

## News from the Chairperson

Recent developments in the world of PiNSA have been as follows... our Medical Advisory doctor, Dr Monika Esser is the incoming President of ASID (African Society for Immunodeficiency) and the ASID congress will be in Cape Town in May 2013. Congratulations to Dr Esser on her achievement and it is great news for PID in South Africa and Africa that our country is hosting the congress.

The ASID congress was held in Tunisia quite recently and Dr Esser was there as well as IPOPI (International Patient Organisation for Primary Immunodeficiencies) representation in the form of Jose Drabwell (Chairman) and Martine Pergent (Board member). I am sure Dr Esser will be reporting on this event.

I have just attended an IPOPI Board meeting in Madrid and also the PPTA (Plasma Protein Therapeutics Association) Congress, both courtesy of the PPTA. The Board discussed, amongst other matters, the programme of the up and coming bi-annual joint conference ESID/IPOPI/INGID to be held in Florence in October. See <http://www.ipopi.org> for more information on this event. I will be attending as a Board member and we are endeavouring to send one of the PiNSA committee.

Other news is that IPOPI has given us funding for the printing of

materials as well as reimbursement for the surveys some of you completed last year. I have brought back examples of materials from other countries for us to use and the IDF in the USA have kindly given us permission to use their zebra logo for our fundraising (see right) if we wish to.

We want to celebrate World PI Week – see the poster at <http://www.worldpiweek.org/sites/default/files/WPIW%202012%20Poster.pdf> and also the website itself <http://www.worldpiweek.org/> by issuing a press release.

PiNSA activities still revolve around lobbying the medical aids for recognition of PID as a chronic disease, supporting patients where we can, using opportunities to market the association and so on. We are planning an AGM and will be calling for nominations to the committee but will give you plenty of warning. As always your feedback is more than welcome so please email either myself at [jgrosario@mweb.co.za](mailto:jgrosario@mweb.co.za) or Mariana du Toit, our secretary at [pinsahelp@mweb.co.za](mailto:pinsahelp@mweb.co.za)

Best wishes  
Joy Rosario  
Acting Chair



26 March 2012



### Make a donation!

PiNSA is a voluntary organisation that depends on fundraising to spread the message about PID. We know that thousands of South Africans are still undiagnosed and we have an opportunity to make a difference in their lives. Please inform our secretary at [pinsahelp@mweb.co.za](mailto:pinsahelp@mweb.co.za) if you do make a donation, we would like to thank you formally.

Account name: PiNSA  
Standard Bank  
Account Number: 07 562 322 6  
Branch: Rondebosch  
Branch Code: 025009  
Swift Number: SBZA ZAJJ  
NPO Fund Number 028-020



*Left: Members of the IPOPI Board (left to right) Sven Fandrup, Dragana Karuga, Martine Pergent, Johan Prevot, Executive Director, Teresa Espanol, Jose Drabwell, Marcia Boyle, Roberta Pena and Joy Rosario*

## Conference inspires new ideas

### **Katharine McKenzie reports back on the African School for Primary Immunodeficiencies at the ALLSA conference.**

The African PID Teaching School was held in October 2011 and formed a parallel session at the Allergy Society of South Africa (ALLSA) Congress. I represented PiNSA at the school and would like to thank the committee for the opportunity.

One of the highlights of the PID School was the presence of Dr Andrew Cant, the clinical director of children's services at Newcastle Hospital, one of the two main centres in the UK that treats children with PID. Dr Cant, who is a world authority on PID, shared a lot of information, and challenged some assumptions about the costs of treating rare diseases. He has promised to address a PiNSA patient meeting next time he is in South Africa

### **The South African PID Registry**

Dr Monika Esser discussed the SA PID Registry. The registry currently includes 188 patients. By comparison the European registry has 14 000 patients registered and compares with PID incidences estimated at between 1 /4000-10000 in the USA, 0.08 of the population in Latin America and 0.6% of the population in Australia. Marcia Boyle of the IDF estimates that PID incidence is as high as 1:1200 of the population. It was pointed out that South Africa should be diagnosing between 10 and 20 SCID patients a year.

Dr Esser highlighted the important role that the register plays in carving out a niche for PID in the South African context where we have quite an advanced medical care system, but one that is beset by major



*Delegates at the African School for Primary Immunodeficiencies.*

epidemics including HIV. The registry also provides data which enables patients to be tracked over time.

Of the 188 patients on the SA register, 134 are in the Western Cape, 28 are in Gauteng (even though Gauteng is the biggest province with 11 million residents), 7 in the Free State, 5 in KZN (with a population of 10 million), 5 in the Northern Cape, 9 in the Eastern Cape and none in the Northern Province and North West. The demographic breakdown of patients suggests that the majority of South African PID patients are unreported and probably undiagnosed, contributing to South Africa's extremely high infant mortality. Currently 52% of the registry is white, 35% coloured and 13% black reflecting the need for a different healthcare system in South Africa so that diagnoses can be made and healthcare provided to patients with PID in all provinces and communities.

Of the 188 patients on the registry 75 receive IVIG treatment and 3 receive subcut treatment which is administered off label.

### **PID cases in SA**

A number of PID cases in South Africa were presented, spanning the private and the public sector, and including rural provinces. It was inspiring to see the care and dedication of South African medical practitioners working with children and young people with rare diseases. It was also instructive to see that there is a growing awareness of PID and that diagnoses are being made, even in resource poor communities, opening the way for education and support for families who are affected by genetic diseases.

### **Implementation of the National Health Insurance**

Dr Motsoaledi, the Minister of Health addressed the conference over dinner. He highlighted the key features of the policy paper on the National Health Insurance that is being proposed for South Africa.

Some of the motivation for the NHI as identified by Dr Motsoaledi strengthens the case for more resources being dedicated to the diagnoses and treatment of PIDs in

South Africa.

In particular the Minister highlighted South Africa's dismal infant and child mortality rates, which mean that South Africa is unlikely to achieve Millennium Development Goal 4 to reduce child mortality. The Millennium Development Goals are developmental targets set for different countries through the United Nations system.

Current levels of under-five mortality rates indicate under-five mortality rates of 104 per thousand live births in 2007 and 97 per thousand live births in 2001. The current level of under-five mortality in South Africa is far higher than the international set target for South Africa which is 20 per thousand live births.

### **Bone Marrow Transplant**

Dr Cant's presentations made a number of references to bone marrow transplants, pointing out too that not all PIDs are appropriately treated with this. There has been little experience of using bone marrow transplant in the treatment of PIDs in SA.

Dr Cant challenged South Africa to think differently about BMT. He noted that a resource poor country like Jordan has successfully carried out 80 bone marrow transplants over the last five years. He also pointed out that while they are expensive and require specialised expertise, having severely ill children in the public or private health system is also very costly – not to mention the toll this exacts on the child and the family. In other words BMT is cost effective when contrasted with the ongoing cost of treatment.

Dr Cant also observed that transplant success rates were now about 90% and that BMTs were no longer seen as a risky intervention. In the United Kingdom there are two centres for bone marrow transplant in London and Newcastle.

### **Birth screening**

Screening babies at birth for PIDs is becoming more common in

developed countries. Currently highly specific birth screening is conducted in five American states, including the US's most populous state, California. Germany and the UK are also looking very closely at the possibility of introducing birth screening.

### **Sub-cut vs IVIG**

Dr Esser presented on subcutaneously administered immunoglobulins, referred to as 'subcut'.

The advantage of this method is that the Ig is self-administered and can therefore be done at home, eliminating the necessity for a hospital visit. Also steady Ig levels can be reached with more frequent and smaller infusions.

Subcut is now the standard treatment in Sweden and Norway (and has been for the last 20 years) where IVIG has been phased out.

Several quality of life improvements have been linked to the use of subcut including patients feeling healthier and compliance is reported to be better.

Dr Cant said that he was 'incredibly impressed' with subcut, and noted too that it could be administered to babies and was very well tolerated in younger children.

In South Africa sub cut is not yet supported by medical aids as it represents off label use. It would though result in considerable cost savings for the medical aid industry as it does not require hospitalisation to be administered.

### **Laboratory work**

A number of other items of interest were also discussed including the laboratory work that is done in the state National Health Laboratory Service. Dr Melinda Suchard's presentation about laboratory diagnosis pointed to the specialist skills that are required to reach accurate diagnoses. It was encouraging to see that laboratory scientists are very committed to assisting patients to reach an accurate diagnoses, even in public hospitals.

### **Lobbying for PID patients**

It was interesting and inspiring to attend this conference, and there is real value in patients and parents attending gatherings like this. Some things stand out which merit more discussion and focus from PiNSA:

1. The case studies presented on new patients in rural South Africa raise some questions about what it means to play a supporting role for newly diagnosed patients and their families who are not well resourced and who depend on the public sector for their health care. This is something that we should start talking about in PiNSA so that we are not only representative of the minority of patients who have access to private medical aids.
2. It is very unfair that subcut is not more widely available in South Africa. We need to campaign to get the medical aids to support the use of sub cut technology and to ensure that the equipment needed is readily available.
3. We have an opportunity to develop a bold vision of the kind of care that we would like children and all patients to receive in the context of the proposed National Health Insurance (NHI). The child mortality figures in the country are unlikely to change unless there is a child-patient centred approach to care, and the availability of more specialised facilities, not only for children with PID, but for other rare conditions as well. While PID may be perceived by some as a fringe issue in the context of HIV, we know there are several thousand undiagnosed children who could be getting good care and looking forward to a better quality of life. The NHI policy documents make a strong link between the quality of health services that exist in a country and its potential for economic growth. PiNSA should engage with the NHI policy documents, lobbying for national resources in support of PID and including the need to develop South Africa's capacity to carry out bone marrow transplants.

## What you need to know about Prescribed Minimum Benefits (PMBs)

**P**rescribed Minimum Benefits (PMBs) refer to conditions that if left untreated could compromise the health of an individual; of these, a chronic disease list specifies 26 chronic conditions (see list below for PID conditions).

Currently a court case between the Council for Medical Schemes and the Board of Healthcare Funders (medical aids) is underway to determine whether medical aids can stipulate the rate that treatment of PMBs should be charged at (PMBs are blamed for dramatically increasing the costs for medical aids since they were introduced six years ago), or whether the cost should be paid in full.

Regardless of this debate (PiNSA will update you on the outcome) PMBs remain important for most of us with PIDs (see the ICD10 list below).

It is important for members to know that:

1. According to the Medical Schemes Act, PMBs should be covered (in line with prevailing practice in state hospitals).
2. Medical savings should not be used to cover PMBs' diagnosis, treatment and care.

3. Your physician will probably need to have your treatment plan authorised (annually or even more often), with a code that describes your condition (see list below).
4. This authorisation, following a diagnosis (most likely confirmed by blood test results), may be through hospitalisation or chronic care, or both (possibly changing from time to time – medical aids' procedures can be erratic in this regard). You may also need to fill out a PMB application form.
5. Your medical aid may stipulate designated service providers for the treatment.
6. Be aware of your rights and enforce them when necessary; do not hesitate to call your medical aid/broker and query non-payment of claims or payment out of your medical savings. Always try to make sure you have received authorisation for treatment before you receive it.

**D80.0** Hereditary hypogammaglobulinemia

**D81.0** Severe combined immunodeficiency [SCID] with reticular dysgenesis

**D81.1** Severe combined immunodeficiency [SCID] with low T- and B-cell numbers

**D81.2** Severe combined immunodeficiency [SCID] with low or normal B-cell numbers

**D81.4** Nezelof's syndrome

**D81.5** Purine nucleoside phosphorylase [PNP] deficiency

**D81.6** Major histocompatibility complex class I deficiency

**D81.7** Major histocompatibility complex class II deficiency

**D81.8** Other combined immunodeficiencies

**D81.9** Combined immunodeficiency, unspecified

**D82.0** Wiskott-Aldrich syndrome

**D82.1** Di George's syndrome

**D82.2** Immunodeficiency with short-limbed stature

**D82.3** Immunodeficiency following hereditary defective response to Epstein-Barr virus

**D82.4** Hyperimmunoglobulin E [IgE] syndrome

**D82.8** Immunodeficiency associated with other specified major defects

**D82.9** Immunodeficiency associated with major defect, unspecified

## Are you on the PID registry?

All South African patients with PID are encouraged to provide their details for inclusion on PID Registry. The PID Registry is part of an ethically approved confidential research project at UCT and Stellenbosch universities. Dr Monika Esser, one of the doctors responsible for the register highlights the important role that this information

plays for research and lobbying. 'The registry provides objective evidence needed to change policies, inform medical aids and improve patient outcomes through data sharing'.

*For information about the registry, or to submit data contact Rina at [rina@sun.ac.za](mailto:rina@sun.ac.za) or fax number for enquiry 021-9389138/4005 or 0722100206 (mobile).*

## A useful resource

PiNSA's tech-savvy chair, Joy Rosario, has created a great online resource which compiles magazine and news articles from around the world about PID.

Check it out at <http://www.scoop.it/t/primary-immunodeficiency>



# World PI Week

22nd - 29th April 2012

Change the lives of millions this week.

Speak out about primary immunodeficiencies (PI) and help us diagnose and bring access to the right treatment for people living with PI.



**Primary Immunodeficiencies** (PI) are hereditary and genetic defects in the immune system that cause increased susceptibility to a wide range of infections, affecting the skin, the ears, the lungs, the intestines and other parts of the body. These infections are often chronic, persistent, recurring, debilitating, and in some cases, fatal.

Thousands of organisers from across the globe will join forces during World PI Week **to take these diseases out of the shadow** by hosting events to raise awareness amongst the medical profession, parents, schools, day care centres, and nurses and encourage governments to take the necessary steps to ensure that every baby, child and adult is diagnosed early and receives the appropriate treatment to lead a normal life.

For more info, visit [www.worldpiweek.org](http://www.worldpiweek.org)

**Baxter**



**CIS** Clinical Immunology Society



European Federation of Immunological Societies



**jm**

Jeffrey Modell Foundation  
Curing PI, Worldwide.

