

e-Update

July 2006



The global organisation working to improve the quality of life for people
with primary immunodeficiencies.

IPOPI is a Charity registered in the UK, registration number 1058005

The new LOGO

Did you spot the difference on the front cover? Yes, we have a new logo! Our old logo has served us well for many years now but during our various and many conversations about the Strategic Plan over the past year and more we kept coming back to the fact that our logo fails to convey the message that we are about people! So, we have kept the globe – our international emblem – in a new format and put in the middle of a ring of people! We have also introduced a new colour – they are orange tones and when you come to Budapest you will receive our new leaflet, a new poster and a card folder – all with our new messages upon them! So there you are – another reason for coming to Budapest

Budapest 2006

In October you have a excellent opportunity to learn new things about primary immunodeficiency, meet and discuss with people from around the world, share other cultural experiences, find out what IPOPI is doing and planning to do – as well as electing the Board who will carry forward the work of IPOPI through to 2008.

As a Board we are all very excited at this opportunity to meet and share and exchange thoughts and ideas for the future and we want YOU to be there too! Budapest is a beautiful

city and we will all benefit from being in the wonderful city!

So, if you have not booked yet, do so soon and make sure you catch up on old friends as well as making new friends! Go online today at www.esid2006.com and register!!

The following members and emerging members of IPOPI received grants to enable them to attend Budapest: India, Argentina, Brazil, Iran, Serbia-Montenegro, Morocco, Estonia and Germany (IMMI).

The IPOPI AGM will be held during the meeting in Budapest. Very soon all NMOs will receive a copy of the papers for the meeting. It is important that they are discussed and considered very carefully. There will be the Audited Accounts and the other usual pieces of paper – but there will be other new material as well!

So do make sure that the relevant people set aside reading time to consider a new proposed Rules of Procedure and – maybe more controversially – a proposal for membership fees from NMOs. Now, before you get too excited, the subscriptions from members will be modest and will not account for a major part of IPOPI's income!! The reason for seeking them is that our funders feel that modest contributions from NMOs show commitment to IPOPI and also demonstrate that IPOPI is on a professional footing. You can still be a member of IPOPI and not pay your NMO membership fee, but by not

subscribing it could be that there would be no voting rights or ability to stand for election to the Board. Those papers will be coming and in order to save time we would like to hear questions in advance so that we can answer them and not leave things hanging over for another two years!

IPOPI's role in EMEA

During the past few months IPOPI became recognised as an organisation appropriate to work alongside the European Medicines Evaluation Authority (EMA). Although we have done this for many years it is now a requirement that all such organisations must fulfil certain criteria in order to continue this work. Our regular delegate to EMA meetings is Mrs Jose Drabwell. Not only living in the London area – where EMA is situated – she also speaks several European languages which makes her role the more interesting and valuable to us.

Jose recently attended a two day meeting at EMA at which the use of immunoglobulin was the subject. Jose writes:

“David Watters and I attended a workshop on IVIG in London at the EMA offices on the 5th and 6th July. The aim of the meeting was to discuss the use of IVIG in primary immunodeficiencies as well as the need for this preparation to be used in other conditions. Separate presentations were made with regard to the use of IVIG in paediatrics, where of course the main issue was to improve the health of children.

It is important that the guidance for the use of IVIG should be internationally consistent, but this should most certainly be the case in Europe. Again the absolute need and usage of a registry of primary immunodeficiencies was pointed out. It is an essential tool when information is sought or if a clinical trial needs patients with a specific condition. If diagnoses and treatment are to progress there is a definite need for more clinical trials.

Further discussions took place about the frequency and dosage of IVIG in relation to recurring infections and hospital admissions. Safety and side effects stimulated a lively round of questions and answers. Naturally it is essential that any adverse effects are reported both speedily and effectively.

The demand for immunoglobulin is growing and there is no synthetic replacement. Although Ig is interchangeable, one must not forget that there are slight differences in the various preparations. David spoke about the potential for shortage of Ig and presented all the participants with an early copy of the “draft EUPID consensus statement”.

Many positive comments were received about this document.

Presentations were made for the use of IVIG to be included as an established treatment in neurological/secondary immunodeficiency conditions. More results from clinical trials are called for.

After two days of impressive presentations and lively debates just on IVIG, David and I came to the conclusion that it was a very important meeting and we were very pleased to be included in this workshop as the representatives of all the patients who have a life-long dependence on immunoglobulin”.

Jose Drabwell

IMMUNOGLOBULIN and WHO Essential Medicines List

In our last Update – in April! – we reported on our work on this important topic. Since then we have been to a meeting in Geneva with the relevant WHO officials and held several telephone conferences and face to face meetings in order to take this work forward. We are happy to report that the work is well advanced and on target and we are especially grateful to Charles Waller and Johan Prevot from PPTA; Theo Evers and Ann Clarke at IPFA; Dr Helen Chapel representing ESID and our very own Dr Teresa Espanol for their enormous input to this task.

We will very soon be in a position to share an excellent submission with all our NMOs and other readers and by mid-October it must be sent to WHO and it will be on their website for wider comment until the Committee meets in March 2007 to consider the application. We will be able to make a short verbal presentation to the Committee before they discuss the application in camera.

IPOPI exists to:

- **Work to improve access to early diagnosis and optimal treatment and care**
- **Promote the establishment and support the work of National Members Organisations (NMOs)**
- **Be a strong effective voice for NMOs**

5th ESID Spring Meeting Prague 2006

IPOPI Board Member Eva Soergel writes:

IPOPI was again invited to participate to the ESID Spring Meeting for immunologists from Eastern Europe in Prague.

I was very proud to have the opportunity to once again represent IPOPI. This year I spoke about the "Perspectives of IPOPI" to inform about many changes in IPOPI in the last year. The Board members



of IPOPI have been working on the new Strategic Plan which will be presented at the Budapest

meeting in October. One of the changes consists in the new and broader vision of IPOPI. It says: IPOPI – the global organization working to improve the quality of life for people with Primary Immunodeficiencies.

Participants of the Meeting came from the Czech Republic, Great Britain, Russia, Poland, Hungary, Slovakia, Lithuania, Romania, Estonia and Germany.

Dr. Sediva from the Prague Motol Hospital gave information about the reimbursement for intravenous immunoglobulin in the Czech Republic.

The Meeting took place in a wonderful friendly and familiar atmosphere – like every year.

highlighted how an early diagnosis and appropriate therapy can modify the prognosis and the quality of life for these patients. The aim of the round table was to learn from the experience of "sister organisations" how to become an effective organisation.

Doris Theato, from the German Organisation DSAI, talked about her experience as a mother of 3 children with agammaglobulinemia and of her involvement in DSAI.

The Italian Organisation, AIP, was represented by the Chairman Michele Del Zotti, Gloria Beretta and Bianca Pizzera. Michele described the work of AIP and their collaboration with the national network for PIDs and stressed the importance of being part of an international organisation like IPOPI that bring together so many different experiences and cultures but with the same intent: to fight primary immunodeficiencies.



MUSIC, CULTURE & SOLIDARITY IN VIENNA

A concert followed by a round table took place on the 5th and 6th of May at the Italian Institute of Culture in Vienna. The event was organised by OESAI, the newly established Austrian patients organisation for PIDs, with the help of Rita, a Young Italian researcher who lives and works in Vienna and who was the inspirer of the event.

The purpose of the evening was to provide information about OESAI and to raise awareness on PIDs among both patients and the general public.

During the evening performances of classic music, madrigale, bossa nova and a chorus, alternated with several speakers. In particular Prof. Hermann Wolf provided an overview of PIDs as rare disorders which vary in incidence and severity and

AIP SUPPORTS RESEARCH

Thanks to private and corporate donations and to the intense fundraising activity of our members from all over Italy, AIP has financed important research projects:

- *Study of the molecular mechanism underlying primary defects of humoral immunity: Supervisor Prof. Plebani;*
- *Development of molecular test for the diagnosis of PIDs: Supervisor Prof. L. Notarangelo;*
- *Study of pathogenetic aspects of PIDs: Supervisor Prof. A. Plebani;*
- *Study of the trascriptional factor AIRE and Foxp3 in the development of autoimmunity: Supervisor Prof. L. Notarangelo;*
- *Study of the molecular mechanism of PIDS: Supervisor Prof. A. Plebani*



PRIMARY IMMUNODEFICIENCIES

European Primary Immunodeficiencies Consensus Conference

19 – 20 June 2006
Paul-Ehrlich-Institut, Langen, Germany

DRAFT EU PID CONSENSUS STATEMENT DRAFT

Together, the attendees of this conference agreed a Consensus Statement on PIDs with the intention of using this, as well as the conference's forthcoming recommendations and report, to:

- Communicate to EU governments the extent of the negative impact PIDs currently have on healthcare systems and undiagnosed patients.
- Demonstrate the disparities of care and treatment that exist for people with PIDs across the EU.
- Provide examples of immediate actions and initiatives that EU Member State governments can take to reduce the burden of PIDs.

The following Consensus Statement focuses on three key areas where priority action is needed:

1. Awareness and Education
2. Screening and Diagnosis
3. Treatment and Management

1. Awareness & Education

Consensus Statement

PIDs are widely under diagnosed.

Early identification of PIDs will:

- Save lives,
- Improve health, quality of life, and lifespan in identified patients through adequate treatment,
- Enable genetic counseling and prenatal diagnosis within the family.

Tools for identification of PIDs are:

- Diagnostic guidelines for recognition of symptomatic patients,
- Appropriate immunologic and genetic laboratory tests,
- Screening tests for suitable diseases.

Recommendations and tools needed

I. Gathering information

Clinical protocols are needed to reliably identify PIDs; these can be created by development, implementation and evaluation of:

- Diagnostic guidelines on a scientific basis,
- Standardized diagnostic criteria for PIDs.

Assessment of the impact of PIDs on the community is needed; this is enabled by epidemiologic studies to assess:

- The prevalence and incidence of PIDs in the population,
- The impact of PIDs on public health,
- The impact of PIDs on health care costs.

International PID registries enable future diagnostic processes by identifying:

- The pattern of clinical presentation of these diseases,
- The natural history of the various PIDs (morbidity, mortality, complications),
- Relationships between clinical disease patterns and genetic backgrounds.

II. Appropriate diagnostic tools

Practical tools for efficient diagnosis of PIDs are needed in every country; this is enabled by availability of:

- Simple diagnostic tests at the local level,
- Immunologic tests in specialist diagnostic centres at the national level,
- Elaborate tests through networks of excellence across Europe.

Appropriate evaluation of diagnostic tools is needed; this is enabled by:

- Development of age-matched reference values for all diagnostic immunologic tests,
- Regular quality control of immunologic laboratories.

Research on the feasibility of screening programmes for PIDs is needed to prevent damage, including:

- Development of suitable tests,
- Assessment of costs and benefits,
- Evaluation of ethical aspects,
- Development of management guidelines for identified patients.

2. Screening & Diagnosis

Consensus Statement

General public

- There is a lack of awareness of PIDs amongst the general public.
- There is misunderstanding of the impact of PIDs on schooling, work and social life of individual patients. This is especially important in terms of lack of avoidable loss of working days.
- The huge differences between PIDs and HIV/AIDS are not understood
- Lack of infectivity of PIDs is not appreciated and has to be presented as genetic even if multiple genes like diabetes.

Healthcare professionals

- Due to a failure to include applied Immunology within professional training programmes, there is a lack of awareness of PIDs by:
 - First line healthcare (family GPs, doctors, nurses, midwives) i.e. lack of awareness of symptoms
 - Secondary healthcare (doctors in community and teaching hospitals) i.e. lack of understanding of availability and efficacy of treatments
 - Allied professionals (physiotherapists, dieticians, genetics nurse specialists, pharmacists, psychologists, dentists).

Healthcare policy makers and implementers

- There is a lack of awareness of PIDs in healthcare policy makers, at national and EU level.
- Information for immunisation campaigns for those individuals whom vaccine fail to protect

- There is a lack of awareness of PIDs in healthcare implementers i.e. managers, insurers and pharmaceutical companies (antibiotics, vaccines).

Recommendations and tools needed

General public

To increase awareness of PIDs, public health campaigns and educational programmes are needed; this is enabled by development, implementation and evaluation of:

- Updated, translated (for non-native speakers) and adjusted (for special groups) material used for the recognition of potential patients,
- Material suitable for primary school curricula, including books, leaflets, letters for parents and information for school nurses to distribute.
- Material suitable for public health campaigns world-wide; this might include a “Beat the bugs day for primary immune deficiencies” annually, hiring a public relations company to provide appropriate branding such as TV banner headlines for websites, standard advertisements (for ease of recognition) to be used (with translations) in all EU member states
- Inclusion of a PIDs story line in national TV soap-operas

Concentrating on:

- The difference between PIDs and HIV/AIDS
- Emphasising the lifelong nature of PIDs and their treatability.
- Infections: appreciation of the natural frequency of infections over a lifetime and how to recognise abnormal numbers / severity of infections
- Basic understanding of the mechanisms of immunisation, understanding the components involved and the implications of vaccine failure.

Strong patient groups can help to achieve this.

Healthcare professionals

To increase awareness of PIDs, better education is needed; this is enabled by:

- Provision of standards for basic and applied immunology training in the core content for medical & nursing schools, with particular emphasis on PIDs,
- Coupling nurse education with protocols for vaccine failures and recognition of excessive numbers of infections,
- Integrating basic and applied immunology teaching, particularly alongside immunisation, into programmes for training fellows in general paediatric, internal medicine, rheumatology, respiratory medicine, and infectious disease,
- Distribute information used for education of all groups on accessible websites
- Enabling accrual of educational credits from shared material
- Reciprocation of information on PIDs, including guidelines and education, at professional meetings of related medical specialties.
- Including PIDs as a topic in continued professional development for related medical specialists in career posts, physiotherapists, nurses and midwives.

Healthcare policy makers and implementers at EU and national levels i.e. EU level: Institutions, Parliament, Member states, EMEA. National level: regulators, legislators, national advisory bodies (such as NICE), Insurers. World-wide level: WHO, pharmaceutical companies, vaccine manufacturers

To increase awareness of PIDs by:

- Studies on impact of diseases and therapy, coupled with epidemiology, public health impact and cost effectiveness studies to demonstrate savings and improvement in quality of life,
- Strong patient organisations in all EU countries, with identification of prominent patient advocates,
- Easily accessible information for health managers/insurers (e.g. leaflet)
- Regular publications from national registries

3. Treatment & Management

Consensus Statement

Effective therapies for PIDs exist.

Early treatment saves lives, prevents morbidity and improves quality of life.

Experts have reported that early treatment of PIDs is cost effective.

Treatment safety is a priority.

There is a significant disparity of care within and across EU Member States:

- There is a lack of specialised care in many countries,
- There are wide variations in the availability and funding of existing therapies,
- The availability of self treatment at home is inconsistent throughout the EU.

There are not enough trials for new therapeutic strategies.

Variation in methods in post marketing surveillance trials of products makes effective comparative analysis difficult.

Recommendations and tools needed

Develop and implement European guidelines which provide equal access to treatment within the EU, assuring a optimum standard and quality of patient care in the appropriate treatment setting.

Cross country initiatives should be developed to allow the exchange of expert experience and education in order to:

- Organise specialist nurse/midwife training courses in the EU,
- Fund medical & nurse specialists to visit other immunology centres,
- Educate related healthcare professionals.

EU treatment centre networks should be established to develop methods in order to determine disease outcomes through:

- Standardising clinical trials and post marketing surveillance,
- Using the online professional registry facility from ESID.

Adequate funding should be made available to provide:

- Optimum treatment and care in each EU Member State,
- Safe treatments,
- Support for the on-going development of the ESID registry.

Encourage and support the appropriate supply of treatment, specifically immunoglobulins, for PID patients requiring this life saving therapy.

The above is not rocket science – it is simple, practical suggestions that are adaptable wherever you are in the world – and it will be available as a full report (on disc) at the Budapest Congress on October 4-7 2006.



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