Fabry support group Australia

7 November 2014

Dear Committee

Re: Life Saving Drugs Programme Review
Fabry Support Group of Australia Submission

Introduction

The Fabry Support Group Australia (FSGA), 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 Life Saving Drugs Programme (LSDP) has allowed sufferers of Fabry Disease 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. These benefits are the quest that the FSGA would like continued.

FSGA would like to thank the Department of Health; Felicity McNeill and Kim Beswell for making time to meet with representatives from FSGA and other rare disease patient organisations in the meeting facilitated by Rare Voices Australia on 4th September 2014 in Melbourne. It was both helpful and insightful to be updated in the process of the review of the Life Saving Drugs Programme (LSDP) and are pleased to hear that the Australian Government has indicated that it supports, and will continue to support the LSDP to allow the current rare diseases including Fabry Disease to access expensive but vital treatments.

Following the Minister of Health's announcement in April this year that the Post-market Review of the LSDP would proceed, and the focus is to ensure that Australians with very rare conditions continue to have subsidised access to much-needed, expensive medicines was welcomed by FSGA.

On this point, FSGA is very supportive of a review that brings change to the benefit of patients living with this very rare disease to ensure that they can access as needed very expensive and innovative therapies for their very rare life limiting disease.

FSGA understands the review will examine important issues such as access and equity, value for money and the future administration of the programme.

As was highlighted in the FSGA Submission to LSDP in both 2010 and 2008, FSGA are most appreciative of the LSDP for the following reasons:

- LSDP relieves the cost of Enzyme Replacement Therapy (ERT) for the Fabry patient allowing access to vital treatment.
- LSDP allows Fabry patients to experience an improved quality of health and life.
- LSDP allows Fabry patients the ability to contribute to the local community.
- LSDP allows ERT to be localised and in some cases receive ERT at home via ERT home infusion programme.

Treatment of Fabry Disease

There are many valuable symptomatic and protective/preventative treatments recommended for Fabry. As well as ERT there are many treatments in development. Presently people who present with Fabry disease are treated by a Fabry Specialist, often a Metabolic Geneticist or Renal Specialist. Fabry 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, Pain Management Team and Dermatologist. Patients with Fabry Disease are treated by all their specialists annually via the Fabry Treatment centres that are located in most states of Australia. Fabry 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 see their

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Fabry specialist twice a year if they are on ERT and once a year if not on treatment 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. ERT at this stage is the only safe effective treatment that is available to Fabry patients who meet criteria to have it funded by the LSDP. Treatment for Fabry is available via ERT when the patient meets the selection criteria under guidance and consultation with the Fabry state clinician.

Fabry Support Group Australia

The FSGA was founded in 1994 and recently celebrated its 20th Anniversary. A major milestone but also a sad one, as the group reflected the loss of life to those who received late diagnosis of this rare disease, and were unable to receive the benefits of receiving access to therapy in their life time or accessing the therapy in late disease progression.

One of the problems of suffering a rare disease such as Fabry is that an individual doctor is likely to have limited experience in treating the condition. In setting up Fabry Clinics many aims were achieved, such as:

1. To improve the medical service to Fabry 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 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 disease research

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

The years that followed saw the Fabry Support Group formulate a

Mission Statement

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

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

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

ERT is an expensive treatment and the FSGA reiterates its support that it continues to be financed under a specialised programme such as the LSDP. 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 programme either.

2. LSDP allows Fabry 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 programme. 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 Fabry patients opportunity to experience an improved quality of health and life.

The FSGA have a lot of personal contact with patients that have Fabry Disease. Many people are receiving treatment and it has changed their lives. 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. Fabry 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 Fabry sufferer an opportunity to operate as any other individual who does not have the disease.

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

The treatment has allowed Fabry 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. ERT relieves some of the typical symptoms of Fabry in day to day life allowing them to function as a 'normal' person.

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 Fabry patient lives close to their major state hospital treatment centre.

6. Critical need for the LSDP to continue

The current ERT treatment is essential to the lives of patients with Fabry disease. The FSGA members who are receiving ERT are so grateful and thankful that the LSDP have ensured Fabry patients receive their vital ERT. Funding of ERT 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. 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. For example, the current UK guidelines for instituting Enzyme Replacement Therapy in Fabry disease patients includes:

- **Gastrointestinal symptoms** such as pain, vomiting or altered bowel habit which are significantly reducing quality of life and not attributable to other pathology.

The following symptoms may occur in Fabry patients, but in adults would not be sufficient by themselves to warrant ERT **but they may be sufficient in children**:

- Episodic vertigo interfering with quality of life
- Hearing loss

It is 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.

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 FSGA 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.

The current Australian guidelines do not allow for children suffering with gastrointestinal problems to be eligible for ERT, unless they take maximum doses of appropriate analgesia and antiepileptic medications for peripheral neuropathy. This may mean that children who are not necessarily struggling on a day to day basis with neuropathic pain are required to take maximum doses of medications that they may not need before they are allowed access to ERT for their gastrointestinal problems.

2. Access to new Fabry disease treatments:

Background: There are currently two available subsidised ERT treatments for Fabry Disease. FSGA are very interested in the clinical trials taking place whereby new therapies are being investigated and Australian Fabry patients have taken part in such trials. The treatment being investigated is exploring treatments that are administered orally which have shown to be safe and effective for particular mutations of Fabry Disease (those who make some enzyme) and will be an oral medication only. There is also investigation into combination therapies for patients who make absolutely no enzyme and therefore they need to receive both ERT (but in less frequency as the current practice of ERT administration) and oral medication.

This may alleviate the need for a patient to have fortnightly infusions, either in the hospital or at home. This would allow the patient would be able to lead a more normal life.

Proposal: FSGA would like the assurance from the LSDP that as new treatments are approved by the Food & Drug Administration (FDA) in the U.S. and European Medicines Agency (EMA) that the Therapeutic Goods Administration (TGA) Australia will follow suit and allow these treatments to be made available to Australian patients.

3. 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 the FSGA 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 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.

4. 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 their Fabry expert and/or an appointment made to discuss the results. The FSGA are happy to abide by the monitoring conditions, but we'd also like to see the results for our efforts.

5. Direct costs incurred by the Fabry Patient.

Background: Patients who have Fabry Disease need to stay involved in their own treatment as this is an important part of staying well. Fabry 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 continues. Not to mention hospitalisation, operations, medicines, prescriptions, specialised treatments and rehabilitation. A few may have been skipped! The FSGA knows of one Fabry patient on the ERT programme in rural NSW, who is being charged courier fees by her local hospital to get the ERT drugs to the hospital.

Proposal: Fabry patients are rebated for their direct costs that they incur as a result of treating Fabry Disease, especially having access to the ERT drug at the most convenient hospital to the patient.

Summary

The Australian Fabry community is very grateful to the Australian Government for the funding of ERT treatments 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, the FSGA believe the programme 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.

As new treatments become available on the international market FSGA strongly recommends they be considered for reimbursement on the LSDP. Taking into account the benefits that can be made available to the Australian Fabry patients to ensure they are able to access safe, effective treatment for their rare disease.

The diseases currently attracting funding in the LSDP 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.

The FSGA welcomes the review of the LSDP and hope for positive outcomes for all our members.

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