NEWS @ ESID

Newsletter Issue 2 | 2012

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ESID Summer School 2013
25 September - 29 September
Hersonissos, Crete, Greece
APPLICATIONS OPEN - click here

ESID Biennial Meeting - Florence, Italy
3-6 October 2012
The ESID 2012 Meeting was a great success with a record number of 1'840 participants & we would like to thank all of you who attended!
Access the abstracts - click here
View pictures - click here

Baxter - ESID Fellowship Award
2-year grant in the value of $ 100'000
Congratulations to Dr. Margje Haverkamp!
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Participate in the Stem PAD study
A study on the outcome of transplant for antibody deficiency and CVID in collaboration with EBMT Inborn Errors WP and ESID BMT and Clinical WP.
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Molecular analysis of PIDs
The ESID Genetic Working Party wants to help to connect and support the collaborative clinical, genetic laboratories and research units associated with the ESID society.
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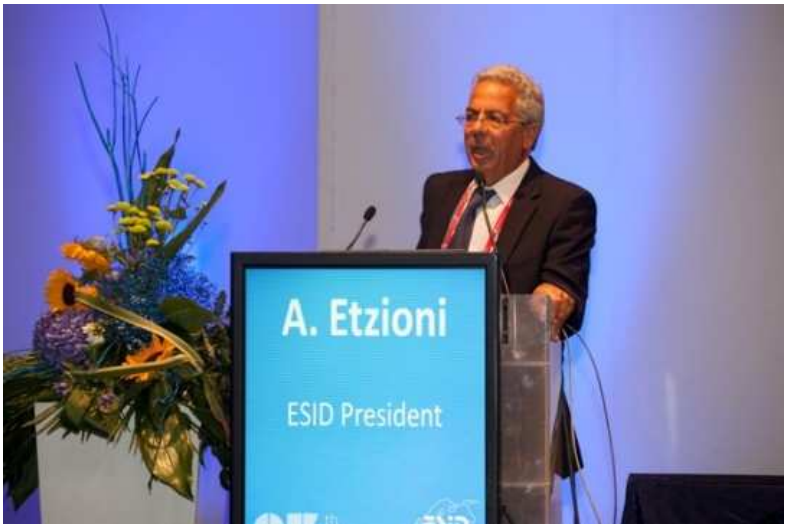
NEWS @

President's Corner



Dear ESID members,

First I would like to thank you all for reelecting me for the presidency of our society and will try to do my best for ESID.

I take this opportunity to thank all who were involved in the Florence meeting and mainly Eleonora Gambineri, for organizing such an amazing event.
More than 1800 participants (ESID record!) attended the meeting which in my opinion was on the highest scientific level, it was wonderful to see the interaction between people from all over the world and I'm sure many international collaboration will start soon.

This is also a right time to thank Bobby Gaspar (SCT/GT) and Klaus Warnatz (clinical WP) for their excellent work during the last 4 years and to congratulate the new board members especially the president elect – Andrew Cant.

Our aim remains the same: increasing awareness and knowledge regarding P.I.D. We allocated money for each of the working parties to increase their activity, new grants for investigating the various aspects of PID are now available.
We have established strong ties with our American colleges (Clinical Immunology Society - CIS) and soon ESID members will be able to publish in the "ESID corner" in the Journal of Allergy Clinical Immunology JoCI and become affiliated members in CIS. The collaboration with LASID and ASID is also growing and we encourage ESID members to take part in their meetings.
Although our next meeting in Prague is still 2 years away, we already started working on various aspects concerning the 2014 meeting.

Wishing you all greetings for the next year (without Sandy around).

Sincerely,
Amos

ESID President Elect

It is amazing to think that it is over a month since the very successful Biennial ESID meeting in beautiful Florence, so delightful in the early autumn.

I am sure you will agree that it was a great success and many congratulations to Eleonora, Amos, the board and the scientific committee who put together a first rate programme. It was exciting to learn of so many new primary immunodeficiencies as well as expand our knowledge of known conditions and learn of better ways of treating PID.
My only regret was not being able to attend more than one parallel session at once.

We are very privileged to work in such a fast changing field. When I attended my first ESID [then EGID] meeting in 1990, very little was understood about the causes of PID, proper immunoglobulin therapy was rather new and bone marrow transplantation only just moving from being an experimental procedure.

We now understand the molecular basis for more than 150 primary immunodeficiencies and have much more effective treatment, so patients enjoy greatly improved survival and much better quality of life.

So what next? A great deal I would suggest! Powerful new genetic techniques may mean we can identify one ‘new’ PID a month, but there is still a lot of work to be done on discovering the biological effect and the implications for patients and their treatment. Moreover as there are rare disorders, further work needs a forum for exchanging ideas and results, and collaborating using

registry data to accurately delineate the exact clinical picture for each genetically defined PID.
Beyond this, accurately assessing new treatment, whether scIg, new HSCT methods or new methods of gene correction, can only be done on a multi-centre basis.

ESID already has an excellent track record for acting as a catalyst for such ventures and each working party focuses on different areas where such collaboration can be fruitfully developed. However, the results from each working party's endeavours are so dependant on participation by ESID members. So can I urge you to support Andy Gennery as he seeks to build on the superb work undertaken by Bobby Gaspar, Stephan Ehl as he continues so well with the demanding work on the registry, Despina as she takes up the clinical working party chair, Capucine as she develops genetic collaboration, for which a workshop is being planned, Esther as she continues to co-ordinate the excellent range of educational activities, Marta with her imaginative work for the juniors and Anna, who is much involved in both PID care in development and the next ESID meeting.

We are privileged that such talented ESID members freely give up their time and effort to lead our working parties!

With very best wishes for the holiday period and New Year.

Professor Andrew J Cant

Secretary's report

Dear ESID members,

The **Biennial ESID meeting** is just finished and all of us have enjoyed great science, the wonderful and perfect venue. Congratulations to Eleonora Gambineri for this great success!!

Our ESID family is now increased: 710 ESID members, 235 out of them are Junior members that means a lot of energy and new ideas.

During the **General Assembly**, we have announced the results of ESID board vote. Two new ESID Board members have been elected:

- **Despina Moshous** (Hospital Necker-Enfants Malades, Paris) who will chair the Clinical Working Party
- **Andrew Gennery**(Great North Children's Hospital, NewCastle) who will be the Chairperson of the WP Bone Marrow and Gene Therapy

They will replace **Klaus Warnatz and Bobby Gaspar** respectively at the end of their mandate. I wish them four years of intensive work and at the same time I take the occasion to thank Klaus and Bobby for their important contribution during these four years.

Finally, **Andrew Cant** (Great North Children's Hospital, NewCastle) has been elected as "President Elect" when Amos Etzioni will conclude his mandate.

During the General Assembly (GA) several **issues have been raised:**

- **ESID website**: Three different options have been presented to the GA: the first one proposed by JL Casanova was to maintain the current website; the second choice was to improve ESID website and third possibility was the generation of a new ESID website.

  The GA has voted the second option. Marta Rizzi will be in charge of the new website restyling with the final aim to make it more attractive and user friendly. Marta will show the new version of ESID website next ESID board meeting that will be hold next January.

  During the GA, Desa Lilic from NewCastle has suggested the **introduction into the website of a new topic describing the list of European Centers offering new assays used for the diagnosis of PID.**

  I encourage members to send their suggestions that are important to design a website that can satisfy the needs of every member.
- **ESID Endorsed Meeting**: ESID will provide the logo to educational activities that fulfill the scope of ESID's mission and objectives.
  You can find all the details on the website.

Briefly, the main points are as follows:

The details of the meetings (name/venue/dates organizer)
The organizers are active ESID members
The Preliminary Scientific Programme, including invited speakers

The candidate should submit the proposal to the ESID Board at least three months in advance to give enough time to the Board members to examine and understand whether the endorsement is appropriate.

During the ESID board meeting held in Florence, the Board **has approved to endorse:**

- 2nd Workshop on Diagnostics of Immunodeficiencies 2013 organized by Dr. Klaus Warnatz held in Freiburg next June.
- 3rd Meeting on Primary Immunodeficiencies for residents and fellows organized by Dr. Pere Palacin in Barcelona next May.

Importantly, financial support will be offered by ESID only to Societies such as ASID and PAGID to support travel tickets of invited ESID board members.

Finally, the President has informed ESID members about the interest of **Clinical Immunology Society (CIS) to have a dedicated corner to ESID in Journal Clinical Immunology.**

Call for Next Biennial meetings:
As Bids for biennial meetings take place four years in advance, during the General Assembly we have illustrated the three applications received from :

- Barcelona
- Manchester
- Moscow

The vote is now open until November 10th 2012 until 12:00 noon (CET).

We count on your participation to help us identifying the destination to our 2016 Biennial meeting!

Please let me know if you have any comments or suggestions. Our society is the result of your contribution!

Anna

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ESID 2012 Biennial Meeting

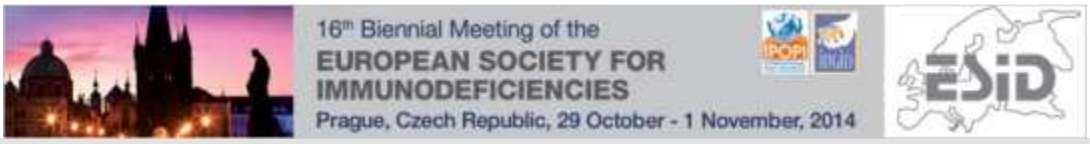
The 15th ESID meeting in Florence attracted 1837 participants from 74 countries.

The sessions were well attended and the INGID and IPOPI meetings had full halls.

We hope that you learned a lot during the meeting and also enjoyed the beautiful surroundings and atmosphere of Florence.

We are looking forward to welcoming you in Prague for the 16th ESID meeting.

Sincerely yours,
Eleonora Gambineri
ESID 2012 Congress President



See you in Prague 2014!

News : ESID Working Parties

BMT & Gene Therapy

Farewell note by Bobby Gaspar - End of mandate & Welcome Andrew Gennery

Dear All,
Its been a pleasure to Chair the BMT and Gene therapy working party over the last 4 years. I think we made some significant advances, most notably the publication of the transplant guidelines which are now used in may centres worldwide. During the last 4 years there have been many advances in transplant and gene therapy and these have been discussed and developed at our annual meetings. I would like to thank all my colleagues for their contributions and participation in studies, protocols and meetings. Your entusiasm and collaborative spirit have been extremely important to the activity and success of the Working Party. Finally, I would like to wish my successor Andy Gennery from Newcastle, the very best in this role.

Best wishes
Bobby

New BMT&Gene Therapy WP Chair A. Gennery

For those that do not know me, my name is Andy Gennery, and I work in Newcastle upon Tyne, UK, in Paediatric Immunology and HSCT.

Firstly, my heartfelt thanks to Bobby Gaspar, who has done a tremendous job over the last few years in leading the group. To have achieved consensus over conditioning guidelines is a remarkable achievement, and to modernise and integrate the data collection forms is also tremendous – thank you Bobby.

No doubt there will be many exciting opportunities as we move forward, but in the immediate future I am looking forward to (hopefully) the introduction of newborn screening, with the challenges that will bring, our closer ties with the American PID group and the possibility of collaborative studies and prospective trials, and of course the integration of developing centres.

I look forward to meeting established friends, making new ones, and together advancing our knowledge and improving our treatment of Primary Immunodeficincy diseases.

Andrew Gennery

Clinical

Farewell note by Klaus Warnatz - End of mandate & Welcome Despina Moshous

Dear all,

I would like to thank again for the time you have entrusted me as the head of the clinical working party for the last four years. It was a great honour to serve with the colleagues on the board of ESID. Many projects are still in progress and will continue.

- Diagnostic workshops: After an excellent workshop party on “Diagnostics in combined immunodeficiency” Capucine Picard, Mirjam van de Burg and I decided that we will put the information together in a short publication.

  On the 17.-19.june 2013 we will hold the second workshop on Diagnostics of Immunodeficiency in Freiburg, endorsed by ESID. Information is available at http://www.uniklinik-freiburg.de/cci/live/events/DiagWorkshop2013.pdf.

  I still feel very strong about the potential of clinical surveys under the umbrella of ESID although the path was longer and harder than anticipated.
- Splenectomy in CVID:
  Finally, thanks to the work of G. Wong and many others the manuscript: Outcomes of splenectomy in patients

with Common Variable Immunodeficiency (CVID): a survey of 45 patients has been accepted with Clinical Experimental Immunology.

- BCGitis in PIDD:
  Soon to be published (S. Rosenzweig and N. Rezaei).
- NEMO study: The multi-institutional survey of the Clinical and Immunological PHENOTYPE of NEMO-deficient patients by Capucine Picard, Jordan Orange and Jean Laurent Casanova is recruiting, please contribute all your NEMO patients.
  e-mail: Capucine PICARD
- PBSCT in CVID: The survey has been prepared and introduced at the last EBMT and ESID meeting. Marta Rizzi and Claudia Wehr (CCI Freiburg) retrospectively collect all available data on indication, management and outcome of PBSCT performed in adult CVID patients. Please contribute your patients.
  e-mail: Claudia.wehr@uniklinik-freiburg.de
- Good syndrome: The survey collects clinical and basic immunological data on patients with Good syndrome. Please contribute your patients to Anna Simon (Nijmegen)
  e-mail: a.simon@AIG.umcn.nl

Thanks again and I am happy to have a great successor.

Despina, I wish you all the best for your term and I am happy to support wherever I can.

With best regards

Klaus

New Clinical WP chairperson D. Moshous

Dear all,

Thank you for placing your trust and confidence in my abilities to chair the Clinical Working Party of the ESID. It is a significant commitment, but I am more than motivated to support and promote your interests in Clinical Immunology. On this occasion I would like to thank Klaus WARNATZ for his important work in the Clinical WP over the last 4 years. Some of the projects he launched are on-going and you can still contribute. Now the chair turns in the hands of a paediatrician, and I'll do my best to foster also joint paediatric-adult surveys.

Some information about myself: after my medical studies in Germany and partly in France (exchange programs with Strasbourg, Paris, Montpellier), I interrupted my residency in Paediatrics in order to perform basic research in immunology in the INSERM U429 laboratory (headed by Pr Alain Fischer in Paris), within the group of Dr Jean-Pierre de Villartay. I was particularly interested in the molecular basis of yet genetically undefined forms of T-B-NK+ SCID in humans due to problems in V(D)J recombination/DNA repair. The identification of the Artemis gene was the major achievement during my PhD thesis. I then completed my training in paediatrics and gained my first clinical experiences in the field of primary immunodeficiency and haematopoietic stem cell transplantation in several specialized centres for Paediatric Immunology/Haematology throughout Europe, and I had the chance to work in the departments of Pr Alain Fischer in Paris, Pr Luigi Notarangelo in Brescia, and Pr Ulrich Göbel in Dϋsseldorf.

Since November 2005, I am assistant professor in the unit for Paediatric Immunology, Haematology and Rheumatology at Necker Children's Hospital in Paris (Pr Alain Fischer). For the last three years, I am fortunate enough to work again part time in fundamental immunological research in the unit where I did my PhD (now called INSERM U768), but I continue to work in the clinical unit for Paediatric Immunology and Stem Cell Transplantation.

It is thus only natural that I am very interested in the clinical translation of basic immunology. It is an increasing challenge to transpose research timely from bench to bedside, and there is much room for collaborative trials on innovative treatment options. Improved understanding of the pathophysiology of immunological disorders and the development of specific immunotherapy opens the way for new diagnostic and therapeutic approaches for patients. One example is the management of Familial Lymphohistiocytosis. We have gained very encouraging results by using Alemtuzumab (Campath-H1) as first line treatment in patients with Familial Lymphohistiocytosis in a series of now more than 15 patients treated in our unit in the Necker-Hospital. A nation-wide protocol is about to start in France. If you are interested to use this approach, I will be more than willing to share our experience with you. This study will take part in "TREAT-HLH" initiated by Stephan Ehl.

ESID lives by the exchanges between its members, so I will be happy to get your feedback on topics that are of special interest to you. Any ESID member can propose trials to be placed on the ESID website. Each WP chairperson has now a small budget to put forward the activities of its working party, so I will be able to extend the former activities of the Clinical WP.

Looking forward to hear from you.

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Education

Dear all,

We can look back upon a very successful Educational Day
in Florence on Wednesday October 5, visited by many juniors, but many seniors as well. During the plenary lectures the sessions were made interactive by using a remote voting system. Not all participants used the voting system, and those who did not always answered every question. 114 juniors/trainees and 313 seniors/established doctors & scientists participated in the voting.

The participants were asked to indicate their primary occupation: 57 participants indicated their work was in the paediatric clinic only, 73 participants worked in the paediatric clinic & laboratory/research, 21 participants indicated their work was in the adult clinic only, 38 participants worked in the adult clinic & laboratory/research, and 73 participants indicated their work was limited to laboratory/research only. Of those who indicated their background, 51 were ESID members, 34 ESID non-members, 117 INGID meeting participants, 36 IPOPI meeting participants, and 23 participants were related to a sponsor.

So, the Educational Day does not only attract doctors and scientists! The parallel sessions moderated by seniors gave juniors the opportunity to present and discuss their work with the audience in an informal way; the same was true for the junior's poster walk, where the juniors could present the posters they had specifically submitted for the Educational Day program.

On Thursday, October 5, the Educational Working Party had organised a workshop on how to increase awareness for PIDs together with the PID-care in development Working Party, INGID and IPOPI. Short presentations were given covering the subject from various angles, ranging from the Dutch www.altijdziek.nl initiative (Esther de Vries), a website with information in separate portals for patients/parents, primary health care and secondary/tertiary health care professionals, to the efforts of a handful of immunologists in Malaysia that increased the number of identified patients dramatically within a few years (Lokman Mohd. Noh).
The remote voting system was used in this workshop as well, and showed that at least 12 ESID, 18 INGID and 34 IPOPI participants were present, as well as 6 people related to a sponsor.

Anna Sediva presented the European Immunoglobulin Map 2012; the participants showed they expected the use of subcutaneous immunoglobulin substitution to increase substantially in the coming years. Jose Drabwell discussed the great achievements made within a few years with theWorld PI Week; the next one being held April 22-29, 2013. Almost all participants indicated they were somehow involved in this initiative in their country. Mikko Seppänen conducted an extensive search of how PIDs are dealt with in 16 large medical text books of other specialties, showing the potential for a lot of improvement there. Peter Vickers stressed the potentials of modern electronic communication and social media.

Our next large event will be the ESID Summer School 2013 in Hersonissos, Crete, Greece from September 25 tot 29. Please make sure that you apply in time click here, or advise your juniors to apply in time if you are a senior!

Best regards,

Esther de Vries,
Chair Educational Working Party

Genetics

ESID Genetic Working Party (WP)
wants to help with connecting and supporting the collaborative clinical, genetic laboratories and research units associated with the ESID organization. The exchange of genetic diagnostic tools will improve the diagnosis and the genetic counselling of patients with primary immunodeficiencies (PIDs), which are important for the patients' support and care.

First, we would like to be able to help **direct our fellow ESID members**
when they have questions regarding laboratories that can be contacted for genetic testing of potential PIDs. In this regard, we would be grateful if you could answer the two questions below if you have not already done so, **by return email (capucine.picard@inserm.fr):**

- **Do you perform genetic testing for primary immunodeficiency diseases?**
- **If yes, for which genes are you performing genetic tests?**

Thank you in advance for your feedback. All information provided will be made available on the ESID website.

Second, in the context of the **technological advances in genomics, we want to start a PID next-generation sequencing (NGS) consortium**
to efficiently implement and exchange experiences with all the laboratories that use or want to use the new NGS technology to diagnose PIDs. The ESID Genetics WP would like to organize a working party meeting in March 2013 at Amsterdam with laboratories already using these new tools. If you are interested in participating in this consortium, **please contact Capucine Picard (capucine.picard@inserm.fr).**

Our best,
Capucine PICARD for the Genetics WP of ESID

Registry

Progress of database reorganisation

The Registry Steering Committee which was formed at the ESID workshop in December 2011 (see previous ESID Newsletter) has agreed on a new system for documentation. There will be three levels of documentation. Level 1 will consist of a baseline form at initial registration and a yearly follow-up short form. Level 2 will extend level 1 by a diagnosis form and a current visit form, which is specific for each IUIS cathegory. The latter shall be filled at initial registration and at follow-up once a year. The level 2 datasets were available on the ESID website for two months so that the ESID community could review them and send their comments to us. Level 3 forms will be added later for dedicated studies. A key feature is the follow-up, i.e. each patient is documented once a year.

The new documentation structure was presented at the ESID meeting in Florence as part of the "Networking in PID" session. There was a lively discussion, and we will take the points raised there into consideration during the further course of the project.

The steering committee members will meet again in December 2012 to finalize the level 2 datasets for and discuss the next steps to be taken. One major issue that has not been completely resolved is the question to which extent data that

has been entered so far can be transferred to the new database.

Publication guidelines

For the first time, the registry steering committee has defined rules for publication of data from the ESID Database. These rules define the necessary steps for different kinds of publications, e.g. publications that use some limited statistics from the ESID Database or publications that are mainly based on data from the ESID Database.

These rules have been published on the ESID website in June 2012 and are available in the Registry section of the website

Moreover the application process for study initiatives using data from the ESID registry has been formalized. We provide an application form for study initiatives in the same section of the website.

Stephan Ehl
Chairman of the ESID Registry Working Party

ESID Juniors

Dear all,

After the great and inspiring meeting in Florence we are back to work full of ideas and enthusiasm, ready to get more involved in the society and to deepen our knowledge in primary immunodeficiency.

Let me start thanking all of you for renewing my mandate as chair of the ESID Juniors. The past 2 years have been a great challenge, and at the same time a beautiful experience where I had the possibility to work together with many of you on different projects. When I started I called for help and participation and many showed availability to help, and many gave their contribution. We experienced the net that we are constructing in Florence.

We had a very well visited Summer School reunion, 85 Juniors participated to the session ESID Juniors activity, and we filled up a bar in Florence in our informal get together on Wednesday night. And more informal meeting have been organized just among you guys, meeting again friends made in one of the past schools or workshops. Collaborations have been started, information have been exchanged, advices taken and given, at the core of the ESID Juniors inspiration.

We are growing, we are growing, Juniors!

Back to the facts now! The last 6 months have been very exiting a full of learning and networking events for the Juniors, let me sum up:

- **May 2012: Freiburg workshop 'Methods in PID'.**
  The ESID Junior WP in collaboration with the Center for Chronic Immunodeficiency in Freiburg hosted 30 participants from 21 different countries and 7 faculties.

  The workshop wanted to address basic research questions in PID. How to study B cells (A. Cerutti, Barcelona and NY), T cells (F. Annunziato, Florence), Innate immunity (T. Kujpers, Amsterdam), next generation sequencing (C. Bacchelli, London), databases for PID research (B. Grimbacher, Freiburg), functional analysis of genetic defects (H. Eibel, Freiburg), diagnostic workflow in PID (H. Ochs, Seattle), those were the themes treated during these 4 intense days of workshop.

  The contribution of the faculty was enriched by the projects and data of the students. The informal atmosphere and the availability of the mentors made it a great learning and interactive event. Here you can see some picture.

- **May 2012: Prague Spring meeting** organized by Anna Sediva, you can find the report in the PID care in development page of the website.

- **October 2012: ESID Biennial Congress in Florence.**
  Eleonora Gambineri our treasurer and congress president did a wonderful job.
  In close collaboration with Esther de Vries, and with the help of 2 Juniors, Ales Janda (Freiburg) and Margje Haverkamp (Leiden) we organized the EDU day, which preceded the 'official' congress on Wednesday.

  The program was appreciated not only by the Juniors, but from all participants to the congress, surely were there more than the 250 'official' ESID Juniors! During the EDU Day, Juniors had the possibility to present their work in oral presentation, or during the poster walk.

- We had also the **Summer School reunion**, in a designated area, surrounded by the beautiful art work of the PID art exposition (thanks again Crina Samarghitean and Maaike Kusters for organizing it! See the report of Crina in the newsletter!), we had time and location to connect again to our old fellows and catch up with them.

  I am proud to say that 85 people participated at the ESID Juniors activity session, 65% of which were newly interested people, new Juniors faces.

  During this Juniors session we could use a voting system, so I had the chance to assess the past projects and collect info on the future ones.

As for the past activities:
The map of center, located in the ESID Junior part or the website, is not yet very well known and used, but ca. 97% of us think that this is a useful instrument. Therefore we will update it, so that the info available is still valid and we will advertize and promote it.

As many of you may know, we started a year ago a closed group in facebook, called 'ESID Juniors', who want to have access to it has to look for it, ask to become a member, and the administrator will allow then to enter the group.

To date we have 71 members and a lot of info exchanged. The facebook platform has been used to post free positions, to look for collaborators, to inform over grant possibilities, to advertize congresses and workshops, and to exchange pictures and impression after Junior events.

56% of us think that this a good instrument for quick information and communication, and would really invite all of you to become more active and join it!
https://www.facebook.com/groups/esid.juniors/

About 50% of us don't read the newsletter. Of course you are not in this 50%...But in the newsletter is located an important Juniors contribution 'the young researcher corner'.

During the meeting we decided we should go on with this initiative, but we will publish all the info not only in the newsletter but also into the facebook page so these will be available to download. After the ESID meeting a motivated young researcher joined the team, welcome to Stefania Giannelli (Milan), and thanks to Sara Ciullini (Florence) and Imma Brigida (Milan) for their engagement, competence and motivation!

And now, two more exciting years are ahead of us and many ideas and projects are fighting to become real…

First of all ESID Juniors will make a proposal to improve and re-organize the ESID website, please contact me if you have any idea, or suggestion.

Then we plan to track Summer School alumni to start a 'Peer Mentoring in PID': practical advice and help in early career step from young colleagues to young colleagues.

2013 will have several networking and learning events for Juniors:
17-19 June 2013 'Workshop: Diagnostic for Immunodeficiencies'
http://www.uniklinik-freiburg.de/cci/live/events/DiagWorkshop2013.pdf
organized by Klaus Warnatz in collaboration with the Center for Chronic Immunodeficiency and the ESID Junior WP

NEWS @

3-8 March 2013: ‘Advanced School in PID’ San Paulo Brazil
http://www.icr.usp.br/ESPCA-PID/

May 2013: Prague Spring meeting organized by Anna Sediva
25-29 September 2013 Hernissoss, Crete the one and only ‘ESID Summer School’!
http://www.esid.org/home-esid-summer-school-2013-apply-now-466

High quality events to improve our knowledge in PID, to get in contact with the experts in the field and to build up a net of young doctors and investigators in PID!

Some of you approached me during the ESID congress willing to be more active in the society. Any contribution is welcome! We collect time, ideas, criticism, support and enthusiasm; everybody can find a suitable way to contribute to the society. Get involved, contact me!

Wishing you all a great end of the year, and looking forward for a sparkling 2013!

Marta Rizzi
ESID Juniors WP Chair

PID Care in Development

ESID PID Care in Development WP continues with an effort to increase awareness about the field of primary immunodeficiency diseases across Europe and beyond.

To achieve this goal, we work in cooperation particularly with **ESID education and ESID juniors WPs, with J-project activity organized by Laszlo Marodi and with IPOPI**. Main activity for this summer was concentrated on already announced EUROPE IMMUNOGLOBULIN MAP.

Those of you who attended **ESID biennial meeting** in Florence Awareness Workshop could see interim results presented there. Current status of a map is attached as a presentation, and final status of 2012 edition will appear on ESID homepage. The survey brings a unique and interesting view on a situation with immunoglobulin treatment of PIDs in individual European countries and further shows the trend and changes in this area.

In 2012 the situation improved mainly for Eastern European countries. IVIG are available in practically all European countries even if a spectrum of available preparations differs. SCIG are being more and more used, spreading from North and West to East and South. The introduction of SCIG therapy is more pronounced in children population, with adults sticking to IVIG therapy. There is a number of other detailed information in Europe Immunoglobulin Map on subtly different strategies used in individual countries. European countries mostly respect ESID recommendations for a dose and frequency of applications that are uniform across the continent.

On the other hand there are substantial differences in price and spectrum of immunoglobulin preparations. This pan-European survey showed to be a very useful and interesting tool. We thank to all who replied to the questionnaire, it is very much appreciated. We plan to issue such map on an occasion of our biennial congress, with next edition appearing thus in Prague 2014.

J-project meeting series continues in 2012 with incredible 23 meetings in Eastern Europe and beyond. It is a wonderful activity that helps with diagnosis and treatment of PIDs and we thank Laslo Marodi for his continuous effort (see

http://jproject.dote.hu/ and separate chapter on ESID homepage).

We are looking forward to the year 2013 and as always we welcome all your comments and suggestions.

Kindly also find the **2012 "Europe Immunoglobulin Map" below.**

For ESID PIDCD WP Anna Šedivá
Prague, October 26, 2012

EUROPE IMMUNOGLOBULIN MAP 2012 (1351k)

Young Researcher Corner

by Sara Ciullini Mannurita

CELLULAR CYTOTOXICITY AND RELATED PID

Natural Killer cells (NK) and Cytotoxic T Lymphocytes (CTL) are classes of effector lymphocytes able to kill infected cells. Although NK and CTL use different receptor to recognize their target cell, they all contribute through similar mechanisms to destroy the target cell that has been recognized.

NK cells comprise 5-15% of human peripheral blood lymphocytes and are characterized by the expression of the CD56 surface antigen and the lack of CD3. They also express CD16 (FcgRII) a receptor molecule that specifically binds the Fc part of an antibody, by which they mediate antibody-dependent cellular cytoxicity. Indeed NK cells have both cytotoxicity and cytokine-producing effector functions and they are involved in early defenses against both allogeneic (nonself) cells and autologous cells undergoing various forms of stress, such as infection with viruses, bacteria, or parasites and malignant transformation. The cytockines production contributes to initiation of the antigen-specific immune response, making NK cells an important link between innate and adaptive immunity.
NK cell function is regulated by a balance between activating and inhibiting receptor signals. Several types of inhibitory NK cell receptors recognize MHC class I molecules on target cells and prevent NK cell cytotoxicity toward normal cells. Downregulation of MHC class I molecules on target cells may lead to NK cell¡Vmediated lysis. The process of cell target recognition is due to specific steps consisting in cell-cell contact, adhesion, formation of an immunological synapse involving specific activation receptors, granule polarization and exocytosis and target cell detachment.
In particular the mechanism of NK cells killing is the same as that used by the CTL generated in the adaptive immune response: they release cytotoxic granules on the surface of the bound target cell and the effector proteins they contain penetrate the cell membrane and induce programmed cell death. The major constituent of these granules is perforin, a cytotoxic protein able to polimerize and form transmembrane pores in target cell membrane allowing water and salts to pass into the cell that rapidly die. The other class of cytotoxic proteins are granzimes, serine proteases which activate caspase cascade and apoptosis once in the cytoplasm of the target cells. The core of lytic granules is surrounded by a lipid bilayer that contains Fas ligand and lysosomal-associated membrane glycoproteins (LAMPs). Degranulation by cytotoxic cells results in depletion of intracellular perforin and LAMP-1(CD107a) appearance at the cell surface.

NK cell deficiency states are associated with a wide range of diseases and infection susceptibility. The deficiency can be due to absence of NK cells or absence of NK cells activity.
Analysis of NK cell population by flow cytometry will overlook the majority of NK cell deficiency states. Assessment of NK cell cytotoxicity, antibody-dependent cellular cytoxicity (ADCC), cytokine responsiveness, NK cell subject phenotype and cytokine production will provide a more complete understanding of a patient¡¦s NK cell status.

A number of gene defects are associated with an impairment of NK cells cytotoxicity, and are responsible of familiar hemophagocytc lymphohistiocytosis (FHL). Defined causes include defects in proteins linked to the pathway of granule-mediated cytotoxicity: perforin (FHL2); Munc13-4 (FHL3) involved in priming of the secretory granules and their infusion into plasma cell membrane; syntaxin-11 (FHL4) involved in regulating intracellular protein transport between donor and target cell; Munc18-2 (FHL5) involved in the regulation of vescicle transport to the plasma membrane by the interaction with syntaxin 11 .
Sensitive flow-based screening assays have been developed for all subtypes. Intracellular staining for perforin focused on cytotoxic lymphocytes (CD8 T cells and
NK) can accurately identify patients with PRF1 (perforin) mutations as well as carriers.
NK cell cytoxicity is a functional immunologic parameter that has been commonly evaluated, however the assay has several limitation. The classical protocol to study NK cytotoxicity is labour intensive, usually involves radioactivity and is not widely available. It is based on testing polyclonal NK-cell populations in a 51Cr-release assay for cytolytic activity against the human myeloid leukemia cell line K562, characterized by the absence of MHC expression. Briefly, K562 are labeled with 51Cr- and plated with various concentration of patient PBMCs, the co-colture are incubated for 4hr at 37„aC and the supernatants are collected to evaluate 51Cr release by gamma scintillation The percentage of lysed target cells is proportional to 51Cr release by target cells.
Recently a new assay was set up to evaluated NK and CTL function. It is based on measurement of cell degranulation through quantification CD107a surface expression by NK and CTL using flow cytometry. Diminished expression of CD107a by NK cells

following stimulation and degranulation has been shown to predict defects in the genes encoding Munc 13-4 or syntaxin 11 and Munc18-2, providing a useful and rapid screening tool for FHL3 and FHL4 and FHL5. Two variants of this method can be identified, one to them is based on co-colture on cytotoxic cells with cell line target cell and subsequently evaluation of CD107a expression, the second assesses CD107a expression on IL2-activated PBMcs after stimulation with PHA or anti-CD3. The latter protocol has the advantage that can be easily performed in most routine immunology and hematology laboratory as no cell lines and radioactivity are required.

Moreover other genetic conditions may cause a clinical syndrome that largely overlaps with hemophagocytc lymphohistiocytosis, although they have some additional and specific features.
Griscelli Sindrome Type 2 (GS2), Chediak-Higashi Sydrome (CHS), and Hermansky-Pudlak syndrome type II, caused by muntation in RAB27A, LYST and AP3B1 respectively, display impaired cytotoxic activity.
X linked lymphoproliferative disease (XLP) it is caused by mutation in SH2D1A encoding the protein signaling lymphocyte activation molecule-associated protein (SAP) and BIRC4, the gene encoding for X-linked inhibitor of apoptosis (XIAP). In this case an overt defect in lymphocyte cytotoxicity, however intracytoplasmatic staining for SAP and XIAP can provide a rapid diagnosis.

Sara Ciullini Mannurita

REFERENCES

Bryceson YT, Rudd E, Zheng C, et al. Defective cytotoxic lymphocyte degranulation in syntaxin-11 deficient familial hemophagocytic lymphohistiocytosis 4 (FHL4) patients. Blood (2007) 110:1906–1915.

Janka GE. Familial and Acquired Hemophagocytic Lymphohistiocytosis Annu. Rev. Med. (2012) 63:233–46

Johnson TS, Villanueva J, Filipovich AH, Marsh RA, Bleesing JJ. Contemporary diagnostic methods for hemophagocytic lymphohistiocytic disorders. J Immunol Meth. (2011) 364: 1-13.

Kogawa K, Lee SM, Villanueva J, et al. Perforin expression in cytotoxic lymphocytes from patients with hemophagocytic lymphohistiocytosis and their family members. Blood (2002) 99:61–66.

Marcenaro S, Gallo F, Martini S, Santoro A, Griffiths GM, Arico´ M, Moretta M and Pende D. Analysis of natural killer-cell function in familial hemophagocytic lymphohistiocytosis (FHL): defective CD107a surface expression heralds Munc13-4 defect and discriminates between genetic subtypes of the disease. Blood (2006) 108:2316–2323.

Wheeler RD, Cale CM, Cetica V, Arico' M, Gilmour KC. A novel assay for investigation of suspected familiar hemophagocytic lymphohisticytosis. Br J Haematol. (2010) 150:700-730.

Wood SM, Ljunggren HG, Bryceson YT, Insight into NK cell biology from human genetics and disease association. Cell. Mol. Life Sci. (2011) 68:3479-3493

PIDart Expo 2012

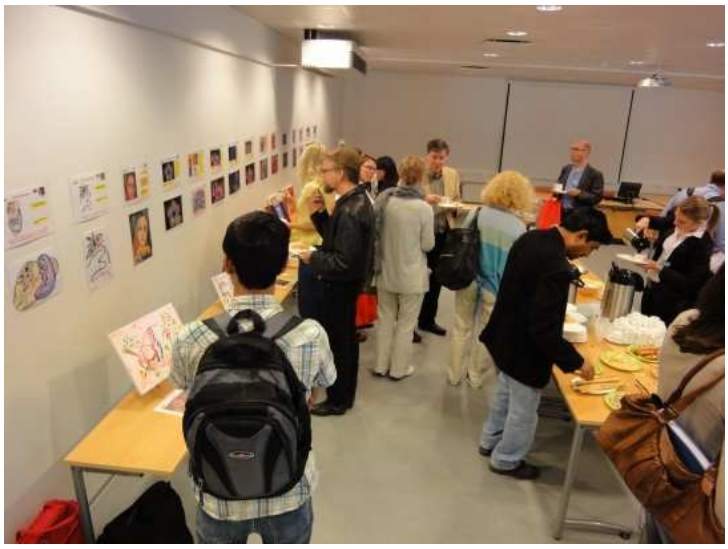
15th Biennial Meeting of the European Society for Immunodeficiencies, ESID
Florence, Italy, 3-6th October 2012

For the **'Art & Photo exhibition/competition 'Visionary PIDs'**
the organizers invite all interested conference attendees to submit images ("artworks") that have been generated as part of a research project or were inspired from the clinical work in PIDs/Immunology field. They are also soliciting images resulting from creative efforts that involve scientific concepts or employ scientific tools and methods. Artists are also invited to stretch their imagination and have a projection of ´how the PIDs/Immunology field will evolve in 2020. Most original artworks will be rewarded.
Participate in this exciting event to surprise and inspire us, to open our eyes and minds and let's learn more about PIDs in a new way!

Deadlines

- May 1, 2012 Call for Art & PIDs Exhibition Opens
- August 30, 2012 Artwork Submission Deadline

NEWS @ ESiD



PIDart Committee
Crina Samarghitean, Finland (Chair)
Maaike Kusters, Netherlands (Co- chair)

All questions pertaining to the Art & PIDs exhibition/competition should be directed Crina.Samarghitean(at)uta.fi and maaikekusters(at)hotmail.com

Exhibition
The Art & PIDs exhibition/competition will take place in Educational Day. A space has been set aside where the submitted artwork can be viewed in a relaxed atmosphere. Images will be shown both in physical form and projected as part of a slide show during the conference breaks.

Contest
Images presented in physical form will automatically enter a contest for the best PIDart award unless the artist indicates during the submission process that the work should be excluded from the contest.
Conference delegates and chairs will select the winner of the contest through majority vote based on their personal preference. Each delegate will have one vote.

Submission Guidelines

An artist can be an individual or a group of individuals. Each artist may submit one image for presentation in physical form and projection. The artist has to indicate during submission of images if the image is to be presented in physical form. If the artist submits multiple images for presentation in physical form there is no guarantee that all images will be displayed based on exhibition space.

Space for physical presentation of images is limited and will be assigned on a first-come first-served basis.
If you can not attend the conference but still have artworks to present on the Art & PIDs exhibition/competition please contact Crina.Samarghitean@uta.fi and maaikekusters@hotmail.com

Content
The artwork may deal with any theme in Science/Imunology but the organizers are particularly interested in work that is reflecting on topics related to Primary Immunodeficiencies (PIDs). While artistic freedom is a primary concern of the organizers the nature of the event requires certain restrictions on the content of the artwork. Artwork that contains sexual, political, religious themes or created for commercial purposes will be rejected.

Materials
The final artwork has to be submitted for review before the deadline through the online submission system. Together with the artwork a brief description of the work has to be submitted as well as a title for the work. The description must not be longer than 150 words.

Images to be presented in physical form must be smaller than 43 x 43 (109.2 cm x 109.2 cm). All images must be submitted in their final form and resolution. In case of images employing non-digital media a high-resolution digital photo of the artwork has to be submitted. Images can be submitted in PNG, JPEG, GIF, TIFF or PDF format. The file size must not exceed 20 MB. Artists whose images are accepted into the exhibition for presentation in physical form will have to bring the final images to the conference. For projection images will be scaled down to a resolution of 1024x768 pixels.

Submission Form
Kindly download the form below

Review
All submitted artwork will be reviewed for compliance with content restrictions as well as technical requirements. Artwork out of compliance will be rejected. Furthermore, the organizers reserve the right to reject artwork for any reasons.

Copyrights and Permissions
The artist must be holding the copyrights for all parts of the artwork or have permission from the copyright holders to use their works. Copyright holders other than the artist must be attributed accordingly in the abstract accompanying the artwork.

NB:By submitting artwork to the 'Art and PIDs' exhibition/competition the artist gives permission to the organizers to display this work and reproductions thereof during ESID Biennial Meeting 2012 and in materials associated with this.

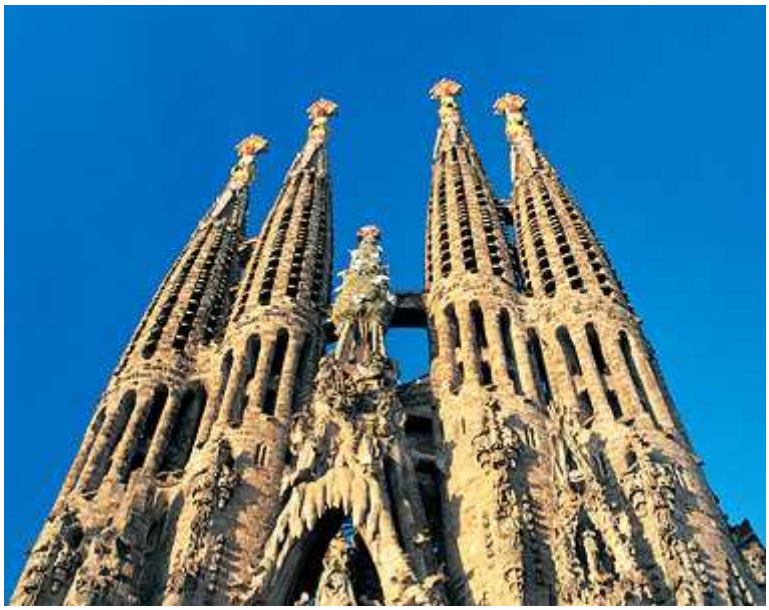
PIDArt 2012 Submission Form (45k)

ESID Elections 2012 & Call for bids ESID 2016

Meet the new ESID Board Members and find out more about the winner of the ESID 2016 Meeting Call for bids

ESID Call for bids 2016 - Congratulations to ...

Barcelona!
The ESID, INGID and IPOPI members have voted and the location of the ESID 2016 meeting will be Barcelona!
We would like to thank all bidders for their tremendous efforts and the ESID Administrative Office will keep you updated on the next steps!



The online polls for the ESID Call for bids have been completed and kindly find the official ESID/IPOPI/INGID votes on the ESID 2016 Bid below:

Total score (incl. ESID,INGID,IPOPI)

Barcelona 185 (n=votes) 45 %
Manchester 139 (n=votes) 34 %
Moscow 88 (n=votes) 21 %

ESID Elections 2012 - Meet the new board members & see the final results

NEWS @ ESID

2012 ESID Board Elections - Final Results (1001k)

Congratulations to Dr. Despina Moshous & Andrew Gennery for joining the ESID Board in 2012 as WP Chairperson Clinical & BMT & Gene Therapy!
Read their introductory messages to ESID members

Dr. D. Moshous

Dr. A. Gennery

ESID Summer School 2013

ESID SUMMER SCHOOL for Primary Immunodeficiency Diseases
25 September – 29 September, 2013
Hersonissos, Crete, Greece

The ESID Educational Working Party is pleased to announce the **2013 Summer School for Primary Immunodeficiency Diseases!**
This event, which has received very positive feedback from many participants in the past who found the course invaluable, will run from September 25 to September 29, 2013 in Hersonissos, Crete, Greece!

Faculty
For five days a well known international Faculty will be leading case-based discussions and give lecture presentations on primary immunodeficiencies. Educational Working Party Chairperson Esther de Vries and Marta Rizzi as the ESID Junior representative, together with Andrew Cant, Helen Chapel, Mirjam van der Burg, Steve Holland, Georg Hollander, Eleonora Gambineri, and Klaus Warnatz will be some of the names appearing in the Summer School faculty.

Accelerate Your Career
If you are working on a career in primary immunodeficiencies, whether in the clinic, the lab, or a combination, this will be an excellent opportunity to update your knowledge from teachers at the forefront of PIDs, hear about the latest diagnostic tools, and learn from leaders in the field who see and manage patients with the whole spectrum of primary immunodeficiency, while also networking and socialising with your colleagues.

Summer School 2011 - Barga

A little scoop…
The detailed Summer School programme will be finalized at a later stage but as ESID member you will receive web mails with updates and more detailed information on a regular basis.

Location
The Aldemar Cretan Village with its traditional and white-washed Greek architecture is situated beside the beautiful golden beach of Anissaras, Crete. Its two-storey houses provide comfortable accommodation, while the premises offer great leisure possibilities and cultural sights – click herefor more information

NEWS @

Final Selection ESID Summer School

The following participants were selected

Africa:
Leila Jeddane
Wesal Mohammed
Jonathan Peter
Nesrine Radwan

North America:
Niti Chokshi

South America:
Illeana Moreira

Eastern Europe:
Lucie Grodecka,
Veronika Kanderov,
Peter Kopac

Far East:
Yunfei An

Western Europe:
Kristian Assing
Abdullah Baysan
Stephan Borte
Giorgia Bucciol
Maria Carrabba
Tanya Coulter
Beate Hagl
Stefanie Henriet
Jenny Lingman
Myriam Lorenz
Timi Martelius
Jose Luis Martin Alonso
Tania Nicole Masmas
Beatriz Morillo
Peter Olbrich
Heidi Schaballie
Arianna Troilo
Elizabeth Ralph
Natacha Santos

ESID Biennial Meeting - Florence, Italy

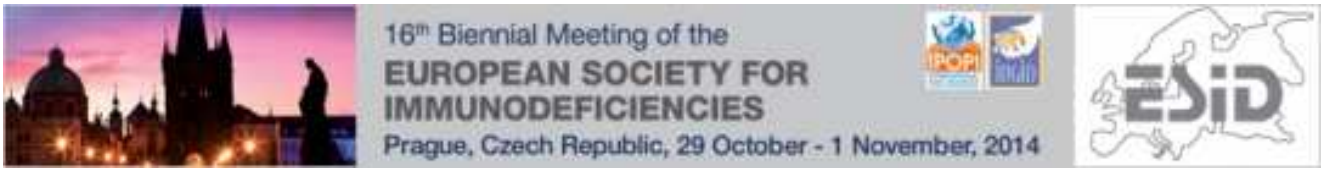
The 15th ESID meeting in Florence attracted 1837 participants from 74 countries.

The sessions were well attended and the INGID and IPOPI meetings had full halls.

We hope that you learned a lot during the meeting and also enjoyed the beautiful surroundings and atmosphere of Florence.

We are looking forward to welcoming you in Prague for the 16th ESID meeting.

Sincerely yours,
Eleonora Gambineri
ESID 2012 Congress President



See you in Prague 2014!

Picture Gallery

Please click on "Photo gallery" for a photo impression of the 15th ESID Meeting in Florence, 3-6 October 2012
Photo Gallery

Baxter-ESID Scholarship Program 2013 Fellowship Award

The European Society for Immunodeficiencies (ESID) and Baxter BioScience are pleased to announce the second time Fellowship Award, which offers an award to one individual for a 2 year fellowship program with a concentration or focus on primary immunodeficiency. The 2013 Fellowship Award will provide one individual with a two-year grant in the amount of $100,000 in total.

The objective of the grant is to support the development of academic clinical immunology research careers of advanced fellows-in-training who have demonstrated a commitment to the study of primary immunodeficiency as a career path in Europe. Funding is to support the education and research endeavors of this individual.

The deadline for the award application is May 1, 2012 (12:00 pm CET) APPLICATIONS CLOSED

Guidelines and application form can be found below.

For further information or questions, kindly contact:

Susanne Schmitt (ESID Administrative Office)
esid.admin@kenes.com

Instructions & Guidelines

Baxter ESID Fellowship Award 2013 Instructions_Guidelines_FINAL (100k)

Application Form

Baxter ESID Fellowship 2013_Application Form_FINAL (103k)

Congratulations to Dr. Margie Haverkamp

Participate in the StemPAD study

A study on the outcome of transplant for antibody deficiency and CVID in collaboration with EBMT Inborn Errors WP and ESID BMT and Clinical WP.

Dear All,

Please find details of this important study from Freiburg on the outcome of transplant for antibody deficiency and CVID.

A letter explaining the study and the study questionnaire are attached below for download.

It is likely that there are only a few patients with these conditions that have been transplanted but its important to ensure that as many as possible are captured by the study.
Please address any queries regarding the study to Marta Rizzi marta.rizzi@uniklinik-freiburg.de

Kind regards
Bobby Gaspar
Chairperson ESID WP BMT

StemPAD_sketch_V1_2 (81k)

StemPAD questionnaire V1.2 (225k)

Molecular analysis of PIDs

The ESID Genetic Working Party wants to help to connect and support the collaborative clinical, genetic laboratories and research units associated with the ESID society.

In order to facilitate access to information for genetic evaluation on primary immunodeficiencies (PIDs) to the clinicians and ESID members for their patients, please find below a list of laboratories which are caring out genetic explorations for PIDs. The list of genes performed by this laboratories are available in the excel file enclosed and availabe for download below. Prenatal diagnosis can be done in Ulm and Paris laboratories, if the mutation of the index patient is known.

Laboratories:

- Institute for Clinical Transfusion Medicine and Immunogenetics, University Hospital of Ulm Dept. of Molecular Diagnostics, Molecular Therapy and Experimental Transplantation, Ulm Germany.
  Contact: k.schwarz@blutspende.de
  Tel. +49 (0)731/150-599, Fax: +49 (0) 731/150-645

- Medical Immunologist, University Medical Center Utrecht, Dept. Medical Immunology UTRECHT, The Netherlands.
  Contact: lcorput@umcutrecht.nl
  Tel.+ 31(0)88 - 755 6573
- Study center for primary immunodeficiency (CEDI), Necker Hospital, Paris, France
  Contact: capucine.picard@nck.aphp.fr or capucine.picard@inserm.fr
  Tel. + 33 (0) 1 44 49 50 88

Useful links:

Databases for immunodeficiency-causing mutations
The portal for rare diseases and orphan drugs
RAPID: Resource of Primary Immunodeficiency Diseases
USIDNet
CEREDIH

List of genes performed by these laboratories (33k)