

LSDP Review
By Megan Fookes
On behalf of the Fabry Support Group of Australia

Introduction

The Fabry's Support Group (FSG), on behalf of its members, welcomes the opportunity to provide comment on the current arrangements for supply of enzyme replacement therapy (ERT) to Australian Fabry patients.

The LSDP has allowed sufferers of many diseases to lead a better life. For some patients this may mean just arresting symptoms but for others being on treatment allows them to move forward and normalise their life. Basically it is the LSDP which has helped Fabrys patients throughout Australia. These benefits are the quest that the Fabrys Support Group would like continued.

In this submission I will review:

1. Fabrys Disease – Symptoms in childhood, teens and ageing
2. What is Fabrys Disease?
3. Treatment of Fabrys Disease
4. Fabrys Support Group – its role and aims
5. Life without ERT – My father's journey of Fabrys prior to ERT
6. Positive aspects of the Life Saving Drugs Program – what is working
 - Cost – unaffordable without LSDP
 - Improves quality of life
 - Allows Fabrys patient to contribute to local community
 - Localised treatment
7. Opportunities for enhancement of the LSDP include
 - Home Infusions
 - Process of funding ERT
 - Guidelines for treatment of Fabrys Disease
 - Designated treatment centres
 - Communication from doctors
 - Direct/Indirect costs incurred by patient on ERT

Do you recognise me?

Source: National Fabry's Disease Foundation website



Has anyone in your family or a family you know suffered at an early age from kidney disease, kidney dialysis, kidney transplant, heart arrhythmias, left ventricular hypertrophy, heart attacks, transient ischemic attacks (TIAs) or strokes, or passed away early in life due to kidney failure, heart attacks or strokes? Many physicians, parents, teachers, family members and friends know someone with Fabry's disease who doesn't realise he or she has the disease. While disease

manifestations vary greatly from one individual to another, these are some of the common signs and symptoms. I have family members who have been diagnosed with heart disease, kidney disease, and central nervous system disorders and/or have suffered with or lost their lives to heart attacks, strokes and kidney failure at an early age in life (30-50s).

As a child:

- I complain a lot about burning pain in my hands or feet.
- I either don't participate in strenuous physical activities or don't last as long as others.
- I often just don't feel well or have a general body ache.
- I often don't want to go to school or participate in sport activities.
- I overheat easily because I don't perspire much, if at all.
- I have unexplained high fevers sometimes resulting in clinic or hospital visits.
- I have small reddish-purple spots on my trunk region and umbilicus
- I sometimes have mild to severe stomach cramps and diarrhoea after eating.
- I sometimes have low self esteem and lack confidence because I can't do the things the other kids can do.

As a young adult:

- Many of the other symptoms continue, and:
- The small reddish-purple spots on my body are more visible and more abundant.
- I am not very tolerant of hot and cold temperatures or of temperature changes.
- The pain episodes are limiting and often debilitating, and I get shooting pains in my extremities.
- I have proteinuria and I sometimes have abnormally high levels of albumin in my system.
- In an optometry exam, I have an opaque streaked or whorled pattern on my corneas.
- My diarrhoea is worse (often after eating) and often disrupts my daily life.
- I spend a lot of time in the bathroom because of diarrhoea
- Sometimes I have severe stomach cramps, and nausea and vomiting.
- I often have significant swelling in my lower legs, ankles and feet even without evidence of kidney or heart dysfunction.
- I am developing a progressively worsening chronic cough.
- I have low energy levels, fatigue and sometimes depression about my situation.
- Physicians cannot determine what is causing the various symptoms I exhibit.
- I often am unable to work or work sporadically because of my poor health.

As I age:

- Many of the other symptoms continue, and:
- Other people can hear me breathing more; I progressively wheeze quite a lot.
- I have mild to severe ringing in my ears and I have progressive hearing loss.
- I get tired very easily and often tend to sleep a lot.
- My Cardiac ECG is abnormal indicating I have Left Ventricular Hypertrophy.
- I am having cardiac problems such as rapid or erratic heartbeat.
- I sometimes have attacks of atrial fibrillation at an early age.
- I've had one or more heart attacks at an early age.
- I have kidney disease at an early age but I often don't have diabetes.
- I am on kidney dialysis.
- I am on a kidney transplant list or have had a kidney transplant.
- I've had a Transient Ischemic Attack (TIA/mini-stroke) at an early age.
- I've had one or more strokes at an early age.
- I am at an increasing risk of premature death and need someone to recognise my condition to allow me the option of getting diagnosed, evaluated and treated before it's too late.

Even though I have many of these common symptoms, I have a diminished quality of life and I am at risk of a shortened life-span, a treatable disorder.

NO ONE REALISES I HAVE FABRY DISEASE

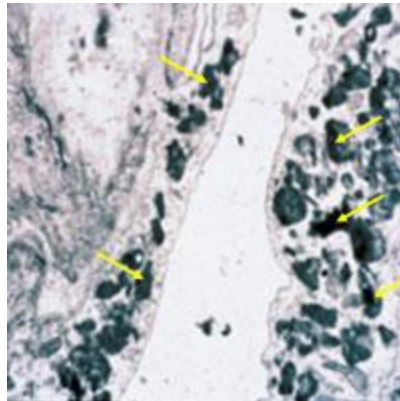
While some individuals exhibit most of these common symptoms to varying degrees, some individuals exhibit only a few of the symptoms and are surprised at the appearance of significant heart, kidney and central nervous system impacts in their 30s, 40s, and 50s.



*Example of angiokeratomas seen on a young Fabrys patient.
Source: Lysosomal Diseases Australia Newsletter Nov 2000*

What is Fabrys Disease?

Fabry's disease is an inherited condition caused by a malfunctioning gene in the body. Because of this in-born error in the body's genetic makeup, an essential enzyme known as α -galactosidase A, or α -GAL is either missing or is deficient in the amount necessary to breakdown specific fatty substances. Without this enzyme, certain fatty substances (primarily one called globotriaosylceramide, or GL-3) are not broken down and removed from the body, and instead stay in the cells. The result is a build-up of this fatty substance in the walls of blood vessels and other tissues over time. It is this accumulation that starts in early childhood that causes most of the problems associated with Fabry's disease.



Vascular Endothelium- GL-3 accumulation

Because the accumulation occurs in blood vessels throughout the body, it leads to a narrowing and eventually a blockage of the vessels found in some of the major organ systems involving the heart, kidney, and brain. By the age of 40, organ damage can be severe and life threatening. The clinical presentation as a result of this progressive accumulation varies from individual to individual; this is true even with affected members from the same family.

If the signs and symptoms of Fabry's disease are recognised early, doctors can help manage them better by treating the patient with the missing enzyme from the patient's body. This is infused into the body via what is commonly known as Enzyme Replacement Therapy. As a result the patient can experience an improved quality of life.

Treatment of Fabry's Disease

Presently people who present with Fabry's disease are treated by a Fabry's Specialist, often a Metabolic Geneticist or Renal Specialist. Fabry's patients also see many other specialists at least once a year and more often when necessary to treat any of the 40 or more symptoms that may occur. These specialists include: Cardiologist, Nephrologists, Neurologist, Gastroenterologist, Ophthalmologist and Dermatologist. Patients with Fabry's Disease are treated by all their specialists annually via the Fabry's Treatment centres that are located in most states of Australia. Fabry's is so rare it is terribly important that patients receive treatment from specialists that understand this disease and have treated other patients that have this condition. Many patients who have Fabry's see their Fabry's specialist once a year and have a series of different tests that enable doctors to see how the disease is progressing. It is via this information that specialists can decide what treatment is best for the patient. Treatment for Fabry's is available via Enzyme Replacement Therapy when the patient meets the selection criteria.



Photo of the Committee of the Fabry's Support Group Inc taken at FSG's first Conference held in October 2007 in Brisbane.

[Fabrys Support Group](#)

In 1994 my Father was diagnosed with Fabrys Disease at the age of 48. After researching and finding very little information about such a rare fatal condition, my mother wrote to the Australian Women's weekly with an article featuring my father and his new predicament. As a result many other people who had been diagnosed with this fate came in contact with my parents and the group was formed. The first FSG meeting was held on 4th June 1994 at the Murdoch Institute, Royal Children's Hospital, Victoria with over 55 people in attendance. It was an outstanding success and the consensus of the meeting was that a Fabry's Support Group be formed. Fabrys Support Group was officially incorporated from 20th June 1994. It was at this time that the Royal Melbourne Hospital Nephrology Department agreed to form a central Fabry's Clinic at the Royal Melbourne hospital and to do some Genetic Research at this clinic. One of the problems of suffering a rare disease such as Fabrys is that any individual doctor is likely to have limited experience in treating the condition. In setting up the Fabrys Clinic many aims were achievable such as:

1. To improve the medical service to Fabrys patients
2. To improve the knowledge of the condition by expanding clinical experience
3. To set up a database documenting the clinical features of Fabry's disease in the Australian population.
4. To centralise care so that new treatments can be initiated when they become available
5. To keep abreast of advances in Fabry's disease research

Fabrys patients saw their Fabrys Doctor at this centre privately and were bulk billed.

The years that followed saw the Fabrys Support Group formulate a

Mission Statement

"To provide support for those affected directly or indirectly by Fabrys disease throughout Australia. Increase recognition, awareness and understanding of Fabrys disease, its effects and potential solutions."

Aims and objectives of FSG

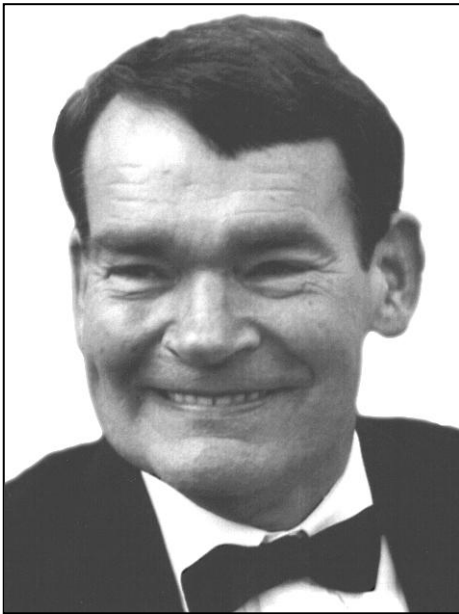
1. Communication of Information on Fabry's Disease and available treatment, to Fabry's Patients, their carers and the Australian Community.
2. Communicate with similar Fabrys Associations in other countries to enable us to ensure awareness of progress worldwide.
3. Obtain sufficient funding to ensure the FSG is able to operate effectively.
4. To ensure that all Fabry's patients receive the most effective treatment, support and care available.
5. To ensure adequate financial support is available to all Fabry's patients, to cover the cost of their treatment, ongoing care and well being.
6. To ensure Enzyme Replacement Therapy is made available to all Fabry's patients.
7. To ensure all Fabrys patients are given opportunity to receive Enzyme Replacement Therapy through home infusions.
8. To raise awareness and understanding of Fabrys disease amongst the medical profession.

Fabrys Support Group funded a medical student to research Fabrys Disease and the members of the FSG happily volunteered themselves to be studied and tested for this project. ERT trials of drugs Replagal and Fabrazyme began in the year 2000 but were mainly available to those patients who were experiencing vital organ damage. This program was also available to some interstate patients but not all. In 2004 funding of the treatment of Enzyme Replacement Therapy became available for 55 patients with Fabrys Disease via the Life Saving Drugs Program.

Let me illustrate what life without hope or treatment for a patient diagnosed with Fabrys Disease. The patient in this illustration is my father who unfortunately did not quite make it to treatment. He died from Fabrys Disease just months before ERT began here in Australia.



*Photo of oldest FSG member celebrating his 60th birthday
He has been on ERT for 9 years!*



[My Father's Life with Fabrys without Enzyme Replacement Therapy Treatment](#)

My father suffered many years of unexplained elevated temperatures, very painful hands and feet and spots around his midriff of which many doctors saw and could not explain. The pain was somewhat worse in the summer months when he would always try to keep cool. In 1967 Dad had a Cervical Sympathectomy which according to the surgeon would warm his hands and hopefully eliminate the hand pain. This surgery was unsuccessful. He then went on a campaign of visiting alternative professionals i.e. hypnotherapists, chiropractors, naturopaths, Chinese herbalists etc., all to no avail. As he got older the pain seemed to diminish and so did his circulation. I remember his hands were always so cold and a funny bluish colour to look at.

In 1991 Dad discovered his legs were badly swollen and contained excess fluid. His doctor referred him to a renal specialist who, after consulting him and seeing red spots on his lower body, diagnosed him with Fabrys Disease. Finally at the age of 48 years he was very relieved to find that all the pain and suffering from his fairly miserable childhood was not in his head, but in fact caused by this Fabrys disease.

As a result my mum and Dad were keen to find out as much information as they could about Fabrys. Mum then formed the Fabrys Support Group in Australia and had an article published in May 1994 in the Australian Women's Weekly Magazine. The response to this article was incredible and the Support Group was up and running. I have attached a copy of this article.

In 1993 Dad suffered a stroke which affected his left side and after rehabilitation he attempted to resume his working career as a Property & Services Manager for a large Pharmaceutical company where he had worked for 22 years. He walked with the aid of a walking stick, his brain was good, but his thought processes weren't as quick anymore as a result of his stroke. He soon realised that he could not cope with this high powered position and after negotiation, retired on a Disability Pension. In 1994 he was haemorrhaging from the bowel and after 8 months of tests and monthly transfusions he had become very anaemic when a growth was diagnosed in his bowel. He was hospitalised again and underwent major bowel surgery.

In 1995 he discovered a terrible abscess on his left leg that simply was not healing. He was hospitalised again and he was diagnosed with a blood clot in his left leg. The clot stopped any circulation in his leg and resulted in an above knee amputation. He spent six months in hospital in rehabilitation with other elderly stroke victims and paraplegics and quadriplegics, He seemed so sad, helpless and alone! After moving to a private rehabilitation unit where he was amongst a different clientele he worked hard to rehabilitate with the aim to walk again! He didn't walk well with his prosthesis due to his stroke and weakened left side. But he tried very hard and never gave up! In 1996 Dad proudly led me, his only daughter down the aisle to marry Mark. He did this so beautifully using his prosthesis which he found so incredibly challenging! There wasn't a dry eye in the room! A couple of years later due to such poor circulation of the left side of his body where the small blood vessels which supply blood to the extremes of the body, the "stump" on Dad's left side became very infected and after dressing his badly infected stump and taking very strong medication every day, it was useless, Dad would need further amputation of his stumpy leg. Dad at this stage had become very weak and was not keeping a lot of food down.

Gastrointestinal problems were a daily occurrence. After many meals Dad had terrible diarrhoea and vomiting. He lost a great deal of weight and became very weak and susceptible to sickness and common viruses. Fortunately he came through the long surgery managing not to take too much of his stump off. It was so painful to watch a positive and cheerful person reduced to a pain-racked invalid.

Dad's vascular system was quickly deteriorating. Added to these problems was the affect that the Fabry's had on his heart, bouts of pneumonia with longer stays in hospital - one such stay he had septicemia, golden staph and pneumonia, but again he fought back and came home. Although Dad was confined to a wheelchair his sense of humor and determination to enjoy life shone through. The local council made disabled access to the footpaths in his area so that Dad could drive his motorized scooter easily from place to place.

One morning he rang to tell Mum that he was having champagne and nibbles with neighbors 6-7 kms away! He set off on the bike track with his scooter enjoying life.

It was in 1999 that Mum and Dad decided to have a holiday away from hospitals. They had originally planned to travel to the United States and visit Mt Sinai Hospital and meet up again with Professor Desnick the forefather of ERT, but soon realised that this was a huge ask, so instead holidayed in Queensland. They had two lovely weeks enjoying the warm weather, peaceful surroundings and enjoying a well-earned rest from hospitals.

However it was too short lived, in November Dad was rushed to hospital again. This time his heart had deteriorated and the doctors said that unless he had open-heart surgery he would not survive. Dad's fighting spirit was inspirational. He was so proud to hear the news from me that he was to become a grandfather for the first time and we all thought that this would help to get him through this terrible time. It was not to be. David died whilst undergoing heart surgery on November 8, 1999.

The Enzyme Replacement Trials commenced in Australia only months after Dad died. It was too late for Dad but he was contented with the news that it was very near and that his grandchildren would benefit from the therapy.

This is one of very many stories that unfortunately I could tell you of life with Fabry's Disease and no treatment. As all available data from published studies from both Shire (TKT) and from Genzyme, confirms that enzyme definitely shifts ceramide from tissue deposits and helps symptoms, particularly nerve pain. Improvement continues as the years after commencing enzyme – after all, the individual person's ceramide load did take a lifetime to accumulate, and people starting enzyme at highly variable ages and disease "loads". When access to Fabry's enzyme replacement programs in Australia began it was an exciting but anxious time in many ways for members of the Fabry's Support Group. It was frustrating for those sufferers who were waiting to start enzyme, sometimes boringly repetitive for those who had commenced regular therapy, disruptive for long-suffering partners, children and work colleague. A challenge for all concerned!

Positive aspects of the Present Life Saving Drugs Program (LSDP)

1. Life Saving Drugs Program relieves cost of Enzyme Replacement Therapy (ERT)

Enzyme Replacement Therapy is an expensive treatment and the FSG reiterates its support that it continues to be financed under a specialised program such as the Life Saving Drugs Program. It costs on average \$400,000 per patient per year for treatment. However this is no comparison to the cost of open heart surgery, kidney dialysis, and kidney transplant, rehabilitation after stroke and heart failure and not to mention many years of hospitalisation and loss of productivity in the workforce! Most people simply could not afford to receive ERT without the help of the LSDP. State Governments certainly don't have this money in their budgets to subsidise such a fantastic program either.

We understand that recent governments have made big savings within the PBS. The FSG believes that it is morally and medically justified that some of these savings continue to be attributed towards supporting patients with diseases that have been inherited through no fault of our own. It's not like we're seeking treatment for lung cancer or a liver transplant after a lifetime of smoking or drinking.

In addition to funding ERT for Fabry disease, the LSDP supports also ERT for some other lysosomal storage diseases. From our research on the internet and discussions with other support groups, it is our understanding that there are only a limited number of these diseases with treatment available now or in the near future. We believe that each of these patient populations are small and well defined and that by using the current model of setting guidelines for patients to be approved for treatment, there is little likelihood of the cost of the ERT programs blowing out like they have for some other well known drugs on the PBS.

2. LSDP allows Fabrys patients to receive ERT

Many people with Fabry Disease who are currently receiving treatment would not be alive today if it wasn't for this program. If they are not receiving ERT they simply would not outlive what Fabry Disease throws at the average sufferer. Many people who have Fabry Disease have endured many major health issues. Fabry affects every major organ of the body. Fabry patients have to endure health issues such as: acroparesthesia (constant tingling pain that affects the hands and feet), Fabry crises (episodes of intense excruciating burning pain), impaired sweating (hypohidrosis or anhidrosis –causing frequent fevers, overheating with exercise and sensitivity to hot weather), skin rash (angiokeratomas) found around the groin area, corneal patterns, gastrointestinal problems causing diarrhoea and nausea and major organ system damage such as; reduced kidney function, kidney failure (kidney dialysis and kidney transplant), enlarged heart, malfunctioning heart valves, irregular heartbeat, heart attack, heart failure (pacemakers and bypass surgery), Central nervous system problems such as dizziness, head pain, premature stroke (many patients are on blood thinners) and emotional issues such as feelings of depression, hopelessness, alienation and denial of their symptoms. It has been clinically proven that ERT has reduced the severity of this long list of symptoms for the average Fabry patient. Put quite simply, if Fabry patients don't receive ERT, bad things happen to them before they die, but they do die.

3. LSDP allows Fabrys patients to experience an improved quality of health and life.

As President of the Fabrys Support Group, I have a lot of personal contact with patients that have Fabrys Disease. Most people are receiving treatment and it has changed their lives. The oldest living Fabrys Support Group member was told at a very young age that he would not live to see 45 years. Today he is 61 years old! He has received treatment for the past 9 years. He claims that he would not be alive if it wasn't for his treatment. He has had less Fabrys attacks, (vomiting and pain in the hands and feet) and no more deterioration of his heart or kidneys. Many people on treatment are functioning as per 'normal'; the treatment has allowed their health to remain stable and not deteriorate and to function in their day to day lives as 'normal' citizens of society. Fabrys patients on ERT are enjoying their improved quality of life and able to socialise out of the home, enjoy a better quality of life with family and loved ones. It allows the Fabrys sufferer an opportunity to operate as any other individual who does not have the disease.

4. LSDP allows Fabrys patients ability to contribute to the local community

The treatment has allowed Fabrys patients opportunity to contribute to their local community and function in the paid work force as their fellow peers, friends and family do. Not only does this allow more productivity in society, it enhances the patients self esteem and level of self-worth. Another patient who has Fabrys and is a father of two young children said that without this treatment he probably wouldn't be alive today. It has contributed to his health, allowing him to provide for his family and continue working in the paid work-force. He also says it is better for the kids not to see him so sick and unwell all the time. ERT has relieved some of the typical symptoms of Fabrys in his day to day life allowing them to function as a 'normal' father. Before treatment he suffered daily from the usual Fabrys stomach attacks and these have reduced significantly. They have recently returned from an overseas holiday which they timed around treatments. They certainly would not have been able to travel before he received treatment.

5. LSDP allows ERT to be localised. Patients are receiving treatment closer to home

When ERT was first conducted here in Australia the only treatment centre that conducted the therapy was Melbourne. Now ERT is conducted in most states of Australia, beginning for the first 6 months at an LSDP-nominated State referral hospital. In more recent years ERT is conducted at patient's local hospitals after their treatments were established to be safe by their referral centre hospital. The referral centres still oversee the patient's progress and are monitored every 6 months to watch their progression with the disease and their treatment. Localising treatment has released the burden of travel for the patient, their carers and families and their employers. It allows the patient to receive some control of their treatment and care. After all not every Fabrys patient live two bus stops from their major state hospital treatment centre!

6. Critical need for the LSDP to continue

I would hate to think what life would be like for the patients that have Fabrys Disease without their current ERT. Compared to what my own father went through in his Fabrys journey of countless operations, constant hospitalisation and enduring so much on a day to day basis not to mention the emotional strain on family, friends and loved ones who watched helplessly as my father endured so much. The medical practitioners and professions did what they could base on so little they knew about this fatal condition. Dad was a living 'guinea pig' who was prepared to try anything to help ease the symptoms of this terrible disease. I don't want to see anyone go through what my family endured. Another FSG member has just celebrated his 41st birthday which he never thought at all possible! He believes that his ERT is very much responsible for this mile-stone. He has received ERT for 5 years and for the past 18 months has received his treatment at home. He says *"this treatment has changed my life in so many ways. It's like I have been given a new lease on life! I am the oldest living Fabrys male in my family. My two uncles passed away in the last five years due to Fabrys. One aged in his 50's and the other in his mid 40's. I thought my fate would be the same! I have experienced many new things thanks to my improved health. Things just as simple as being able to play sports with my boys as I never had the energy and could not stand the temperature of being over heated as I could not sweat. I still can't play as long or as hard as they can, but it is good just to be able to do a little with them and they understand when and why I will have to sit out after a bit. Before my treatment I would not even think about going outside with them. I would hate to think what my life would be like without this treatment and often wonder whether or not I would still be here. I would hate to think what my life and the lives of so many other sufferers that are receiving ERT would be like if we could no longer receive treatment. I am lead to believe that if treatment were to stop that you would not only go back to where you were health wise prior to treatment but you would be in a state a lot worse than before and would deteriorate at a rapid pace. I could write pages and pages about how my life used to be when I was younger. About the endless trips to hospital in agony and hours of waiting to be told it's all in his mind go home and take a couple of aspirin. About the months of worry after I had a stroke .As I'm sure everyone else suffering the same disease could but I don't like living in the past and have a great new outlook on the future.*

Like so many members of the Fabrys Support Group who are receiving ERT they are so grateful and thankful to the Life Saving Drugs Program that have ensured Fabrys patients receive their vital ERT. Funding of enzyme replacement therapy gives hope to the sufferers of this disease and their families and for those who have already experienced the benefits of therapy, life without ERT would be very grim.

Opportunities for Enhancement of the LSDP

Whilst the Australian Fabry community are thankful for the provision of ERT to Australian Fabry patients who meet the current funding guidelines, the following comments represent areas which we feel could be improved.

1 Home Infusions

Background: In some cases, FSG members are able to attend a more convenient hospital for infusions after they've have their first 6 months of treatment at the designated treatment centres. This is wonderful for the convenience of the patient, but even better would be giving patients access to home infusions. Indeed, some patients are receiving their ERT via home infusions. This is particularly helpful for patients who have had strokes or are on dialysis, as it allows the patient to pick and choose time of day that suits them to receive their treatment in the comfort of their home. It also reduces the stress and burden on patients who may have too far to travel to the ERT treatment centre. Please bear in mind that most Fabry patients are still in active employment and the time they spend travelling to hospital, coupled with an infusion of 2-4 hours, means that patients can miss a considerable amount of work each fortnight, therefore home infusions can be administered at a time that is less disruptive to the work and social circumstance of the Fabry patient. Home infusions are common given overseas in US and UK for many Fabry patients. Children who are on ERT can receive their treatment at home which allows them to do this after school – causing less disruption to their school fortnight – very important for teenagers who cannot afford to miss too much school.

Proposal: Home infusions should be supported for all patients whenever practicable. This would lead to greater quality of life, greater productivity, and higher compliance – thereby maximising the benefit of therapy.

Benefits of Home infusion of ERT for Fabry patient:

- i. **Time Factor** - No travel to and from hospital placing less strain on the patient who is burdened with doing this for themselves or having a carer take them.
- ii. **Health and well-being of patient** - No travel also means less physical strain on the patient. Travel is very tiring and exhausting for someone coping with a chronic disease such as Fabrys
- iii. **Employment** - Work force would be less affected by the patient requiring time off their employment every fortnight. Most patients take the day off to allow for travel to and from treatment and recovery after treatment.
- iv. **Hospital strain** - No hospital also means a lot less strain on an already over burdened hospital system.
- v. **Risk of infection** – by keeping ERT in the home the Fabrys patient is less susceptible to risk of common in house hospital infections that are extremely fatal for a Fabrys patient.
- vi. **Treating children** with Fabrys is less stressful in the home environment and less threatening for a young sufferer who has a lifetime of treatment to endure.
- vii. **Cost Effective** – Home infusions are more cost efficient via home infusion. Use of Hospital beds and equipment is costly!

2. Process of funding ERT

Background: Two forms of ERT have been available to Australian Fabry patients with funding from the LSDP since July 2004. We greatly appreciate that funding has been provided by the government. However whilst working closely with the pharmaceutical companies to support the listing of their products on the LSDP, we became aware of the long and seemingly unstructured process to list Fabrazyme and Replagal on the LSDP. We are sincerely thankful to Genzyme and TKT (Shire) who made “compassionate use” drug available to some of our more severe members for a number of years in the lead-up to government funding. Some of these members may have suffered further and irreversible damage to their organs had the compassionate program not been available. As we have made clear in this submission, Fabry disease is a life-threatening disorder – one in which the first major event may result in death by stroke, heart attack or kidney failure. We felt like we were sitting on a ticking time bomb when funding was being sought, wondering if and when we would ever get access to funded treatment. For us it was a slow and fearful process. Without a properly structured process, we are concerned that there might be undue delays for the introduction for new alternatives to the current ERT products, should any arise in the future.

We also understand that funding for ERT has historically been announced in the May budget announcements, for funding to begin on July 1st. As we have stated in this submission a number of times, Fabry disease is a life-threatening disease, and any delay in receiving treatment could lead to irreversible damage and even death. If the LSDP is truly “Life Saving”, the treatments need to be made available as soon as a funding decision is made, and patients not made to wait until an arbitrary date that is convenient to the government.

Proposal: The process for the review of funding applications for the LSDP needs to be time-limited and the process structured and published in publicly available documents. This includes reviews of funding for the two currently available products, as well as for any new products that may be recommended for listing on the LSDP. Transparency through all steps of the funding process between companies and Government, patient groups and Government would be welcomed by all concerned.

3. Guidelines for the treatment of Fabry disease:

Background: The current guidelines for receiving ERT in Australia seem to be much more stringent than those used in other advanced nations such as the US, UK and Germany.

The current US guidelines for instituting enzyme replacement therapy in Fabry disease patients are as follows:

- Adult males (>16 yr): At time of diagnosis of Fabry disease
- Paediatric males: At time of development of significant symptoms or if asymptomatic, consider at 10–13 yr
- Females (all ages) Monitor; institute if significant symptoms or evidence of progression of organ involvement

These are much more relaxed than the Australian guidelines. It's our belief that there are still Australian patients who are suffering from Fabry Disease who are made to wait until their vital organs deteriorate before they can receive ERT.

For example, I myself have some Fabry symptoms, as does my child. Should we both be forced to wait until near-organ failure until we get access to funded therapy? We know from some of the clinical studies that patients with more severe disease do not respond as well to therapy as those who receive treatment earlier.

Fabry's needs to be likened to diabetes – Fabry patients require their ERT just like diabetics require their insulin. Diabetes patients take regular medicine to prevent the terrible symptoms from occurring. Fabry's is exactly the same. Once the disease is more established later in life it may be too late to reverse the damage that has already occurred to vital organs.

For example one young male Fabry sufferer who is now 20 years of age was diagnosed with Fabry's at age 17. At the age of 8 he would get burning pain in his feet, brought on by exercise, the heat and fevers. During his fevers he screamed in pain and this was put down as poor circulation. He went undiagnosed until the age of 17 and a half when finishing high school. At this point the burning pain increased and spread to his hands and feet and the lesions increased in number. He now is diagnosed with having Fabry's Disease and is in University but is still yet to receive ERT. When Fabry's patients meet one another within the group and tell their story of how they came to realise that something was not right and naming their symptoms the stories all read so similar to this one I've just relayed.

ERT needs to be available to all patients who have the disease, including children and females, with no difference in the criteria between males and females. Research suggests that it is most important that ERT is administered to all patients who have the disease to help prevent renal failure, heart attack/failure, and strokes.

Proposal: The FSG would like an immediate review of the Australian guidelines to bring them in line with world's best practice. The current guidelines wait too long before ERT can be initiated, which may lead to irreversible symptoms and major organ damage.

4. Designated treatment centres:

Background: The LSDP has nominated a major hospital in each state to give the first 12 infusions of ERT and to also perform an evaluation of treated patients every six months. Whilst we agree that there are certain benefits to keeping a base of expertise at central locations, there are a number of issues with this that have created difficulties for some of our members:

- **Logistics:** Particularly in the larger cities such as Sydney and Melbourne, the designated treatment centres can be a long way away from home. This makes it difficult for patients to be dropped off at the hospital by family members, thus committing the patient to 6 months of travel and high parking costs (another issue in itself).
- **Doctor choice:** Unfortunately not all patients see eye to eye with their doctors. Under normal circumstances if this occurs, the patient changes doctor. Under the current structure decreed by the LSDP, this fundamental right of the patient has been removed. We know of members who are unhappy seeing their appointed doctors who would greatly appreciate the opportunity to visit alternative experts. Patients need to feel comfortable and supported by their Fabry's treating doctor – any conflict in this long-term relationship between the doctor and patient could jeopardise the patient's treatment.

Proposal: More than 1 centre needs to be established in each of the larger cities – in particular Sydney and Melbourne. This would offer the patients a choice of the most convenient location for infusions and would also offer another choice of Fabry expert for patients not happy with their original doctor.

5. Communication from doctors

Background: A number of members have commented that the communication between the Fabry expert and themselves, or between the Fabry expert and their local doctors, has been quite poor. They do not receive much information on their test results or how their treatment is going. Most patients want to be more actively involved in monitoring their health – this counts for both treated and untreated patients.

Proposal: After each 6 month review the patient should be sent a report from the Fabry expert and/or an appointment made to discuss the results. We are happy to abide by the monitoring conditions, but we'd also like to see the results for our efforts.

6. Direct costs incurred by the Fabrys Patient.

Background: Patients who have Fabrys Disease need to stay involved in their own treatment as this is an important part of staying well. Fabrys sufferers visit a range of specialist Doctors and physicians for the varying symptoms they endure. These specialists include: Geneticist, Renal Specialist, Cardiologist, Dermatologist, Ophthalmologist, Nephrologists, Neurologist, and the list continue. Not to mention hospitalisation, operations, medicines, prescriptions, specialised treatments and rehabilitation. I am sure I have skipped a few! After speaking with a fellow Fabrys patient who is on the ERT program in NSW I was surprised to hear the ongoing costs incurred by the Fabrys patient. The male individual is 41 years of age and a father of two young children living in Sydney NSW. His family has top private health cover. After Medicare rebates and health fund rebates his medical costs per year are somewhere around the following figures: prescriptions \$1200, Specialist Doctor visits for kidneys and eyes \$500, General Practitioner \$300 and Lymphatic treatments (massaging, bandaging of swollen legs toes to groin) \$3000. Total direct cost to this Fabrys patient per year after rebates is approximately \$5000. This is just medical costs not his wife and two children!

Proposal: Fabrys patients are rebated for their direct costs that they incur as a result of treating Fabrys Disease.

7. Indirect costs to patients and families.

Background: There are many ongoing costs relating to the operation side of Fabrys Support Group Inc. The FSG represents over 70 people from across Australia affected with Fabrys Disease. This isn't all the people who have Fabrys Disease in Australia. Some do not wish to be in the group and others do not know that it exists. The group has been operating for 14 years and is challenged with providing support to many patients across the country. FSG communicates with its members via newsletters which are sent via email, mail, phone and meetings. FSG meetings are held 3-4 times a year.

Unfortunately FSG can only meet via conference call due to the high travel costs for sending its committee to meet one another face to face. FSG did fund travel costs for FSG committee members to meet but costs were as high as \$10,000. The Association must by law meet 3-4 times a year to represent FSG members. Committee members include: President (NSW), Vice President (Vic), Treasurer (NSW), Secretary (Qld), two Ordinary Members (NSW) & (Qld) and Public Officer (Vic). At the moment FSG committee meets once a year only which limits what support can be made available to Fabrys patients across Australia. FSG also has a website but this needs to be updated and rebuilt. The FSG needs to look at renaming itself to include 'Australia' in its title as FSG is a member of other global Fabrys organisations and associations and it is difficult to locate us due to this. Hence FSG needs an injection of funds to re-develop itself after 14 years of being in operation. The overall costs of operating FSG is as high as \$15,000 this does not include any office supplies, computer running costs, phone costs, electricity costs etc. On average as President of FSG I spend on average 3 hours a day operating FSG.

Proposal: Fabrys Support Group be eligible for funding to assist with the running and new website set up costs of the Fabrys Support Group Inc which represents the Fabry patient community of Australia.

Summary

As mentioned earlier, the Australian Fabry community is very grateful to the Australian Government for the funding of ERT via the LSDP. In Australia the introduction of ERT funded by the LSDP has significantly changed the lives of the patients as presented throughout this submission. The patients who are fortunate enough to receive therapy could not afford to pay for it themselves, so they are very thankful.

However we believe the program as it currently stands is treating patients too late and that the guidelines should be modified to allow for less severe patients to be treated. Patients should also be given a choice of doctor and infusion centre, and should also be given the option of home treatment once they are able to tolerate infusions outside of the hospital setting.

The diseases currently attracting funding in the Life Saving Drugs Program are rare and fatal conditions with a poor prognosis. The funding mechanism needs to address the urgency and the severity of these conditions and fund in a more appropriate timeline with a clear indication of the process for all those concerned.

We welcome the review of the LSDP and hope for positive outcomes for all our members.