PiNSA
PRIMARY IMMUNODEFICIENCY NETWORK OF SOUTH AFRICA
September 2013

New momentum for PiNSA

Janet Grab, Chair of PiNSA reports back on a busy year to date

PiNSA has certainly been active since our last newsletter, and it is great to see such enthusiasm and life in our organization! The PiNSA meeting supported by IPOPI at the ASID conference from 6 to 9 June 2013 was a real highlight.

The Congress was attended by 11 PiNSA members and two representatives from IPOPI, Mrs Josie Drabwell (Chairperson) and Johan Prevot (Executive Director). It was wonderful to meet patient representatives from Uganda and Morocco as well, and their experiences certainly gave us all some food for thought. You can read more about the PiNSA-IPOPI meeting in the IPOPI report on ASID included in this newsletter.

PiNSA had a table at the conference at which we displayed our awareness flyers, together with other resources for patients. There was a great interest from the medical community, especially in the booklet "Our Immune System" a wonderful resource written by Sara Le Bien and recently translated and printed by PiNSA. Bob and Sara Le Bien generously agreed to fund the translation of this booklet into Afrikaans, Xhosa and Zulu, and so this wonderful resource is now accessible to many more South African PID patients.

The PiNSA delegates held a very productive meeting at which many ideas were discussed on how the organization should work towards achieving our objectives. It was great to have fresh input and enthusiasm from the delegates. Some of the ideas include more frequent provincial patient meetings, patient information packs for new and newly diagnosed members, fundraising initiatives, awareness initiatives including use of social media to increase awareness of PID. You will hear more about these initiatives in this and future newsletters.

Myself and Dr Monika Esser were due to meet with the Minister of Health on the last evening of the Congress, but unfortunately he cancelled his visit at the last minute. The key issues that we were planning to raise with him included the necessity for Clinical Guidelines on Diagnosis and Treatment of PID, Adding Immunoglobulin therapies to the Department of Health's essential drugs list (EDL) for treatment of PID (IVIG is currently only on the EDL for neurological indications), adding PID to the list of chronic diseases described in the Prescribed Minimum Benefits legislation, advocacy and awareness of PIDs (distributing our flyers to clinics and hospitals) and training for doctors and nurses on PID. These issues will now be addressed to the Minister in a letter from PiNSA and supported by our Medical Advisory Panel doctors.

Lastly, I would like to invite you to let us know how PiNSA can best serve your needs as a person living with PID, or family member of a person living with PID. As always, please feel free to contact us with any ideas you have or initiatives you would like to get involved with. You can contact our secretary Mariana du Toit at pinsahelp@mweb.co.za or you can contact me directly at janetgrab@gmail.com.

Best wishes
Janet Grab
PiNSA Chair

PiNSA and IPOPI delegates at ASID, Sun City.

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PiNSA members reflect on the ASID conference

A sizeable delegation of PiNSA members attended the recent African Society of Immunodeficiencies congress at Sun City. It was the first time that so many patient representatives attended a gathering of this nature. Two of the delegates reflect on the experience.

Outstanding sessions enriched patient participation

"Where does one start with saying thank you. What a huge privilege to be invited to attend the ASID conference. Maybe I need to start at the beginning, driving with Joy and Mariana to Sun City. I learned so much from them about how PiNSA started out and what they have achieved up till now. Joy's example and passion for PiNSA is really admirable. It was such a privilege to meet all the other members as well as the committee members. My Thursday started out with lectures from key opinion leaders from around the world. The sessions I attended were absolutely outstanding, and realising that there has been major progress in the PID field. I also realised that the challenges and the need for more education and support is an enormous task that is lying ahead for clinicians and caregivers.

"The Ampath session I attended presented by Dr Cathy van Rooyen and Dr Sylvia van den Berg about 'Appropriate investigation for PID in South Africa' was absolutely one of the best lectures throughout the whole four days. The case studies presented and the treatment and the insight into the difficulties we as a country are faced with was insightful and practical for anyone working on a daily basis with patients. Being given the right tools to evaluate patients and also what the right steps are in regards to diagnosis was of enormous value. As a patient and as a caregiver I was really encouraged by them.

"All the clinical sessions I attended were of great value and talking to some of the doctors made me realise how many opportunities there are to make a difference in patients' lives living with PID. Although PIDs are not as rare as previously believed I do believe the task ahead is enormous. PiNSA is of upmost importance in helping patients and leading them in the right direction.

> *"All the clinical sessions I attended were of great value and talking to some of the doctors made me realise how many opportunities there are to make a difference in patients' lives living with PID."*

"The session with IPOPI was also very insightful and just realising how far ahead the European countries are in regards to PID. We can learn a lot from them and I believe their experience can help as in future.

"In short thank you PiNSA once again for the opportunity I learned so much and appreciate the change to have been able to be a part of PiNSA and the ASID conference."

Karin Ludwig
Registered nurse and PID patient

Karen Ludwig.

Delegates from Uganda and South Africa at the IPOPI session, ASID.

Hope for the future...and new friends

"Having attended this conference has really changed the way I view my disease. The week before we went I had my treatment - 50g of IVIG over 4 to 5 days – a slow and gruelling process for me with lots of side effects. I felt alone and that there was nobody who understood what I was going through. I felt that doctors and nurses do not focus on PIDs because they are rare.

> *"I had my latest treatment last week and it felt so different to me this time. I was filled with hope – I felt like I had more options in terms of my treatment and I felt like I was more knowledgeable about what was going on in my body."*

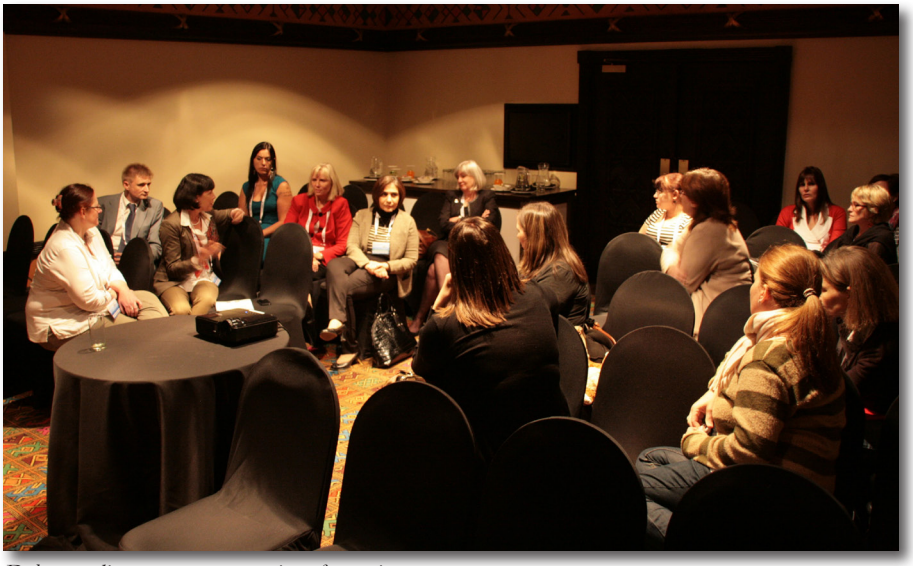
"I had my latest treatment last week and it felt so different to me this time. I was filled with hope – I felt like I had more options in terms of my treatment and I felt like I was more knowledgeable about what was going on in my body. I had new friends messaging me, ones who really knew what I was going through and I didn't feel as alone as I had felt before. I also knew how much research and work was being put into our diseases so I didn't feel as despondent about the future.

"My birthday fell on the last night that we were there, and I got to celebrate it with new friends. Something really eye opening was that there were doctors and professors also dancing and enjoying themselves on the last night that we were there. It kind of reminds you that at the end of the day, they are also human and they also let their hair down and relax sometimes. I think if can remind ourselves of this, we can bridge the gap between doctor and patient it would allow for a deeper understanding of the patient and their needs, and provide a better relationship.

"I would definitely recommend more fundraising to raise money for more members to go to the next conference. The more people that can benefit from this, and in turn pass on the knowledge to others the better."

Allyson Ben-Israel

Allyson Ben-Israel.



Delegates discuss treatment options for patients.

Mohammed Fahil of Hajaar, the patient organisation in Morocco; PiNSA chair, Janet Grab, and Karin Ludwig.

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PiNSA welcomes new Medical Advisory Panel Doctor

In June 2013 Dr Jonathan Peter joined PiNSA's Medical Advisory Panel. The Medical Advisory Panel is a panel of doctors who work closely with and provide specialist advice to PiNSA's national committee and members.

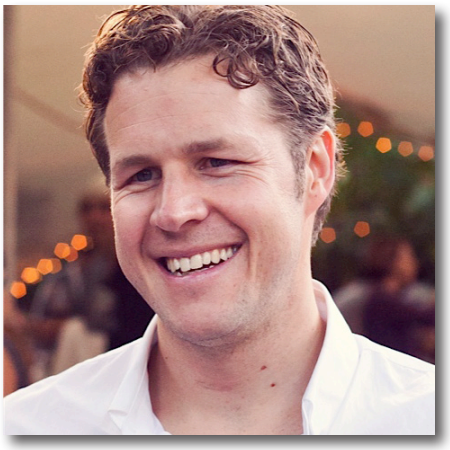
Dr Peter is a physician, specialized in adult internal medicine at the University of Cape Town. He is the recipient of several awards for medical training including the Suzman Medal for the best candidate in the Fellowship of the College of Physicians, South Africa in 2009. His clinical and research interests include: primary immunodeficiency diseases, the immune-pathogenesis and therapy of TB including TB vaccines and the development and validation of TB Diagnostics suitable for use in resource-limited, HIV-endemic settings. Jonathan has over 30 publications in high impact factor journals and has received support from several prestigious grants.

> *"I am committed to improving the care of South Africans affected by PID and being an advocate for their specialist needs and requirements. Serving as a medical advisor for PiNSA would be an important part of this commitment."*

Jonathan is currently an Oxford Nuffield Medical Fellow with a joint appointment to the department of Clinical Immunology, John Radcliffe Hospital and the Jenner Vaccine Institute, under the supervision of Dr Siraj Misbah and Professor Helen McShane. During his two-year Nuffield fellowship he will undertake further clinical immunology training in areas including primary immunodeficiency, severe allergy, and autoimmune diseases, while at the same time conducting a laboratory project to investigate a human BCG challenge model for potential use in novel TB vaccine development.

Dr Peter says, "Primary immunodeficiencies are rare but complex medical conditions. Specialist knowledge and training in their management is essential for optimal patient care and outcomes. In South Africa, PID remains under-diagnosed and under-resourced. I would like, together with organisations such as PiNSA, to help improve this. I am committed to improving the care of South Africans affected by PID and being an advocate for their specialist needs and requirements. Serving as a medical advisor for PiNSA would be an important part of this commitment."

We welcome Dr Peter to the PiNSA Family and look forward to working together in the years ahead!



Dr Jonathan Peter.

Why South Africa's PID registry is so important

Patient registries are on-going, exhaustive systems of data collection of patients with the same diseases from a geographically defined population over an extended period of time. By collecting patient data, patient registries constitute key instruments in supporting health service planning, increasing knowledge on rare diseases and supporting research by pooling data in order to achieve a sufficient sample size for epidemiological research, clinical research, surveillance of drugs used off-label, and post-marketing orphan drug surveillance. This is the view of Eurodis, the alliance that champions the rights of people with rare diseases in Europe.

"Although very thorough and effective research infrastructures, patient registries are tools that require significant time and human resources as well as financial investment and sustainability. As such, collaborative efforts are paramount at the regional, national and international levels to establish, manage and derive outcomes from patient registries," says a Eurodis factsheet about patient registries.

"The motivation to share data in the field of rare diseases is high for all stakeholders. For many rare diseases, patient registries are the only existing sources of information about the natural history of the disease (i.e. symptoms, different patient profiles and evolutions), its epidemiology, optimal clinical and social management and quality of life outcomes. For rare diseases where a registry exists, there is evidence that quality of care and life expectancy improves dramatically," says Eurodis.

South Africa's PID registry now has 216 registered patients and is managed by three collaborating universities – Stellenbosch University, the University of the Witwatersrand and the University of Cape Town.

For information about the registry, or to submit data about yourself or your child contact Rina Nortjie at rina@sun.ac.za, fax (021) 938 9138/4005 or 072 2100 206 (mobile).

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Networking with the PID community in the United States

Joy Rosario reports on her trip to the Immune Deficiency Foundation Congress in Baltimore from 27 to 29 June 2013.

My daughter Gabrielle (who has PID with a diagnosis of autosomal recessive Hyper-IgM) and myself were fortunate to visit the Unites States of America recently. The main reason for the visit was to attend the Immune Deficiency Foundation (IDF) Congress in Baltimore and this was per kind invitation of Marcia Boyle, President and Founder of IDF.

I sit on the International Patient Organisation for Primary Immunodeficiencies (IPOPI) Board with her and we have become good friends. Attending the IDF was a lifetime opportunity to meet with the PID community in the USA, there were approximately 1 200 delegates and these included PID people and their families and also medical specialists and industry. Other than the IDF, the trip was self funded and Gaby and I took the opportunity to visit New York City, we both love that city so this was an extra bonus.

Gaby has started her own project for people with chronic illness (see separate article) and we visited the Jeffery Modell Foundation in NYC so that she could present the idea to Vicki and Fred Modell. The objective was to receive guidance from them on how to take it forward, the JMF is an international initiative and operates in over 70 countries, their Facebook page has 25 000 likes!

When we arrived in Baltimore we were invited to join Marcia and her husband John for dinner with Bob and Sara le Bien. Bob was instrumental in starting IPOPI and Sara wrote the PID booklet that we had translated in South Africa so it was a very special evening.

The Congress itself, themed Zebra of course, was beautifully organised and jam-packed with workshops ranging from Healthcare and Life Management sessions to Specific Primary Immunodeficiency Disease Sessions (see the programme link – www.idfnationalconference.org). Industry was also well represented and Gaby met up with the Freedom 60 infusion pump people from whom she buys her sub-cut needles.

The social events were significant especially when, amongst other esteemed people who received awards we met Carol Ann Demaret, mother of David (the 'boy in the bubble'), who died of PID at the age of 12. There were many tears in the audience that night.

Anyway, I gathered as much as I could that would benefit PiNSA, (seven kg worth of material in fact, it's a good thing I travel lightly!) and also chatted to people in industry around strategic relationships between our national member organisation and themselves.

In conclusion, this was an important and quite emotional journey for us to have taken. We realised what amazing support there is in the USA for people with PID, that despite the sense of isolation in South Africa we belong to a much broader community, that the medical community is generous with knowledge and particularly how much the American spirit of philanthropy is so evident at an event such as this – caring and sharing. God Bless America!

For more information see:

- http://www.primaryimmune.org/wp-content/uploads/2013/03/IDF_Advocate_Spring_2013.pdf
- https://primaryimmune.org/idf-publications/patient-family-handbook/
- http://www.primaryimmune.org/about-primary-immunodeficiencies/relevant-info/additional-resources/

Carol Ann Demaret and Gaby Rosario.

Marcia and John Boyle and Sarah and Bob Le Bien with Joy Rosario.

Donate to PiNSA!

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 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

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MY POSSIBLE

MyPossible is an initiative to encourage anyone who is living with a challenging chronic or invisible illness to shift their attention from that illness onto achieving something physical that they believe is Possible for them.

We are entertained with media stories of people achieving major accomplishments (some even with disabilities or survivors) but what about those people out there who just are coping with day to day setbacks, troublesome health & the unknown?

Health is a luxury. For those born with or who have encountered illness that throws curveballs and countless hospital visits, that requires ongoing sometimes frustrating treatment, that is isolating and lonely against the backdrop of healthy people doing amazing things, **the health you do have needs to be protected and invested in.** Too often we choose to focus on and indulge 'the illness', instead of focusing on building what you already have.

Everyone has a story of overcoming hardship in whatever format, but sometimes it seems that all the healthy types are achieving big things like climbing mountains or paddling around islands in a canoe or doing IronMan over and over, and it can seem so unattainable. It is so far removed from where you may be right now, in a sterile room, stiff white linen with the hospital logo all over it, a grumpy nurse, a stressed parent and a drip stuck in your arm.

Gaby Rosario challenges all of us to do something 'possible'.

The question is simple. **What is Possible to you?**

Is it summiting Everest base camp? Is it doing a gym class? Is it walking around the block? What ever you deem Possible to you, no matter how insignificant it may seem to others – why not set out to achieve it?

In 2008 MyPossible was to cycle the Argus. I failed. I lost confidence, my health went into a spin and it just didn't work out. I did manage to complete a 44km cycle race though, so I know that I'm capable.

And now in 2013 I've decided to pick up on it again. MyPossible this time is the 5150 Triathlon in November.

The next steps now for me are to help find support (in whatever way it is offered) to achieve MyPossible – and then begin the training. It is not about the race day – it is about making sure that I do **whatever I can to preserve my health and ensure it is intact before, during and after race day.**

This blog is going to track MyPossible journey and there is Facebook too to stay connected if you're keen to help out, support & enjoy a good laugh or two.

But most importantly **#MyPossible is an invitation to create your own Possible.**

If you are suffering or lonely as you cope through an illness that seems isolating, this is a community of relative positivity, compassion, motivation, no pity parties and an encouragement to join in and create your own Possible.

Tweet me the answer to the question and I'll support you all the way – What is your Possible? Or find more of me on @gabyrosario.

Visit the MyPossible blog at http://mypossible.wordpress.com/

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PiNSA fundraiser .....raises the roof

Conrad and Joanne Amm, uncle and aunt of Seth, hosted a magnificent fundraiser in support of PiNSA at their home in Constantia, Cape Town.

Organised by Sue Gordon, mother of Seth, the 'whisky, wine and canapé' evening was a stylish and fun event. Paediatrician Dr Paul Sinclair addressed the evening, highlighting the rights of children with PID to access quality health care.

Delicious food was served by Extreem Kwizeen, who donated their services and food to PiNSA free – serving an array of different dishes throughout the evening. Music was provided by Dream Wedding DJ Brent. For the adventurous some of the Cape's best wines and sparkling wines and superb single malt whiskies were on offer for tasting. Several lucky draws resulted in several people leaving the event with lovely goody bags!

PiNSA would like to thank Sue Gordon for all her efforts in organising a sterling event – that was enjoyed by all, that raised awareness of PID and which raised more than R33 000 for PiNSA through ticket sales and a blind auction.

We also thank Intra for whisky donations, Wade Bales, Nico and Anita Breed and Groot Constantia for wine donations, and Gavin, Heather and Matthew of GW Photography, and Joanne and Conrad Amm and their family for hosting the evening and welcoming everyone to their home.