. Ilyin and J. Degtjarova (Moscow, Russia), prof. L. Cerempei (Moldova), Velbri Sirje (Tallinn, Estonia).

The first part of meeting was started with the information about J-project and overview of XII ESiD Meeting in 's-Hercogenbosh, the Netherlands. M. Belevtsev from the Belarus Research Center for Pediatric Oncology and Hematology (Minsk) has reported on the possibilities of Immunological and genetics diagnostics of primary immunodeficiencies in Belarus. Prof. L. Cerempei from Moldova and Sirje Velbri (Estonia), as well as T. Prokofjeva shared their knowledge on the situations with PID in their countries. The Hyper-IgE diagnostics, clinical aspects and immunopathology were presented by Prof. L. Marodi and M. Guseva.

A series of presentations from local speakers in the second part of the conference were dedicated to SCID and CVID. The lecture by Prof. Nadezhda Shabashova from Institute of Medical Mycology, Medical Academy of Postgraduate education (St. Petersburg, Russia) Chronic mucocutaneous candidiasis was very interesting and important for pediatricians.

The case report about family with Hyper-IgM syndrome obtained genetic confirmation with the help of the collaboration with Debrecen Jeffrey Modell PID Reference Center. L. Marodi just brought genetics results: two sons have a c.511C>A non-sense mutation on exon 5 of the CD40L gene; the mutation resulted in the stop of codon and of the CD40L protein at amino acid position 184 (p.S184X). Mother is the carrier (heterozygous). Two daughters have no mutation.

The clinical presentations in the third part of the meeting, such as Chronic granulomatosis disease, T-cell lymphoma in a child with Nijmegen breakage syndrome, The first registered case of Wiskott-Aldrich syndrome in Latvia, demonstrated our own experiences in this field.

Among social events, there were visits to the Old Town of Riga and Lido Recreation Centre.

We would like to thank all the participants for their contribution. The experience shares in the meeting will undoubtedly improve the situation of diagnostics and management of patients with primary immunodeficiency in Latvia.

To sum up, we suppose that the J- project is essential and useful; especially, for small countries with limited financial resources. The collaborations with the East-Central European Infectious and Pediatric Immunology centre, Debrecen, Hungary and others give us possibilities to improve the level of the primary immunodeficiencies diagnostics in Latvia and we are looking forward to continuing the successful cooperation.

**Tatjana PROKOFJEVA**



## News & Views—Continuing

### REPORT The 28th J Project Meeting

The 28th Meeting of J Project on Primary Immunodeficiencies was held on 18-19 March 2009 in Timisoara and Buzias (Romania), two cities located in the western part of the country, along the borders of Hungary and Serbia.

More than 70 participants, dedicated immunologists but also young doctors, representing all the historical regions of Romania ( Banat, Transylvania, Muntenia, Oltenia, Moldova, Bihor and Maramures ) attended it. A colleague from the Republic of Moldova also joined us. The meeting was organized within the framework of the J Project, under the role of Prof.dr.Laszlo Marodi, who together with Melinde Erdos were participants from Hungary.

The agenda of the conference was generous It comprised two parts:

A pre-conference educational session was organized dedicated to post-graduate education of young physicians

- The first held in Timisoara
- Preconference educational session
- Patient's Group activity and
- The second held in Buzias
- Bone marrow transplantation in PIDs
- PID registry in Romania
- PID case presentation

(PhD students, residents, general practitioners, pediatricians, laboratory specialists) aiming at providing a useful tool for a comprehensive approach of immunological processes and their deficiencies.

It was followed by patients group discussions regarding specific difficult and controversial clinical situations (Chediak-Higashi syndrome, CVID syndrome with severe atypical mycobacteria lung infection, cluster of autoimmunity in the family of a child with CVID syndrome) focussing on diagnosis and differential diagnosis, concluding on the importance of bio-molecular investigation for a definite diagnosis in the field of Primary Immune Deficiencies.

The second day meeting took place in Buzias. Professor Marodi presented a review on the history of J Project, highlighting its achievements, development and perspectives. Its hallmark is the steadily increasing number of participant countries; started in Hungary, implying at first the neighboring countries; it succeeded to expand the project farther in Asia (Jekaterinburg) and Africa (Cairo). The J project spread out knowledge and skills in the field of diagnosis, intending an improved approach to therapy of these rare diseases. All these involved countries maintained an active connection with Debrecen, the center of J Project, and could share progress in this field, having access to a biomolecular diagnosis.

Prof. M. Serban, president of the local organizing committee, presented general data regarding BMT in PID – an important and the only curative treatment for many severe life threatening PID. The review was of interest for PID ,the participants taking into account the present preoccupation in extending BMT in the field of PID therapy.

Dr. Mihaela Bataneant, the secretary of the local organization committee, informed the audience about the progress and changes of the National PID registry in Romania: a number of 209 patients, with more accurate diagnosis than in the previous registry 6,2 % of them with a bio-molecular signature.

In the following, there have been reported interesting case reports, new experiences in the field: first PNP identified in Romania, Wiskott Aldrich syndrome, XLP syndrome, Hyper IgD syndrome ,aso

As deliverables, all participants received a booklet with the main abstracts of the presentations made at the meeting and an electronic course material based on ESID recommendations for diagnostic approach of PID.

The meeting was officially recognized by the local Medical Collegium and accredited with 10 CME credits.

The meeting was well appreciated by the participants and feedbacks received have ascertained the importance of such an event for the intellectual and scientific development of the medical community in this region of Europe. All such meetings will be welcomed in the future and periodic workshops on PID will definitely benefit all those involved in the respective domain to benefit of the latest development and advancements in the field of Primary Immunodeficiencies.

**Prof. Dr. Margit SERBAN and Dr. Mihaela BATANEANT**

### Florence hosts ESID Meeting in 2012

Florence has been elected to host the 2012 ESID/IPOPI/INGID meeting!!!

I would like to thank you all very much for your trust and support!

I am looking forward to seeing you there in 2012!

**Eleonora GAMBINERI**





## ESiD Prague Meeting - May 11-12, 2009

Dear all,

It is our pleasure to announce the 8th **ESiD Prague Spring Meeting** that is going to be held in Prague on May the 11<sup>th</sup> and 12<sup>th</sup>, 2009.

The meeting takes place at the Department of Immunology of the 2<sup>nd</sup> Medical School, Charles University and is generally organized in the same way as in the previous years. This is a very active, flexible and friendly meeting, interactive and small and EACH participant is expected to present a short talk on a project he/she is currently involved in, results of previous research or an interesting case report (10-20 min talk). We can accept non-presenting participants only as an exception!

We set up two key themes of this year – B cells in CVID and hyper IgE syndrome, but there will be enough space to discuss any topic related to PID and we hope that it will be in quite an informal way as we experienced during the previous workshops.

Klaus Warnatz and Esther de Vries promised to come as the invited speakers and the members of ESiD board. The co-operation with American ACAAI was established this year, two speakers – the members of this society will participate on our meeting (will be specified later) and ACAAI will support us.

Also this year we would like to provide a space for presentations of members of *ESiDJuniors working party*, however, we welcome applications from all ESiD members. Crina Samarghitean – the chairperson of ESiD Junior Working Party will give a talk about the latest developments and the future activities of ESiD Juniors WP, so we expect broad discussion of the attending *Juniors*.

If you are interested, please, **fill in the online registration form** that you can find on our website <http://imunologie.lf2.cuni.cz/> at the section “ESiD workshop”. Information on the previous workshops can be found there as well. The programme and all the available presentations and material of this year’s meeting will be placed at this webpage.

Since there could be only about 30-35 participants, the applications will be accepted on the first-come-first-served basis. So, do not hesitate and respond as soon as possible. Please, specify a topic that you would like to share with the audience as the detailed programme will be composed according to the submitted papers. Submission of short abstract (max 250 words) of the talk is required this year.

### The essential workshop information

**Conference dates:** **May 11 and 12, 2009**

**Conference venue:** Department of Immunology  
2<sup>nd</sup> Medical Faculty,  
Charles University and University Hospital Motol  
V úvalu 84- 150 06 Prague 5  
  
Olympus, sro.  
Evropská 176  
160 46 Prague 6 - Czech Republic

**Conference fee:** **FREE OF CHARGE**

**Refreshments:** lunch and snacks during both days will be provided **FREE OF CHARGE**



**ESID Prague Meeting - May 11-12, 2009 - Continuing**

**Accommodation:** This year the accommodation costs are not covered by the organizers and it is each participant's responsibility to reserve and pay for it. The list of webpages where you can find hotels, hostels and guest houses is shown below. The conference venue is at quarter "Prague 6". So this area would be most the suitable one for your stay. FREE OF CHARGE for a limited number of participants in a university hostel (double rooms, quite basic service). **Please, let us know your choice and place where you plan to stay (write it in "message for organizers" box).**

<http://www.praha-hotel.eu/de/Default.aspx>

<http://www.accommodations-prague.cz/>

<http://www.traveliana.com/prague-hotels-apartments.html>

<http://www.levne-ubytovani-v-praze.cz/>

<http://www.appartamento.cz/>

<http://www.avehotels.cz/hotels/denisa.html>

**Travel grants:** Please contact Rita Louro via email to apply for the Travel Grant: [esid.admin@kenes.com](mailto:esid.admin@kenes.com)

**Social programme:** On Monday evening - will be specified later and provided **FREE OF CHARGE** (usually welcome dinner and then the theatre or concert) we plan the bicycle tour (2 - 3 days on bikes) after the meeting from Prague to Brno (another immunodeficiency center in the capital of Moravia – region of the Czech Republic) – everyone is invited (will be also specified later)

**Official language:** English

**Contact:** [andrea.polouckova@lfmotol.cuni.cz](mailto:andrea.polouckova@lfmotol.cuni.cz)  
[anna.sediva@lfmotol.cuni.cz](mailto:anna.sediva@lfmotol.cuni.cz)

Please, feel free to contact us in case of any inquiry.

We are looking forward to meeting you in Prague in May.

With best regards,

**Andrea POLOUCKOVA and Anna SEDIVA**

Department of Immunology,  
2<sup>nd</sup> Medical Faculty, Charles University  
University Hospital Motol  
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fax: +420-224-435-962





## Working Party Reports

### REGISTRY WP

The year 2009 began with good news for the ESiD Database for Primary Immunodeficiencies. We finally managed to incorporate data on more than 1'000 patients from the "Registro español de inmunodeficiencias primarias" (REDIP) into the ESiD Database. Spanish centres will still continue to document their patients in REDIP, but have the possibility to view their patients in the ESiD Database and complete the more comprehensive ESiD datasets.

We would like to thank REDIP for making this transfer possible, in particular Nuria Matamoros, Javier Navarro and Isabel Caragol who were of great help.

After the good came the bad news: External funding for the database project was reduced sharply, which means that we do not have sufficient funds to continue paying bonus payments for data entry. We will still pay the bonus for the June 2009 deadline, but there will be no more annual bonus payments afterwards. We will inform all centres individually of this decision and hope that it will not affect the great and continuous commitment that the participating centres have shown so far.

However, we will be able to maintain the publication award. From now on, there will be a maximum of five awards for publications using data from the ESiD Database annually. The winning publications are selected by the ESiD Board. The best publication receives EUR 5.000, the second best EUR 4.000 and so on, so that the fifth publication receives EUR 1.000.

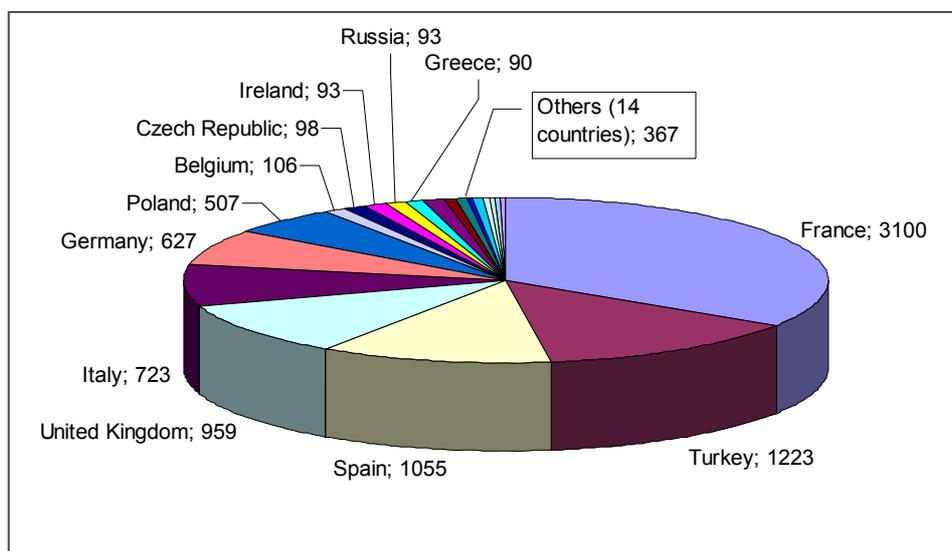
We would like to invite all researchers participating in the project to make active use of the data they document and collaborate with other centres to produce results. In case you need help, please contact the Database Administration at [registry@esid.org](mailto:registry@esid.org)

The total number of patients in the ESiD Database as of March 30th, 2009 was 9'041. The share of deceased patients was 722. France was still the largest contributor with 3'100 cases, now followed by Turkey (1'223) and Spain (1'055). A pie chart with the country distribution is given in Fig. A. Fig. B show the distribution of patients on the main PID categories.

3'213 patients received immunoglobulin replacement. Immunoglobulins were administered intravenously in 2'456 patients (76.4%) and subcutaneously in 738 patients (23%). 707 patients had undergone haematopoietic stem cell transplantation (HSCT). In 297 of these patients, the transplant caused major complications.

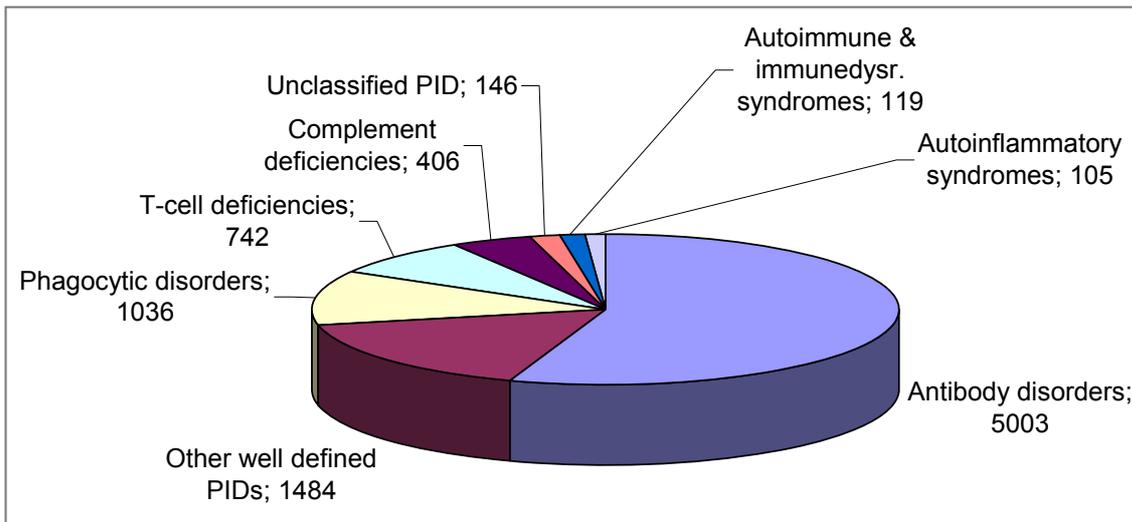
Further routinely updated statistical information on categories, diseases and age distribution as well as numbers on Ig-replacement are available at [www.esid.org/statistics.php](http://www.esid.org/statistics.php)

**Database  
Administration**  
[registry@esid.org](mailto:registry@esid.org)



**Fig. A** Distribution of patients in the ESiD Online Database by country. The absolute figures shown are the absolute numbers of patients per country.

## REGISTRY WP—Continuing



**Fig. B** Distribution of patients in the ESID Online Database by main PID category. The absolute figures shown are the absolute numbers of patients per category.

## SCT &amp; GT WP

Dear All,

I am writing this from the EBMT meeting in Goteborg where we have had a number of interesting sessions on transplant for PIDs.

1. Our regular business meeting was mainly dominated by discussions regarding the transplant guidelines. This is an important document, which although not prescriptive, is used by many centres to guide transplant protocols. We last revised these in 2004 and so we are going through the process again and hope to have a new set of guidelines later this year. I would like to take the opportunity to thank all those who have looked at the guidelines for the various different diseases and their input.
2. The WP session involved a number of talks on outcomes for different diseases. We heard about outcomes for XLP, ADA, SCID with radio sensitivity disorders and we also had two talks from Saudi Arabia where they have relatively large transplanted cohorts of children with Griscelli disease and SCID respectively. All these are retrospective outcome studies and although this is not ideal, given the rarity of the conditions we treat, we need this information in order to inform future practice. I hope all of these studies will lead to future publications and help with forming our guidelines and recommendations.
3. There is also a lot of activity in gene therapy for PID with European groups leading the way. Currently trials are ongoing for ADA in Milan and London and the Milan publication in NEJM in January was a big boost to the gene therapy field. Two trials are also due to start later this year. A new study for SCID-X1 will start in London and Paris and uses a self-inactivating retroviral vector which should avoid the adverse events seen in the previous LTR based retroviral studies. A new trial of gene therapy for Wiskott-Aldrich syndrome using a lentiviral construct is also scheduled for late 2009 and this is coordinated between London, Paris and Milan. These promise to be very exciting developments and may offer significant advantages for patients who lack well matched donors.

With best wishes,

**Bobby GASPAR**





## Working Party Reports

### ESID JUNIORS WP - Educational Day survey results

As you know in December 2008 we set up a web-survey to evaluate the Educational Day within the 2008 ESID meeting. The survey was successfully filled by 62 participants. The results of the survey were presented and discussed at the ESID board meeting in January 2009.



In summary, the survey clearly shows that the quality and selection of keynotes speakers was good and so was the general impression. Educational day is useful especially for communicating science and in clinical practice and the most important aspect of Educational day are the keynotes talks. The survey also shows that people think it is important to have activities targeted at students/juniors at large conferences and are interested to have career related events and local events organised by ESID juniors. A vast majority indicate that people want the Educational Day to be on the first day of the ESID conference. For a full report of the survey please go to [http://www.esid.org/downloads/Survey\\_Results.pdf](http://www.esid.org/downloads/Survey_Results.pdf).

### Meetings

Preparations for ESID Prague meeting and ESID Summer School 2009 have started. You have already received the announcements for these meetings. I hope to see many of you there and to have the chance to talk about our future initiatives. For more information about these meetings or other please go to <http://www.esid.org/symposia.php>

Any ideas or topics you want to discuss don't hesitate to send to us to be included in the program of these meetings.

In the end of May and most part of June I will be in UK, mainly Newcastle upon Tyne Hospital, to accomplish the first part of the Short terms program award, initiated by ESID juniors WP in 2008. Besides research and clinical activities we are trying to organise an ESIDjuniors WP here and other career related events. People interested to get involved in organisation please contact me as soon as possible.

### Travel grants

Two up to three travel grants will be awarded to attend the Prague Spring meeting. For more information about the application process please contact Rita Louro at [esid.admin@kenes.com](mailto:esid.admin@kenes.com), using the subject Prague Meeting-Travel Grants Application.

We would like to thank again to all the ESID board, especially our Treasurer Eleonora Gambineri and Chair of the Educational WP Andrew Cant and all the sponsors for their support in our activities.

### ESID newsletter

We want to thank Lucia Bianchi for the great job she has done taking care of the Young Researcher Corner and welcome Sara Ciullini Mannurita as the new person in charge for this section. I hope also to see more ESID juniors willing to play an active role in the Editorial board of ESID newsletter.

I wish you good luck in all your endeavours and I'm looking forward to hear from you soon!

Have a great spring!

**Crina SAMARGHITEAN**

## Interesting Papers

### Gene hunting season continues!

#### RNF168

The biological response to DNA double-strand breaks acts to preserve genome integrity. Individuals bearing inactivating mutations in components of this response exhibit clinical symptoms that include cellular radiosensitivity, immunodeficiency, and

cancer predisposition. The archetype for such disorders is Ataxia-Telangiectasia, caused by biallelic mutation in ATM, a central component of the DNA damage response. Here, the authors report that the ubiquitin ligase RNF168 is mutated in the

RIDDLE syndrome, a recently discovered immunodeficiency and radiosensitivity disorder, and demonstrate that RNF168 is recruited to sites of DNA damage by binding to ubiquitylated histone H2A (Stewart et al. Cell 2009 Feb 6, 136(3): 420-434).

## AK2

Reticular dysgenesis is an autosomal recessive form of human severe combined immunodeficiency characterized by an early differentiation arrest in the myeloid lineage, impaired lymphoid maturation and bilateral sensorineural deafness in affected newborns. Lagresle-Peyrou et al. identify biallelic mutations in AK2 (adenylate kinase 2) in seven individuals affected with reticular dysgenesis. These mutations result in absent or strongly decreased protein expression. Restoration of AK2 expression overcomes the neutrophil differentiation arrest, underlining its specific requirement in the development of a restricted set of hematopoietic lineages. AK2 is specifically expressed in the stria vascularis region of the inner ear, which provides an explanation of the sensorineural deafness in these individuals. These results identify a previously unknown mechanism

involved in regulation of hematopoietic cell differentiation and in one of the most severe human immunodeficiency syndromes (Lagresle-Peyrou et al. *Nat Genet* 2009, 41: 106-111).

Pannicke U et al. also show that the gene encoding the mitochondrial energy metabolism enzyme adenylate kinase 2 (AK2) is mutated in individuals with reticular dysgenesis. Knockdown of zebrafish ak2 also leads to aberrant leukocyte development, stressing the evolutionarily conserved role of AK2. These results provide in vivo evidence for AK2 selectivity in leukocyte differentiation and suggest that reticular dysgenesis is the first example of a human immunodeficiency syndrome that is causally linked to energy metabolism and can be classified as a mitochondriopathy (Pannicke U et al. *Nat Genet* 2009 41: 101-105).

## G6PC3

The main features of severe

congenital neutropenia are the onset of severe bacterial infections early in life, a paucity of mature neutrophils, and an increased risk of leukemia. In many patients, the genetic causes of severe congenital neutropenia are unknown. Boztug et al. performed genomewide genotyping and linkage analysis on two consanguineous pedigrees with a total of five children affected with severe congenital neutropenia. They found out that G6PC3, the candidate gene encoding glucose-6-phosphatase, catalytic subunit 3, have a homozygous missense mutation in exon 6 that abolished the enzymatic activity of glucose-6-phosphatase in all affected children in the two families. The authors also identified seven additional, unrelated patients who had severe congenital neutropenia with syndromic features and distinct biallelic mutations in G6PC3 (Boztug et al. *N Engl J Med.* 2009 Jan 1;360(1):3-5).

## Avenue for new therapeutics

The way in which bacteria that cause bacterial meningitis mimic human cells to evade the body's innate immune system has been revealed by researchers at the University of Oxford and Imperial College London. They looked at how one protein in the outside coat of *Neisseria meningitidis* enables the bacteria to avoid being attacked and killed by the complement system. The researchers found that the protein in the bacterial coat mimicked the sugars on the surface of human cells precisely, enabling the bacteria to bind factor H in the same way as human cells. The study could lead to the development of new vaccines that give better protection against meningitis B. (Schneider MC et al, *Nat.* 2009, Feb 18)

BTHS, a sometimes fatal di-

sease, is a serious genetic disorder occurring predominantly in males that leads to infection or heart failure in childhood. The new study shows the benefits of targeted intervention with an iPLA2-VIA inhibitor that prevents a major symptom of the disease- cardiolipin deficiency. (Malhotra A. et al. *PNAS* 2009 Feb 17).

Using a novel approach that combines a statistical tool that identifies genes interacting on the same biological pathways with highly automated gene-hunting techniques that scan the whole genome, an international team of researchers has discovered new genes involved in Crohn's disease. Crohn's disease is a chronic and painful condition caused by inflammation of the gastrointestinal

tract. The researchers say their approach broadens the power of gene discovery studies to ferret out potential targets for disease treatments. (Wang et al. *The American Journal of Human Genetics* 2009, 84: 1-7, March 13)

We are looking for volunteers to join the editorial board of the ESID newsletter for the Interesting Papers section. People interested, please contact me or the Editor in Chief, Esther de Vries, at [esid@estherdevries.nl](mailto:esid@estherdevries.nl).

Wish you a wonderful spring, good ideas and inspiration for many interesting and challenging papers!

**Crina SAMARGHITEAN**

**[Crina.Samarghitean@uta.fi](mailto:Crina.Samarghitean@uta.fi)**



## ESID SUMMER SCHOOL

### Primary Immunodeficiency Diseases 2-6 September 2009, Bled, Slovenia

The ESID Educational and Juniors Working Parties are pleased to announce its **7th Summer School for Primary Immunodeficiency Diseases!** This event, which has received very positive feedback from many young doctors in the past who found the course invaluable, will run from the 2-6 September 2009 in Bled, Slovenia!

For five days a well known international Faculty will be leading case-based discussions and give lecture presentations on primary immunodeficiencies. Andrew Cant, Helen Chapel, Jacques van Dongen, Anders Fasth, Steve Holland, Georg Holländer, Esther de Vries, Eleonora Gambineri and Crina Samarghitean, as the ESID Junior representative will be some of the names appearing in this year's Summer School panel.

If you are considering a career in immunology, this will be an excellent opportunity to update your knowledge from teachers at the forefront of clinical immunology, hear about the latest diagnostic tools and learn from leaders in the field who see and manage patients with the whole spectrum of primary immunodeficiency, while also networking with your colleagues.

#### A little scoop ...

The detailed Summer School programme will be finalized nearer the time, but we can already tell you that three of the main topics will be T-cell disorders, hypogammaglobulinemia in children and adults, and innate immunity.

#### Location

Slovenia has only one island but its uniqueness makes it more attractive than many an archipelago. Sheltered by picturesque mountains, the island reigns in the middle of an Alpine lake. Its charm has made it a symbol for centuries of a town to which guests from all over the world love to return. This town, which has already existed for a thousand years, is **BLED**.

#### How to apply

The official language of the Summer School is English and applicants must be ESID members. Each year the Society selects between 30 to 35 young doctors based on their background and interest in PID. For successful applicants ESID will cover accommodation, meals and onsite transfers.

For further information on the Summer School and how to apply please contact Rita Louro at [esid.admin@kenes.com](mailto:esid.admin@kenes.com) or visit <http://www.esid.org/home.php?sub=2&id=237>

**Application Deadline is 15 May 2009!**

*We are very fortunate to have received support from the following companies:*

**CSL Behring**  
Biotherapies for Life™

**Baxter**

**GRIFOLS**

**octapharma**

### ESID Educational Working Party '10,000 Euros' Scholarship

The scholarship will be awarded to an ESID member, who is a physician / scientist under specialist training - interested in pursuing a research project in the field of primary immunodeficiency.

The scholarship should be used for laboratory or clinical research work for at **least 6 months**.

The application should include a personal letter with a statement of career goals and plans on how to achieve those, a project plan, curriculum vitae, list of publications, a letter of invitation from the accepting institution, and a letter of support from the applicant's head of department or tutor.

*Please send your application not later than **May 30, 2009** by e-mail to: Rita Louro via e-mail: [Rlouro@kenes.com](mailto:Rlouro@kenes.com)*

**Andrew J CANT**, Chairman of the ESID Educational Working Party