



### Editorial Board

Eleonora Gambineri

Sara Ciullini Mannurita

Ales Janda

Gustavo Lazo

Nima Rezaei

Crina Samarghitean

ESID members are invited  
to publish in this newsletter



[treasurer@esid.org](mailto:treasurer@esid.org)

### In this issue

Letter from the editor (p. 1)

Note from the president (p. 3)

Secretarial report (p. 4)

Treasurer's report (p. 4)

ESID 2014-Prague (p. 5)

ESIDOnline  
Board elections (p. 9)

End of mandate letters (p. 21)

News & Views (p. 23)

News in images (p. 28)

Working Parties reports (p. 29)

Interesting papers (p. 31)

Young researcher's corner  
(p.33)

### Letter from the editor

Dear ESID members,

I hope you are enjoying the summer time!

This issue of our ESID Newsletter is very important since it will introduce you more in depth to the upcoming happenings related to the next ESID Conference in Istanbul.

Most of all, ESID board member elections! You will find all the details on the candidates and the electronic election that will take place before the meeting. You will also find the end-of-mandate notes from Gerhard Kindle, head of Registry WP and Andrew Cant, head of Educational WP.

Moreover you will see the results from the ESID Publication Award sponsored by PPTA. Many congratulations to the awardees!! Then Dr. Reem Elfeky, travel grant recipient for the APIDD Summer School in Australia, will share with us his exceptional experience on the event.

Finally you can see all the updates from the working parties' heads with the latest news on the ongoing activities of the society.

I would like to remind you that the Newsletter is always seeking for novelties and news. If you have interesting stories/life experiences/clinical topics/research activities or proposals to share with the community please contact me ([treasurer@esid.org](mailto:treasurer@esid.org))! The newsletter is a tool available to every member and any contribution you might have is very welcome!!

Enjoy reading and see you all in Istanbul!

**Eleonora Gambineri**  
Editor-in-Chief

#### NOT A MEMBER YET?

BECOME A MEMBER AND ENJOY THE FOLLOWING BENEFITS:

- **Reduced fee to the Istanbul ESID congress**
- **Reduced fee to FOCIS meetings**
- **Access to a network of PID**
- **Eligibility to receive awards**
- **Eligibility to participate in ESID PID Schools**
- **ESID Newsletter (quarterly)**
- **Access to ESID website members only area**

OCTOBER 6- 9, 2010



[WWW.ESID2010.ORG](http://WWW.ESID2010.ORG)

#### ESID Administrative Office



## A note from the President

---

Dear ESID members,



Time flies, and this is my last letter as President of ESID ! It has been a great privilege and honour to serve the ESID community as a President for four years. I had the difficult task to follow Luigi Notarangelo, who had been a wonderful President. And I am relieved to know that Amos Etzioni, the current President-elect, will pursue our efforts with imagination and ambition. During these four years, I had the luck to work with a fantastic group of people on the ESID board, including Bodo Grimbacher as secretary and Eleonora Gambineri as treasurer. What have been the important decisions made and implemented during this term? First and foremost, we have at last secured a democratic process: ESID decisions are now made by electronic voting and they are truly democratic. The best evidence was provided by the vivid discussions and clear votes revolving around the constitution draft. The whole process took place in the open sky and the results of the vote were implemented in the final constitution. Indeed, our second major achievement is the ESID constitution. Our third important decision was to solicit a company to externalize and run the ESID meetings. We selected Kenes after having rigorously compared various offers. The first months have been very promising, and the organization of the Florence meeting between Eleonora Gambineri and Kenes is going very well. Besides these radical changes, the ESID board has attempted to pursue the development of ESID by promoting its core values: collaboration between centres instead of competition and confrontation, bedside to bench and bench to bedside research in the field of PIDs, awareness of PIDs both in the medical community and among lay people, open and friendly collaborations with our colleagues in Africa, Asia, Latin America, North America, and Oceania. It was especially rewarding to watch the birth of the Latin American Society for Immunodeficiency (LASID) and the African Society for Immunodeficiency (ASID). We hope similar societies can emerge in North America and the various regions of Asia, as I am sure we will strive with them to build a Federation of continental societies. But this is a dream and it will be one of the missions of Amos Etzioni!

I would like to close by thanking you all warmly for your trust over the years. If anything worth was done in my term as a President, it was so thanks to your generous support and endorsement!

Finally, as you know, the Istanbul meeting this autumn will be a huge success. I invite you all to spread the word and thank Necil Kutukculer and his team, who are doing a superb job!

I very much look forward to seeing you all in Istanbul!

All best wishes,

**Jean-Laurent Casanova, MD, PhD**

ESID, President 2006-2010

## Secretary's report

---



We have started the final countdown to the next ESID biennial meeting in Istanbul this October 2010! With a high quality scientific programme, this meeting is expected to gather participants from all over the world. We hope to see many of our ESID members there to take part in our society's most important event!

As you know, ESID members benefit from a reduced registration fee to the biennial meeting. This year we have also decided to offer 10 travel grants to Junior members who wish to attend the meeting. The deadline for applications was July 1, and we have received many. ESID will select the 10 successful awardees who will be notified during summer time.

We are also very happy to announce that our list of countries eligible to receive reduced membership to ESID has been updated! This list is based on the World Bank Countries Classification reviewed every year. ESID reduced membership list incorporates all low, lower-middle and upper-middle income countries. Details can be found in the membership section of the ESID website.

Moreover ESID membership is growing every month and we have now reached more than 500 members! We expect that the community of PID professionals will keep developing and that ESID continues to offer activities and benefits that follow the needs and interests of our members!

Most importantly, we would like to remind you that we will have **Board elections** this year and that your participation in the process is vital for the future of our society. Further in this document you will find the list of candidacies, and respective applications, in preparation for the online voting that will open on September 8<sup>th</sup>.

As a last note: Bids for biennial meetings take place four years in advance, therefore we have for the past few months been calling for applications. We have been approached by a few countries although we have only received so far one formal application from Prague. Please contact Rita Louro at Kenes [rlouro@kenes.com](mailto:rlouro@kenes.com) if you are interested to host the ESID 2014 meeting.

Please see all submitted applications further in this document

### Bodo Grimbacher

ESID Secretary

## Treasurer report

---

Dear ESID members,



I am pleased to announce that the membership campaign promoted by Kenes is very fruitful indeed. We have already increased the number of ESID members by about 25% as compared to the same time 2 years ago!! And I am sure that more will join the society soon at the upcoming meeting in Istanbul! However, if you haven't renewed your membership yet, and you wish to do so...you are still on time! Please visit the ESID website ([www.esid.org](http://www.esid.org)) and follow the instructions.

I would also like to highlight the ongoing vibrant activity to support our young trainees. Baxter is kindly supporting a fellowship award, which is meant to sustain the development of academic clinical immunology research careers of advanced fellows-in-training who have demonstrated a commitment to the study of primary immunodeficiency. Moreover, for this year our budget will enable us to support short-term and long-term fellowships for young physicians/scientists to enable the exchange and the collaboration among different countries!

So keep joining the community!

Best wishes

**Eleonora Gambineri**

ESID Treasurer



## ESID 2014

### Received application – Prague 2014

On behalf of Czech members of ESID, with support of the Czech Immunology Society (ČIS) and Czech Society for Allergy and Clinical Immunology (ČSAKI) we are applying for being the host of the ESID congress in 2014.

While Prague has become well known destination for the small and educational ESID Prague Spring Meeting during the last decade, it has never hosted the ESID biennial meeting.

With the very traditional and prestigious history of Prague, its oldest University in the Central Europe (founded by Emperor Charles IV in 1348), famous Prague Immunology School tradition, Prague represents an ideal location for bringing together immunologists and other specialists in the field for a fruitful meeting.

We would appreciate very much if ESID would consider our invitation to meet at the heart of Europe.



### **Prague Immunology School**

The Czech Immunological Society started its existence in sixties and seventies as informal immunological centre known later as the Czech (Prague) Immunological School represented by Prof. Milan Hasek and Prof. Jaroslav Sterzl. Prof. Hasek's work brought him close to the discovery of immunological tolerance, a phenomenon later credited by the Nobel Prize to P. Medawar and M. Burnet. Substantial contribution of Milan Hasek's group was reflected in anniversary article in Nature Immunology 2003, on an occasion of 50 years since the original Hasek's publication (Ivanyi J. Milan Hasek and the discovery of immunological tolerance. Nat Rev Immunol. 2003, 591-7). Jaroslav Sterzl was a close and dear friend with Robert Good who frequently travelled behind the iron curtain and brought immunology and immunodeficiencies in particular to former Czechoslovakia. Czech immunologists also greatly contributed to the development of mouse models for immunodeficiencies. Nude mice lacking thymus were discovered for immune studies and were first bred by Miroslav Holub (1923-1998), a distinguished Czech immunologist and a renowned poet (his poem Brief reflection on Accuracy, very actual thought on scientific methods, is attached to this application).

Another contribution to the world immunology with Czech origins is represented by Jan Vilček, Czech scientist later working at New York University School of Medicine. His team studied cytokines which led to the development of Infliximab, a monoclonal antibody against tumor necrosis factor widely used in the clinical practice.

### **Prague and ESID**

The constitution of ESID as European activity was well known and welcomed in former Czechoslovakia. A number of leading clinical immunologists became members of the society and benefited from ESID activities. Since 2001 annual ESID Prague Spring meeting is organized and over the decade that event became a respected ESID platform for its junior members (<http://imunologie.lf2.cuni.cz/en/esid-workshop.php>).

### **Prague: a city and congress venue**

Prague – the capital of the Czech Republic situated on both banks of the Vltava river is a beautiful city with a rich history. In the course of its thousand-year history, Prague has always been the political, cultural, and business centre of the country. Prague, often called “Golden” or “Hundred-spined”, belongs to the architecturally unique European towns, attractive for tourists from around the world. Visitors find themselves enjoying a living museum of European architecture from Romanesque time to the present.

Prague has 1,200,000 inhabitants and stretches over approximately 500 square kilometres. The dominant feature of the city is Prague Castle, which houses the gothic St. Vitus's Cathedral. The castle had been the seat of Czech kings since 1087, until 1918 when it became the seat of presidents of the Czechoslovak Republic, and since 1993 it has been the seat of the president of the Czech Republic. Prague has one of the oldest universities in Europe; the Charles University which was founded in 1348.

With a widely opened pro-market economy, stable democratic government, stable currency and well-known working skills of the people, this country provides a great opportunity to host a conference in order to meet scientists and business people from all over the world.

### **Congress venue**

#### **Visa**

The Czech Republic became part of the European Union (EU) as of May 1, 2004. Furthermore, as of December 21st, 2007, the Czech Republic became part of the Schengen Area. For visa requirements see:

[http://www.learn4good.com/travel/czech\\_republic\\_visa.htm#req](http://www.learn4good.com/travel/czech_republic_visa.htm#req)

So far there are no major overlaps with other events foreseen in 2014. Potential dates are therefore October 8.-11., or alternatively 15.-18. 10. 2014, covering period from Wednesday to Sunday.

**Travel**

Located in a heart of Europe, Prague is a very comfortable destination covered by a number of major airlines, connecting Prague by non-stop flights with many European destinations. Prague airport report flights to/from 130 destinations in 51 countries and covers traffic of roughly 12 millions of passengers a year. Travel by bus and train is also available and advantageous for some nearby countries.

**Accommodation**

Prague has large capacities in all types of hotels. Delegates can choose different styles from luxury five- star hotels to three-star hotels (or two stars hotels and hostels). All of the suggested hotels are easily accessible – at most 5 - 20 minutes by underground, bus or tram ride away from the Prague major Congress Centre and/or other potential congress sites.

**Congress facilities**

One of the possible venues, Prague Congress Centre is one of the modern architectural landmarks of Prague and the largest convention and culture centre in the Czech Republic. In Congress Centre, there are 8 main halls (total capacity up to 5,000 people) and 30 smaller meeting halls (total capacity up to 1,500 people). Other smaller sites such as Clarion congress and hotel and others are available for 2014 event.

**Dates**

So far there are no major overlaps with other events foreseen in 2014. Potential dates are therefore October 8.-11., or alternatively 15.-18. 10. 2014, covering period from Wednesday to Sunday.

**ESID Prague 2014 programme outline**

ESID/INGID/IPOPI educational day

Disorders of innate immunity

Cytokine disorders

B cells disorders

T cells disorders

Immune regulation disorders

New and emerging diseases

Diagnostic and laboratory section

Vaccination and new methodologies

Novel treatments, gene therapy

Optional - Invited ESID guests – European Society of Human Genetics (prof.Milan Macek from Prague is president-elect of ESHG, active in 2014)

**Scientific committees:**

ESID board nominees

ESID Prague Spring Meeting invited guests 2002-2010

Local committee: ESID members Anna Sediva, Jiri Litzman and members of their groups, representants of Czech Immunological Societies.

**Social events**

Opening ceremony on site or in other representative buildings/palaces in Prague center  
Program for accompanying persons

Visit of the concert of popular or classical music

Prague Autumn Classical Music Festival

Jazz, Night Jazz clubs

Ballet or opera performance in State Opera

Sightseeing tours (half or whole day) - Prague Castle and the Royal Garden, Prague's Old Town and Jewish Ghetto, Art Nouveau Prague, Mozart Prague, nearby castles, Karlovy Vary, Brno and George Mendel Monastery

Sport events, active sport sites – golf, tennis, outdoor activities, walks

**Poem**

Brief reflection on accuracy  
BY MIROSLAV HOLUB

*Fish*

*always accurately know where to move and when,  
and likewise  
birds have an accurate built-in time sense  
and orientation.*

*Humanity, however,  
lacking such instincts resorts to scientific  
research. Its nature is illustrated by the following  
occurrence.*

*A certain soldier  
had to fire a cannon at six o'clock sharp every evening.  
Being a soldier he did so. When his accuracy was  
investigated he explained:*

*I go by  
the absolutely accurate chronometer in the window  
of the clockmaker down in the city. Every day at seventeen  
forty-five I set my watch by it and  
climb the hill where my cannon stands ready.  
At seventeen fifty-nine precisely I step up to the cannon  
and at eighteen hours sharp I fire.*

*And it was clear  
that this method of firing was absolutely accurate.  
All that was left was to check that chronometer. So  
the clockmaker down in the city was questioned about  
his instrument's accuracy.*

*Oh, said the clockmaker,  
this is one of the most accurate instruments ever. Just imagine,  
for many years now a cannon has been fired at six o'clock sharp.  
And every day I look at this chronometer  
and always it shows exactly six.*

*Chronometers tick and cannon boom.*

"Brief reflection on accuracy" from Poems Before and After by Miroslav Holub.  
Published in 2006 by Bloodaxe Books.

[www.bloodaxebooks.com](http://www.bloodaxebooks.com)



## ESID - 2010 Online Board Elections

Online voting opens on September 8<sup>th</sup> and closes on October 8<sup>th</sup> at 12noon CET

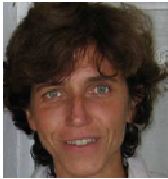
As you know we will need to elect or renew the term of office of the members of our Executive Board. Some of these members will integrate the ESID Board for the first time, whereas others are entitled to a second term of office and thus wish to continue to serve the society.

Although, the online voting will go live on the ESID website [www.esid.org](http://www.esid.org) on **September 8th 2010** - one month prior to our biennial congress -, and will be closed on **October 8th 2010** at 12noon CET, we would like to invite you in the meantime to take a look at the applications of our candidates.

### *First Mandates*

#### **Applications for Secretary: Anna Villa and Christoph Klein**

Applicant: **Anna Villa**



My name is Anna Villa and I would like to candidate myself for the position of the next ESID Secretary. I am MD researcher (since 1986) and I have spent many years in the research field of primary immunodeficiencies. In particular, I have focused my studies on the molecular and cellular bases of Omenn syndrome and more recently I have addressed my analysis to the cellular bases and gene therapy of Wiskott Aldrich syndrome. Probably some of you had the opportunity to know my work on the molecular dissection of the pathogenesis of another rare disease, the Autosomal Recessive Osteopetrosis, which is treated in many transplant centres.

I am Senior Scientist at National Council of Research and Head of Unit at the Telethon Institute for Gene Therapy (San Raffaele Hospital in Milan, Italy). In the past, from 2002 until 2006, I have been an active member of ESID board as Chairman of the Genetic Working Party. During that period, I coordinated the European study on the genotype-phenotype correlation in Autosomal Recessive Osteopetrosis, analysis now concluded with the precious help of Despina Moshous and Ansgar Schultz in the framework of EBMT. After four years of serving ESID, I decided to step down to allow new candidates to bring novel ideas that can improve the society. Indeed ESID has had an important role in my scientific life. I am aware that most of my studies are the results of collaborations arisen within the ESID society. I am very grateful to the familiar atmosphere that I have always found among ESID members. Thanks to this network of collaborations, I am now in contact with various Transplant and PID centres all around Europe. Since I am involved in basic science but at the same time I am in close contact with clinicians and bone marrow centres, I do not have any work relations with companies that could cause conflict of interest. Thus I will be able to contact Sponsors without being influenced by any personal interest.

Finally, I would like that our Society could be closer to our scientific and professional life. ESID Newsletter is a great communication tool but it often arrives too late on our desks. I think that many of you would like to know ESID initiatives in advance. I propose to start a monthly update of ESID activities by sending an e-mail describing the new main activities to the ESID members. I think that this correspondence might improve and at the same time make closer the ESID board to the members. ESID is not only a society of scientists and physicians but it is also a place where good friends discuss about science and medicine.

**Anna Villa**

Applicant: **Christoph Klein**

Dear ESID members,

I would like to serve ESID as secretary of the society for the next election period.

My scientific interests are in the areas innate immunity, in particular in congenital neutropenia and selected autoaggressive disorders. Furthermore, my group is involved in developing novel therapeutic strategies.

I am currently the coordinator of the German Primary Immunodeficiency Network and of a European Research Network for neutrophil disorders. These interdisciplinary and international networks are closely linked to many academic centers in numerous emerging countries.

I am convinced that the future of our society will greatly benefit from a strong scientific focus, associating international and interdisciplinary research activities. This effort should include not only a strong representation of European members but also of members in adjacent geographic areas. Furthermore, ESID may benefit from state-of-the-art communication platforms by further developing its web-based presence.

In a clinical perspective, I am sure that our focus in inherited disorders of immunity will teach us many lessons applicable to a wide array of inflammatory and malignant disorders. ESID has been instrumental in promoting science and clinical care for patients with primary immunodeficiency disorders – and I would be honored if elected to assist the future president of ESID in programmatic issues and to further advance science and clinical care for our patients.

Kind regards,

**Christoph Klein MD PhD**

Professor of Pediatrics

Chair, Department of Pediatric Hematology/Oncology

### **Applications for Educational Working Party Chairperson: Esther de Vries and Laszlo Marodi**



Applicant : **Esther de Vries**

Dear ESID members,

Having been ESID Treasurer for 8 years, Chief Editor of the ESID Newsletter for 9 years, and President of the ESID biennial meeting in 's-Hertogenbosch in 2008, I know many of you well, and many of you know me!

Besides that, I have also been active as Faculty Member of the ESID Summer Schools since 2003, and this was one of the ESID activities I actually liked most. It is very inspiring to spend nearly a week with young clinicians and researchers in immunodeficiency, all of them eager to learn and good company as well. Coming from so many different backgrounds inside and outside Europe, they have taught me a lot as well during those years.

I started taking care of just the opening session, introducing the ESID diagnostic protocol for suspected immunodeficiency, ourselves, and the students to each other, and generally 'breaking the ice'. I still do that, but have through the years taken more tasks upon my shoulders, such as selecting the applicants, helping selected students improving their case presentation, and – in Bled – selecting and negotiating about the venue and gala dinner site, helped by Eleonora Gambineri, our current Treasurer.

Sponsors are very important for the success of the ESID Summer Schools, and I have shown with the 's-Hertogenbosch meeting that I can handle that too. I think I am a good choice to take over from Andrew Cant, our current Educational WP Chairman, and look forward to the chance of continuing his work.

I sincerely hope you will entrust me with your vote as Chairman of the ESID Educational Working Party!

My best regards,

**Esther de Vries**

Applicant : **Laszlo Marodi**

Dear All,

the purpose of this letter is to provide the ESID Board, ESID members, and Kenes with the information about my decision to apply for the chairmanship of the Educational WP in the next 2 years after the Istanbul meeting.

Having been developing an efficient physician education network (The J Project) over the past five years in Central and Eastern Europe, and starting with a similar project as the head of the PID in Development WP in Western EU countries, I feel that the Educational WP would provide an appropriate frame as well as a new challenge for me to continue further my activities and work to serve ESID.

We have now at least five separate activities in Europe which are focusing on education of physicians and scientists to better understand the importance of PID patient care and research in clinical medicine. These are as follows:

- 1) ESID Educational WP (ESID Summer School and ESID Meeting Educational Day program set up are the major activities).
- 2) ESID PID in Development WP (Organizes Awareness Meetings focusing on different regions of "Western EU" countries).
- 3) Regular Prague Spring meetings (Organized always in Prague).
- 4) Winter PID meetings in Poland (Organized always in Poland).
- 5) J Project meetings (focuses mostly on East European countries and organized by the ECE IPI CTR in Debrecen).

I think these activities should be better coordinated and harmonized under the auspices of ESID in terms of programs, funding, and publicity. Importantly, such an European-wide, harmonized educational activity should be developed together with the involvement and partnership of the Jeffrey Modell foundation, the IPOPI, and INGID, as well as EFIS and national immunology societies in Europe. Feed-back about each educational activity from our membership and from outside ESID should provide the tool to further develop or modify individual activities and programs. Such a coordinated educational network would further strengthen ESID, as a Society, and should improve PID patient care all over Europe along with an increased activity in PID clinical research.

**László Maródi**

## Genetics Working Party Chairperson: Bodo Grimbacher and Capucine Piccard



Applicant : **Bodo Grimbacher**

Dear colleagues and ESID members,

With this application I would like to ask you to support my bid to become the Head of the ESID Genetics Working Party starting in October 2010.

Being interested in the genetics of primary immunodeficiencies since my postdoc period at NIH in Jennifer Puck's laboratory, I feel however, that the ESID Genetics Working Party would be a good place to place my future ESID activities. I would like to suggest for the years to come to:

1. To bring people together.

Currently many laboratories across Europe and the wider ESID community which actually spans across the whole world carry out genetic testing in primary immunodeficiency patients. My last count was that there are more than 160 genes involved in Mendelian traits causing PID. The concept how centres tackle this testing is very diverse. Some centres provide mutation detection for many different conditions; others focus on more specific traits they know. In the next term I would like to bring together interest groups on

mutation detection in PID and also people interested in specific genes to improve the quality of mutation detection and especially mutation reporting within ESID groups.

Why do I think that this is foremost importance? I had diagnosed a patient with STAT3 deficiency because we detected a missense mutation in STAT3 which we did not find in 200 controls and which also has not been reported in any database. The computer modelling assigned this change a "possibly damaging". Several months later though, we found in this patient an even more profound genetic deletion in Dock8.

These things will never be published, however we all can learn from these experiences. This is why sharing information is so important in the field.

2. To provide ESID members over the ESID website with a listing of where you can get genetic testing.

In recent years I searched for collaborators capable of competently evaluating for me genes like APECED or EVER1&2. I am very well networked, so I found good laboratories, but it was hard. From this experience I conclude that ESID should offer a listing of where to get genetic testing done. This shall be an inclusive approach giving a good and as complete as possible overview on all interested laboratories involved in genetic testing, this shall not be an approach to support selected centres.

I am aware of the Finnish database IDiagnostics curated by Mauno Vihinen, and I think that the Genetics Working Party should make more use of this resource.

This list may also be linked to the ESID registry and the current IPID efforts of the Clinical Working Party.

3. Develop guidelines for gene-specific testing and counselling.

Genetic testing and counselling are subject to local laws and regulations. Some laboratories are accredited, others are not. Therefore, for the time being, guidelines need to be locally implemented. In some countries and in some institutions there are already guidelines in place and ESID may be able to make these available to centres who are still in search for them or struggling to implement them. I hope that through the ESID genetics Working Party, ESID can give some advice as of how to do this. This will again require a lot of work and a lot of help from the ESID community is needed, but I am sure we can all learn from each other.

Warmest regards,  
**Bodo Grimbacher**

Applicant : **Capucine Piccard**

Dear colleagues,

I write to solicit your votes during the upcoming election to the chair of the genetics working party on the ESID board. It would be for me an honor to serve ESID as I strongly believe in European collaboration to foster care, research and training for primary immunodeficiencies (PIDs).

Since November 2005, I am the head of the 'study center of immunodeficiencies' (CEDI) at the Necker-Enfants Malades hospital in Paris. This diagnostic laboratory is responsible for the immunological and genetic investigation of over 80 PIDs. Through this activity, I have established numerous collaborations in Europe and beyond, some of which have led to the description of new PIDs, such as the genetic defect in STIM1 (New England Journal of Medicine, 2009). I have also a weekly PID clinic consultation within Alain Fisher's Pediatric Hematology-Immunology department at the Hospital Necker-Enfants Malades, where I had served earlier for three years as a clinical fellow (2002 to 2005).

My main research activity follows up on my Ph.D. which I did in the Laboratory of Human Genetics of Infectious Diseases, at the Necker Medical School in Paris, under the supervision of Jean-Laurent Casanova (2000-2004). In the course of my PhD, I discovered the first human PID affecting the TLR and IL-1R signaling pathway (Science 2003). I have since been leading a research group in the laboratory, exploring new PIDs predisposing to infections caused by pyogenic bacteria. We described the genetic, immunological, and clinical features of various defects of NF-kB-mediated immunity. The most significant contributions included the discovery of MyD88 deficiency (Science in 2008) and the through immunological and clinical description of IRAK-4 deficiency (JEM 2007 and Medicine 2010).

I think that my expertise in clinical immunology, in biological immunology, and in molecular genetics of PIDs, provide me with a solid background to take responsibility of the ESID chair of the genetic working party. In the context of the technological advances in genomics, it is an exciting challenge to develop these applications to diagnosis of and research on PIDs.

Thank you for your attention to my candidacy.

Kind regards,

**Capucine Piccard, MD, PhD**

Head, Lab of CEDI

Research Investigator, Lab of HGID

Consultant, UIH

**PID Care in Development Chairperson: Raffaele Badolato and Anna Sediva**

Applicant: **Anna Sediva**

Dear ESID members,

I have been working as a clinical immunologist in Prague since 1995. Before that I trained in pediatrics from 1985 to 1995 and I have carried out research in pediatrics and immunology in Prague, Czech Republic and in NY, USA {1992; 1995}. Currently I have a particular interest in Di George syndrome and I have a passionate commitment to teaching and training the next generation of Immunologists, and with this in mind have, for the last 10 years, organized the very successful Prague Immunology Spring School. This has given an opportunity for young clinicians and laboratory scientist to meet and interact with eminent immunologists from around the world. In particular the themes of recent summer schools have been B cells in primary immunodeficiencies and innate immunity including autoinflammatory disorders. Sited in Eastern Europe the Prague Spring School has meant that many young doctors from Eastern Europe

If elected to the ESID board I would be very keen to further develop opportunities for learning and exchange of ideas for young immunologists from less favoured countries, in Europe and beyond. I would particularly stress the importance of attracting young immunologist to the field of primary immunodeficiencies. I would like to extend the opportunities to join summer schools for medical graduate students and give them the chance to pursue the PhD in our field.

Best Regards,

Anna Sediva, M.D., PhD.  
Institute of Immunology,  
University Hospital Motol  
V uvalu 84  
Prague 5, 150 06  
Czech Republic



Applicant : **Raffaele Badolato**

Dear Friends and Colleagues,

We have assisted in the last few years to a rapid increase of the awareness of Primary immunodeficiencies in European countries. The PID care in Development working party has provided the occasion to improve patient care and access to appropriate diagnostic procedures and genetic testing for many PID patients living in European countries. These achievements offer the concrete basis for the follow-up work that this working party should carry out in the next future.

I believe that we need to move the next step in the accomplishment of standard of care of PID in every European country. I am confident that our goal for the next years will be to achieve availability of genetic testing and of the most suitable treatment options for patients suffering of PID all over Europe. These objectives will require well-built interactions of the PID care in Development WP with the all the other working parties and with the President to achieve full realization of this plan.

My past work at the Pediatric Immunology Unit of University of Brescia provided a strong expertise in the implementation of genetic testing for PID, in the primary care of patients and in the establishment of national and international working groups. For all these reasons I am submitting my application to serve as Chairman of PID care in Development working party.

I have no doubt that we can achieve these goals, but the joined efforts of every ESID member will be required. Should I be elected, it is my intention to put the maximal endeavor to this purpose.

Yours sincerely,

**Raffaele Badolato, MD, PhD**

## Registry Working Party Chairperson: Stephan Ehl

Applicant : **Stephan Ehl**

Dear colleagues,

I would like to apply for the position as Chairman of the ESID Registry working party.

I am a Pediatric Immunologist from Freiburg, Germany and I am currently serving as the scientific director of the newly founded "Centre of Chronic Immunodeficiency" (CCI). My specific interest is immunodeficiencies associated with impaired immune regulation, in particular FHL, ALPS and combined immunodeficiencies, in particular including "leaky" SCID.

The establishment of the ESID registry has been a formidable achievement and the yearly growth of the database is a success for the initiators of this registry, the patients and the treating physicians. However, I think that the potential of this registry as a scientific tool has not yet been fully explored. In my opinion this can only be achieved, if the maxim is "scientific question first". This means that each data entry must be justified by a sound scientific question, that has been evaluated from clinical, immunological epidemiological and biostatistical viewpoints. Reducing overall data entries and directly connecting the entries to specific studies should increase motivation for full registration without compromising the excellent service of the registry as an overall PID documentation platform.

For a time as head of the Registry working party, I would propose to remodel the database for combined immunodeficiencies as a model project following these principles. Experience with this platform will benefit other disorders. These efforts would be coordinated with our colleagues from North American, in particular with Gigi Notarangelo, in order to assure common standards and definitions. Preparatory efforts have already been made for a "leaky" SCID registry. The integration of Gerhard Kindle (the current head of the registry) and Benjamin Gathmann into the CCI will help to realize this endeavour here in Freiburg.

Beste Grüße

**Prof. Dr. Stephan Ehl**  
Wissenschaftlicher Direktor

## ESID Junior Working Party Chairperson: Marta Rizzi )

Applicant: **Marta Rizzi**



My name is Marta Rizzi, I am an Italian medical doctor currently working as a PostDoc in Freiburg, Germany, in the Center for Chronic Immune deficiency. I would like to candidate to become the next ESID Junior WP representative.

My interest in primary immunodeficiency started in Freiburg, during the last year of residency in Immunology. In Freiburg, I have found a very lively scientific and clinical environment where very competent and motivated doctors take care of patients and creative researchers try to better understand the immune mechanisms of PIDs.

I had the chance to participate to the ESID summer school in 2009 in Bled (Slovenia) and to the Florence workshop in January 2010. In both occasions, I appreciated the spirit that animates the ESID Junior working party. Lively and open discussion, direct interaction with senior faculties, and the enthusiasm in exchanging experiences. There I decided to get more involved in the Junior working party, and to actively give my contribution to ESID.

As chair of the Junior WP in ESID I would like to:

Promote and expand mentoring of juniors interested in primary immunodeficiency

Many of us are not only interested in PID, but also trying to develop an academic or clinical career. In the field of PID are many excellent senior medical doctors and scientist, a real treasure of experiences. I intend to promote formal and informal occasions in which the juniors can meet the seniors and discuss, ask advice, support, and direction for their future career. More in concrete, I would propose to insert a 'mentoring session' in the ESID summer school, and following the example of Eleonora Gambineri, organize a second workshop on PIDs.

Improve juniors networking

ESID already offers short-term fellowships, but also single Institutions offer guest positions for short, middle and long term stays. I want to promote a better advertisement of the positions available and a better exchange of information of what can be done and experienced in each center, so that the juniors can expand their network and their career opportunities. Furthermore, dedicated ESID juniors informal meeting should be organized at each congress where the juniors are present.

Help in career development

List of grant, prize, fellowships available for juniors in PID, to be listed in our website.

More space to junior PID researchers:

Many of our juniors are not clinicians, but researchers working in the lab. The ESID summer school is at the moment very clinically oriented, even if includes all the latest in research. I would like to organize a junior dedicated research meeting where the junior researchers in PID will have the chance to present their data and discuss them within the pair group and with senior faculties.

Fund raising for ESID junior activities

I will try to get companies involved in support our initiatives, in particular to finance a new workshop.

Support of family friendly career development.

In PID we have lots of good example of how a brilliant career can be coupled with the family. I would like to bring the experience of the faculty closer to the juniors, to better support and promote successful family and career development of both male and female clinicians and researchers.

I hope that the ESID Junior WP will become more and more a useful and used instrument for all the young clinicians and researchers willing to pursue their career in PID.

Best Regards,

**Marta Rizzi**



### ***Renewal of mandate letters***

#### **SCT/GT Working Party Chairperson: Bobby Gaspar**

Applicant : **Bobby Gaspar**

Since election to this post in two years ago, the main achievements of the BMT/Gene therapy and Inborn Errors WP have been to:

1. Establish retrospective data on outcome of HSCT for specific immune deficiencies including LAD, Cartilage Hair Hypoplasia, XLP
2. Update the outcome from the SCETIDE database
3. Initiate new studies for PNP, Reticular dysgenesis, DNA repair defects
4. Revise the EBMT guidelines for HSCT in severe immunodeficiencies
5. Organise two highly successful Working Party meetings in Sirmione, Italy and Cambridge, England
6. Forge links with US transplant physicians with the aim of conducting collaborative studies

I ask your support for re-election to the post of Chair for the following reasons:

I aim to complete retrospective data analysis for all forms of SCID and severe immunodeficiencies on the basis of underlying molecular defects;

I aim to disseminate the EBMT guidelines more widely;

I aim to encourage further discussion on the use of HSCT and gene therapy and to establish guidelines for their use for specific immunodeficiencies;

I aim to create better links with the Paediatric Working Party of EBMT to help establish paediatric specific education on HSCT;

I aim to revise the IEWP database/registry forms so that these are now brought up to date;

I aim to create greater links with the US transplant group and conduct collaborative studies for specific diseases;

I aim to encourage greater participation from transplant centres worldwide so that our WP is recognised as the major source of information and guidance on HSCT for severe immunodeficiencies;

I thank you for your support!

Kind regards,  
**Bobby Gaspar**

#### **Treasurer: Eleonora Gambineri**

Applicant : **Eleonora Gambineri**

Dear all,

I am coming to the end of my first term as Treasurer, and I would like to give you an update on my activity.

I remember that when I was standing for election in 2008 I was a bit nervous but also excited by the idea of being Treasurer. Managing money and getting everything organized is a great responsibility and takes a lot of effort, but most of all needs a lot of dedication on top of the everyday work. Nevertheless, I must say that these past 2 years have been very challenging, stimulating and exciting at the same time and I have enjoyed fully.



I had the opportunity to interact with many of you for all sort of different matters. It is really fulfilling to meet or have a word with lots of people from different countries that, like me, share this great interest and commitment to the field of PID.

Given my background as previous head of the ESID Junior WP, one of my aims was to try to solidify/increase the ESID budget to support financially activities designed to promote the education and the professional growth of young trainees. I am glad that during the past 2 years new activities have been developed, as the newly established "ESID winter workshops". The ESID junior Educational workshop, held in Florence, Italy during the last winter has been a unique opportunity to bring together previous ESID Summer School attendees and young trainees from different countries. A two-days of lectures on different interesting topics followed by vibrant discussions...a great combination to reinforce the network! This new event gave me the chance to approach new sponsors (i.e. Kedrion) interested in the society and willing to support further activities. New sponsors mean more money to invest in the future generations! Consequentially, it was also possible to develop more travel grant and fellowship opportunities for trainees.

Finally, also with the valuable support of Kenes, the number of members has increased of about 25% compare to last year! A great achievement! This indicates that the society is growing and will be facing new important challenges!

It was an honour for me to serve ESID as a Treasurer in the past two year. It has been an exceptional experience and I have learned a lot, but mostly I grew up together with the society. I would love to face the new challenges and to promote the future of this excellent organization.

I am very motivated to continue my work with enthusiasm and commitment and I will do my best to accomplish my duties. Thus, I hope I can count once again on your vote and your support for the upcoming elections!

**Eleonora Gambineri**  
ESID Treasurer

### Clinical Working Party Chairperson: Klaus Warnatz



Applicant: **Klaus Warnatz**

Dear colleagues,

When I applied nearly two years ago, I had expressed the following 5 goals.

- 1) Developing a platform for immunophenotyping of primary immunodeficiency (IPID), as I had announced in the ESID newsletter 2/2008. This platform is meant to serve as a reference website to look up typical changes of immune phenotype associated with certain primary immunodeficiency syndromes.
- 2) Defining, together with you, the most urgent clinical questions in the different fields of immunodeficiency and setting up working groups to develop specific recommendations.
- 3) A special interest of mine has always been the coexistence of immunodeficiency and autoimmunity. At this time, there are no satisfactory, let alone evidence-based, recommendations for immunosuppressive therapies for most immunodeficiency syndromes. In the first phase, current disease specific treatment protocols will be collected from participating centres, which subsequently will build the basis for clinical trials in selected disorders..

- 4) Encouraging and supporting initiatives from you, which will serve the aim of improving diagnosis and treatment of PID patients.
- 5) Evaluating the possibility with the ESID board, and especially the future treasurer, for creating a funding system by the ESID, allowing for small grants to initiate clinical multi-centre trials, in order to achieve the goals mentioned above

Looking back, I feel that I have accomplished only some of these aims. IPDnet.org has been launched. It is still growing and thanks to the help of several of you (I would like to especially mention Kimberly Gilmour, Mirjam van der Burg, Toni Bonilla, Thanks!) it is taking shape and I hope it will be helpful to many of you.

I sent out a survey last year within one of the newsletters in order to define the most burning clinical questions. The very poor response made me realize that this is not the way to develop answers to these questions. I am very happy for suggestions as to how to improve. Meanwhile, I was pleased to support the survey by Sergio Rosenzweig and Nima Rezaei on BCGitis and Esther de Vries's call for an update on the ESID diagnostic protocol on PIDD.

We have initiated a multi-centre survey to retrospectively analyze the use and outcome of immunosuppressive therapy in CVID and I have just uploaded a retrospective analysis of the outcome of splenectomy in patients with CVID.

Since I still believe in the necessity of good clinical trials (retrospective as well as prospective) I would like to continue to encourage and support your initiatives in this field. This is essential and due to the rare disease character requires the collaboration between several centres, which can easily be facilitated by the Clinical WP of the ESID. We have not implemented a funding system by the ESID for such initiatives. Therefore, I will continue working on this opportunity with the new board in case I am re-elected. The other major goal will be the update and new development of diagnostic protocols and recommendations for various PIDD. This call has already been stated in the last newsletter and will be the ongoing focus of the coming years.

Please feel free to contact me with questions, suggestions,

With best regards

**Klaus Warnatz**

### **ESID Junior Working Party Chairperson: Crina Samarghitean**



Applicant: **Crina Samarghitean**

Dear friends and colleagues,

The last two years have been very successful for ESID juniors working party and the future looks even brighter. After the ESID juniors working party in 2008 from den' Bosch, Netherlands, when officially I started my first term as a chair of this young party, we have been able to set up a survey to find out what ESID juniors want and to help shaping future ESID meetings. We had the chance to meet, discuss interesting clinical cases and exchange research ideas in ESID Prague meeting 2009 and 2010 and in ESID juniors workshop in Florence, 2010. All these meetings have been characterized by the same spirit, innovation, communication and friendship. After each of these meetings I have been more inspired to initiate new activities, I was more motivated to improve our services and to increase public awareness for PIDs.

A wonderful opportunity for ESID juniors to learn and interact with each other was in ESID summerschool 2009, in Bled, when 30 brilliant ESID juniors from all over the world had the chance to meet leaders in the field and got an assigned mentor. I have been actively involved in the organisation of the ESID summerschool and also have been part of the mentoring process. After the evaluation survey of the summer school in Bled I realised that a lot of work still is needed to be done to bridge the gap between clinicians, lab people and computer people. With this aim in mind a new initiative of the ESID juniors working party have been proposed in May 2010 and get a positive feedback from the ESID board. The negotiations with the sponsors companies have been started and we expect to have the first ESID juniors symposium in 2011, in Finland. First day will cover the clinical approach in PIDs diagnosis, followed by lab diagnosis approach and then by a demo session of a computer-aid diagnosis. The second day will focus on most recent therapies and biotechnologies on PIDs and the essential role of ESID registry and national PIDs patient registries. This meeting aims not only to close the gap between clinicians, lab people and computer people but also to encourage the development of national PIDs registries and the active participation of young doctors in PIDs field.

The numbers of ESID travel awards for ESID juniors, both for short and long term visits have been increased during these 2 years and I was happy to see many young ESID juniors actively involved in these activities.

In research ESID juniors have been also very active. I was very excited to read and recommend for our trimestrial ESID newsletter interesting papers in which ESID juniors have been involved. Some of these papers have been awarded ESID publication award 2008 and 2009 and some have been even selected and evaluated in international literature awareness database which identify the most significant papers in medicine.

Now, we are before another important meeting for ESID community, ESID 2010, in Istanbul, Turkey. We will have besides outstanding lectures and presentations also interactive session in the Educational Day. Here, ESID juniors will find the latest advances in the field and will have the possibility to get direct feedback from their mentors. In the second day 7.10.2010, it will be the ESID juniors working party, where ESID juniors will have the chance to interact with each other, brainstorm new ideas and celebrate not only the winners of the ESID juniors awards but also our 'golden mentors' for 2009 and 2010. A short career advice will be given and we will open for the first time an art exhibition 'Visual reflections on PIDs'. This art exhibition with works created by young doctors/researchers, patients with PIDs or any other ESID member willing to participate, will nurture our eyes and make us reflect to the amazing job we are doing. After the biannual ESID juniors working party we will have a social event in which the music and dance of 'the city of 4 elements' will cross the boundaries and unite all spirits.

If I will be re-elected I will continue to seek out new funding opportunities for ESID juniors; introduce and refine innovative bioinformatics techniques for PIDs; increase the awareness of PIDs in those countries in which PID are not well recognized; and foster collaboration between ESID juniors and other ESID working parties, as well as with European and non - European societies dealing with clinical immunology. Short meetings between young doctors and PIDs patients will increase the communication between the doctors and their patients.

I thank you for your support and hope to see you all in ESID 2010, Istanbul, Turkey!

**Crina Samarghitean MD, MSc**

## End of mandate letters

---

### Registry Working Party - Gerhard Kindle

Dear ESID members,

It was a pleasure to serve as the chairman of the Registry working party, following Lennart Hammarstrom, who gave the idea to an international database and Bodo Grimbacher, who established the fundamentals for the current form of an internet based online register.

It was also a very instructive and valuable experience to be part of the executive board of ESID for four years and thus being able to follow the fortunes of ESID first hand.

I should like to thank all the people, who have supported me in running and improving this important project. Without the dedication of so many of you the success of the Registry would not have been possible. First of all many thanks to all who have documented into the database and helped to make it such a valuable resource for research. I should also like to thank my team here in Freiburg: Stephan Rusch and Daniel Fix, who are working inwardly on the programming and administration of the back-end of the system, Sabine El-Helou, who has joined the team in November 2009, with her main focus on improving the documentation of the German patients and most of all Benjamin Gathmann. A lot of you know him since many years as the ESID database coordinator, who played an essential role in the growth and success of the project. And last but not least many thanks to PPTA for their enduring support since the start of the project that formed the basis for the set-up and the continuous operation of the Registry since 2003.

However, with all the achievements in the last years, I'd like to endorse that the Registry is merely a tool that has to be used for studies and analyses. We have already witnessed a steady increase in this matter and we know that more publications are in the pipeline, but we are convinced that the value and impact of the Registry can be even further expanded through the means of intensive use for all kind of research projects. On this note I hope that you will continue to broaden the range of use and consider running your research projects within the Registry.

After four years I have now to step down as chairman of this working party according to our constitution. I'm confident that my successor in this function can rely on your continuous support and wish him great success and all the best for his term of office.

Thank you very much

Yours  
**Gerhard Kindle**

### Educational Registry Working Party - Andrew Cant

#### Achievements over the last few years

##### Summer Schools

The Educational Working Party has been organising biennial summer schools for 10 years. Through these highly interactive workshops with about 30 students and 6 faculty members we have sought to enthuse, inform and encourage the next generation of leaders in PID practice and research. It is so encouraging to see how former summer school students now have staff positions in PID centres. The most recent ESID summer schools (26-30 September 2007, Malaga and 2-6 September 2009, Bled) brought together young doctors and scientists from countries around the world and covered the diagnosis, pathogenesis and treatment of primary immunodeficiencies. For 5 days a well known international faculty led case-based discussions and gave lecture

For those considering a career in immunology, the summer schools provide an excellent opportunity to update 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 within the whole spectrum of primary immunodeficiency, whilst also providing the opportunity to network with colleagues. Feedback received from the summer schools has been extremely positive; and individuals have found the course to be invaluable.

### **Educational Days**

As part of the biennial ESID meeting the Educational Working Party has led the planning and running of educational days. The ESID educational day on 16 October 2008, 's-Hertogenbosch, The Netherlands was a fun and stimulating day with lots of participation from ESID juniors and plenty of lively interaction with the faculty. The only problem was that too many ESID "seniors" also wanted to participate! Poster sessions and discussion were included as well as "state of the art" mini lectures and case presentations. Following the, s'Hertogenbosch meeting there was a lot of discussion as to how best enable more people to get more from the education sessions and so at the forthcoming ESID 2010 Meeting 6-9 October 2010, Istanbul, with the ESID Board's support and encouragement, it was agreed to integrate an expanded programme within the main congress programme with some sessions more suited for lecture style presentations to large groups and others for smaller groups for more participants. To achieve this it is planned to have 4 plenary education sessions, 11 interactive education workshops including presentations and discussion, and 9 'sunrise' education sessions giving updates on specific "hot topics" in a practical and relevant way, with plenty of time for lots of questions and discussion. We look forward to seeing everyone!

### **Winter workshop and spring meetings**

The Educational Working Party has also supported the ESID Prague Spring meetings (May 2007, 2008, 2009, 2010) organised by Anna Sediva, and the ESID Juniors Educational Workshop (Florence 2010) organized by Eleonora Gambineri, all extremely successful!

ESID Educational Working Party Scholarships – provides a wonderful opportunity for trainees to visit other centres to pursue a research project and further develop their clinical skills within specialist areas. In particular yearly fellowships have been awarded to young trainees in the field of PID, and more opportunities will be available soon for exchange programs.

It has been a real pleasure to be the chairman of ESID's Education Working Party for the past four years. Unfortunately my term of office has come to an end but I believed that my successor will continue the successful initiatives developed so far and implement new and exciting ones in the future.

Thank you for your trust!

Best wishes,  
**Andrew Cant**

## News & Views

### The ESID Meeting in Istanbul, Turkey October 6-9, 2010



Dear ESID members and contributors of ESID 2010 Istanbul congress,

Thank you so much for all your contributions to ESID 2010 Istanbul Meeting. A record breaking number of abstracts have been submitted during regular abstract submission time. A total of 618 abstracts have arrived and late-breaking abstracts are coming in every day.

**Abstract numbers in different categories are as follows :**

Record breaking number  
of submitted abstracts!

		Oral	Poster	Abstract only	Total
A	T-Cell and combined immunodeficiency	47	60	7	114
B	B-Cell immunodeficiency	44	79	7	130
C	Deficiencies of innate immunity (Phagocytic cells)	25	39	2	66
D	Deficiencies of innate immunity (Complement and TLR's)	13	15	1	29
E	NK Cells; NKT cells, Tregs and Th17 cells in immune regulation	13	15	1	29
F	Stem cell transplantation/gene therapy	30	13	0	43
G	Immunoglobulin and cytokine therapy	11	32	1	44
H	Genetics/Genetic counseling/Genetic predisposition to infection and autoimmunity	19	42	1	62
J	Quality of life, long term management in PID and PID registries	24	37	2	63
K	Animal models and Basic Immunology	10	8	0	18
L	INGID Abstracts	6	12	0	18

Oral abstract evaluation will finish on July 1st and then we will inform all participants for their acceptance and category. For receiving an acceptance letter, at least one of the authors must have done their registration.

Late-breaker abstract  
submission deadline is  
August 15th

**Deadline for late-breaker abstract submission is August 15th, 2010 and deadline for regular registration fee is October 1st, 2010.**

I hope to see all of you in İstanbul and I promise you that you feel better on the 10.10.10!

With my best wishes

**Prof.Dr.Necil Kutukculer**  
Congress President

**Congress registration fees for ESID Members:**

### ESID/PPTA Publication award 2009

	Before 15 June, 2010	After 15 June, 2010	After 1 October, 2010
ESID MEMBERS	EUR 350,-	EUR 450,-	EUR 500,-
ESID MEMBERS less than 30 years old	EUR 250,-	EUR 350,-	EUR 400,-

It is our pleasure to announce that **five publications have been awarded by the ESID publication award 2009, funded by PPTA**. This award is dedicated to publications related to the Registry Project. If you have published or will publish any papers in 2010 that are based on data from the ESID Registry please remember to apply for the 2010 award. (As usual ESID-Board members involved in any of the publications did abstain from voting).

#### The 2009 laureates are:

1. Ballow M, Notarangelo L, Grimbacher B, Cunningham-Rundles C, Stein M, Helbert M, Gathmann B, Kindle G, Knight AK, Ochs HD, Sullivan K, Franco JL. Immunodeficiencies. Clin Exp Immunol. 2009 Dec;158 Suppl 1:14-22. (PMID: 19883420)

2. Tóth B, Volokha A, Mihás A, Pac M, Bernatowska E, Kondratenko I, Polyakov A, Erdos M, Pasic S, Bataneant M, Szaflarska A, Mironska K, Richter D, Stavrik K, Avcin T, Márton G, Nagy K, Dérfalvi B, Szolnoky M, Kalmár A, Belevtsev M, Guseva M, Rugina A, Kriván G, Timár L, Nyul Z, Mosdósi B, Kareva L, Peova S, Chernyshova L, Gherghina I, Serban M, Conley ME, Notarangelo LD, Smith CI, van Dongen J, van der Burg M, Maródi L. Genetic and demographic features of X-linked agammaglobulinemia in Eastern and Central Europe: a cohort study. Mol Immunol. 2009 Jun;46(10):2140-6. Epub 2009 May 5. (PMID: 19419768)



3. Gathmann B, Grimbacher B, Beauté J, Dudoit Y, Mahlaoui N, Fischer A, Knerr V, Kindle G; ESID Registry Working Party. The European internet-based patient and research database for primary immunodeficiencies: results 2006-2008. *Clin Exp Immunol*. 2009 Sep;157 Suppl 1:3-11. (PMID: 19630863)
4. Samarghitean C, Ortutay C, Vihinen M. Systematic classification of primary immunodeficiencies based on clinical, pathological, and laboratory parameters. *J Immunol*. 2009 Dec 1;183(11):7569-75. Epub 2009 Nov 16. (PMID: 19917694)
5. Samarghitean C, Vihinen M. Bioinformatics services related to diagnosis of primary immunodeficiencies. *Curr Opin Allergy Clin Immunol*. 2009 Dec;9(6):531-6. (PMID: 19779331)

### **Report - APPID Summer School, Australia 9-14 May 2010**

It was a marvellous experience to attend the APPID summer school this year in Australia, where I received intensive courses on primary immune deficiency (PID). Giving me the chance to get in contact with other colleagues from Australia and with eminent immunologists was an incredible opportunity to me. In Egypt, we have many patients and families with PID due to a high consanguinity rate in Egypt. Unfortunately, we do not have the facilities to perform genetic studies. Reading in genetics of immune deficiency was very difficult until I attended the APPID summer school where Professors of immunology; Luigi Notarangelo, Helen Chapel, Mary Ellen, Steve Holland, Stuart Tangye and Melanie Wong simply clarified molecular genetics of almost all PID diseases. This has greatly changed my understanding of many of PID diseases such as hyper-IgE, and recurrent warts where we should consider the possibility of DOCK8 or STAT3 mutation in such cases.

Also, I was very much privileged to have Professor Luigi Notarangelo as the supervisor on my oral presentation. He is a great immunologist. I had the opportunity to discuss with him some atypical PID cases in my country. In fact, the APPID summer school was very well-organized in all aspects. All juniors working in PID had the opportunity to communicate with colleagues from other centres. We also, received a booklet about the indications and methods of administration of immunoglobulin therapy in PID. Hand-outs of all lecturers and oral presentations were available. When I returned back to my centre at Ain Shams University in Egypt, I was interested to transfer my experience and the summer school hand-outs to my colleagues in order to spread the valuable knowledge I received among registrars interested in PID and to improve the diagnosis and management of PID patients in Egypt.

I am grateful to my professor Shereen Reda who directed me to join the ESID junior members and encouraged me to apply to the APPID summer school. I am very grateful to Jill Smith; executive officer and to Stephen Bowditch; BSL biotherapy for preparing for me a comfortable flight to Australia. Last but not least I am grateful to the ESID junior working party who encourage juniors from all countries and especially from developing ones to build-up their careers and improve research in their countries.

Kind regards,

**Reem Elfeky**, Lecturer of Pediatrics, Ain Shams University, Cairo, Egypt

## Report - 6<sup>th</sup> Ukrainian J Project Meeting on «Primary Immunodeficiencies»

According to the East-European Initiative “J-project” the 6<sup>th</sup> Ukrainian Meeting «Primary Immunodeficiencies» has taken place in Kharkiv (Ukraine) at May 20, 2010. The meeting was organized by initiative of Chief Pediatric Immunologist of MOH of Ukraine Liudmyla Chernyshova and Chief immunologist of Kharkiv region O. Savvo.

Kharkiv is the second largest city in Ukraine after Kiev with the population of about 1,5 million. In the 1920-ies Kharkiv was the capital of Ukraine. The immunologic center was organized there many years ago, now it serves as a center for 3 regions of Ukraine (Kharkiv, Sumy, Lugansk). The number of diagnosed PID patients is to be increased. The definite diagnosis is still a problem in these regions, as well as the use of up-to-date treatment. The main problem is that doctors of first line contact have limited knowledge about PID. Among other possible reasons is insufficient laboratory base for diagnostics of primary immunodeficiency. For this reasons educational programs are necessary to improve the understanding of PID.

The goal of the seminar was to increase awareness about PID and its diagnostics, to give the benefit of experience of treatment of such patients in the Kharkiv region. The leading immunologists from different regions of Ukraine and about 120 pediatricians from Kharkiv region have taken part in the Meeting.

Achievements in diagnostics of primary immunodeficiency in Ukraine and elsewhere, demonstration of clinical cases of primary immunodeficiency (antibody deficiencies, Nijmegen breakage syndrome, Wiskott-Aldrich syndrome, ataxia-teleangiectasia), the possibilities of genetic diagnostics and treatment of primary immunodeficiencies, were the basic directions of the presented reports. László Maródi noted the great contribution of Liudmíla Chernyshova in the development of clinical immunology in Ukraine and international cooperation, and also handed to her the award “Pro Collaborazione” conferred in 2010 by Debrecen Regional Committee of the Hungarian Academy of Arts and Sciences. Besides, in his presentation L. Maródi acquainted the participants with the latest advances in genetic diagnostics of PID and the discovery of new diseases and mutations.

On patient's section during this seminar patients and their parents (in particular the patient with XLA, father of two boys with XLA, father of a boy with WAS) talked about their history of disease, path to diagnosis and treatment problems. Their speeches were memorable and impressive.

At the end of the seminar was held a workshop on the diagnosis of the most common primary immunodeficiencies.

In her report L.Chernyshova stressed that J Project Meetings gave much help in improvement of PID detectibility in Ukraine which is now supported by statistical data from the Jeffrey Modell Foundation presented also by L. Maródi. The number of PID patients in Ukraine are now approaching 2000 which includes transitory hypogammaglobulinemia and selective IgA deficiency.

**Liudmyla Chernyshova**  
Professor and Chair  
PID Center,  
Kiev, Ukraine

## Report - ESID Prague Spring Meeting, May 10 And 11, 2010

It was a big honour for me to be chosen as a participant of the ESID Prague spring meeting and I would like to thank Rita Louro for all her assistance and ESID for giving me that opportunity.

This meeting was organized under to auspices of the Department of Immunology of the 2nd Medical School, Charles University and Faculty Hospital Motol and hosted by Anna Šedivá and Andrea Poloučková in Olympus Headquarters in Prague. My special thanks for Andrea Poloučková who answered very thoroughly to all my questions by e-mail and who help me in all steps of my visa process and accommodation booking in the city center in Hotel Karolinum in historical building of the Charles University.

The main topics of ESID Spring Meeting were B cells in CVID and Treg and Th17. The program incorporated practical aspects and guidelines, but also state-of-the-art understanding of primary immunodeficiency. The first day of meeting was pointed to CVID and related disorders. Klaus Warnatz (Freiburg, Germany) gave us a fascinating opening lecture on CVID topic, which was followed by two presentations from our colleagues from Brno, Czech Republic - Šárka Růžičková and Jan Stuchlý and a lively discussion. After the lunch Lennart Hammarström (Stockholm, Sweden) presented us new achievements in CVID treatment, Amairelys Belen Barroeta Seijas (Rome, Italy) - very young, emotional and at the same time very competent PhD student told us about TNFRSF13B genetic characterisation of pediatric patients with hypogamaglobulinemia, Anna Šedivá (Prague, Czech Republic) gave us a brief but very informative presentation about B lymphocytes classification in Schnitzler syndrome and Aleš Janda (Freiburg, Germany) presented the data concerning Clinical and immunological overlap between autoimmune lymphoproliferative syndrome and CVID.

Bodo Grimbacher (London, UK) opened the second day of the meeting with the detailed and pointed lecture on CMC. It is always impressive when I am listening to Prof. Grimbacher lecture since he could speak about so serious topics keeping smiling all the time. This day we also discussed allergy and clinical presentation of HIES with Andrea Poloučková (Prague, Czech Republic); Clinical characteristics and molecular analysis of central European patients with CGD with Gašper Markelj's (Ljubljana, Slovenia) help; Rudolf Horváth (Prague, Czech Republic) reported us about Disturbances in the homeostasis of Th17 lymphocytes in patients with HIES an CGD and Tomáš Freiburger (Brno, Czech Republic) presented the data about FcRn in health and disease. After the lunch Crina Samarghitean, ESID Juniors WP (Tampere, Finland) gave us the latest information concerning ESID junior's activities and latest advances in PIDs knowledge services and then she invited all young participants to present their data on the different aspects of PID.

Case report session gave many PhD and medical students and junior members an opportunity to show their work to everybody, share their problems and look for the solution. I would like to say thank you for all my colleagues and special thanks to Klaus Warnatz for their helpful comments and suggestions regarding my patient with CVID whom clinical case I've presented during the meeting.

The room was full throughout the whole meeting and discussions were always informative, friendly and emotional. Faculty excellence and intimate and collegial atmosphere allowed plenty of space for such discussion and permitted participants to profit from this event, auguring significant theoretical and practical knowledge in PID. It is well known fact that we could find the answer even on very difficult question if we would work on it together.

The ESID Prague spring meeting was not only a scientific event. The cultural and social program was also exceptional. In spite of rainy weather we spent a fascinating time in one of the most beautiful city in Europe – Prague. On the first day late afternoon there were an Opening exhibition – Paintings of Immune system painted by the students of Academy of Fine arts in Prague with the welcome presentation of Anna Šedivá and professor of Fine arts. Then all participants were transferred to the restaurant Petřínské terasy in the centre of the Prague where we could enjoy delicious Czech food and drinks in a company with our new art friends. All participants were invited to the hiking tour in Krkonoše Mountains and the end of the meeting. It was so nice to find yourself in a wonderful and silent place after the crowded and a little bit noisy city.

When I came back home I decided to share the received completely new information with my colleagues and I had out of expectations opportunity for that: our Allergy and Occupational Health Department provided a special seminar for general practitioners were I gave a presentation about initial diagnosing of PID in general practice; I also send a short report about CVID with clinical cases for the conference which would be held in October in my Academy and developed new lectures on PID for clinical immunology short course which took place at our Department. I think it would be a good practice to present the detailed information concerning the PID for our clinicians at the Allergy and Occupational Health Department.

Best regards,  
**MD Larisa Fedarushchanka**  
ESID Junior member

## News in images

---



Our President Professor Jean-Laurent Casanova received *Honoris Causa* degree at the University of Debrecen, Hungary, for his outstanding scientific achievements in the field of genetics of infectious diseases.



In recognition of outstanding contribution to the success of the J PROJECT, Liudmyla Chernishova (middle) and her group in Kiev, Ukraine received PRO COLLABORATIONE Award from the Debrecen Regional Committee of the Hungarian Academy of Sciences. Also pictured, L. Maródi (left).

## Working parties - Reports



### Report from the ESID BMT& Gene Therapy Working Party

**Bobby Gaspar**

As well as the ESID meeting in Istanbul, we will also be holding our annual ESID/EBMT Inborn Errors Working Party meeting later this year. The meeting is scheduled for 24-26 November and is to be held on the island of San Servolo in Venice. The location is really spectacular and as ever we have an equally exciting meeting schedule. This year we hope to highlight the outcome of HSCT in a number of different forms of PID as well as looking at newer immunodeficiencies such as IL-10R deficiency and the role of allogeneic transplant in autoimmune conditions. There will be sessions on gene therapy, immune reconstitution and discussion of optimal conditioning protocols. The meeting remains an essential calendar date for those involved in the transplantation of severe immunodeficiencies and I hope to see many old and new friends at the meeting. I contact all those interested in the WP by e-mail but if you are not on my e-mail list and want to be involved in the WP and to receive further news of the meeting and other activities, please contact me at: [h.gaspar@ich.ucl.ac.uk](mailto:h.gaspar@ich.ucl.ac.uk)

### Report from the ESID Juniors Working Party

**Crina Samarghitean**

Dear ESID juniors and other ESID members,

First, I want to thank you for your support and all your great work for the ESID juniors working party. During these 2 years I have been fortunate to meet many of you and establish new collaborations, which I hope will continue in the next years too. Despite of our achievements in 2008-2010 there are still some activities, like launching of the first decision support system for PIDs (**PIDexpert**), developing of new factfiles for **IDR**, developing of **IDdiagnostics**, a genetic and clinical test database for PIDs, which have to be better integrated within the ESID registry and within other ESID working parties activities. That's why, I decided to continue to serve further ESID and stand for a new re-election hoping that I will have the same support from you.

#### The ESID Istanbul meeting

As we know already, in **October 2010**, in Istanbul, will be the **14<sup>th</sup> meeting of the ESID**. A great program for ESID juniors and all ESID members have been already set up by the organizers. Beside outstanding lectures, interactive sessions in the first day of the meeting, in **7<sup>th</sup> October** will be the **Dinner workshop of the ESID juniors working party** and a **Social Event**, exclusively organized by ESID juniors with great help from Rita Louro (Kenes) and Ilke Incekara (Topkon). I would like to encourage you to mark already your calendars and joins us in these two events where for the first time not only the most active ESID juniors will be awarded but also their 'golden mentors'. A short career advice from them will motivate the ESID juniors to get more involved in future ESID juniors activities and in PIDs field. The night will end with a brainstorming session and the opening of the art exhibition '**Visual reflections on PIDs**'. More details about the art exhibition will be on the ESID website and in the email list. After the **ESID juniors workshop** we will have a **social event** in which the music, singing, dance and the good mood will be the main themes. So, don't hesitate and get actively involved in these new initiatives of the ESID juniors working party: send your ideas for the brainstorming session, enchant us with your inspired paintings and with your artistic talents and make **ESID juniors workshop 2010** another unforgettable event!

#### **The ESID Prague meeting**

Recently, another successful ESID Prague meeting excellently organized by Anna Sediva and Andrea Polouckova took place in May this year. Beside excellent lectures and presentations ESID juniors have been delighted and inspired by the opening of an art exhibition – **Paintings of Immune System**, painted by the students of Academy of Fine Arts in Prague. We hope to see some of these paintings also in Istanbul, at the **ESID juniors**

October 7th  
Dinner workshop and Social Event !

'Visual reflections on PIDs' – Art exhibition opening  
  
Please visit the ESID website to learn more about this initiative and on how to apply

This year, ESID kindly awarded a **travel award for Prague meeting** to **Larisa Fedarushchanka**, from Belarus. Congratulations to the winner!

**Hope to see many of you at the ESID Juniors working party and the Social Event in Istanbul 2010!**

**Crina Samarghitean MD, MSc.**  
Chair ESID Juniors Working Party

## Report from the ESID Registry Working Party

Gerhard Kindle and Benjamin Gathmann

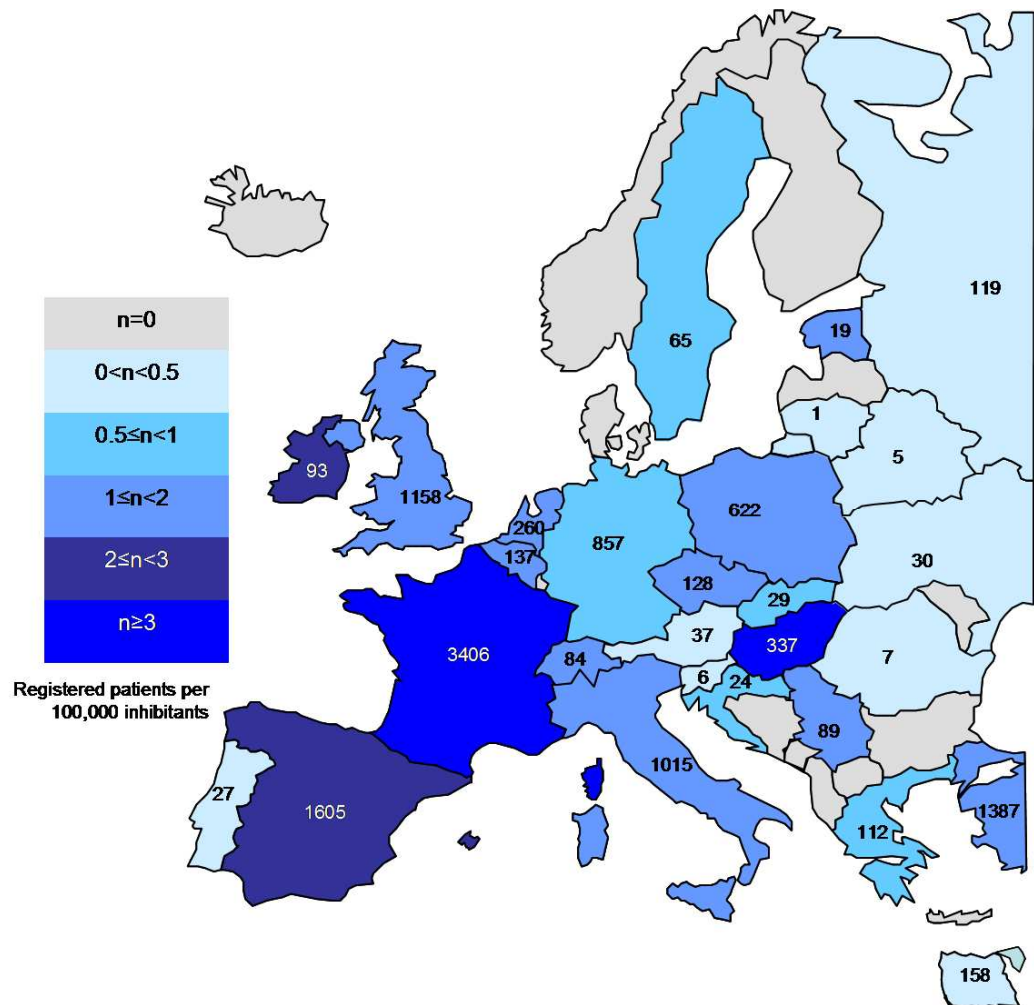


Fig. 1 Registration progress by countries. The absolute number of patients is displayed in the map. The colours indicate the documentation rate in relation to the country population.

Before the upcoming ESID congress in Istanbul, we would like to take a short look back at what we have achieved over the past two years.

### Documentation progress

In patient numbers, we have seen an increase from 7,550 registered patients at the end of 2008 to now 11,817 patients (June 2010). This is an increase of 4267 registered patients within less than two years.

To our knowledge, the ESID Database now represents the largest single collection of data on PID patients worldwide. This would have not been possible without establishing successful collaborations with existing databases and national registries such as Spain and Italy. Data is transferred electronically to the ESID Database regularly from a total of six databases. Furthermore, the ESID Database has been instrumental in setting up new national registries in a host of countries which include the United Kingdom, Germany, Sweden, Austria, Switzerland, the Netherlands, Belgium and the Czech Republic. The documentation progress is displayed in Fig.1.

We would like to note that this map and a lot more information are visible on the ESID website ([www.esid.org/statistics.php](http://www.esid.org/statistics.php)) as well. However, we have modified the map on the website recently so it shows the actual documented minimal prevalence rate per 100,000 inhabitants. The numbers in the statistics pages are updated once a month, so it is always worth having a look.

### Data quality

To ensure data quality, such as completeness and integrity, we have implemented many additional features to the data entry forms in addition to regularly running plausibility checks on the data. Furthermore, we now offer standardised patient questionnaires on the ESID website which can be used to collect in particular quality of life data before or during patient visits. We encourage all participating centres to use these questionnaires. We also plan to translate the questionnaires into as many languages as possible so they can be used all over Europe.

### Database aims

The core aims of the Database project are to create a data collection to answer questions on PID epidemiology on the one hand, and to foster the use of the database as a platform for multi-centre studies on the other.

As for PID epidemiology, we have fared even better than expected. Several peer-reviewed publications are proof of this. The latest of these, resulting from the close collaboration between ESID and the French national centre CEREDIH, is:

CEREDIH: The French PID study group. The French national registry of primary immunodeficiency diseases. *Clin Immunol.* 2010 May;135(2):264-72. PMID: 20399414

We expect similar analyses by other national registries soon and will keep you informed via the ESID website and by e-mail.

## Interesting papers

---

### Reviews

**Good syndrome (GS)** is a rare association of thymoma and immunodeficiency first described more than 50 years ago. This syndrome still remains a mystery to clinicians. A recent systematic review includes all the clinical, laboratory and immunologic findings from 152 patients with Good syndrome. The syndrome has a worldwide distribution and approximately half of the cases (47%) have been described in Europe. The diagnosis of thymoma preceded the diagnosis of hypogammaglobulinemia, infection, or diarrhea in 42% of patients whereas in 38% of patients the diagnoses were made almost simultaneously within 2 months of each other. The authors found significant mortality in patients with this syndrome (44.5%). Further studies are needed to elucidate this clinical entity. An early recognition of this syndrome and mortality prevention may be increased by clinical acumen and increased awareness of the clinical and immunological profile of this syndrome (**Kelesidis T, Yang O, Clin Immunol. 2010 Jun;135(3):347-63.**)

In a recent review the authors described three groups of PIDs that seem to impair immunity to primary infections without compromising immunity to secondary and latent infections. Patients with mutations in ***IL12B*** or ***IL12RB1*** present mycobacterial disease in childhood with a favorable progression thereafter. Patients with mutations in ***IRAK4*** or ***MYD88*** suffer from pyogenic bacterial diseases, including invasive pneumococcal diseases in particular. Finally, mutations in ***UNC93B1*** and ***TLR3*** are associated with childhood herpes simplex encephalitis, which strikes only once in most patients, with almost no recorded cases of more than two bouts of this disease. Although life-threatening in early childhood, these new PIDs are associated with a favorable outcome in adulthood. They provide proof-of-principle that infectious diseases of childhood striking only once may result from single-gene inborn errors of immunity (**Bousfiha A, Picard C, Boisson-Dupuis S et al, Clin Immunol. 2010 May;135 (2):204-9**).

### Guidelines/Therapies

**Severe combined immunodeficiency (SCID)** has been under consideration for population-based newborn screening (NBS) by many NBS programs. Recently, SCID has been recommended for inclusion in the US uniform panel of newborn screening conditions. A marker of SCID, the **T cell receptor excision circle (TREC)**, is detectable in the newborn dried blood spot using a unique molecular assay as a primary screen. The New England Newborn Screening Program developed and validated a multiplex TREC assay in which both the TREC analyte and an internal control are acquired from a single punch and run in the same reaction. Massachusetts then implemented a statewide pilot SCID NBS program. The authors describe the rationale for a pilot SCID NBS program, the screening test algorithm, the screening follow-up algorithm and preliminary experience based on statewide screening in the first year. The Massachusetts experience demonstrates that SCID NBS is a program that can be implemented on a population basis with reasonable rates of false positives (**Comeau AM, Hale JE, Pai SY, Bonilla FA, Notarangelo LD et al, J Inherit Metab Dis. 2010 May 20,** ).

### Hemopoietic Stem Cell Transplantation (HSCT)

A complete list of definite, as well as possible, indications for hemopoietic stem cell transplantation in primary immunodeficiency is provided in a recent article. Included are: severe combined immunodeficiency, profound T cell defects, autoimmune and autoinflammatory syndromes, innate immune defects, hemophagocytic disorders, and other conditions. Some causes and limitations of **HSCT** are discussed (**Roifman CM, Fischer A, Notarangelo LD et al, Immunology and Allergy Clinics of North America, May 2010 30(2): 261-262**).

**Therapeutic monoclonal antibodies** often target molecules involved in innate and adaptive immune responses and are used to treat patients with various immunological conditions such as inflammatory and autoimmune diseases and haematological malignancies. In a recent comment appeared in **Nature/Reviews Immunology** the authors propose that the study of primary immunodeficiencies (PIDs) may help to identify, monitor and prevent potential adverse effects of therapeutic antibodies, in particular infectious diseases (**Maródi L, Casanova JL, Nat Rev Immunol. 2010 May;10(5):299-300**).

**Warts hypogammaglobulinemia infections myelokathexis (WHIM)** syndrome is an autosomal dominant primary immunodeficiency disorder caused by mutation in ***CXCR4***. Affected patients suffer from recurrent infections of the respiratory tract and soft tissues, and marked susceptibility to warts caused by human papilloma viruses from the second decade of life.



**Granulocyte colony-stimulating factor (G-CSF) and intravenous immunoglobulin (IVIG)** have been used as therapy but the patients continue to develop infections. None of the patients reported earlier has received **hematopoietic stem cell transplantation (HSCT)**. A recent successful allogeneic **HSCT** in a girl with WHIM syndrome have been described. One year after the transplant, the patient has near normal hematologic and immunologic parameters. Thus, the authors propose allogeneic **HSCT** as a curative treatment option in patients with WHIM syndrome if G-CSF and IVIG support fail to prevent infections (**Kriván G, Erdos M, Kállay K et. al Eur J Haematol. 2010 Mar;84 (3):274-5**).

Hope you enjoyed the papers selected for this issue. Wish you a relaxing summer with good ideas and inspiration for many interesting and challenging papers!

Do you have interesting articles you want to share with the whole ESID community? Don't forget to send them to us.

Please, write your comments and suggestions to **Crina.Samarghitean@uta.fi**

## Young researcher's corner

Dear ESID Juniors,

*First of all I really hope to meet all of you in Istanbul on the occasion of the XVth ESID meeting!*

*And now I'm going to introduce you to a short review on laser micro dissection:*

LASER ASSISTED MICRO DISSECTION: METHODS AND APPLICATIONS

Laser-assisted micro dissection (LAM) is a powerful instrument for obtaining pure cell samples from a tissue or a cell culture. This is very useful because the quality of data obtained from genetic or proteomic analysis using tests based on tissue extracts will often be dependent on the cellular homogeneity of the study samples. For this purpose recently LAM has been introduced enabling rapid procurement of homogeneous cell samples from both frozen and formalin-fixed, paraffin-embedded tissue sections and

from cytological smears, suitable for gene analysis of DNA, RNA and proteins.

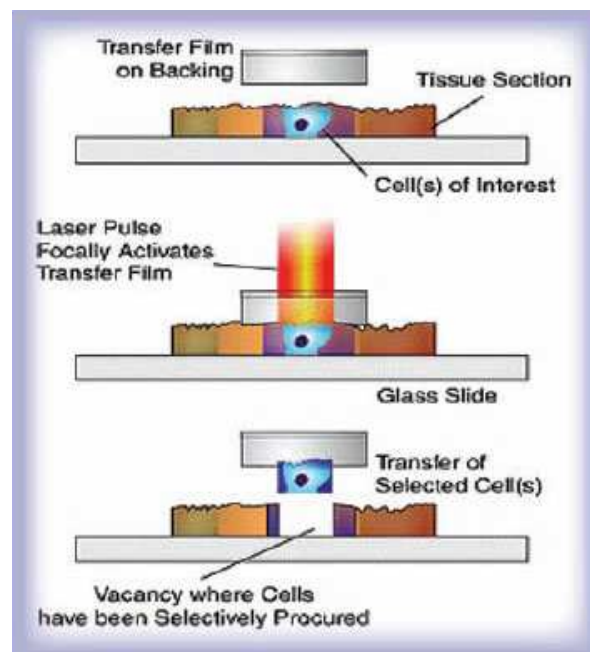


Fig. 1: Representation of Laser capture micro dissection steps

Two systems are available in LAM: Laser capture micro dissection (LMC) and Laser beam micro dissection (LMM) or Laser pressure catapulting. The former utilizes a low-powered infrared laser to activate an adhesive thermoplastic film on the cap of a micro centrifuge tube that has been placed over the cells of interest. Through this activation, the film directly "captures" the cells on the cap, and they can then be analyzed further (Fig. 1).

The latter uses a pulsed ultraviolet (UV) narrow beam focus laser directed through the microscope objectives to cut around cells of interest that are mounted on a membrane-coated

The excised cells then are either ejected or dropped via gravity into a receptacle (commonly a micro centrifuge tube), in which they can be studied further (Fig.2).

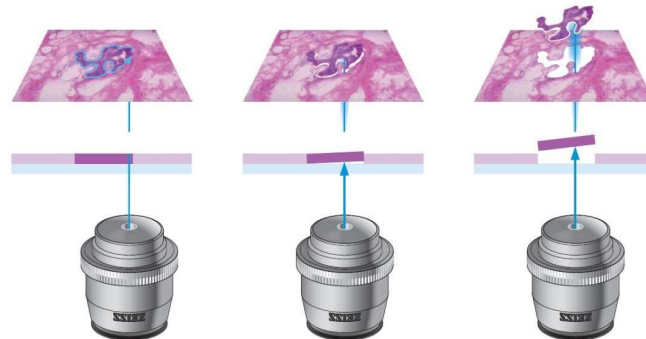


Fig. 2: Laser pressure catapulting system: Laser cutting and laser catapulting processes

LAM has several advantages over current tissue micro dissection approaches: it is simple, requires no moving parts, involves no manual micro dissection or manipulations, and enables one-step transfers. Moreover in LCM the transferred tissue on the film retains its original morphology, thereby allowing microscopic verification of the specificity of the captured material. The use of sterile, disposable transfer films minimizes potential contamination, which is particularly important for PCR-based analyses.

LAM-procured samples can potentially be subjected to any method of molecular analysis. Genomic analyses have been the most common to date, such as real-time RT-PCR, differential display, gene microarray, loss of heterozygosity, and mutation analysis, with only a small number of studies examining proteins. The preference for nucleic acid over protein analysis is largely due to the methodologies that allow nucleic acids to be amplified from the typically small yields of material collected in LAM studies. However several reviews are available for protein analysis after laser micro dissection. The successful use of LAM to procure cells can be technically demanding, with the three key factors (tissue morphology, capture success, and maintenance of molecular integrity) being dependent on numerous variables such as tissue fixation method, staining procedure, slide temperature, and section processing. Furthermore, with a number of LAM microscopes available to researchers, each with their own principles of operation, the impact of these variables is dependent on the instrument being used. If the interest is based on studying RNA expression is convenient to work with frozen, non-embedded sections to maintain RNA integrity performing LAM with standard histochemistry (e.g. H&E staining) methods. It is often the case that morphology of frozen tissue sections is not sufficient for conclusive identification of cells of interest, and the use of wax-embedded tissue sections is preferable. Wax-embedded sections allow for easy section preparation, and can be used with conventional staining techniques, including immunostaining, quite readily. The main obvious difficulty with wax-embedded tissue sections is a reduction in RNA yield and quality. Thus the use of wax-embedded sections is a compromise between better morphology and RNA yield and quality. Nonetheless, there are some studies that have successfully obtained and analyzed RNA from wax-embedded testis tissue sections.

Summarizing the laser assisted micro dissection is a powerful and flexible method to collect specific samples without alter or damage the morphology and chemistry of the sample, nor the surrounding cells and to allow to analyze them with classical technique of molecular biology.

## REFERENCES

- Hunt JL, Finkelstein SD. (2004) Micro dissection techniques for molecular testing in surgical pathology. *Arch Pathol Lab Med.* 128:1372-1378
- Gjerdrum LM, Lielpetere I, Rasmussen LM, Bendix K, Hamilton-Dutoit S. (2001) Laser-assisted micro dissection of membrane-mounted paraffin section for polymerase chain reaction analysis. *Journal of Molecular Diagnostics,* 3: 105-110

Kaneko K, Okiji Tkachi, Kanero R, Suda H, Nor E J. (2009) Gene expression analysis of immunostained endothelial cells isolated from Formaldehyde-fixated paraffin embedded tumors using laser capture micro dissection – A technical report. *Microscopy Research and Technique*, 72:908-912

Pinzani P, Orlando C, Pazzagli M (2006) Laser-assisted micro dissection for real-time PCR sample preparation. *Molecular Aspects of Medicine*, 27:140–159

*What do you think about these methods?*

*I will be glad to receive any suggestions or comments from you on specific applications!*

**Sara Ciullini Mannurita** ([s.ciullini@meyer.it](mailto:s.ciullini@meyer.it))

University of Florence,

Department of Sciences for Woman and Child's Health

“Anna Meyer” Children Hospital

Florence

Italy

### **Take part in the Newsletter!**

We are looking for volunteers to join the editorial board of ESID newsletter. People interested, please contact Editor in Chief, Eleonora Gambineri at:

[treasurer@esid.org](mailto:treasurer@esid.org) or Rita Louro [esid.admin@kenes.com](mailto:esid.admin@kenes.com)

Wish you good ideas and inspiration for many interesting and challenging papers! Do you have interesting articles you want to share with the wholeESID community?

Don't forget to send them to us!

### **Contact us:**

For queries related to ESID initiatives please contact Rita Louro by email at [esid.admin@kenes.com](mailto:esid.admin@kenes.com) or directly by phone at +41 22 906 91 49

For queries related to your membership please contact Justine Peleg at [esid@kenes.com](mailto:esid@kenes.com)

**ESID Administrative Office**

C/o Kenes International | 1-3 rue de Chantepoulet - POB 1726 | 1211 Geneva 1 | Switzerland  
Ph- +41 22 906 9171 | Fax. +41 22 732 26 07 | Email: [esid.admin@kenes.com](mailto:esid.admin@kenes.com) | Website: [www.esid.org](http://www.esid.org)