IDFNZ intouch
KIDS FOUNDATION
April 2016

Dates for your diary

Circus Quirkus
Tuesday 24th May
Napier Boys' High School - 6pm

Wednesday 25th May
Gisborne War Memorial Theatre - 6pm

Family Conference 2016 at Butterfly Creek
25th-26th June

The Boatshed
Monday 27th June
Christchurch morning tea

Family Conference 2016

We're excited to be planning our Family Conference for 2016, with the theme of 'Living Well'.

This is a free, informative, family-friendly event with a range of interesting and informative talks from medical speakers and patients. We also offer child minding so you can get the most out of the conference - see you there!

REGISTER NOW!!!

22 - 29th April

Join IDFNZ In Celebrating PI Awareness Week

IDFNZ joins with patient groups across the globe promoting awareness of Primary Immunodeficiency (PI) and the challenge faced by patients living with these disorders.

In New Zealand we are privileged to have treatment available for the conditions associated with Primary Immunodeficiency. However, the main problem we encounter is lack of awareness of the warning signs and achieving the initial diagnosis from a specialist. To combat this issue, we developed seven warning signs that help in the diagnosis of PI.

These seven warning signs were developed to assist in early diagnosis and intervention. The cartoon poster of the early warning signs of PID has been instrumental in promoting awareness of the rare medical conditions linked to PI. All too often individuals struggle with repeated ill health and multiple infections before medical professionals reach an understanding that there is an underlying health condition responsible. The warning signs act as a reminder to look beyond the obvious symptoms as well as to be aware of what certain unusual symptoms may be indicating. Early diagnosis of these PI conditions help medical professionals in their treatment of the deficiencies, as well as enabling parents and patients to understand a little more of what they are going through.

Leading up to PI week, IDFNZ will be distributing the 7 Warning Signs posters and other PID information to GP surgeries across New Zealand for display in waiting rooms. Members can assist by requesting additional copies of posters to hand deliver to their surgery manager and suggest it be

displayed to raise awareness. This will allow patients to read about warning signs while they are sitting in waiting rooms and in doing so recognise if they may have any of the signs of PI. Other posters will be produced and given to the GPs themselves so that they are able to have them on hand in their offices and be reminded of the 7 warning signs when they are diagnosing patients. There will also be E-Training available for GPs to participate in given by ASCIA (Australasian Society of Clinical Immunology and Allergy Limited) to help GPs refresh and update their knowledge of Primary Immune Deficiency disorders.

Discover more about the amazing world of Immunology during World Immunology Week - 22-29th April

What better time to visit some cool interactive websites to learn more about immunology and how the human immune system is designed to work!

Discovering how the immune system works has been a major achievement by scientists in the 20th century and in turn has helped doctors understand why faults sometimes occur with resulting immune disorders , leading to better treatment outcomes for the 6 million patients world-wide diagnosed with PID.

Immunology is a heroic branch of leading edge science; scientific research into the immune system has saved countless human lives – some of the pioneer scientists have been awarded Nobel Prizes in recognition of their efforts and breakthroughs. Visit **www.nobelprize.org** to learn the history about these particular Nobel prize winners and their medical and scientific discoveries; an inspiration for today's budding scientists.

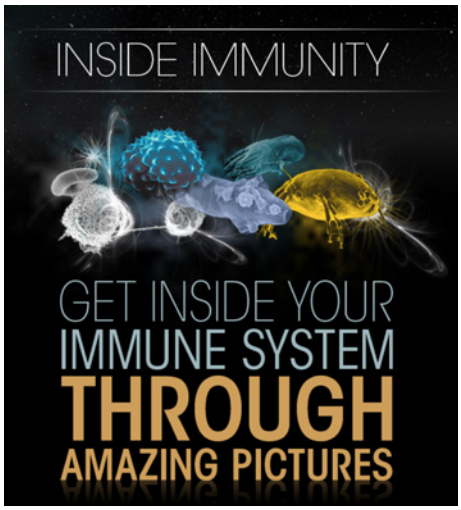
Understand the significance of immunology breakthroughs

http://www.nobelprize.org/educational/medicine/immuneresponses/

This section of the same website is a great interactive education resource for all ages; you don't have to be a genius to understand the work of the Nobel Laureates.

The site also contains fun games and simulations, based on Nobel Prize-awarded achievements. This is a great resource for all ages to read and explore interactively. We recommend you play the Immune defender game on this site - a fun way of experiencing the battles fought by the immune system.

This website also has good information on Blood Typing and the Genetic code – highly recommended!



Want to really get inside the Immune System?

See a powerful web exhibition of the immune system, created by EFIS (European Federation of Immunological Societies) with INSERM and The Institute Pasteur – visit http://www.inside-immunity.org/#explication

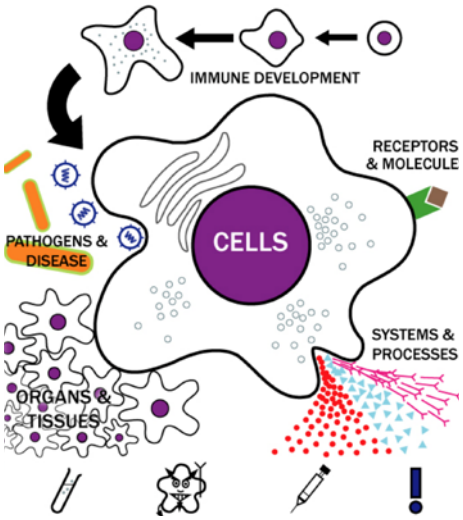
Looking inside the Immune System this way, you can see the most amazing pictures of the Immune system components and method of protection at a celluwlar level- this exhibition is virtual, accessible for everyone and specifically targeting a large audience. Great for school presentations kids!The same **EFIS** website also has an excellent downloadable book **'Your Amazing Immune System'** explaining how the immune system works.

Want printed information about the normal immune system?

Contact the IDFNZ office for a free printed copy of our Investigate Immunity information booklet series and other resources listed on our website www.idfnz.org.nz

If you have children aged 4 - 12 that enjoy online games, take a visit to the IDF website arcade of games such as Whack-a-Germ, Phagocyte Force and Jigsaw Puzzler. Based on the immune system, these games are a great way to have fun, while learning about the immune system. Although created for small children, it can be enjoyed by all ages! There is also a great video – **In Tune with Your Immune System Battle of the Bands.**
This video for older teens compares the human immune system to a rock band - it needs all the parts functioning to make great music! A battle of the bands occurs when the bad band - featuring bacteria, viruses and fungi - arrives and tries to prove who the best band is.
To view "In Tune with Your Immune System–Battle of the Bands" go to IDF's teen website, IDF Common Ground, www.idfcommonground.org, and click on the first video listed on the video player. You can also access it via IDF TV at www.primaryimmune.org/idf-tv

Visit the **British Society for Immunology** for more good information on the immune system, including both an overview and detail of specific components. This link http://www.immunology.org/Page.aspx?pid=1054 is especially suited for older students, or those seeking more specific information. This includes an online booklet giving a really good **overview of the immune system** plus **'Bite sized Immunology'** - a comprehensive series of powerpoint summaries on specific cells of the immune system for those really interested in the finite detail; What is immunology?, T cells, TREGS, Invariant NKT Cells, Natural Killer Cells, Regulatory Cells, Basophiles, CD8 + T cells, CD4 + T cells, Dendritic Cells, Eosophils, Gamma Delta T cells, Macrophages and Mast Cells etc. in scientific detail.



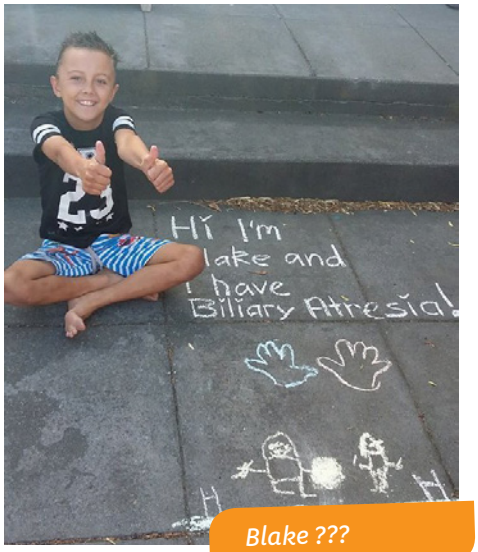
Hands Up for Rare Disease Day

Well done to all those who submitted their Hand Up for Rare Disease Day chalk drawings. It was wonderful to see so many patients putting up their hands and naming their disease. Not only was their support for the Rare Disease Day cause shown, but they also used their chalk drawings to express themselves and how they felt about their condition. Those who got creative with chalk art had a lot of fun promoting particular rare diseases and raising awareness. Thanks also to all of our amazing members who joined in with the Rare Disease Day Hands Up Campaign on Facebook – showing support by naming their particular condition was very courageous. It helped us to explain more about PID or liver disease and the important early warning signs which can assist others to be diagnosed and treated.

Congratulations to Phoebe Jaquiery (Dunedin) and Megan Shannon (Dargaville) who are our joint winners of the 'Get Arty' chalk drawing competition!

Megan Shannon

Phoebe Jaquiery



Blake ???

Harry McPhail

Family Conference 2016

25th - 26th June

Put this important date into your diary now!

This conference will be held at Butterfly Creek, Auckland, over two days filled with interesting and informative talks on the theme of 'Living Well'. This will include understanding and coming to terms with your illness and coping strategies and information to make the most of life, as well as useful tips on coping with hospital life, stress and anxiety. Our medical speakers will offer the latest medical information, and you will have the opportunity to ask questions of both our medical and patient speakers.

This is an informative, family-friendly event with formal talks for parents/adult patients. A children's programme will be running alongside to entertain 3 – 14 year olds while their parents, adults and older children are encouraged to use their time to listen to the information given without the worry of keeping younger children occupied.

The event is free to attend but members need to formally register through the website or the registration forms in this newsletter to secure a place. This is a great opportunity to learn new information and meet other families sharing a similar journey – there is strength in connecting and sharing.

Butterfly Creek is an amazing venue with fun activities for families. The conference is structured so that parents/children can have free time together to explore these at the end of each day.

We hope to see as many families as possible registering for this event. Registration forms are enclosed in this newsletter and available on the IDFNZ website.

Our People

Amber Comer

After months of the warning signs of failure to thrive, multiple and recurring infections and repeated trips to the hospital, Amber Comer was finally diagnosed with a Primary Immune Deficiency. She is symptomatic of C2 deficiency, which means she cannot fight encapsulated bacteria. These are the bugs that bring on sepsis, meningitis and skin infections, bacterial ear infections, staph, strep, Hib and meningococcal, etc. This is her story as told by her mother, Jodi Wrightson.

Amber was induced at 38 weeks' gestation due to small growth. She was born 5lb 9oz in December 2010. During Amber's first four months of life, she was diagnosed with prolonged jaundice. At 5.5 months of age, one morning about 10am Amber started to scream a high-pitched scream out of nowhere. No pre-illness, no signs of anything wrong prior. Her breathing got rapid, she had the sweats, and she refused her bottles. We did what any other parent would: Pamol and observe. A few hours later, after we realised observing and Pamol was not helping, we decided to get her in to the local medical centre. The local doctor, being rural, was out on a call at that time, so we had to wait for a call to bring her in. At 2pm that day we had the call and arrived at the medical centre to have them call an ambulance, which shipped us off to our local base hospital. Amber's heart rate was up, temperature, and breath rate, along with her inconsolable crying, and lack of interest in food showed she was not a well little girl. She went through numerous tests and we finally learnt our girl had bacteraemia plus bacterial pneumonia and pleurisy (streptococcus). In the two-week admission with many other tests, more IV lines as they didn't hold well, antibiotics, more x-rays and care, she was not getting much better. We were braced for the possibility of an airlift to Starship for a chest drain. We were extremely lucky that we dodged this, with only sheer luck and her white-blood count dropping a bit where our doctors were willing to watch and wait and continue doing what we were all doing. Two weeks later Amber was discharged and sent home on oral antibiotics. However, approximately one week after Amber finished her oral antibiotics, as scripted, she became unwell again needing medical attention. This time there was no fever, but a viral chest infection needing inhalers, steroids and a lot of care and love. This went on for six weeks. Not long after that, there were ear infections, croup and bronchitis, one after the other. She never really got a break from illnesses or needing medical attention for one thing or another. Antibiotic scripts were frequent in the months after this.

One evening in early December 2011 at 11.5 months old Amber went to bed like normal, after her meal, bath, and bedtime story. Happy, and healthy. If only we knew what was to come. At 10pm Amber woke with a high-pitched scream. We went in to check on her, made sure all was well with her and her surroundings like any other parent, before popping her back down to sleep. An hour later she woke again, this time with a high-pitched scream accompanied by vomiting. Our first reaction was, "Oh no, we have a tummy bug". Why would it be anything else? Over the night, she became sleepier, vomiting in between naps on me, curled up on the chair in the lounge. By 4am we rang Healthline. They told us she would need to be seen by a doctor. So at 7am I showered and got stuff ready, while she slept on the bed.

As soon as the GP opened we were in the door. This is when my world started to spin. The doctor did her checks, and then clear as day I remember her saying, "Have you got your car? She needs the hospital, you will be quicker than an ambulance, I think your child may have meningitis". The doctor and nurse got her prepped for the trip by giving her an intramuscular injection of antibiotics in her thigh while I rang my partner to let him know what was happening. They rang ahead and the hospital was waiting for us to arrive. My heart racing, we set off to the local hospital 85km away. I have never driven so fast (yet within the speed limit) as on this day. My heart was racing, adrenalin was pumping and I had the words of "possible meningitis" echoing through my mind the whole trip. I kept checking on her and it was the scariest drive I have ever made, knowing I was alone with a child so sick in the back, yet it was the fastest way to get her medical attention.

We arrived at the base hospital and were taken through and seen immediately. Amber had developed a petechial rash on her face on the trip over. This was when once again blood tests, IV lines, x-rays and urine bags were done, along with a lumber puncture. She was so unwell she just lay there for them, while they stuck a needle in her back. My heart broke. My baby! How can you not complain of such a thing? I sat in the isolation room waiting on my own, my partner at work, our other children at school, hoping, praying, wondering. Then the paediatric consultant who did the lumbar puncture walked in, sat on my bed, and said, "You have a very sick little girl, she has meningitis". I broke, like any other parent would. Emotions ran wild, shock overcame me. The lumber puncture confirmed Amber indeed had BACTERIAL MENINGITIS (Hib E) plus bacteraemia. A nasty infection that does indeed take life most of the time. Antibiotics were started and we were placed in an isolation room. IV lines were being replaced time and again, and with a lot of treatment needed it was decided a PICC line was the best way to go. While Amber was under general anaesthetic getting her PICC line to help with antibiotic treatment for meningitis, one of her lungs collapsed. Amber ended up in ICU for 24 hours with the diagnoses of bacterial meningitis, secondary bronchitis and a collapsed lung. It was a traumatic experience for us to witness and watch our child fight through. Yet through it all, when she could, she would always smile, as if to reassure us.

Two weeks in hospital, and Amber celebrated her first birthday on the ward. What a trooper to make her first birthday! Celebrations were held in the ward. We sure had life to celebrate. We were discharged just over two weeks later.

We went home four days before Christmas 2011 and saw in the new year. Finally, it was over… or was it? You wouldn't believe how amazing it was having our whole family home and living. We cherished every moment.

Two weeks later, in early to mid-January, once again Amber out of nowhere got a temperature. She went off her food, and I did not hesitate getting her medical attention. I was not taking any risks. We took her to the after-hours clinic, where they observed her for a few hours. It was decided she was on the decline, and an ambulance was called yet again. When at the hospital she was sleepy with a temperature, heart rate elevated, fast breathing and off her formula and food. Once again, all the tests under the sun were done, including another lumber puncture. This time it was confirmed septicaemia (blood poisoning) of a different bacterial bug.

Three times in her short year of life, she had fought three life-threatening illnesses. Why? This was the question the doctors were asking also. Once treatment was underway we were taken to Starship via ambulance where, for five days her treatment continued. She underwent MRIs, ultrasounds, and many other tests. We saw doctors, immunologists, infectious diseases consultants, all with different ideas and thoughts on what may be going on. There were different medical terms talked about from all different directions. Blood work, oh the blood work. Needles we stopped counting when we were in the high hundreds, yet she was only 13 months old.

Finally, we were transported back to our base hospital where we completed treatment, and she was discharged home. From that moment, we went into isolation, as each time we took Amber out into the community, she would succumb to something. It was no longer a risk we could take. We used hand-sanitiser, face-masks, and reduced contact with the outside world. Our only venture into the outside world was to attended medical appointments. We lost many friends, along the way; this path is not easily comprehendible. Working was no longer an option for me. My partner juggling everyday living as a dairy farmer trying to make ends meet and provide for our family, along with our other two children, who were walking this path with us. It was never nice to say, "I'm sorry, but XXXX can't come over to play" or "I am sorry I cannot watch your races or attend things at school that I am extremely proud of, due to the high risk that goes with it". It broke our hearts; we were doing the best we could with the situation that was handed to us.

Amber would still get sick, as she had siblings out in the community; we juggled life as well as we could. Many admissions meant dropping things out of nowhere and heading off to the hospital. Children rising from a night's sleep, or getting home from school, to be greeted by Dad, as once again Mum has had to run Amber in to be seen, or treated.

At the age of 18 months old, our immunologist put her on prophylactic antibiotics daily, to help keep the bugs at bay. We got a new vaccine schedule, with specialised vaccines to try and help build up some sort of immunity. At age two, once Amber was within her immunisation schedule, we started to venture out. One trip out would mean hand-sanitisers, and to an outdoor activity, where not many other children were playing or people were around. She was two years of age before she played on her first playground. For 48 hrs afterwards, I sat, fretting and waiting, for what may arise. Slowly but surely, we were able to venture out more, and although there were times we did end up back in hospital, we have much shorter admissions. Who would have thought a childhood fever sometimes can't be observed and treated with Pamol on the couch? We have learnt that we need to move fast, getting her help at the first sign of illness and getting her treatment, as this could mean life or death.
With the help of the special schedule immunisations, the prophylactic antibiotics daily, and our personal house rules, we started to do a few things as a family, friends could come and play again and we could go on family outings.

Amber was diagnosed with C2 deficiency, which means her body can't fight bacteria; the bugs that bring on sepsis, meningitis and skin infections or bacterial ear infections, staph, strep, Hib and meningococcal etc. This will be a life-long juggle for her and our family. However, with having a great medical team from Starship and our local base hospital, getting her seen with any sign of illness, not delaying treatment if presenting unwell, keeping up her prophylaxis, special vaccines, working with her medical teams and educating others that may come her way, keeping her away from sick people, and having understanding that things are a little different and her body cannot fight bugs like you or I, Amber can live a more normal life. She sure knows what life is all about and what it means to smile.

Written by Amber Comer's mother.

Jodi Wrightson

Meet the Board

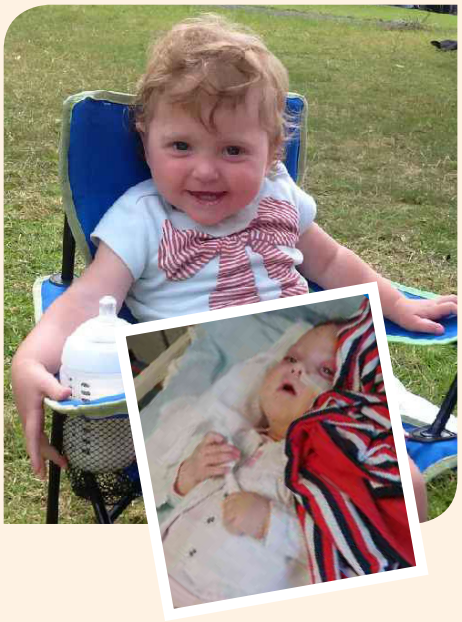
Dr Miriam Hurst works as a clinical immunologist and immunopathologist in Auckland. She graduated from the University of Otago and completed her immunology training in Newcastle-upon-Tyne in the UK and Sydney, Australia. She is interested in primary and secondary immunodeficiency as well as anaphylaxis.

Her spare time is mostly taken up with two small children but she does enjoy theatre and reading.

Kahn grew up on the North Coast of NSW, Australia. He completed a science degree at the University of NSW in 2000 before going on to study medicine at the University of Sydney. After graduating in 2004 he competed the majority of general paediatric training at John Hunter Children's Hospital in Newcastle, Australia. He moved to Auckland at the beginning of 2013 to complete specialist training in Paediatric Immunology and Allergy. He now works as a consultant Paediatric Allergist / Immunologist at Starship Children's Hospital.Kahn has a research background in both immune deficiency and allergy diagnostics. He conducted an honours thesis Severe Combined Immunodeficiency and has published primary research on new diagnostics for common childhood food allergies, an area where he has ongoing research endeavours. He has contributed to the published cases of interesting or unusual presentations of immunodeficiency and has collaborative research involvement with world leading immunology labs around the world.

Kirsten Foster

Early warning signs of unusual long lasting infections and failure to thrive alerted Kirsten's parents that all was not well with their baby girl. Parents Sarah and Rob reflect on the family's traumatic journey that led to Kirsten being diagnosed with SCID.

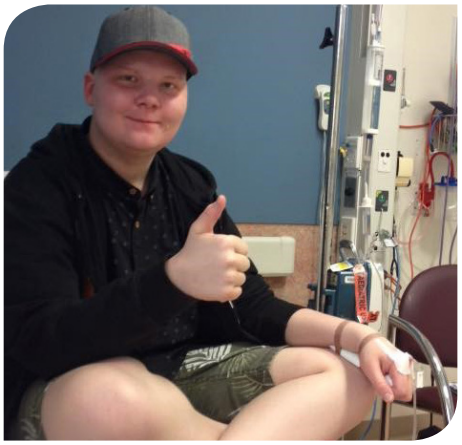


On 6th June 2014 our second daughter, Kirsten Victoria Foster, was welcomed into the world. She was a big, healthy looking baby that continually gained weight and was very happy. I was a little surprised when she started getting her hormonal spots at only two weeks old. A week later, instead of the spots clearing, they had totally covered her face, were spreading down her torso and getting worse. It was just awful, she looked terrible and they were like a hot, angry rash. We took her to the doctor and they sent us to hospital to get her checked out. This happened twice and both times we got sent home, told it was just the average hormonal rash and it would clear. It didn't clear and finally we went to another doctor who took one look at her and put her onto antibiotics. This mainly cleared it up but then she seemed to get what looked like eczema. I had to change my washing powder and be careful what we cleaned her with, and keep creaming her etc.

Then at 5 months she got her first cold. The symptoms were same as any child but it just seemed to hang on. Then she started vomiting up her feeds and becoming lethargic. I took her to the doctor and they seemed somewhat concerned by her and said she had a chest infection. They gave us antibiotics for her, which she couldn't keep down.

Liam Watts – Auckland PID member

Unusual infections and bleeding problems were early warning signs leading to Liam being diagnosed with a rare Primary Immune Deficiency.



At fourteen years old, Liam Watts has had to deal with pain and sickness that most of us never even have to imagine. He was diagnosed with Thrombocytopenia when he was only one year old. Doctors investigated and managed the symptoms for many years, however the bleeding problems became worse over time. Liam had to contend with frequent nosebleeds lasting 10 hours as well as painful rashes and the discomfort of eczema breakouts. In 2014 Liam was given a diagnosis of Wiskott-Aldrich Syndrome (WAS).

Wiskott-Aldrich syndrome (WAS) is a rare and unique primary immunodeficiency disease that affects only about 1 in 1 million males. In addition to being susceptible to many different infections, patients have problems with abnormal bleeding due to unusually small, dysfunctional platelets (blood cells that play an important role in the formation of blood clots). For patients with WAS, this leads to unique health challenges such as increased susceptibility to bacterial, fungal and viral infections and the increased possibility of cancer. They need platelet infusions to help manage these health issues.

The only cure was a Bone Marrow Transplant – which Liam and his family prepared for in August 2014.The treatment is brutal with the patient having to undergo chemo to destroy their own marrow before the donor bone marrow is inserted into their blood stream through a Hickman line in the chest. Liam had to leave school and spend the majority of a year in and out of hospital. He also faced complications that included GVHD (Graft-versus-host disease) where the immune cells of the transplant see the patient's body as 'foreign' and so attack the body's cells causing anything from hot itchy rashes on the skin to attacking the body's organs. Liam spent the rest of 2015 in and out of the bone marrow transplant unit at Starship supported by his amazing parents, Nick and Amy who took time out of work to care for him and be with him every step of the way.

Liam was discharged from Starship just in time for Christmas; he has come a long way but is still dealing with complications and ongoing health issues. Visits to hospital are still too frequent and Liam is struggling to recover his health. Liam has had amazing courage in the face of acute pain, nausea, and complications due to bad reactions to medication as well as the donor marrow fighting against his own body. This young man has a wonderful strength of character and, despite everything he has gone through, has kept his zest for life and his ability to smile no matter what. At fourteen years old, this teen has been through something others twice his age couldn't even imagine.

Follow Liam's journey at https://www.facebook.com/OperationBone/?ref=hl

The next morning, I picked her up and she just wasn't right. Her stomach was heaving in and out, she had a head nod going on and was floppy and seemed exhausted. I rang the health line and they called the ambulance. Her oxygen levels were in the 80s and she was working very hard to breath. That was the first of many, many, days and nights in hospital. There we got told she had bronchitis and she was on oxygen and had to be suctioned out a lot.

Between 5 months and 7 months, we lived a terrifying life with her. Back and forward to hospital, her respiratory condition never got back to normal. Slowly deteriorating, she was taking two steps backward and one forward. Many, many tests, blood tests, x-rays and scans were done, and nothing came clear. Finally, we were sent home with an outpatient nurse either coming each day or calling us. It was so scary. We were apparently put on the list to see the Starship specialists, but we felt we didn't have much time left and that if we waited, we might lose her before it was known what was wrong. In the meantime, I had to have help each morning at home (as this seemed to be when she was the worse) as I couldn't mind Kirsten and my other daughter. Each morning she would go floppy, her respiratory rate would be up to 90 breaths per minute and she would vomit. I couldn't stand it any longer and so we drove to my parents in Auckland and called the ambulance from their house, so we would get in with Starship.

This was the best move we ever did, and helped save her life. She was admitted immediately and within two weeks they had diagnosed her condition as SCID. We were told that someone with this condition wouldn't survive past two years of age without treatment. From there we got transferred to the transplant ward 27B, where we lived for the next six months. Kirsten had a bone marrow/ cord blood transplant six weeks after diagnosis. She continued to have a virus affecting her lungs causing severe respiratory distress, which she couldn't clear until she had her new immune system.

Although the transplant went well, the ride was a rocky one because of the state of her lungs. Having a virus on board for such a long time had caused lung disease, which landed Kirsten down in ICU on a few different occasions. The care at Starship was absolutely amazing and we felt safe and confident with the specialists handling her, even though it was a mighty rare condition. Many prayers later, in September, Kirsten was finally discharged, and has continued to make steady progress ever since. She is two in June and is a beautiful, happy, thriving little girl. We would like to extend a huge thanks to Nyree, Nicola, and the transplant team, the immune team, IDFNZ, and the nurses in wards 26 and 27, for their incredible care over the whole time.

Rob and Sarah Foster

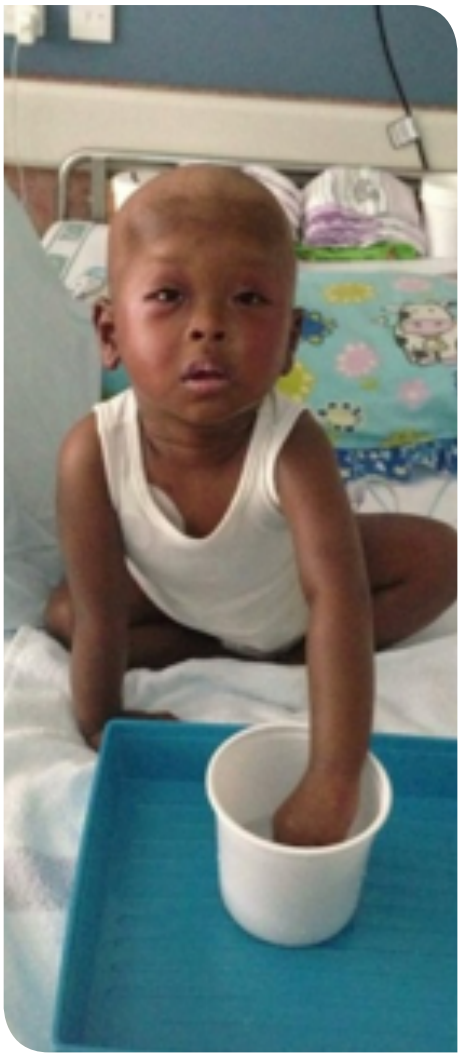
Franky Seng

– PID Auckland Member

Franky Seng was only a few months old when he was diagnosed with the very rare NEMO Deficiency Syndrome in 2011. After weeks of experiencing severe diarrhea and numerous doctors' visits and episodes where he couldn't breathe, he was rushed to Middlemore Hospital with unexplained pneumonia. At six weeks old he was transferred to Starship and a barrage of tests began leading to a PID diagnosis.

Franky's mum explains that he was a happy healthy little baby boy until two weeks of age. That's when the diarrhea started. At first the doctors told his parents that he was allergic to the formula he was being fed but even after it was changed to a different type the diarrhea didn't stop. He was taken to hospital and for the next four weeks the medical professionals that were caring for him experimented with different formulas to try and see if that was what was causing his upset stomach. Sent home when Franky was six weeks old, the Seng family were back at Middlemore within a few days as Franky started to deteriorate. He couldn't breathe and it was soon discovered that he had contracted Pneumonia. From there the doctors started testing him and found that he had NEMO Deficiency Syndrome.

Read more of Franky's story on next page.



Franky's story continued.

NEMO is linked to mutations in the X-linked NEMO gene and means that the patient is susceptible to infections anywhere in the body. It was discovered later that Franky's mother carries the mutations and that it can be passed down to any male child she has. Because of NEMO, Franky's immune system doesn't work as it should and so he spent six months in hospital before his first birthday.

When Franky was two years old, his parents were told that without a Bone Marrow Transplant, he wouldn't live to see seven years of age. And so, in 2012, the search for a compatible donor started. It took two years to find a donor and to make sure that Franky was healthy enough for the procedure.

In 2014 a donor in Taiwan was found and their bone marrow was shipped to Starship for Franky. After a round of Chemo to kill of his bone marrow, Franky's new marrow was introduced into his body through tubes inserted into his chest. He spent three months in hospital after his transplant and then was able to go home with his family.

Mrs. Seng speaks lovingly about her little boy explaining that he loves to play with his cars and talk when he has good days. Franky has also started kindergarten and attends two days a week.

However, because of his illness when he was so young he has never learned to eat and swallow normally and so his mother feeds him through a Mickey button in his stomach every four hours. Another part of Franky's everyday life is the nine medications that he has to take every day; however, as he continues to grow stronger, some of these medicines, such as his medication for low iron, can be stopped.

Now, at almost six years old, Franky is getting ready to start school as soon as a teacher aid can be found to care for him and feed him when he is at school. This little fighter has already been for a school visit and even though he was shy and quiet his mother knows that he will learn to love school as soon as he has made some friends.

Franky Seng has lived nearly all his life with a chronic illness that could have killed him. But now, thanks to the generous anonymous donation of bone marrow and the care of many medical professionals at Starship, he has the opportunity to live a relatively normal life and enjoy the simple pleasure afforded to other children his age of going to school and making friends.

Ava-Grace Makasini

Born in November 2013, Ava-Grace Makasini is a beautiful little girl with an infectious smile. However, at just over two years old she has been through more pain and sickness than most of us experience in a lifetime.

Ava-Grace was diagnosed with Megacystis Microcolon Intestinal Hypoperistalsis Syndrome or MMIHS. This is a rare congenital disease with 71% of patients being female. Her abdomen was distended, her colon is very small and she has little to no intestinal movements which means she is unable to eat in the traditional way and must rely on a feeding tube and parenteral nutrition.

At just a few days old, she underwent the first of a series of operations to try and correct her condition. Having thought that she was getting better, the doctors' allowed Ava-Grace to be fed a few mils of milk every few hours but after doing tests it was found that the milk sat in her stomach and wasn't moving through her digestive system and so she had to undergo another surgery to try and find the blockage.

By the time she was three months old she had undergone four major operations, the fourth was to connect her bowels to her stomach to see if she could digest her food that way. After this operation the Makasini family were able to take their baby girl home for short visits. By March 2014 Ava-Grace was going in for her sixth operation as doctors continued to try new ways that would allow her to process food naturally. In the mean time she was given a Hickman line so that she could easily be given her medications.

After six months in Starship Hospital, Ava-Grace was able to go home with her family. However, she was in and out again with fevers, dehydration and cultures positive for infections. Despite this she has beaten the odds and is thriving and growing stronger, with only a few bumps along the way. Multiple infections have meant that Ava-Grace is still being admitted into hospital every few months so that she can be given medication and looked after by the medical professionals there.

By January this year she had endured sixteen major operations on her digestive

system and to replace lines that help when she is admitted to deliver medicine into her system so that her body can fight the many infections and fevers she contracts. In February she went in for another operation on her bowel to try and repair it and is now back home with her family and doing well. Having stayed at Starship Hospital for long periods of time, Ava-Grace and her family have the medical professionals there to thank for the special care they have given this small girl who has fought to survive this rare disease.

Next Steps: Ava-Grace is to be assessed by Melbourne Children's Hospital to explore the possibility of a small bowel transplant to correct her condition. Kids Foundation will be assisting friends and family raise funds to support Ava-Grace and her family throughout their lengthy time overseas.

Should Ava-Grace be accepted for transplant surgery, MOH will cover all medical and transplant related costs. This fundraising campaign will subsidise family and friends who are to support Ava-Grace and her family through the months leading up to and after surgery.

Anybody wishing to donate can visit www.givealittle.co.nz

Ella-Rose Meagher

– Auckland PID member

Congratulations to the winner of our 2015 Writing Competition! Ella-Rose Meagher's inspiring story about how she has grown up with Hypogammaglobulinemia is both well written and uplifting. We've published Ella-Rose's story here to inspire others living with similar conditions to keep going despite the pain or sickness they are living with. Well done Ella-Rose and keep inspiring others with your story – we hope you enjoy spending your Whitcoulls voucher!

'You Shouldn't Let Something You Can't Control, Control You'

School. Drama. Music. Singing. Weekends. Friends. That's what I define my life as and what I let my life define me as. Hypogammaglobulanema (Primary Immunodeficiency) comes under the 'homework' section of my life. Something I have to deal with and adjust to, and something that has to be done, just like actual homework. It's important, but it's not who I am. Every six days however I am reminded of this 'homework', with my routine of setting up my blood products. I then find a quiet space in the house and put a butterfly needle in my stomach. Following that I usually sit for the next hour while my pump does its thing and do homework, watch TV, sometimes have dinner or on a rare occasion go out. I suppose this is what I like to think my life is. It's an occasional reminder every six days. Sadly I know this is not exactly true. I have doctor appointments, with about every specialist you can name. Blood tests. Operations. Then there are the conditions that come off this umbrella term of 'Immunodeficiency'. Sick days, hospital days, days off school. Fed up days. All of these happen on a regular basis, but so do my days with friends, days at school, days at the movies or days spent out shopping. I just have to fit in the other, not so happy days.

I get sick a lot, more than an average teenager. However due to Evogam, my blood products, which are kindly donated by many New Zealanders, I am able to stay relatively well throughout the year. Without Evogam normal teenage things would be hard. I wouldn't be able to have sleepovers, go out as much or do as much at school, let alone be at school. One thing that has truly shaped me was going on a school camp at the end of last year. I was able to go on a five-day sea kayaking camp with my friends from school. Sleeping in a hut and in tents. Kayaking in rain and wet weather. One of the most amazing experiences of my life and one I didn't come home sick. Another reason this camp was so special to me was that the previous year, 2013, I had spinal surgery to correct my scoliosis. This, however, is completely unrelated to my immunodeficiency and was pretty much just bad luck (and bad genes).

With two of my best friends on either side of me, during the School Musical this year.

I never thought I'd be able to do so much after the surgery, but nothing medical has every defined me before, so I wasn't going to let a few rods and a handful of screws do that…and no I don't set off the metal detectors at the airport!

One great thing about Evogam is that I can take it wherever I go, visiting family overseas, or going on holiday. It can be packaged up in our blue chilly bin and taken with us. Something that I am truly looking forward to is a trip overseas next year. I have never been overseas with Evogam without my family before, but next year I am going to be taking it across the world on a school trip. Something that would never be possible without all the plasma donors who give up their time to give people like me and my brother as normal a life as possible. After all what is normal? This is my normal, it's the normal I live my live by.

I think it's important to remember that you shouldn't let something you can't control, control you. It's hard sometimes but no matter what I do I don't rely on any of my medical conditions or let them stop me. One of my favourite quotes is, "Everyone you meet is fighting a battle you know nothing about, be kind always". Most of my friends look at me and see me, Ella-Rose and would not be aware of everything else.

I'm Ella-Rose. I'm fifteen and a half. I have a dog named Molly, I have a brother and a sister, I love drama, singing, hanging out with my friends and family. There just happens to be more to me than you can see.

Ella-Rose Meagher, 15.

PID E-Training for New Zealand GPs

ASCIA (Australasian Society of Clinical Immunology and Allergy Limited) is the leading association for allergy and clinical immunology in Australia and New Zealand. Founded in 1990, they have taken up the responsibility of training and informing doctors and other medical professionals about allergies and immunology diagnosis and treatment. As such they are now offering a new E-Training course to help health professionals renew and refresh their knowledge on Primary Immune Deficiency Disorders.

Released in February 2016, the new PID (Primary Immunodeficiency) e-training enables health professionals such as general practitioners, practise nurses and paediatricians to easily access specific e-training modules to refresh and update their knowledge of Primary Immune Deficiency Disorders. As well as a general update, there are specific modules covering the main disorders, typical case studies and treatment plans which all help these medical professionals to better diagnose and treat allergies and immunology disorders.

ASCIA have also included the 7 warning signs material developed by IDFNZ so that when patients present these to doctors and ask if the seven warning signs apply to them, the doctor will recognise the IDFNZ information and can easily access the necessary specialist medical information underlying the simplified public awareness message. This means that not only will a patient be able to recognise the signs of immunodeficiency, their doctor will also easily be able to find more information on the disorder and in doing so be able to find the right treatment for that patient quickly and easily.

As a patient diagnosed with a specific PID condition, we encourage members to pass on information about this new e-training to your GP and practise nurse. Primary Immune deficiency disorders are rare diseases that medical professionals may not come across very frequently.

The ASCIA training includes specific modules which will allow the GP to familiarise themselves with latest information on diagnosis and treatment at their convenience.

http://www.allergy.org.au/healthprofessionals/hp-information/immune-diseases

Special thanks to our medical professionals

Although being diagnosed with a rare disease is traumatic for both patients and families, the level of care here in New Zealand is excellent.

And so, because of this, KIDS Foundation members wanted to thank their wonderful medical teams for caring and supporting them through dark times. They came up with the idea of creating a token of their appreciation. Two different groups of children got creative and made special "thank you" plates to be displayed in their local hospital. The ceramic plates were painted by the children, with their names added and then glazed. They were then presented by the children to specialists representing the Starship and Christchurch hospital staff whom our families hold in very high regard. The plates will be placed on display in each hospital as a reminder of the amazing work done by the medical professionals there as well as a reminder of the children they have helped.

Rare Disease Day 2016

Thanks to all those wonderful children who created inspiring artwork for Rare Disease Day!

On the 28th of February this year IDFNZ marked Rare Disease Day 2016 with members in Auckland and Christchurch gathering at the Botanic Gardens and Spencer Park respectively. An afternoon of fun was enjoyed by all, including crafts, afternoon tea and bouncy castles. The day ended with yellow balloons being released and 'hands up' signs being made to add to the longest 'hands up sign' for the Facebook competition which was raising awareness of rare diseases. These were great opportunities for families to get together and enjoy some fun and relaxation.

Sponsorship & Fundraising

Cuddle buddies from Whitcoulls

A huge thank you to Leanna Wallis and the team at Whitcoulls, Cuba Street in Wellington for their fundraising over Christmas.

Your generous efforts have meant a donation of George Bear and Bookworm soft toys had been donated to IDFNZ to hand out to young patients. These are a great way to help IDFNZ bring a little light and joy into the lives of these sick children. The toys were very generously donated and will greatly help bring a smile to our youngster patient members who have to go through so much. Being in hospital can be scary but a cuddly friend can make such a difference.

Little Shoppers booklet

The Little Shoppers booklet is a great way to support IDFNZ. With every book of special offer coupons purchased, $10 is donated to IDFNZ. The deals in the Little Shoppers booklet are for children aged 0-11, and also includes discounts and free services. What is unique about these discount books is that while the books are only valid until 31st March 2017, every coupon can be used as many times as the buyer wants. The funds donated from the sale of these booklets will then be put to use to help raise awareness of Immune diseases, support families with sick children and bring a little bit of light into the lives of young patients through gift boxes and toys.

Beware Yellow Round the Bays team

Thank you to the BEWARE YELLOW team who participated in Round the Bays on Sunday the 6th of March.

The team were proudly wearing their BEWARE YELLOW t-shirts and having a great time supporting a good cause.

The more people know about the possibility of jaundice in babies pointing to liver disease, the better. Not only were there nurses and doctors participating but there were also representatives of the families that are affected by liver disease in infants.

Thank you to all of you for being part of this great day and raising awareness of pediatric liver disease.