PiNSA
PRIMARY IMMUNODEFICIENCY NETWORK OF SOUTH AFRICA

July 2012

News from the Chairman

The PiNSA Committee held a national meeting and were very pleased to welcome Este Van Rensburg (Eastern Cape) and Janet Grab (Gauteng) as co-opted members. Janet is a clinical trial specialist and was nominated by the committee to attend the Florence ESID/INGID/IPOPI bi-annual meeting with me. Este is an Advocate and has joined the Lobbying group. Dr Andre Van Niekerk agreed to be an ex officio member of PiNSA, representing ALLSA. His presence will be invaluable as not only will he support our Medical Advisory Panel doctor, Dr Esser, but he will also add more gravitas when it comes to position statements and guidelines. A formal welcome to the three of you – thank you for being involved.

Other news from the meeting is that Este is starting an Eastern Cape branch and Katharine McKenzie a Western Cape one. It was decided that the MAP will produce a document, hopefully ratified by ALLSA, for South Africa on Clinical guidelines for PID. This document will give us the much needed impetus when dealing with the DoH, Council for Medical Schemes and individual medical aids. There is a revised PiNSA position statement on treatment for PID in the interim.

The website is in the process of being completely rebuilt using the IPOPI template for national member organisations (NMOs).

In my capacity as an IPOPI Board member, I attended the EURORDIS Summer School on Clinical Trials in Barcelona. This was funded by IPOPI and EURORDIS (see report). It was an opportunity to meet with other NMOs representing rare diseases and we shared common issues.

Work continues on investigating the options around treatment including sub-cutaneous therapy and the products available.

The meeting proposed that I am voted formally back in the Chair until the AGM. This will be in the form of a teleconference and has been proposed for 24th November. Relevant documentation will be sent to all members three weeks beforehand.

Best wishes
Joy Rosario
Acting Chair

5 July 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 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

Fundraiser

PiNSA information evening

Paediatric geneticist, Dr Mike Urban of Stellenbosch University and Tygerberg Hospital addressed a recent Pinsa information evening in Cape Town. Dr Urban discussed the genetic aspects of Primary Immunodeficiency including inheritance patterns and the possible role of genetic testing.

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EURORDIS Summer School for Patient Advocates in Clinical Trials and Drug Development

By Joy Rosario

Thanks to being an IPOPI (International Patient Organisation for Primary Immunodeficiencies) Board member I was accepted to attend Eurordis Summer School In Barcelona. It was a capacity building programme for patient representatives involved at the European level in the development, approval, information and access to orphan drugs, paediatric drugs and advanced therapies.

There were 37 attendees at the training from 17 countries, representing 28 rare diseases.

The organisation of the event was largely managed by Maria Mavris of EURORDIS. Each person was presented with a very comprehensive file consisting of the contact information, programme, biographies, presentations and glossary of terms.

Amongst other presentations at workshops the programme included:

- Clinical research: life cycle of drug development and phases of clinical trials
- Methodology principles of clinical trials: how to conduct a clinical trial

Introductory statistics for medical research:

- Ethical aspects of medical research involving human subjects
- Regulatory procedures
- Workshop on the review of product information by patients in preparation for potential future consultation
- EURORDIS charter for clinical trials in rare diseases

The programme was well balanced taking delegates new to the concept of Clinical Trials through from the known to the unknown. It really helped having a glossary of terms to study during the course and to make sense of at the end. The most interesting sections for me personally were on understanding the phases of clinical trials, exploring the ethics, the use of statistics in medical research, evaluating the product information and of course the committee for advance therapies, gene therapy particularly.

One was encouraged by the stringent processes in place in Europe that ensure that patients not only receive optimum care but they are also heard – their opinion is seen as an important part of the regulatory process.

Websites include

EURORDIS: http://www.eurordis.org/
EMA: http://www.ema.europa.eu/ema/
ORPHANET: http://www.orpha.net/consor/cgi-bin/index.php
US National Institute of Health website: http://clinicaltrials.gov/
CONSORT: Transparent reporting of trials http://www.consort-statement.org/
BMJ Statistics Notes Series: http://openwetware.org/wiki/BMJ_Statistical_Notes_series

Although the Summer School is for European delegates and I attended because of my position on the IPOPI Board, it was an invaluable experience especially for someone from another country. It is recommended that other national member organisations under the umbrella of IPOPI can benefit through inviting speakers on the topic of clinical trials.

I also recommend that IPOPI/PINSA and other NMO's join the rare diseases social media platform. See http://www.rareconnect.org/en

In conclusion a really important aspect of attending the Summer School was meeting the other attendees and also the presenters. I have come away with new friends who share common issues and have also benefited greatly from advice on South Africa.

I thank EURODIS, IPOPI and PiNSA for allowing me the opportunity to attend.

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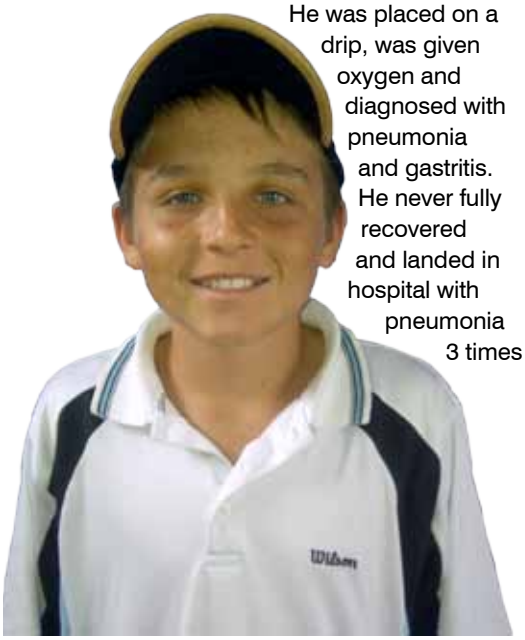
Managing hypogammaglobulinemia as a family

By Esté van Rensburg

Joshua Benjamin van Rensburg was born on 24 February 1999 and was a relatively healthy baby except for having asthma, sinusitis and regular bronchitis. At the age of four our ENT doctor removed his tonsils. Nebulizers were often used on a regular basis.

At the age of 9, he won a national tennis tournament for u/10. The next year he participated in the national tennis tournament in Bloemfontein. The second evening of the tournament, he started with a sore throat and was up all night with gastro. I took him to the doctor who reported that Josh was fine and could play tennis. The next day he collapsed on the court, could hardly move and had a temperature of 40 degrees. We rushed him off to the local paediatrician who immediately admitted Joshua. Within a matter of hours he lost his voice and was struggling to breath. He was placed on a drip, was given oxygen and diagnosed with pneumonia and gastritis. He never fully recovered and landed in hospital with pneumonia 3 times within a matter of 3 months.



After numerous blood tests, Joshua was diagnosed with hypogammaglobulinemia (more specifically CVID) and had to receive IVIG treatment (Polygam) every 4 weeks. He still regularly got ill with bronchitis and pneumonia. We eventually had to take him out of school and started home schooling. After seeing Dr. Monica Esser, she suggested Polygam every 3 weeks. He now receives Polygam every 3 weeks and does not regularly get ill. For two years, Joshua hardly grew (as this often stunts their growth), but he has now finally started growing after being on polygam treatment.

After doing 6 months research on the diagnosis, I insisted that they test Joshua's sibling, Joel, seeing that there's a 25% chance of the sibling having the same. To our shock, Joel was diagnosed with the same. Joel was permanently ill with sinusitis, bronchitis and struggled to breath as a baby and toddler. Nebulizers were a daily routine. In fact, he was worse off than Joshua and struggled with permanent ear infections, but seemed to have got better since the age of 10. The blood tests revealed that his B-cells are "faulty" and don't produce sufficient IgG and IgA. He does not yet receive Polygam, but we are checking his Ig levels on a regular basis and have taken him out of school to prevent regular exposure to germs and illnesses.

At first, I tried to be calm and did so much research, joined every single Immune Foundation in the world and tried to keep their lives as normal as possible. Then, after two years, I had a bit of a breakdown, realizing that medically speaking there is no cure and they will have to live with this for the rest of their lives. My faith in God has picked me up and kept me strong and both boys have learnt that if they want to have a relatively normal life, they also need to take responsibility for their health. They walk around with hand sanitizers and use bactroban in their noses when going out. Their friends know not to have contact with them if they're ill.

Both boys still play tennis and give 100% when they're well. They've had to learn to listen to their bodies and not play tennis when they're not feeling 100%. It is incredible how strong children are and how easy they adapt if we as parents are strong and show trust and faith in their abilities.

Their school, Grey Junior, has been incredibly supportive with the home schooling and often allows the boys to attend school for short periods. Both boys still play tennis for their school. The SANBS (SAn National Blood Service) approached me to assist them with their blood drive. They used Joshua's story and this has helped them to increase their blood donation numbers tremendously. Joshua's school also participated in the blood drive in support of Joshua and others.

Thank you to Joy Rosario and PINSA for all their support.

Esté van Rensburg, Port Elizabeth, Eastern Cape Representative

We as parents need to speak out and make others aware of this as we owe it to our children and others suffering from the same illness. If there is any one in the Eastern Cape whose children have a Primary Immune Deficiency Disorder, please contact Esté on 082 928 1057 or e-mail: careprom@mweb.co.za

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Diagnosis of antibody deficiencies

What are antibodies?

Antibodies, or immunoglobulins, are specialised proteins made by the immune system to help destroy invading pathogens. There are 5 different classes of immunoglobulin (Ig): IgG, IgA, IgM, IgD and IgE and each plays a slightly different role in the immune system.

If the immune system malfunctions and antibodies are not produced or cannot function correctly, the body can be left defenceless and susceptible to infections. Antibody deficiencies are the most common form of Primary Immunodeficiency (PID) and the most common symptoms are severe, persistent, unusual and/or recurrent infections. PIDs are usually hereditary.

How are antibody deficiencies diagnosed?

For the immune system to be assessed, a sample of blood is taken from the patient and sent to a laboratory where specialist diagnostic tests are performed. Most commonly the levels of IgG, IgA and IgM antibodies in the blood will be quantified and the result compared to the normal level expected for a patient of the same/similar age.

Immunoglobulin G (IgG) is divided into 4 subclasses and often the levels of these subclasses are also measured because these antibodies are responsible for destroying different types of pathogen.

The next step is to investigate whether the antibodies present in the patient's blood are able to function properly. This can be done by assessing how the patient responds to a vaccination.

Vaccines work by tricking the immune system into thinking a pathogen is invading and therefore specific antibodies will be produced to destroy that particular infection.

These antibodies remain in the blood long after the vaccination meaning that the individual is protected again further infection from that particular pathogen.

Specialised blood tests are able to identify the quantity of specific antibodies e.g. those made to protect against Tetanus or Pneumococcal infections.Comparing the numbers of specific antibodies before and after a vaccination allows an assessment to be made as to whether the individual can effectively make antibodies when they are vaccinated or infected.

The results from these tests will be used in conjunction with further test results and other findings such as clinical presentation of the patient and family history.

Why is a diagnosis important?

It is important to diagnose an antibody deficiency as quickly as possible to avoid or at least limit any long-term damage caused by the frequent infections.

A repeated or very severe infection can cause irreversible damage to the organ affected which is often the lungs.

A diagnosis of antibody deficiency will usually lead to treatment with replacement immunoglobulins. This gives the immune system a 'kick start' by providing it with the vital antibodies it needs to help fight infections. This treatment usually makes a huge difference to the patient's life by dramatically reducing the illnesses, which in turn can lead to fewer days off school or work and the chance to lead a near-normal life again.

In summary, it is of paramount importance that antibody deficiencies are diagnosed as quickly as possible to limit permanent damage and suffering to the patient. Specialised but relatively inexpensive diagnostic tests form an essential part of the diagnosis of antibody deficiencies which in turn influences the treatment prescribed.

Holly Cox MSc
Product Support Manager
Binding Site

For more information about diagnostic tests available for Primary Immunodeficiencies please visit www.bindingsite.com.

Binding Site

Founded in 1983 and headquartered in Birmingham, UK, Binding Site is a Specialist Protein company committed to the research, development, manufacture and distribution of innovative immunodiagnostic blood tests for the global laboratory market. With extensive expertise in antibody specificity technology, Binding Site gives clinicians and laboratory staff the tools to significantly improve diagnosis and management of patients across a range of cancers and immune system disorders.