IDFNZ intouch
KIDS FOUNDATION
Issue 2, 2017

# Dates for your diary

## PI WEEK
**22 - 29th April**

## PI Adult Group Meetings
**Thursday 20th April**
Wellington Coffee/movie event

**Sunday 30th April**
Auckland Coffee/movie event

*See flyer details on website*

## KIDS Foundation Fundraiser / Auckland Event
**Saturday 17th June**
Despicable Me 3 Movie

*Buy tickets for friends and family to support our fun "Beware Yellow" Fundraiser*

*Tickets on sale from IDFNZ KIDS Foundation info@idfnz.org.nz.*

## 2017 Circus Quirkus
**Monday 8th May** - Queenstown
**Tuesday 9 May** - Invercargill
**Thursday 11 May** - Dunedin
**Saturday 13 May** - Christchurch
**Monday 15 May** - Nelson
**Thursday 18 May** - Palmerston North
**Saturday 20 May** - Wellington
**Monday 22 May** - Napier Boys' High School
**Tuesday 23 May** - Gisborne War Memorial
**Wednesday 24 May** - Mt Manganui
**Friday 26 May** - Rotorua
**Sunday 28 May** - Hamilton
**Sunday 11 June** - Auckland



# World PI Week 22-29th April

**World PI Week is a global campaign which aims to raise awareness and improve diagnosis and treatment of primary immunodeficiency (PI).**

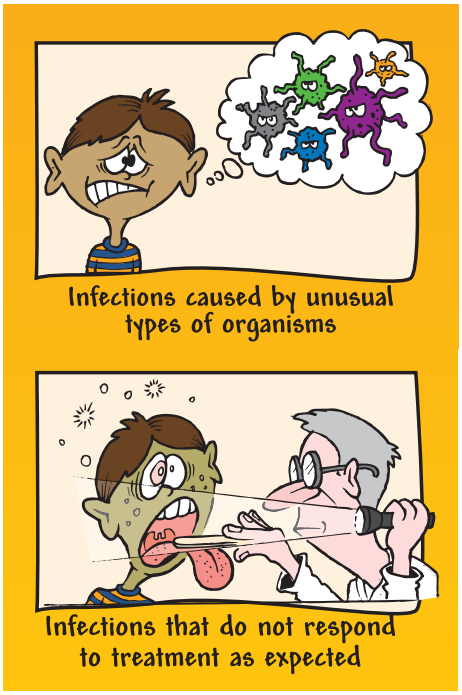
***The Immunodeficiency Foundation of New Zealand, IDFNZ, marks World PI week by promoting the early warning signs of Primary Immunodeficiency.***

Primary Immunodeficiency (PID) is a growing category of 250 different disorders resulting from some form of deficiency of the immune system. Primary Immunodeficiency can, if not treated, be chronic, serious and even fatal.

Many PID disorders are considered 'rare diseases' which has meant that diagnosis and treatment have historically been difficult, but thanks to research and medical progress over the last 60 years, many of these conditions are now treatable. Yet because symptoms are similar to common and recurrent infections, PID often remain undiagnosed, misdiagnosed, or can be diagnosed at a late stage.

Once a diagnosis is established, much can be done for PID patients. At a minimum, the recurring infections can be treated with low or moderate doses of appropriate antibiotics. These can help prevent permanent damage to the lungs and bronchial tubes, thus promoting the patient's long-term survival while improving the quality of life. When appropriate, immunoglobulin therapy is the accepted protocol for a wide range of PID diseases. Advanced treatments such as the interleukins, PEG-ADA, and gamma interferon can help in some complex cases. Bone marrow transplantation and gene therapy may be the appropriate protocol in specific disorders

Thanks to new therapies, greater public awareness, and better access to information, many patients with PI are leading more normal lives - going to school, camp, work, playing sports, and enjoying a better quality of life. There has never been more hope for people who are immunodeficient.




***It has been estimated that 70% of PID patients are undiagnosed.***

New Zealand is introducing new born testing for SCID in 2017 – this is a major step forward for diagnosing this fatal PID condition and will surely save precious lives.

Other PID conditions however remain challenging for clinicians to diagnose – the early warning signs are important.

Greater awareness is the key to ensure more PID patients can be diagnosed, treated and lead productive lives. Help IDFNZ spread this important message.

# Know the signs?

Serious, Persistent, Unusual or Recurrent and difficult to treat infections are hallmarks of Primary Immune deficiency (PID) medical conditions that can affect patients of all ages.

## 7 Warning Signs
When to suspect primary immune deficiency


An unusually large number of infections requiring treatment


Any other unusual symptoms related to infections


Infections caused by unusual types of organisms


Infections in unusual places


A child that does not grow, or put on weight as expected


Infections that do not respond to treatment as expected


Family history of an immune deficiency or abnormal infections


# Know the signs?

## 7 Warning Signs
When to suspect primary immune deficiency


**New online training now available**

Free, specialist e-training for medical professionals wanting to quickly update on primary immune deficiency disorders. https://immunodeficiency.ascia.org.au

IDFNZ supports people in New Zealand living with primary immune deficiencies.

IDFNZ Immune Deficiencies Foundation of New Zealand

The Kids Foundation

For more information, visit www.idfnz.org.nz

# Living With PID

**Being diagnosed with PID (Primary Immunodeficiency) can be a relief but it can also bring up some questions and struggles. Once you are diagnosed you may wonder what the next steps are, how to deal with your illness and the effect it has on your life. You are not alone. IDFNZ patient members can offer useful tips and advice outside of the treatments and support of your healthcare providers.**

PID occurs when certain components of the immune system do not work properly. The immune system helps the body fight off infections, leaving PID patients susceptible to reoccurring and prolonged infections. It can also cause the body to fight itself causing inflammation and pain from an autoimmune response. There are varying degrees of PID from reasonably mild cases to severe with over 250 different known types of PID identified. Patients can be diagnosed with PID in childhood or adulthood; and as a hereditary or genetic defect of the immune system they can occur through generations of an affected family.

PID's are diagnosed by an immunology specialist; these are lifelong conditions so it is important that patients can talk openly with their doctor and understand the management of these conditions. Your immunologist will decide on the best course of treatment for you to live life to the full.

Most newly diagnosed PID patients are referred to IDFNZ – we are able to provide free membership and relevant printed information to further explain your diagnosed condition in "patient friendly" terms, as well as offering the opportunity to meet other patient members and to liaise with support staff. From time to time IDFNZ offers patient conferences and seminars.

Depending on your diagnosis, there are various courses of treatment that may be offered to you; typically these may include Immunoglobulin (IG) replacement therapy and prophylactic antibiotics. Bone marrow or stem cell transplantation may be an option for younger patients with specific PID conditions. IDFNZ can provide information on IG replacement and medical diaries which might be useful to log treatments and appointments.

IG replacement therapy can be done through an intravenous infusion so that the immunoglobulin is given straight into the bloodstream through your veins (IVIG). Alternatively, subcutaneous (SCIG) infusions involve the immunoglobulin being injected into the fatty layer beneath the skin, usually in your arm, leg or belly - using a manual syringe (Push method) or with an infusion pump. NZ PID Patients are generally given access to a number of methods of treatment and product used – allowing a degree of personal preference to fit in with lifestyle and in response to any adverse reactions. Talk to your immunologist or nurse specialist about options available to you.

Where SCIG pumps are required, IDFNZ can supply these for your use via your medical team, if this is a preferred method you might like to trial. IDFNZ can also provide handy chiller bags for patients on home SCIG treatment, and can assist in practical issues such as organising free courier services to deliver non IG consumables direct to home – eliminating the need to collect from the hospital (one less hospital visit each month). Talk to your nurse specialist if you wish to access this service.

Other treatments such as antibiotics are given to PID patients to help fight infections. They are usually taken in pill form but can sometimes be given by injection or infusion. The PID patient typically requires longer and / or higher doses than normal patients to counter infection, and sometimes is offered prophylactic treatments as a preventative measure. Always inform your immunologist if you are scheduled for surgery or dental treatment, being PID can have unexpected repercussions with any invasive treatment for "other" medical conditions - your immunologist can advise if prophylactic antibiotics are necessary prior to surgery.

Routine tests prescribed by your doctor will help monitor how well your PID condition is being managed and if there are any changes to treatment that need to be made. Once diagnosed and under a regular treatment plan, PID patients can expect to meet with their specialist annually. Should you have any questions or concerns between visits, feel free to reach out to your nurse specialist who will assist you in answering questions and determining if you need an earlier appointment with your immunologist. Because PID conditions are life-long you will find that establishing good lines of

communications with your medical team will be invaluable.

Living with PID is a journey. There are sometimes ups and downs; as someone living with PID you need to be able to learn about your disorder and work closely with your doctor to help you continue with your life as normally as possible. Linking with other PID patients and IDFNZ can also provide support to develop coping strategies.

Suffering from a chronic PID condition and dealing with it's effects in your day-to-day life can be draining and disheartening, adding emotional fatigue on top of physical fatigue. These are real medical conditions, but are completely invisible to those around you and so you may feel you are battling alone. There will be times when you may feel completely exhausted, nauseous, alienated, ridiculed or in constant pain with a plethora of other side-effects, symptoms and emotions. IDFNZ PID patients were asked to comment on what some of these challenges have been in their own journey and to offer advice and tips for coping.

## PID's and Fatigue:

**How does PID fatigue affect my everyday activities?**

"It really affects every aspect of my life, even though I am as well now as I can possibly be, I still struggle hugely with overwhelming fatigue. It's the fatigue that affects everything especially in the evenings. I don't go to evening functions, unless I can take time off work in the afternoon to sleep. When I have an active infection, I am just not myself. I try my best to act, but I am about a 20th of the real me. I used to be bubbly, funny, and chatty, up for anything! Now I keep to myself quite a bit. So my friends and family are the people who really miss out. I give everything I have to work, because as a solo parent I have to work, but then there's not much left. Some days I go straight home to bed. I'm also much grumpier with my kids than I used to be. I don't have the resources to be patient. I push myself to do physical things like take the rubbish out or even go and get the mail, because I know it's important to keep moving and using muscles. I make myself walk upstairs when I'm up to it. I always have more energy in the first week after my infusions, so when I'm invited to something important I always look to where I'll be on my infusion calendar. It's disappointing if I see that the event is in my worst week, because I know I'm going to be tired and depleted."

*Linda, a CVID Patient from Auckland.*

Chronic physical tiredness is such problem, ***"hospital visits every 3 weeks for infusions. That day and often the next is a write- off due to tiredness ….Then there are all the other visits - for the asthma, breathing, bones, diabetes, cataracts, melanoma, arthritis etc. etc….All related to PID"***

***"Fatigue - Fight it every day! Some days I'll start a job I've done many times before push and do it badly or give up - HATE IT!!!"***

***"My life is very restricted. I've given up most stuff - my life revolves around my home and garden. I walk when I can"***

*Colleen, a CVID patient from Auckland.*

***"One of my greatest challenges was and still is fatigue from battling the attitude and ignorance of GP's…..there is often very little empathy towards those of us who are living with PID"*** *Linda.*

After diagnosis Linda discovered life can change for the positive however, ***"Having the right medical team and support group around you can help you feel listened to and gives you the courage to keep pushing for more tests, better medication, or even a different approach to your illness."***

IDFNZ can provide education material on PID's diagnosis, treatment and management, designed for GP's –if you are tired of the same fruitless battle we can provide you (or them, directly and discretely), training information written by Specialists, to ease the way.

Laura, a patient member from Nelson, has also found that having a good support system is important and something that is worth investing into developing and maintaining even when you might be feeling low; ***"……as you regain a little energy……reach out to others……..stay in touch with your support group and medical team."***

Making sure that your doctors and support people are sympathetic and willing to listen and help is very important. Also, ***"…reach out to others who are in a similar position to you even if they don't have exactly the same illness or degree of symptoms. The safety and understanding that comes from these steps is invaluable."***

IDFNZ provides 4 respite care holiday homes across New Zealand – these are for patient members of all ages to make use of. By all means have a family holiday in the summer, but remember they are also there if you need a weekend or mid -week break-away off season – a safe escape to enjoy quiet time and a change of scenery, alone or with loved ones. Make use of these valuable resources to recharge your batteries.

## Dealing with pain, exhaustion, nausea and feeling unwell in general:

PID's may involve long periods of feeling unwell, experiencing repeated infections and illness. This constant grind can wear the most resilient PID patient down over time. Assuming the appropriate medical treatment is available infections can be overcome, however PID patients are often slow to recover and fully recuperate. How do PID patient members cope?

Attitude is very important; it is all too easy to be totally self- absorbed when feeling unwell.

Michele, a CVID patient from Auckland, says that it is important for her to not dwell on the negatives ***"……I consciously focus on, and am grateful for, that which I can do…"*** a positive outlook helps Michele deal with the symptoms of her PID.

## Nutrition:

Poor absorption of nutrients as a side effect of PID is a real problem for many patients, as is heightened vulnerability to food poisoning – carefully selected, fresh nutritious foods and adopting safe cooking and preparation methods, are important measures to ensure both food safety and maximum nutritional value. A diet of fast-food and leftovers is not for the PID patient.

***"Eating well – ensuring good nutrition becomes a priority. Looking after yourself physically and emotionally is very important".*** *Laura*

Mary, an Auckland CVID patient says she ***"takes extra time to shop daily for fresh foods, keeps her refrigerator extra clean and cooks just what she needs each day to avoid waste and the problem of potentially unsafe leftovers".***

## Living Well:

For all patients living with PID, learning the limits of what you can and can't do is very important. Live life to the fullest but realise that your normal day may be quite different from someone who does not have PID. You may not be able to socialise in quite the same way but there are always ways of getting around these challenges.

Colleen, from Auckland advises ***"Keeping away from places with large crowds - concerts etc.; Music and dance- I used to dance and sing - no longer possible I really miss even movies!"*** Smaller events in environments you can control can be the answer meet friends at an outside café, picnics in the summer etc.

***"At 3pm a person without PID may be ready for afternoon tea, whereas a PID patient could be ready to go to bed for the night." "Don't push your body too much, realise your limits and work to those".***

***"Rather than try and force your body into something like an intense bike ride with your friends.....meet for a coffee somewhere lovely for a catch up... or perhaps enjoy a beautiful gentle walk".***

*Michele*

***" I remind myself.....I am alive.....I can move, see, hear, laugh, love, and occasionally dance!".***

*Linda*

Consider your environment – is there anything you can do to improve your susceptibility? Where you live and who you socialise with may make a difference.

***"Making the choice to move to a warmer dryer climate helped immensely".***

*Laura*

**"Working through times of illness as a team with those in your support group and family is very important".**

*Michele*

## Challenges of Treatment:

Sometimes unexpected, adverse effects may accompany treatment (IVIG/SCIG/ or antibiotics), these need to be noted and reported to your medical team, but are often an unfortunate side-effect of dealing with PID. Some ways to combat these side-effects is to medicate before-hand with guidance from your doctor; try lowering the flow of your intravenous pump or request to change the brand of immunoglobulin you take. If infusing at home, make sure you are set up properly- make yourself comfortable with a cosy blanket and pillows at hand so you can snuggle up and snooze or relax with a book/ movie whilst infusing. These treatments are important for your health and should be treated as invaluable; the ultimate "me- time" to preserve your health.

On the bright side, PID patients who have found the balance with their treatments agree that they have made life better.

*Linda* writes that she has ***"...more energy in the first week after her IVIG infusions", and that she "...would simply not be able to work... without Intragam P infusions".***

William, a CVID patient from Wellington; noted that ***"that although it took a while for the IVIG treatment his doctor prescribed to 'kick in'; he has enjoyed significantly better health than before diagnosis and treatment".***

As a newly diagnosed PID patient you may feel scared, alone or unable to cope with what you are going through. Just remember that there are others who are going through the same things. You have made invaluable progress, identified that there is something wrong, sought medical help, been diagnosed through tests and the7 warning signs, and received treatment. Even if you are not 100% healthy, that does not mean that you should give up. Treatments can be tailored and refined to work better for you, IDFNZ and support are just a phone call away; your medical team know the best ways to treat and manage your PID, and there is a whole raft of information that can be useful to you only a few key-strokes away.

***"Life isn't about waiting for the storm to pass...It's about learning to dance in the rain."*** *Vivian Greene.*

***"Courage doesn't always roar. Sometimes courage is the quiet voice at the end of the day saying, I will try again tomorrow,"***

*Mary Anne Radmacher.*

# IDFNZ Patient Services Available to all registered patient members


**Support of patient members and families includes:**

- Support visits where requested – at home or hospital
- Information relating to specific medical conditions, treatment, welfare entitlements, advocacy and referrals to other support agencies where needed.
- Networking and connecting with other patient members
- Patient Education events
- Library service , free loan service
- Various Social events for patients (and families/ siblings) in your region

---

**Family grants and financial assistance towards approved medical expenses; all patients can access application forms from Support staff.**

**Financial assistance typically includes:**

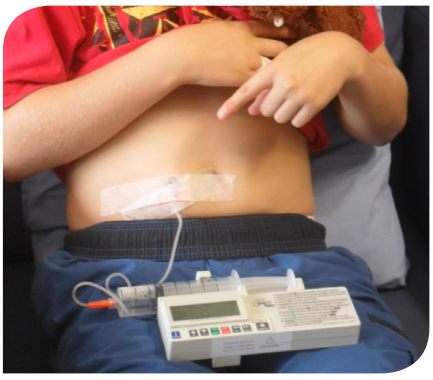
- Parking vouchers to help subsidise hospital car park expenses
- Hospital café vouchers for long stay patients
- St Johns ambulance membership assistance (when available)
- Chemist vouchers towards prescription medications (when available)
- Travel grants for family support and visitation of family members for long stay patients (liver transplant/BMT/etc.)
- Other items / assistance may be considered on a case by case basis
- Patient Education events
- Library service , free loan service
- Various Social events for patients (and families/ siblings) in your region

---


**Approved medical equipment and supplies can also be applied for through Support staff. Examples include:**

- SCIG pump for PID patients (in conjunction with your immunology team)
- Courier service to home deliver SCIG consumables (not Plasma)
- Chiller bags for transport/ storage of SCIG supplies.
- Epipens ( must be prescribed by your GP)
- Medical diaries
- Medical bracelets ( available for purchase)

---



**Respite care facilities are available at 4 locations in New Zealand, families can apply for holiday breaks at greatly subsidised cost. Families recovering from transplant or extended hospital admissions may receive complimentary vouchers for free stays.**

- Red Beach, Auckland
- Mangawhai Heads
- Otaki Beach
- Bannockburn

The Foundation also offers up to 6 selected families Waiheke breaks each year in conjunction with Holiday Helpers Network Waiheke Island

# Our People

# Christine Jones

***My Story***

***When my immunologist first told me I had an immune deficiency my first reaction was one of enormous relief. I had no idea why I took so very long to get over any infection, in particular chest and sinus infections. I'd accepted chronically infected sinuses as my lot in life.***

For years I'd been pestered by well-meaning friends telling me my diet was deficient. My problem was that I was on antibiotics so much of the time, hence weakening my immune system. At their urging, I'd reluctantly tried a multitude of vitamins and things like colloidal silver, only to be told, when they failed to work, that they didn't work because I didn't believe in them! To be told there might be a real reason for all this was a huge relief. I also had an auto-immune disorder called myasthenia gravis which causes extreme muscle weakness after even very moderate use. I'd had to leave teaching as my mouth muscles just couldn't last a whole day and my leg muscles weren't up to running up and down a soccer field or a netball court. I hated this disability, it's a right pain.

When I was told I could have a treatment which involved a regular IV infusion for the rest of my life, I hesitated. I knew it was extremely expensive and, already in my early sixties, I felt the tax payers' money was better spent on someone younger. So, I refused. But a talk with a field officer some months later changed my mind, and when I heard there was a chance the myasthenia might improve, I was sold on the idea. Another strange thing was that I'd had endless dental problems. For instance, every root filling and crown I'd had had become infected, and after spending many thousands of dollars I'd lost all the teeth that were root filled due to abscesses. When I told my dentist I was immune deficient he said it explained much that had puzzled him about my dental problems. Who knew your immune system affected your teeth?

Once I began infusions I waited for the myasthenia to improve. It didn't at first and I was dreadfully disappointed. But, after about five or six months there was a marked improvement, and since then I've had one long remission and although the disorder returned it has never been as bad as it was originally. A miracle! And I've not had one attack of either bronchitis or a respiratory infection since I began the infusions. No longer are my sinuses chronically infected. I do get infections but not like I used to. The infusions changed my life. I do take much longer to get over other infections than is usual but I'm getting older and heh, nothing's perfect. The small inconvenience of regular infusions is worth it a hundred times over. And the staff at immunology are such wonderful people they make the visits a pleasure – almost.

Christine Jones

# Past patient events


*Wellington Group - February meet up.*

# Lisa Fe'ao

### My Story - the short version.

***I grew up in the 60's and 70's on the North Shore of Auckland. In many ways it was a wonderful childhood, except that I was different. I was overweight, frequently had cold sores, sore throats, school sores and a head ache.***

In those days nobody knew about Immunology or Immune Disorders. I remember watching "The Boy in the Plastic Bubble" when I was about 15, and that was the first time I found out I had an Immune system! I eventually went on to study Nursing, and anatomy, physiology and biology were subjects I really enjoyed learning about.

Looking back over my life now that I know what is wrong with me, it is sometimes difficult not to feel angry with the doctors I have seen throughout the years. And there have been many… I have suffered multiple physical problems, especially chronic headaches, chronic bronchitis, uncountable stomach bugs, and skin infections. The list goes on and on… eventually at the age of 45, thanks to an excellent Otorhinolaryngologist, I was referred to an Immunologist because he explained that my sinusitis was so severe there must be an underlying condition. Finally, I was diagnosed at 45 with CVID also known as Hypogammaglobunlinaemia.

By the time I was diagnosed I was in pretty bad shape. As a solo parent I had continued to work full time, but working required all the energy I had, and my three children missed out in their teenage years, on having a mum who could be the mum she wishes she could have been. Taxiing children to sports trainings, games, parties, sleepovers, school events, parent-teacher interviews, plays, shows, etc all those things that parents do for their kids, were extremely hard for me. I was exhausted all the time, but evenings and nights were worse. It would have been so much easier if I wasn't a conscientious mother, but I was, and it will forever be my biggest regret that my condition disabled me from being the mother they should have had.

There have been and still are, many days/nights when depression becomes my unwanted companion – s/he tends to hang around when you live with chronic pain, fatigue, diarrhoea and infection. However, there remains a strong determination to not become an "invalid" (how I hate that word), and to not become a "victim". Of course there are times when it does become overwhelming and then I have a bit of a pity party, but eventually I find a way to overcome that and get back up. Quite honestly, it isn't easy to be positive all the time. I have become quite the accomplished actor!

Since I found and followed sites for people who live with chronic illnesses, my world got better. The stories, quotes, and anecdotes I have read have become a huge source of inspiration and comfort. It's amazing how much easier it is when you know you're not the only one. I have saved many of these to my phone, and I scroll through them when things get tough. The ones that resonate loudly are those that focus on how much strength it takes to accomplish what I do. I used to think of myself as a weak person, but seeing myself as strong, is a positive outcome to being ill. Laughter is also of vital importance, without humour, we are lost.

So here I am now at 53. In the last couple of years my health has been more stable. I take time off to go to the hospital every three weeks for 27 grams of Intragam P, and without that, I would definitely not be working. I doubt I'd be alive! I have a number of Consultant physicians, who between them, work to make my life as good as it possibly can be. I still work full-time, I am fortunate to work for an organisation which offers flexibility and grace. My kids still all live at home, in various stages of blossoming and maturing, and I look forward to seeing where their lives take them. They are amazing people - having a chronically sick mum hasn't been the easiest way to grow up. But they have learned true compassion, even if they don't know that yet. That is a gift.

"One day she finally grasped that unexpected things were always going to happen in life, and with that she realised the only control she had was how she chose to handle them. So she made the decision to survive using courage, humour and grace. She was the queen of her own life and the choice was hers."

- Lupytha Hermin

Lisa Fe'ao


## IDFNZ Going In Style for PI Week

Calling all adult PID patient members – there is opportunity to meet other PID patient members over PI Awareness week. IDFNZ is hosting both a Wellington (Wednesday 20th April) and Auckland (Saturday 30th April) free movie event - to bring adult members together to enjoy a light hearted comedy – "Going in Style".

Please join us - to book your place RSVP now to: Philippa (Wellington, Light House cinema event) piwellsupport@idfnz.org.nz or Janet (Auckland, Hoyts Sylvia Park event) info@idfnz.org.nz.

**Places for both events are limited so be quick.**

# Medical Matters

## *Autoimmunity and Immune Dysregulation in Primary Immune Deficiency. Dr Annaliesse Blincoe, Paediatric Immunology Fellow.*

The main role of the immune system is to protect us against infection by a wide range of pathogens, including bacteria, viruses and fungi. To do so the immune system must be able to recognise "self" from "non-self". In doing so the immune system has a second important role in not reacting to "self", otherwise known as immune tolerance. The development of immune tolerance and regulation of immune function involves all layers of the immune system.

Immune dysregulation occurs when the immune system is not able to control the immune response. Autoimmunity is one form of immune dysregulation, and occurs when the body mounts an immune response against itself, and against what are referred to as self or auto-antigens.

This abnormal immune response can affect multiple organ systems including the haematological (blood) system; the gastrointestinal system, joints, skin, liver and eyes. Further well known examples of autoimmune disease include Type 1 Diabetes, Coeliac Disease, Inflammatory Bowel Disease and Systemic Lupus Erythematosus.

Primary Immune Deficiency (PID) classically presents with severe, recurrent or unusual infections due to impaired mechanisms that normally protect us against infection. It is however becoming increasingly recognised that many PIDs are associated with autoimmunity. In some cases, these autoimmune conditions may in fact be the first sign that a patient presents with before being diagnosed with PID. It can also be difficult to treat patients with PID and autoimmunity as one must balance immune-suppression with the risk of infection in a patient who is already vulnerable.

It has recently been reported that approximately ¼ of patients with PID will also have evidence of autoimmunity [1] (Fischer et al). It seems somewhat paradoxical that patients whose immune systems cannot protect them against infection can mount an abnormal immune response to self-antigens causing disease[2]. Many reasons have been suggested as to how this may occur and it is well known that defects in any part of the immune system including T cells, B cells, innate immune cells (e.g. neutrophils) and complement can result not only in immune deficiency but autoimmunity.

A broad range of autoimmunity can be found in patients with PID. Examples of PIDs that have autoimmunity as the predominant feature include IPEX and IPEX Like Syndrome (Immune dysregulation Polyendocrinopathy, Enteropathy and X-Linked Syndrome); Autoimmune Lymphoproliferative Syndrome (ALPS); Autoimmune Polyendocrinopathy, Candidiasis, Ectodermal Dysplasia (APECED).

Other PIDs commonly associated with autoimmunity include Wiskott Aldrich Syndrome, DiGeorge Syndrome, Chronic Granulomatous Disease (CGD), Common Variable Immune Deficiencies (CVID), Severe Combined Immune Deficiency (SCID) related gene defects such as RAG and Artemis mutations, early complement deficiencies and the more recently described deficiencies in CTLA4 and LRBA and STAT1 and STAT3 Gain of Function mutations.

## *Common autoimmune conditions seen in PID*

| | Autoimmune Disorders | Commonly associated PID | Treatment Options |
|---|---|---|---|
| **Haematological**<br>- red, white blood cells and platelets | **Autoimmune Cytopaenias**<br>- Autoimmune Haemolytic Anaemia<br>- Idiopathic thrombocytopaenia (ITP)<br>- Neutropaenia | CVID<br>CID<br>ALPS<br>Wiskott Aldrich Syndrome<br>CTLA4 and LRBA Deficiency<br>IPEX Syndrome<br>STAT3 Gain of Function | Steroids<br>IVIG<br>Immune suppression<br>Biologic therapy<br>Splenectomy<br>Bone Marrow Transplant |
| Gastrointestinal | - Inflammatory bowel disease (IBD)<br>- Coeliac Disease<br>- Autoimmune Enteropathy<br>- Gastritis<br>- Autoimmune liver disease | CGD<br>IPEX / IPEX-Like<br>CVID<br>NEMO<br>Very early onset IBD – e.g. IL10 and IL10R defects | Steroids<br>Immune suppression<br>Biologic therapy<br>Bone Marrow Transplant |
| Rheumatological | - Systemic Lupus Erythematosus (SLE)<br>- Arthtitis<br>- Vasculitis (vessel inflammation)<br>- Uveitis (eye inflammation) | CVID<br>Wiskott Aldrich Syndrome<br>CGD<br>CTLA4, LRBA deficiency<br>STAT3 Gain of Function<br>RAG mutation<br>XLA | Steroids<br>IVIG<br>Immune Suppression<br>Biologic therapy<br>Bone Marrow Transplant |
| Skin | - Vitiligo<br>- Cutaneous Lupus<br>- Eczema<br>- Psoriasis | RAG mutations<br>CGD (female carrier)<br>Wiskott Aldrich Syndrome<br>STAT 3 Gain of Function<br>CVID<br>SCID/Omenn Syndrome<br>IPEX Syndrome | Steroids<br><br>Immune Suppression |
| Endocrine | - Type 1 Diabetes<br>- Thyroid Disease | IPEX<br>APECED<br>ALPS | Disease specific treatment |

## Autoimmune Cytopaenias

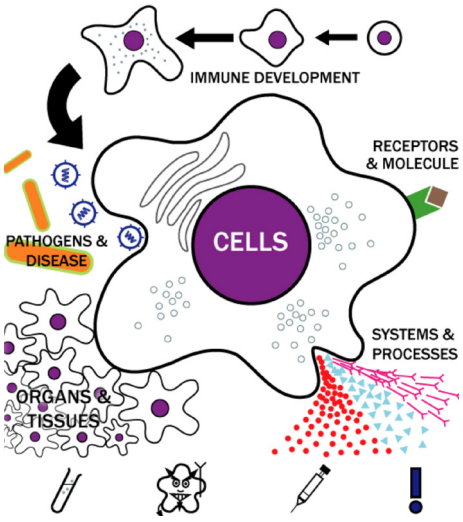
Cytopaenias (low red or white blood cell or platelets) are the most common form of autoimmunity associated with PID with the risk of autoimmune cytopaenia being 120x higher than the normal population and that this may be a presenting feature[1]. One study reported that 13% of patients presenting with autoimmune haemolytic anaemia and 50% of children with a combination of low red/white blood cells or platelets were found to have an underlying diagnosis of primary immune deficiency[3]. Autoimmune haemolytic anaemia occurs when auto-antibodies attack and breakdown red blood cells. This can present with fatigue, dizziness, pallor or yellow discolouration of skin (jaundice). Idiopathic Thrombocytopaenic Purupura or ITP occurs through a similar mechanism to autoimmune anaemia leading to low platelets and presents with bruising, bleeding or small red spots known as petechiae. Autoimmune neutropaenia (causing low white blood cells) can present with fever, infections and mouth ulcers. These can generally be diagnosed with blood tests and specific tests to detect auto-antibodies may be required.

## Gastrointestinal Disease

Autoimmune disease affecting the gastrointestinal tract is also commonly associated with PID. One study reported that children with primary immune deficiency have an 80x higher risk of inflammatory bowel disease compared to the normal population[1]. Further conditions include autoimmune enteropathy, coeliac disease, autoimmune liver disease and gastritis[3]. These can present with symptoms such as nausea, vomiting, diarrhoea and weight loss. Diagnosis may involve blood tests, imaging and endoscopy.

## Rheumatological Disease

PIDs are associated with conditions such as SLE or Lupus (Systemic Lupus Erythematosus), vasculitis (inflammation of the blood vessels), arthritis (joint inflammation) and uveitis (eye inflammation). These conditions can present with symptoms such as joint pain, swelling, weight loss and rashes or visual changes. Diagnosis normally involves blood tests, imaging such as X-Rays or MRI and specialist rheumatological and ophthalmology evaluation.




## Skin Disease

PIDs can be associated with autoimmune skin disorders such as eczema, vitiligo (change in skin pigment) and psoriasis. Skin biopsy and dermatological evaluation may be required.

## Endocrine System

Involvement of the endocrine or hormonal system results in disorders such as Type 1 Diabetes and Thyroid Disease. 70% of patients with IPEX Syndrome will have Type 1 Diabetes, and high rates of thyroid disease are found in patients with conditions such as IPEX and CVID.

## Management

Management of autoimmune disease in patients with immune deficiency can be very challenging. In order to dampen down the abnormal immune response seen in autoimmunity you have to suppress the immune system. This must therefore be balanced against the risk of causing infections in patients that are already very vulnerable to infection. Common treatment options include steroids, immune suppressing agents such as mycophenolate and sirolimus and biologic therapy that is directed against specific cells or immune pathways. In many cases, it is also the treatment of the underlying primary immune deficiency that will help with the management of autoimmune disease. In some cases, this may also include bone marrow transplant.

*References*


1. *Fischer A, Provot J, Jais J et al. Autoimmune and inflammatory manifestations occur frequently in patients with primary immunodeficiencies. J Allergy Clin Immunol 2017 Feb10. Epub ahead of print*
2. *Allenspach E, Torgerson TR. Autoimmunity and primary immunodeficiency disorders. J Clin Immunol 2016;36 Suppl1:57-67*
3. *Walter J, Farmer J, Foldarvi Z et al. Mechanism-based strategies for the management of autoimmunity and immune dysregulation in primary immunodeficiencies. J Allergy Clin Immunol Pract 2016;4:1089-100*
4. *http://primaryimmune.org/about-primary-immunodeficiencies/relevant-info/autoimmunity/*


***Dr Annaliesse Blincoe is the recipient of an IDFNZ CME grant towards her studies in Immunology overseas.***

***Annaliesse is currently engaged as a Paediatric Immunology Fellow at the Centre Hospitalier Universitaire (CHU) Sainte-Justine in association with the Université de Montreal in Montreal, Canada for a two year period.***

***The department at CHU Sainte-Justine is combined with Paediatric Rheumatology. There is significant and increasing overlap between the specialties of Immunology and Rheumatology; of special interest to Annaliesse is the opportunity to gain further knowledge and skills in caring for children with autoinflammatory, autoimmune and neuroimmunological conditions. These are niche areas she is interested in applying to practice on return to New Zealand.***

***Exposure to the increasing use of biologic agents in both rheumatological and immune disorders will also be of significant benefit.The spectrum of primary immune deficiency is increasing, with rapidly evolving methods of diagnosis and investigation of children with immune disorders. International centres are now able to identify ever increasing numbers of disorders using techniques such as Whole Exome and Next Generation Sequencing and significant progress is being made in the management of these disorders, including Haematopoietic Stem Cell Transplantation (HSCT) and gene therapy.***

***Time spent at an overseas centre such as CHU Sainte-Justine will give Annaliesse the opportunity to gain exposure to these advances in diagnosis and management.***

***This will be particularly beneficial as we are due to start Newborn Screening for Severe Combined Immune Deficiency in New Zealand in 2017.***

# June is Beware Yellow Month for IDFNZ KIDS Foundation

***Promoting the early signs of paediatric liver disease is a key mission of the Foundation – Beware Yellow articles and advertisements in the Bounty baby books alone reach over 60,000 parents of new born babies each year. In addition the Foundation prints and sends out free resources to Plunket nurses and Midwives across New Zealand and as far away as Fiji, Tonga and Samoa. Each year we invest in training and continuing medical education grants to support New Zealand nursing specialists, and sponsor seminars, conferences and talks promoting Beware Yellow amongst medical professionals working with young babies.***


## *Help us to raise awareness using the free resources available:*

- Order free copies of **Beware Yellow pamphlets** and posters to hand out in your local community, we have copies in 5 different languages to get the message through
- Host a **yellow cupcake morning** tea for your "baby and mum" or playgroup
- **Like us on Facebook** and send links to all of your contacts when our campaign is running.
- Buy tickets to our **BEWARE YELLOW movie fundraiser – Despicable Me 3**, invite along friends with new babies to share the message with them

# BEWARE YELLOW

## About Liver Disease

Although liver disease remains rare in new born babies, around 1 baby is born in New Zealand every month with a serious liver disease. Often the signs and symptoms are subtle or non-specific and it can take a while for parents and health care professionals in the community to recognize that the baby has liver disease.

There are many different causes of liver disease in babies. Some of these are extremely rare while others, such as biliary atresia, are more common. Each of the different causes of liver disease may have specific signs and symptoms related to that particular disease and it is not possible to mention all of them in one guide. However, there are some general features which are common in most babies with liver disease.

Help us spread the word – feel free to download these posters from the IDFNZ KIDS Foundation website

Remember - Parents know their babies best. If you have any concerns about your baby's health, then you should contact your midwife or GP. Sometimes, a straightforward blood test, usually done as a heel prick is enough to tell doctors whether or not to worry about liver disease.

## Summary


- Jaundice is common in babies and usually harmless but a small number of jaundiced babies have serious liver disease
- Jaundice on day 1 of life and after 2 weeks of age requires investigation
- A simple blood test called split bilirubin can tell whether your baby needs further tests for liver disease
- Baby poo is usually yellow, green or brown – pale poo which is white or cream needs investigation
- Baby urine is usually colourless – if you can see urine in a baby's nappy because it is yellow or brown then your baby needs investigations
- Itching, poor weight gain and bleeding may also be signs of liver disease
- Liver disease is rare in babies but early diagnosis is essential to prevent complications
- In babies with jaundice, pale poo and dark urine, biliary atresia needs to be considered and an urgent referral to a paediatrician is needed
- Do not hesitate to consult your doctor if you are worried about your baby's health


# Our People

## Dominic Joseph-Kura

***By the age of nine months, many children haven't experienced too much hardship in their lives. They haven't been seriously sick, endured hours of surgery, or lived with numerous medications pumped into their tiny bodies to keep them alive and fight off infection.***

Unfortunately, this isn't the case for Dominic Joseph-Kura. He has suffered fevers, pain, biopsies, blood tests, vomiting blood, a liver transplant and has even stopped breathing and has had to be revived. He was the first child in the Cook Islands to ever be diagnosed with Biliary Atresia. Even though he has been through so much in such a short time he still manages to smile. His family want his story to be told to raise awareness and help others know what warning signs to watch for.

Dominic was born a seemingly healthy little boy in May 2016, the first child of a young father and mother and the first grandson in the family. For the first month or so everything seemed to be normal and he was growing and gaining weight. However, his grandmother and family became worried when his jaundice wasn't clearing up, and instead was gradually getting worse and spreading. They waited a few weeks but when the jaundice spread to the whites of his eyes, they knew something was wrong. Dominic was taken to the doctors and spent two to three months in and out of hospital undergoing many tests.

Dominic's stomach began to swell as his condition worsened. He wasn't sleeping through the night because of fevers and his grandmother could only try and sooth him and lower his temperature by feeding him coconut juice and keeping him as cool as possible. As he deteriorated he was only able to drink coconut juice and milk as any solids he ate would be vomited back up or passed whole. This meant that his body wasn't processing the food correctly and was another sign that he was extremely unwell.

By October his family pushed for x-rays and other tests to be done to see if anything was wrong in his digestive system. The results showed that Dominic was in serious danger. Two days later, after getting emergency passports, Dominic was on the way to New Zealand with his grandma and Nurse Kelly.

Once in New Zealand they were taken to Starship where little Dominic was once more put through a series of test to determine exactly what was wrong with him. His grandmother was told that his spleen and liver were swollen and that he suffered from Hyperbilirubinemia (extreme jaundice). The jaundice and pale poo (another of his symptoms) were both warning signs that Dominic's liver wasn't functioning as it should. Little Dominic, at only five months old, was diagnosed with Biliary Atresia. This meant that the duct which flushes the bile out of the liver and into the gallbladder wasn't working. The bile was building up in Dominic's liver and scarring it irreversibly. The only cure for Dominic was a liver transplant.

Dominic's mum was flown to New Zealand to give consent for him to be put on the liver transplant list. He was put onto nine different medications and underwent many different tests to make sure his body was ready for a transplant. His heart, lungs, and bloods were checked every day and it was recommended that he gain at least ten kilograms to maximise his chances of surviving the transplant. Dominic's family needed to keep him healthy and as relaxed as possible during this time.

New Year's Day came around quickly and Dominic's family was cleared to take their little boy to visit extended family in Auckland. Everything was going well until early Monday morning when he went downhill quickly. He started vomiting up blood and was rushed back to hospital and given a blood transfusion. For three weeks he was in PICU, before being discharged to return to the transplant ward in Starship his body swollen from the sickness and his medication. He was discharged from PICU and on the ward for a week before he was taken back down to PICU to clear fluid from his lungs. Unfortunately, on the way down, Dominic stopped breathing and had to be revived by medical personnel. The rest of his family travelled to New Zealand after this happened to pray with and support his grandma and mum. Dominic needed a transplant right away and was moved up the list. His father volunteered to be tested to see if he was a match, however, the day he was to begin testing a suitable organ was found. Dominic had the transplant that same day undergoing an eight-hour surgery. His family learnt that Dominic's defective liver was so swollen it was hard to remove. He spent three weeks in recovery in PICU and had to have a second operation because of a blood and food leak in his oesophagus. After that he had to be fed through a tube.

Dominic loves nursery rhymes on the tablet, the Wiggles and Cookie his Cookie Time bear. He now moves and claps to music and is starting to rebuild his muscles enough to wave. The next step for Dominic is to come off all the tubes which are feeding him and monitoring him, and then once that is done he will need to learn to eat again, as well as doing physical therapy to help build up his wasted muscles. After receiving a donation of clothes his family was able to give his outgrown clothes to Ronald McDonald House to help other families in need. They would like to thank the staff at Starship for everything they have done for Dominic, Ronald McDonald House for accommodation since day one, as well as KIDS Foundation for all their support with food vouchers and even a phone top-up which enabled them to call home to speak to family during times of crisis.

Little Dominic is such a special boy and a true fighter, and his family want this article written to highlight the warning signs of sickness in new babies. They explain that if they hadn't pushed for testing and information about Dominic's results and condition, he could very well not be alive today. Prolonged jaundice and pale poo or dark urine are major red-flags that indicate a child is not well, and should not be ignored. If your baby, or the child of someone you know is exhibiting these symptoms, go to your local paediatric service as soon as possible.

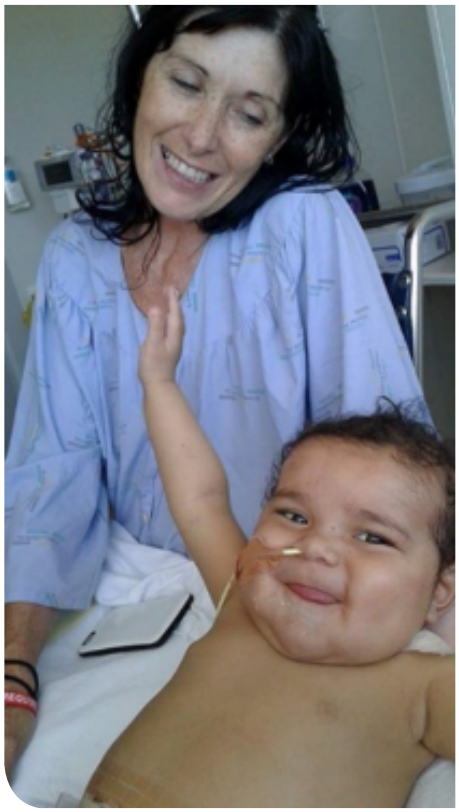
Tangi, Dominic' grandmother, is determined that when she returns home to the Cook Islands she will take Beware Yellow posters and pamphlets to pass onto medical professionals and families to promote the Early Warning Signs of Biliary Atresia and Paediatric Liver Disease.

***"Never give up hope, all you can do is hope."***

Tangi Ruvea, Dominic's Grandma

# Armani Kaifa-Wolters

***Today Armani is a smiley, happy boy of ten months. Born in Christchurch he is the youngest of four children and loves to chat and laugh, and has a good sense of humour. He also loves to eat according to his mum, Nicole.***



However, just a few short months ago, Armani was a completely different little boy.

After a normal pregnancy and birth all seemed well until Armani's traumatic journey began when he was only six weeks old. He had jaundice, as a lot of babies do when they are born. However, when it didn't go away, experienced mum Nicole knew something wasn't right and talked to her Plunket nurse about what could be wrong with her son. This would be instrumental in helping to diagnose Armani. If Nicole had ignored the jaundice things could have been very different for Armani and his family. The nurse suggested that she visit her GP to make sure everything was alright. By then the jaundice had spread to the whites of Armani's eyes and his family was seriously worried about his health.

At the doctor, he was examined and then sent straight to Christchurch hospital. There they performed tests, bloodwork and three biopsies before he was transferred to Starship. His blood results showed that his liver wasn't functioning correctly and his condition was critical.

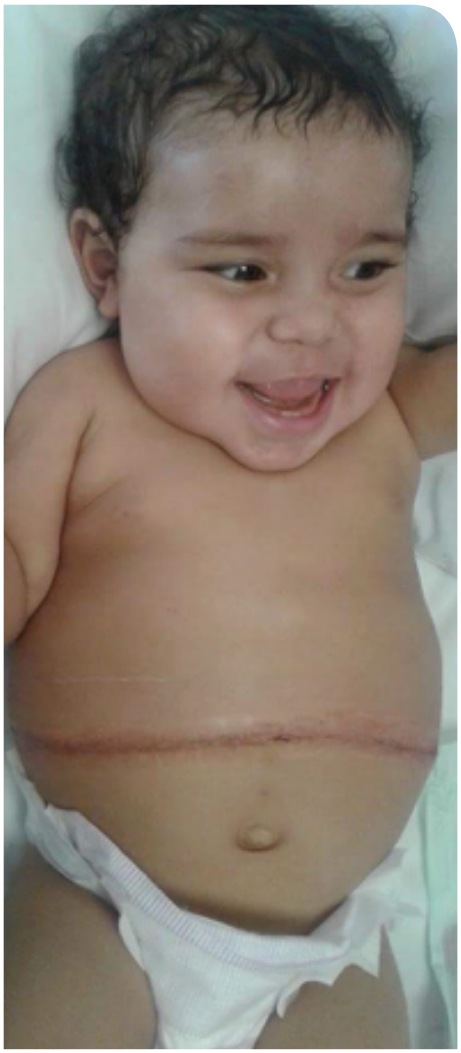
Starship doctors diagnosed him with Biliary Atresia at five months of age. Normally, a Kasai procedure is done to re-establish the flow of bile from the liver into the intestine so that it is not building up in the organ.

However, because of Armani's late diagnosis (he was already five months old by this time), there was too much scarring on his liver. Therefore, his name was put down on the liver transplant list. Returning home to Christchurch was difficult for Nicole as she had no idea how long Armani would wait for a suitable donor organ. Leading up to Christmas this was a very sombre holiday for the family with seemingly endless days in Christchurch hospital and weekly transfusions.

Right away Nicole's mother put up her hand to be tested as a live donor. There was good news ahead, however, when his grandmother was cleared as a donor match, she and Armani returned to Auckland and underwent surgery on the 31st of January. Everything went well and, amazingly, Armani was out of Intensive Care in one day. His grandmother also made a good recovery.

His journey wasn't completely smooth, however. After the transplant, he suffered a blood clot and surgeons had to operate once again to fix the problem. He came through that operation well and has not had another complication or set back since. Armani is currently spending three months recuperating at Starship before returning to his brother and sisters in Christchurch.

The next steps for Armani now are to stay healthy and continue to gain weight. His mum says that he may even be sent home early if he continues to recover and improve as quickly as he has been in the last few weeks. He has the visit of his siblings to look forward to as well. When he returns home, he will continue to have bloodwork done and medication morning and night but he is well on the way to recovering completely and enjoying a full life.



Needing a life-saving liver transplant is every mother's worst nightmare, made even more complicated by having to uproot to Auckland away from her normal family network, Nicole says that she has had the most amazing support. Her mother has given her time and was the first to offer to become a donor for Armani. Nicole says that although she is still in pain, she has been a wonderful encouragement throughout and even through a tremendous amount of pain she has stayed positive and strong.

Her gift has been the seed for a thousand others in Armani's life including his siblings having a healthy brother.

The family would also like to thank KIDS Foundation for everything they have done. The financial support, encouragements and support visits have also been incredible with the Foundation also flying Nicole's three other children up from Christchurch to stay with their mum and brother until they are released from Starship. Nicole would like to thank all the staff at Starship for their help and everything they have done for Armani. She would also like to thank God for all his provisions during this long journey and is thankful that Armani is doing so well now.

Nicole Wolters


*Charnisa (centre) with her Warrior Hero's.*

# Charnisa Kitekei'aho

***My name is Charnisa, I'm 15 years old. I had a liver transplant when I was 6 years old that changed my life. This is my story.***

I was a young, healthy, normal kid doing the things all kids do. Then, one day, I got very sick. My tummy started to hurt so I told my mum. She took me to the doctors for a check-up and they gave me medicine to take. However, as time went by it kept getting worse. One day it got so bad I was rushed to the doctors. That's all I remember. When I awoke from what I thought was a dream, I was in the hospital. My mum and dad were standing by my bedside staring at me through tears. I wondered why.

What I didn't know was that I'd had a liver transplant to save my life. The doctors later told me that my health had declined so fast that they had to operate. I spent three months in hospital recovering, learning to walk and trying to be normal again.

During that time, we lived at the Ronald McDonald House where we met people and made so many friends. Sadly, some of them are no longer with us. I have fond memories of them and miss them.

After my time at Starship, I was ready to go home again to my family and it felt good to be home again. During that time my family and I moved to the South Island.

I attended Marlborough Girls' College in Year 11. I did rather well with my school work despite constantly going to the doctors and hospital for regular check-ups and the occasional mini operation.

Life seemed to be going fine, until 26th October 2016. I was back in Starship again. This time it was different. My liver took a turn for the worse. The liver I'd had since the first operation was starting to shut down. The pain started to become worse and at times unbelievable despite the pain relief I was given.

Even though it had been very hard for me, I still smiled through the pain. Three weeks later I struggled to smile. I felt my strength slipping away and the pain was increasing. My family, especially my mum and aunty, did their best to keep me smiling and focused. But I could see that their hearts were breaking. I needed another transplant and fast. My life was slipping away and my smile was fading.

Finally, we received the news we had been waiting for; a liver had come.

December 5th I went in for the operation and my family waited anxiously for my return and safety. When all went well there was a huge cry of relief as they were told the good news.

Since then I've had many visitors and I know more will still come. I'm not sure what tomorrow will bring, that day is uncertain. All I can do is focus on the now. As I lie here in bed, the nurse busy with her notes, I play Candy-Crush on the iPad. My dad is at my bedside looking at me writing my story through his eyes as he remembers and recounts what I've been through.

This is my story, my life. My smile has returned again.

Charnisa Kitekei'aho

## Charnisa January 2017

Charnisa is a courageous young lady and has continued to recover well from her second liver transplant. Spending three months in Auckland recuperating she has been kept occupied with treats and outings as she regains her strength. Her goal is to return home soon, continue her studies and to have a career as a medical professional.

Charnisa – you will make an awesome Doctor!

# Cycle Ride in memory of Mataru


*Rory Graham receiving a certificate of appreciation for his outstanding support.*

**Rory Graham was devastated when family friend and KIDS Foundation patient member Mataru Taimerua passed away unexpectedly in February this year.**

Mataru aged 5, was a fierce fighter. He was very sickly as a toddler, with quadriplegic cerebral palsy, liver problems and was non-verbal. Mataru spent much of his short life in Starship hospital. In 2014 Mataru received a much needed liver transplant, he later developed lymphoma and underwent chemotherapy which was very hard on his body and later developed dystonia. Mataru was cared for by foster parents Janet and Raj - family life revolved 100% around Matura. Janet and Raj were touched when Rory decided to raise funds in memory of Matura, to benefit KIDS Foundation which had supported him through his medical journey. "Rory's Cycle for the KIDS "was set up on Everyday hero and involved cycling the last leg of the Wellington to Auckland BDO Cycle race.

Rory entered at the last moment but managed to raise an impressive $6,620 for KIDS Foundation, to assist other child patient members facing similar medical challenges.

# From the Starship Liver team

Hi Everyone,

Just a reminder that the flu season is upon us again and already we are seeing cases of influenza A in the community.

Children who have chronic illness or transplant are entitled to free vaccination.

Many adults are also able to access free vaccination so please make contact with your GP practice as early as possible and get your flu vaccine done.

Influenza ( Flu) is not just a common cold and does cause serious illness and death every year in New Zealand.

Ring the CNS Phone 021 837 870 or email us at StarshipliverCNS@adhb.govt.nz if you have any questions!

May those winter bugs stay away!!

Karyn Cate and Meredith

# *Thank you to Holiday Helpers Network Waiheke Island for once again sponsoring a IDFNZ KIDS Foundation family.*

**The Cherry family from Christchurch were able to relax and enjoy some much needed time away. Up until now visits to Auckland have revolved around Luke's liver transplant treatment or Baby sister Amelia's heart surgery.**

It was wonderful to see this amazing family take time out together for a real holiday and to enjoy the last of the summer on beautiful Waiheke Island. As first time visitor's to Waiheke, Megan and Adrian commented on how gorgeous the Island is ,the locals were so friendly and the businesses sponsoring the trip so very generous. "Thank you for the wonderful opportunity and generosity of allowing us to have a trip away as a family where we could rest, regroup and more importantly have some fun! We feel very fortunate to have had this holiday.

Thank you for choosing us." *Megan Cherry.*

# Sponsorship & Fundraising

## YOU ARE INVITED TO A FUNDRAISER EVENT FEATURING A PRIVATE SCREENING OF


### *Beware Yellow – Fundraiser event – Saturday June 17th at 11 am*

Join **IDFNZ KIDS Foundation** for a private viewing of the new Despicable 3 movie – enjoy family time while supporting this worthy cause. Wear an item of Yellow to help raise awareness of the early warning signs of paediatric liver disease.

Tickets are $20 per person and include a drink and welcome gift – contact info@idfnz.org.nz to purchase. Treat the whole family and help raise awareness of the early warning signs.
**HOYTS** - Botany Town Centre, 588 Chapel Road.

The Kids Foundation

BEWARE YELLOW

IDFNZ

*P: 0508 300 600 or +64 9 523 5550 E: info@idfnz.org.nz* ***www.idfnz.org.nz***