"The best advice I can give is this: love your child and know that they love you. Let that love be the light that guides you through whatever lies ahead. Though all of us whose lives have been touched by Pompe's walk a dark path, all the darkness in the world has never yet put out a light."
Kevin O'Donnell, 1994.

At 16 months, Nicole Collicoat (born 2nd March 2004) has exceeded her life expectancy. She has Pompe's disease. Her sister, Pauline, died of the same disease eight years ago at 14 months. More than 17 years ago her eldest sibling was still-born, now considered to have suffered the same disease. Nicole has exceeded her life expectancy because she's one of about 17 babies in the world receiving a new and still unlicensed treatment for Pompe's. The treatment with a drug called Myozyme is currently unavailable to most sufferers because worldwide, Myozyme is licensing approval. Myozyme uses cutting-edge medical technology, and like many drug developments, has its history of politics. And because Pompe's is a rare disease, the development of this potential cure has been an uncompromising battle of persistence by many: sufferers like International Pompe's Association (IPA) Board Member and Australian Pompe's Association (APA) President, Helen Walker; parents of lost children, like Scottish agricultural scientist, Kevin O'Donnell; and scientists like Dr Arnold Reuser and Dr Ans van der Ploeg, the leading world authorities of Pompe's disease, in the Netherlands.

The development of the potential cure has passed through three drug companies and is now with Genzyme, who has thrown what's thought to be more than 300 million dollars into research for Myozyme. Following years of research and testing, Genzyme has now submitted a Biologics License Application to the United States Food and Drug Administration for Myozyme. If approved, Myozyme will be the first treatment for patients with this debilitating and often fatal disease.

But the wait is frustrating. Helen, like many Pompe's sufferers, still waiting for access to Myozyme, says it's like being in the desert dying of thirst. You can see the glass of water but you can't drink it.

Pompe's affects about 5,000 to 10,000 people in the developed world (third world figures are unknown) and is caused by a defective enzyme called acid alpha-glucosidase or acid maltase. Fully functioning, this enzyme breaks down glycogen, a form of sugar stored in muscle cells, which serves as a primary source of energy. Without this enzyme, the glucogen builds up in the muscles, damaging and weakening them. Although the brain is unaffected, every place where a muscle is involved is affected – from breathing and digestion to swallowing and movement. The results of this accumulation of glycogen are primarily the degradation of skeletal, heart and breathing muscles.

Infants with Pompe's disease (infantile-onset) generally die before 12 months of age because their heart muscle is also affected, so therefore in infants the disease is more aggressive than in older patients. Annette Collicoat, mother of Nicole, says her daughter Pauline, who died at 14 months, had classic infantile-onset Pompe's disease symptoms of extreme muscle weakness.

"Pauline was a really floppy baby," says Annette. "She was about 4 months when she was first diagnosed, but we didn't really know anything was wrong until she got a cold." Rapidly Pauline became worse as her heart, liver and tongue became enlarged and weak, eventually leading to heart and respiratory failure.

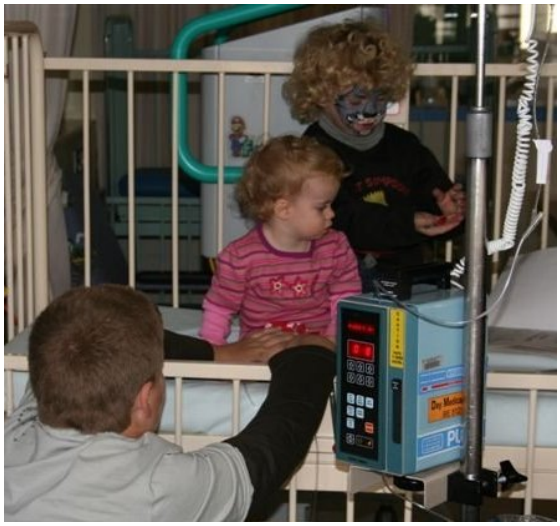
It's a disease which words cannot adequately describe the suffering. Geoff, her husband, was away when Pauline died in Annette's arms. The pictures of her show a gorgeous child with lovely dark eyes. "It's something you never get over," Annette says.

Metabolic physician, Dr Heidi Peters, heads the team administering Myozyme at the Royal Children's Hospital in Melbourne. Her association with the Collicoats goes back to Pauline. And things have changed greatly since then, the first being in early detection. The three children born since Pauline's death, William (6), Phillip (4) and Nicole all underwent testing for Pompe's disease at birth. Nicole was the only one who came back positive.

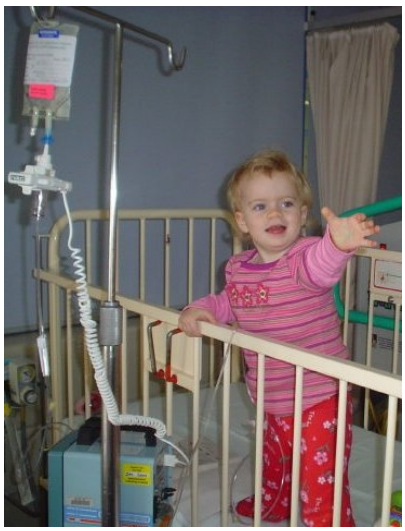
At the time of diagnosis, in what was considered by the Collicoats as hope, was a current world trial conducted by Genzyme with one position left. Called the 1602 trial, to qualify, babies had to be less than six months and patients and their

families had to spend at least six months in the clinical trial site. The 1602 trial has relocated families from Peru to the United States, Turkey to England and Germany to France. In all of these cases, the relocated families were unable to speak the language of the country they were attending. For the Collicoats however, the logistics of relocation were monumental – they have six children and limited finances. There was also the cost of medical insurance to cover Nicole for possible complications, not covered by the trial itself.

Through the collaboration of Genzyme, the Royal Children's Hospital, The Murdoch Children's Institute, the APA and the IPA, treatment was able to commence for Nicole through the Expanded Access Program, which treats patients on compassionate grounds.



During those five hours, Nicole's siblings wander in and out of the room



Treatment means that for every fortnight for five hours Nicole is hooked up to a machine which administers Myozyme.

A team of doctors and nurses are needed: two Metabolic Physicians, a social worker, two dieticians and a team of nurses including a Metabolic nurse. According to Dr Peters, as well as logistics, careful preparation and measurement of the drug, it involves a lot of paperwork which increases valuable research data and aids in approval by world drug authorities.

Nicole is responding well to the treatment and so far has experienced no side effects.

"Medically she is going fantastically" says Dr Peters. "I'm very pleased with her progress. She's certainly past her first birthday and going really well. In fact, it's hard to pick the difference between her progress and that of a healthy child."

Although the future is unknown for Nicole, Dr Peters is cautiously optimistic. "There are no children that have reached later years and that have been on enzyme therapy [Myozyme]," she says. "I hope that she continues to be well and healthy, but we have to be aware that she has this condition and monitor her development, her heart, her respiratory function and hope that she continues well."

Dr. Peter: "I'm very pleased with her progress…In fact, it's hard to pick the difference between her progress and that of a healthy child."

Pompe's disease is genetic and passed to children when both parents have a defective gene. And while Nicole is progressing, it's not just the loss of two children, the Collicoats bear. Knowing what they went through with Pauline, the decision to have more children has brought a good deal of criticism.

"People can be hurtful when they don't understand," says Annette. "Many people [when they found out about Nicole] told me flat out, 'well why have you gone back for more?' and that it's basically our fault for having more children. We both came from big families and wanted a big family. I have had to cut ties with people. It has hurt. And now I often don't bother telling people about Nicole."

In treatment for Pompe's, Nicole is lucky; she's one of only three patients in Australia receiving Myozyme. And because detection and hence treatment was early, she has a greater chance of success. Treatment means that for every fortnight for five hours Nicole is hooked up to a machine which administers Myozyme. It's an arduous ask for any child over the age of one to stay in the one spot, let only have nurses and doctors monitor her every half an hour. But Dr Peters says Nicole handles it well.

Annette and Geoff accept that this trip to Melbourne, every fortnight from their home town of Maryborough will continue for the rest of Nicole's life. If accommodation is unavailable, they'll leave home around six in the morning and arrive back some 13 hours later. At home, in case of an emergency, they also have the support of their local general practitioner, Dr Robert Carson.

The Collicoats are a fully-fledged family unit. The day I met them at the Royal Children's Hospital, all eight of them, were in attendance. During those five hours, Nicole's siblings wander in and out of the room, attend the Starlight room, or help out in such a relaxed way, there's no mistaking that spending time in hospitals is now part of their existence.

On one occasion when Annette is changing Nicole's nappy, the drip cord keeps getting tangled. Next to the cot, 11 year old Shaun, is playing a video game. Annette turns to him and says, "help me Shaun, talk to your sister". Without a word, Shaun pauses his game, steps to the screen, turns it towards Nicole so she can see and resumes his game. His strategy works a treat.

When Phillip, his face painted as a wolf, returns to the room, he's immediately in the cot with Nicole, playing to the camera and letting his sister pull his curly blonde locks.

It's an interesting mix of family participation and I ask Kimberly, the Collicoat's 13 year-old, if she thinks she's different to other kids her age.

"Yes," she says, her painted face from the Starlight room, making her seem younger than her maturity. "I don't complain as much and don't let the little things bother me. I can put up with more things than other kids."

They all help around the house. James, the eldest at 16, cooks. He's hesitant to name a favourite cake or speciality dish, but does admit he likes to cook pasta.

Annette says he always makes a mess. We joke and say that's what men generally do in the kitchen. I wait for James to protest at this alleged unfairness; he doesn't, he laughs and agrees.

James' tenderness and reservedness reminds me of another teenager living with Pompe's disease, 17 year old Marc (Marcus) Leatham. He and his older brother, Dan (19) have been primary carers for their mother, Jayne, since their father walked out three years ago.

Marc (17), Dan (19) with their mother, Jayne

At night they take turns in her bedroom, sleeping on a cot, so they can attend to her during the night. Jayne is now reliant on a respirator, a BiPAP machine, when she lies down, and during the night they make sure it's functioning. If it's not, Jayne cannot breathe, so weak is her diaphragm (the most important breathing muscle). She's also lost the ability to roll over in bed, and the boys now do it for her, instinctively so it seems, for sometimes they can't remember doing so in the morning.

Jayne was diagnosed with late-onset Pompe's disease in 1988, when she was 26. She'd just had Marc, and had trouble carrying him without her legs giving way. As with many adults diagnosed with Pompe's, after diagnosis, many difficulties Jayne had as a child were suddenly explained.

"I've always had trouble getting off the floor and was clumsy," she says. "But I didn't think it was anything, I just thought I was a clumsy child. I could never run or climb up stairs."

As Pompe's disease affects muscles, Jayne's deterioration has been gradual, as the glycogen builds up in the muscles, clogging and weakening them. Now Jayne is confined to a wheelchair, after a long battle to stay out of it. She's exhausted most of the time and is reliant on help for even the most basic of tasks. Despite this, her strength of character is immediately evident and she holds herself with a dignity that belies her condition.

There're no secrets in their family, literally, and for the past few years the boys have dressed her. I ask how she's adjusted.

"I suppose you gradually get used to it," says Jayne. "If it had have been something that had happened overnight then it would be a lot harder in dealing with it. But because it's been a gradual decline, I guess I've had longer to adjust to it. But I still find it very hard."

Even for the most private of tasks, Jayne sometimes needs help. Take going to the bathroom for example. It's not a simple task like it is for most of us.

"I can do it myself, but it takes time and effort and I really have to time it," she says. "Because I'm getting weaker on top, my upper body strength is going and I don't have the strength to pull everything down. Sometimes if I'm really desperate Marc will help."

Jayne has home help for just over 25 hours a week and the boys do the rest with help from the extended family. They cook, clean, do the banking, the shopping and the washing, and Dan drives Marc to work and the like.

At times, she finds this help from her boys difficult to accept.

"Sometimes I pull back from asking. I don't want to ask, I don't want to overdo it with them. I mean they help already so much. Even in the things not directly involving me; Dan drops Marc off at work, where other mums and dads would be doing that."

Although she has little choice, Jayne battles daily to find a balance.

"It goes round and round in your head, balancing between the two extremes. I think it has to do with not being able to do it yourself. And because the boys do so much, it's like I'm onto them all the time. Whatever I chose to do they have to help with. And if I'm asking them to do things that aren't necessary, I get worried about it."

I think of other 17 and 19 year olds and what they are doing. I explain this to Dan and Marc; that from an outsider's point of view, how they care for their mother is exceptional. Jayne agrees, but they shake that down and won't accept it. To them, their help is not extraordinary.

"This is all I've known," says Dan. "I don't see it any differently [to any other mum]. And I don't see I'm doing anything extra. I'm not."

Like the Collicoats, Jayne's parents, Joan and Harry, also have lost two children, then diagnosed with Cystic Fibrosis. With the genetic connection, it's possible, according to Helen, that they may have died from Pompe's.

Of her daughter, Harry says he feels disappointment, sadness, helplessness and frustration. Joan says she feels guilty.

"They say it comes from both sides, but we didn't know," says Joan. "You feel like you've let them down. And it's hard, it's very very hard. And I'm quite sure that Jayne feels the same way with her boys."

Jayne is quick to alleviate her mother's guilt, and says it's not anybody's fault. Although she admits, when it comes to her children, she feels cheated. Despite this, she never gives in.

"I have a theory," says Jayne. "If you stop, everything goes. Sometimes you have to keep going. Sometimes I get really bad and I have to talk to myself. Come on Jayne you can do it and those types of things. Because you have to push yourself through the tiredness, especially in the mornings when it's so hard to try and get up."

There is hope for Jayne. She's just started treatment with Myozyme on compassionate grounds. At first she was told treatment would come in two years.

Then after those two years, she was told it would be two more. And even though Jayne has now received four treatments, she never quite believed she'd get it.

"You need to have hope, but too much hope can be just as detrimental because it plays on your mind and you keep hoping. I suppose if you don't get your hopes up then you can't be let down, you can't be disappointed. I think I just said, look it's not going to come. And it's only because of Bronwyn [her sister] and Helen Walker that I got treatment, otherwise I wouldn't have it now."

Of course getting treatment earlier may have prevented her from being so frail now, but even to receive treatment at all has been a logistical challenge. There were legal components to work through, and St Vincent's Hospital in Melbourne, the treating hospital, had to be assessed by Genzyme as capable of handling the procedure. And there's the paperwork. Even when Jayne takes Panadol, this has to be reported to Genzyme in addition to the collection of other data.

"It's quite an intensive process," says Jayne's sister, Bronwyn Morkham. "Even though Jayne is part of the official trial, Genzyme still requires that they run to trial protocol and they collect the data. So it's a real feather in the cap for St Vincent's that they have taken on this, but also that Genzyme has approved them as a world class facility for ground breaking research. And without their support this would not be happening."

Living with a rare disease also has innumerable other complications and frustrations. Not only is it hard to explain - Jayne, like Annette has given up - but it doesn't fit into any neat categories within the Disability Support System.

"What you need," says Bronwyn, "is persistence. And what's difficult if you are sick is persistence. There's not enough in it [the disability system] so you're put in a position where you have to fight for every small thing that you can get, and fight over the top of others who might be as equally as deserving. But unless you yell hard enough or loud enough (the old squeaky wheel syndrome), you won't get anything. And this ends up putting you in this horrible position where you are constantly fighting the system, which is wrong. It's bad enough to have a disability or a disease, let alone then having to ramp up to the level and have to fight for the very basic support to keep you independent."

Of what is there, it's frustratingly thin, which many people with a variety of disabilities can relate to.

"There's too much demand and too little resources," says Bronwyn, "and they're trying to give a little bit to everyone. And within the system there's the sense that you should be grateful for what you've been given. It's a whole mind set around it and in aged care too. In that if you get two or three hours a week care, you should be grateful for that. Whether it's going to be adequate to support you seems irrelevant."

It's not just the difficulty in getting treatment or lack of resources facing people with a rare disease. Take a look, for example, at what critical illnesses are covered in some insurance policies: cancer, chronic kidney failure, heart attack, organ transplant, artery surgery and stroke. There are no rare diseases.

Despite the frustrations in treatment, access and funding, Pompe's patients continue to wait and hope. They're closer now to the treatment, which may not cure them or stop them dying from the disease, but will certainly make a difference to their quality of life.

If the Biologics License Application is approved, it's expected Myozyme will receive priority review by the U.S. Federal Drug Authority. If approved, it's then hoped the treatment will be fast tracked into Australia and approved by the Australian regulatory bodies and given Federal Government funding. For the average person, and one restricted for years by Pompe's disease, funding their own treatment is financially impossible.

In the meantime, every help is gratefully received. The Paul Newman Charity Fund has just awarded a grant to the APA for nearly $8,000 for six special cushions for wheelchairs. The cushions, designed in the Netherlands, allow the user to pass urine without being transferred to the toilet.

"These pillows will make a huge difference to the independence of six of our members," says Helen. "It's donations like these that keep us hoping that somehow, someway, we will receive the treatment we need."

For more information about Pompe's disease, go to the website of the International Pompe Association (IPA) at: www.worldpompe.org

An article written by: Lisa Louden (Victoria, Australia)