Australian NPC Disease Foundation Inc.
'Finding a cure for NPC Disease'
'PERSEVERE'

29 March 2010

Committee Secretary
Senate Community Affairs References Committee
PO Box 6100
Parliament House
Canberra ACT 2600
community.affairs.sen@aph.gov.au

Dear Sir/Madam,

Submission: Inquiry into Consumer Access to Pharmaceutical Benefits

We write as a representative patient support organisation for a very small group of Australians suffering from a rare and inherited neurodegenerative disease known as Niemann-Pick type C (NPC) and enclose a Disease background flyer as part of this submission.

NPC is a progressive neurovisceral genetic disorder that occurs in 1 in 150,000 births. 80% of affected patients are children, but 20% are adults at the time of diagnosis. NPC predominantly results in neurological difficulties, often initially presenting as problems with coordination and eye movements, and over time affecting walking, swallowing and memory. Children often develop seizures and significant learning problems with illness progression. A significant proportion of adult patients initially develop a major mental illness, and this is often the first sign of the disease. NPC Disease is a terminal disease with no known cure.

Some Quick Disease Facts:

- In an Australian Study, the average age of diagnosis for NPC was 9.3 years (range 0.1 to 37.7) out of 19 diagnoses
- In a US Survey, the average age of death for NPC1 was 16.2 years, with one half of patients dying before the age of 12.5 years.

Symptoms:

Symptoms vary with age of onset and may include:

- Jaundice at (or shortly after) birth
- An enlarged spleen and/or liver (hepatosplenamegaly)
- Difficulty with upward and downward eye movements (vertical supranuclear gaze palsy)
- Unsteadiness of gait, clumsiness, problems in walking (ataxia)
- Difficulty in posturing of limbs (dystonia) leading to contorted limbs making smooth movement difficult
- Slurred, irregular speech (dysarthria) and problems in swallowing
- Learning difficulties and progressive intellectual decline (cognitive dysfunction) leading to memory loss & dementia
- Sudden loss of muscle tone which may lead to falls (cataplexy) in younger children
- Tremors accompanying movement and, in some cases, seizures

NPC Disease is a terminal disease with no known cure, however in February 2010, a treatment breakthrough was approved by the Therapeutic Drugs Administration in Australia for a drug called 'miglustat', which is available under the brand name Zavesca®. Currently this is the only approved treatment available for NPC and is approved for the "Treatment of progressive neurological manifestations in adult and paediatric patients with Niemann-Pick disease Type C".

The difficulty facing patients and their parents is that this treatment is not as yet publicly funded and the cost for treatment of children (and adults) is so prohibitively expensive that it is beyond the reach of almost all NPC sufferers in Australia.

We fully understand that Government and Cabinet will need to consider approval for funding of NPC due to the high cost of treatment, however many serious diseases have differing disease progression pathways, and NPC disease progression demonstrates that any delay in treatment of NPC patients will cause damage that is not reversible once treatment is provided. It is for this reason that Government needs to look at its funding approval process for life threatening diseases, to ensure that any patient damage caused by delays to implementation of disease treatment is as minimal as possible.

Data collected by professionals in both Australia and overseas have strong indications that by treating the patient with NPC with the drug "Miglustat" that progression of the devastating effects of the disease may be prolonged in appearing hence, allowing the patient a better quality and longer life status.

Any delay in obtaining approval for funding with this treatment means that our small number of Australian patients, those patients being our children, are denied their right to life. Progression of the disease has shown patients having a normal day to day diet to vitamised food to eventually peg feeding, to be able to walk independently to being bed ridden or wheel chair bound, to be able to speak, to having no voice, from being socially active to totally unable to socialise appropriately. NPC Disease is a very aggressive disease taking no pity on its victims. Damage caused by the disease is irreversible, and sadly life as it once was can not be regained by administering a drug of any kind. Please take a moment to familiarise yourself with attached photos of Australian patients both past and present dealing with life with NPC Disease and see that they too are everyday people who deserve the right to live their lives to the fullest of their abilities. These faces are our children….. Australian Children, living, lost and loosing.

We look forward to being able to provide certainty of treatment for sufferers of NPC and wherever possible relieve the anxiety and cost burdens on families with children suffering this horrific disease. To this end, we ask the Senate Committee to consider recommendations that provide for timely access to treatment for very rare diseases, where sufferers should have the same right of access to treatment as provided to patients for many other diseases that are more common in Australian society.

We thank you for the opportunity to make this submission and would welcome any further contribution we could make to assist you with your deliberations in this vital area.

Yours sincerely,

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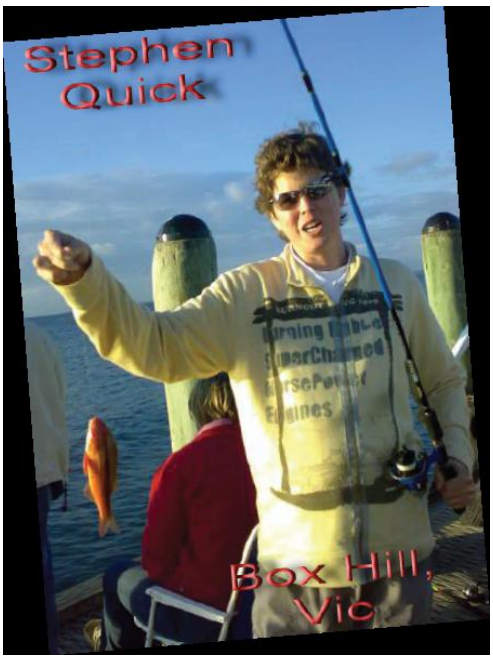

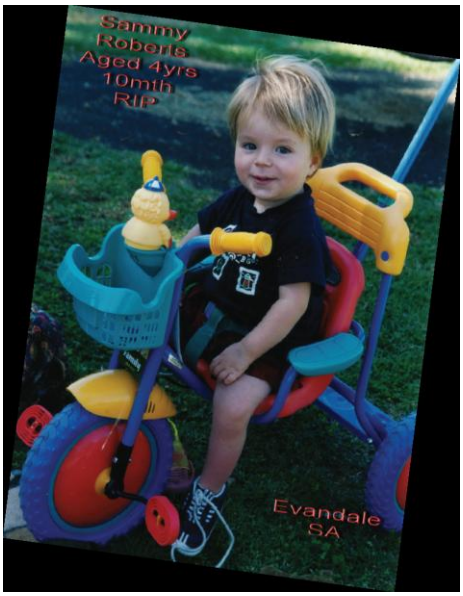

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