

# ESID Newsletter

## Contents

Introduction	...
ESID information	
President's letter	
Secretary's report	
Treasurer's report	
News & Views	
...	
Working Party reports	
...	
Focus on a country: ...	

*The ESID Newsletter* is made for the members of ESID - the European Society for Immunodeficiencies.

It is published under the responsibility of the ESID Board, and at this moment it is edited by Esther de Vries.

Any ESID member who is interested in publishing his or her views, research, new ideas or other material in the ESID Newsletter is cordially invited to submit copy to the Editor. Suitability for publication is assessed by the Editor in consultation with the other members of the ESID Board.

Editor's address:

Dr. Esther de Vries,  
pediatric immunologist,  
Jeroen Bosch Hospital  
loc GZG, P.O. Box 90153,  
5200 ME's -  
Hertogenbosch, the  
Netherlands, tel. +31-73-  
6162965, fax +31-73-  
6162948, email  
esther\_de\_vries\_nl @  
yahoo.co.uk .

*Front page:*  
*ESID Summer School*  
*2003, Algarve, Portugal*

*Dear ESID members,*

The front page shows you the very enthusiastic group of participants of the ESID Summer School 2003 in the Algarve, Portugal. Every single participant became a member of ESID, even the one faculty member who hadn't yet joined! It was a marvellous experience for everybody; you can read more about it in this issue of the ESID Newsletter.

This issue of the ESID Newsletter will bring you ongoing information about the Board's initiative with financing a Registry with the help of PPTA.

The 'Focus on a country' section shows you Germany this time, with Prof. Wilhelm Friedrich from Ulm as Established ESID member, and dr. Ulrich Pannicke, also from Ulm, as Young Investigator.

We also have a new section in the ESID Newsletter, that I will hope you will enjoy reading. It is called 'PID-care in development', and shows you countries where enthusiastic doctors are trying to build up a primary immunodeficiency care network. You will be surprised what a lot of work can be done for but so little money!

If you feel like suggesting a country or person for the 'Focus on a country' or 'PID-care in development' section, want to attract attention to your symposium, express your opinion, write a review, or anything else of interest to the ESID community, **please don't hesitate to contact me at esther\_de\_vries\_nl @ yahoo.co.uk**

Best wishes to all of you!

Esther DE VRIES, Editor



ESID is the European Society for Immunodeficiencies. It was formed in 1994. The forerunner of ESID, the informal European Group for Immunodeficiencies (EGID) was established in 1983. Anyone who is interested in primary immunodeficiency diseases can become a member of ESID. You can find the necessary information to contact the treasurer Esther de Vries at [www.esid.org](http://www.esid.org).

Within ESID, six Working Parties are actively engaged in coordinating the member's joined efforts in patient care and research in primary immunodeficiency diseases: Bone marrow transplantation (chair: Andrew Cant), Pathology (chair Fabio Facchetti), Patient registries (chair: Bodo Grimbacher), Clinical (chair: Jean-Laurent Casanova), Genetics (chair: Anna Villa), and Education (chair: Anders Fasth). Anyone who is interested in participating in one or more of these Working Parties is invited to do so. Please contact the chairman of the relevant Working Party (contact information is available at [www.esid.org](http://www.esid.org)).

In 1994, a main registry of patients with various forms of immunodeficiency in Europe was established. Altogether, data from some 10,000 patients from 26 countries were received until now. In 1995, the first locus-specific immunodeficiency mutation database accessible through the internet was established (BTKbase for X-linked agammaglobulinemia - curators Mauno Vihinen and C.I. Edvard Smith). Since then, several additional locus-specific data bases have been established: ADAbase (adenosine deaminase deficiency - curators Mauno

Vihinen and Michael Hershfield), BLMbase (Blooms syndrome - curator Mauno Vihinen), CYBAbase (autosomal recessive p22 phox deficiency - curators Dirk Roos and Mauno Vihinen), CYBBbase (X-linked chronic granulomatous disease (XCGD) - curators Dirk Roos and Mauno Vihinen), CD3Ebase (autosomal recessive CD3 epsilon deficiency - curators Mauno Vihinen and Jose R. Regueiro), CD3Gbase (autosomal recessive CD3 gamma deficiency - curators Mauno Vihinen and Jose R. Regueiro), CD40Lbase (X-linked hyper-IgM syndrome - curators Luigi D. Notarangelo and Mauno Vihinen), JAK3base (autosomal recessive severe combined JAK3 deficiency - curators Luigi D. Notarangelo and Mauno Vihinen), NCF1base (autosomal recessive p47 phox deficiency - curators Dirk Roos and Mauno Vihinen), NCF2base (autosomal recessive p67 phox deficiency - curators Dirk Roos and Mauno Vihinen), RAG1base (autosomal recessive severe combined RAG1 deficiency - curators Mauno Vihinen and Anna Villa), RAG2base (autosomal recessive severe combined RAG2 deficiency - curators Mauno Vihinen and Anna Villa), SH2D1Abase (X-linked lymphoproliferative syndrome (XLP) - curators Luigi D. Notarangelo and Mauno Vihinen), TCIRG1base (autosomal recessive osteopetrosis (arOP) - curators Mauno Vihinen and Anna Villa), ZAP70base (autosomal recessive severe combined ZAP70 deficiency - curator Mauno Vihinen), WASPbase (Wiskott-Aldrich syndrome - curators Mauno Vihinen and Luigi D. Notarangelo) (information is available at [www.esid.org](http://www.esid.org)).

ESID organizes a biennial congress to facilitate international contact between primary immunodeficiency specialists. The last congress was organised in 2002 in Weimar, Germany; the next congress will be organized in Versailles, France in October 2004, and the one after that will be in Debrecen, Hungary, in 2006.

= ESID Information =

## ***President's letter***

Dear friends and colleagues,

We are half way to the next Meeting to be held in Versailles, France. Alain Fischer is preparing an exciting scientific program. We look forward to another most fruitful ESID Meeting, 20 years since EGID officially started its activity.

During these 20 years, we have been able to work together, exchange ideas, share clinical protocols. By doing so, we have altogether put Europe at the leading front in the elucidation of the pathophysiology and treatment of primary immune deficiencies.

Furthermore, the Meeting to be held in Versailles will offer another opportunity to meet with the patients' and families' associations (IPOPI), and with INGID, and thus consolidate our mutual collaboration.

More specifically, as medical doctors and researchers involved in primary immune deficiencies, it is our own responsibility to foster the promotion of a new generation of basic and clinical scientists that will take over these very same tasks in the years to come. It is under this concept that ESID has started Educational Summer Schools during the last few years, first under the guidance of Helen Chapel, and more recently of Anders Fasth. Following the great success of the Educational Day that preceded the ESID Meeting in Weimar, the ESID Board and Alain Fischer have jointly decided to replicate an Educational Day in front of the ESID Meeting in Versailles.

Meanwhile, I wish to express my greatest appreciation of the fact that the participants at the last ESID Summer School have proposed to create an ESID-Juniors Working Party. This idea, which has been favourably met by the ESID Board, will require a formal approval by the ESID Assembly. In the meantime, I believe it follows the long-awaited request for a new

generation of young trainees and clinical scientists that work together to promote ESID activities.

In this issue of the ESID Newsletter, Necil Kutukculer reports on a scientific Meeting that has taken place in Turkey, and that was very much focused on primary immune deficiencies. **I encourage other ESID Members to report on similar initiatives in their own countries**, and thus contribute to the knowledge that Europe as a whole is continuously involved in the promotion of awareness of primary immune deficiencies and in the improvement of standards of clinical care.

With these expectations, I look forward to an exciting ESID Meeting in 2004, and encourage an active participation from all ESID Members. During the Meeting, we will have to vote for new Chairmen of several Working Parties, and for the President-Elect. **Candidatures are welcome.** Please, do not forget to renew your ESID membership now! Each of you are essential to the life of our Society!

Luigi NOTARANGELO

## ***Secretary's report***

From the last ESID Board meeting on October 27<sup>th</sup>, 2003:

The ESID board met for its second meeting of 2003 on Oct 27 at Frankfurt Airport. The two main topics on the agenda were the next biennial ESID meeting in Versailles, October 21<sup>st</sup> to 24<sup>th</sup>, 2004, organised by Alain Fischer, and the current progress made in establishing a new online-version of the ESID patient registry.

The organisation of the next biannual meeting proceeds with admirable professionalism. Alain Fischer reported that the meeting site in Versailles is easily accessible using public transport and has a good and relatively short connection to the next airport at Paris. The ESID board at this occasion decided that accessibility of the congress site to all ESID members traveling from near and far should also be paid attention to in the organisation of future biannual meetings (i.e. the meeting site should be located not more than one hour away from the next airport). The Palais des Congrès in Versailles represents a magnificent place neighbouring the famous Versailles castle and its gardens, a location that will certainly contribute to make this meeting a remarkable event. The scientific committee headed by Alain Fischer now made suggestions for the scientific program, which were discussed in depth by the ESID board. Topics of the main symposia will be defects in innate immunity, susceptibility to human papilloma virus (HPV) infection, and defects in immune homeostasis. In addition there will be workshops dealing with the pathophysiology, diagnosis and treatment of antibody deficiencies, as well as with stem cell therapy and gene therapy in primary immunodeficiency diseases, immunodeficiency databases and registries, and the pathophysiology and management of congenital neutropenia. Several companies have already expressed their interest to sponsor the next ESID meeting, with hopefully many more to join them in the near future. At the last meeting in Weimar the first Educational Day at an ESID biannual meeting was organised, and the interest demonstrated by the large number of participants was overwhelming. At the next meeting in Versailles Anders Fasth and Esther deVries plan to continue this success story and organise an educational day about defects in innate immunity, phagocyte defects and immune dysregulation, while in the other half day the latest from clinical topics on the diagnosis and management of primary immunodeficiency disease will be discussed, with the aim to give guidelines that help the ESID members in their clinical work with suspected and proven primary immunodeficiency patients. The ESID board unanimously agreed to the proposed scientific program and thanked Alain Fischer and the other members of the scientific committee for their successful efforts in choosing interesting and very relevant topics. Alain Fischer further told the ESID board about the next milestones in the organisation of the Versailles meeting: the scientific program will be finalized soon, a second announcement will follow at the end of the year, while abstract submission (via electronic mail and internet only) and registration should follow in summer 2004. Close communication with INGID and IPOPI regarding the finalized scientific program will enable optimal coordination of the three parallel programs. One important message to the ESID members: the next general assembly should take place on Friday 22<sup>nd</sup> October, 2004, in Versailles at the occasion of the next biannual meeting. All members are cordially invited to attend this assembly, thereby providing as much input as possible to important decisions taken at this occasion, e.g. the election of new members to the ESID board.;

Bodo Grimbacher then updates the ESID board on the most recent developments regarding the realisation of the new online-version of the ESID patient registry. Attending the meeting were Dominic Veith, who is responsible for information technology matters concerning the realisation of the database, and Barbara Frisch, the future database monitor. They were ready to answer questions of the ESID board concerning more technical matters, e.g. database security, equipment, monitoring of the local centers' input etc. Bodo Grimbacher explained to the board the data set of the CVID subregistry, the first subregistry of the ESID database to be realized. While the ESID main registry is more comparable to the existing ESID patient database with important improvements such as updating on a regular basis and more contents, a disease-specific subregistry such as the CVID registry should contain an extended set of

parameters covering important clinical, therapeutic and immunological issues. PPTA still expressed their interest to sponsor the online registry, and the respective contracts are now in their last stage of approval. One crucial point will be whether the ESID members are willing to contribute data to the ESID registry. Bodo reported that as a response to a recent email, more than 100 centers expressed their willingness to share patient data with the ESID registry. In the disease-specific subregistries, only patients that are actively seen will be entered, as the patients' permission has to be documented in a consent form. All objections of the local Ethics committees will be taken care of, including their comments to the informed consent form. Additional ESID-subregistries will then be created on demand.

The ESID board further plans to support a proposition to the WHO to reintroduce immunoglobulins to the live-saving drugs. In addition, the creation of a new working party is proposed by Esther deVries called the ESID-juniors working party, which should especially take care of the demands of younger ESID members. This project finds full support from the ESID board. The chairperson and the other members of this working party shall be elected at the next general assembly.

Hermann Wolf