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2515

Dear House of Representatives, Standing Committee on Health, Aged Care and Sport.

I am writing to you as a mother of an 8-year-old with a very rare liver disease and as the President of the global charity for this disease, The ***PFIC Network***. PFIC stands for Progressive Familial Intrahepatic Cholestasis. This is a devesting genetic liver disease, the hallmark symptom is an unbearable, incessant, and debilitating itch. The itch comes from the inside, there is no cream or approved medication that can take this itch away. PFIC usually results in end stage liver disease and carries a high risk of death. On top this PFIC children have little quality of life as the itch makes it impossible to sleep for any substantial length of time. The itch is so consuming that it is hard to participate in school and life in general, resulting in a very poor quality of life.

The ***Inquiry into approval processes for new drugs and novel medical technologies in Australia*** is exciting news for Australian PFIC patients. Since my daughter's diagnosis of PFIC at five months old I have advocated and fought for her and all PFIC patients in Australia to have access to the best possible treatments available. I even spoke at Parliament House in 2014 on behalf of Rare Voices Australia, for the need for easier and more efficient access to treatments in rare disease.

Currently there are two highly promising medications about to seek approval for the treatment of PFIC internationally. One with the FDA and the other with the EMA. Both trials are in stage three and both have reported reduction in itch and therefore the slowing of end stage liver disease. There are also several cases now reported where children have such a dramatic improvement in symptoms, they are taken off the liver transplant list.

I would like to refer specially to the fourth term of reference in the inquiry:

Without compromising the assessment of safety, quality, efficacy or cost-effectiveness, whether the approval process for new drugs and novel medical technologies, could be made more efficient, including through greater use of international approval processes, greater alignment of registration and reimbursement processes or post market assessment.

PFIC patients are beyond desperate for approved and safe treatment options. Most children will face liver transplant – which is by no means a cure. In fact, we see PFIC specific complications post-transplant in PFIC type 1 and PFIC type 2**.**

Currently the approval process for novel medications in Australia have multiple and complex approval pathways that mean the reimbursement process for rare disease patients is lengthy and uncertain. As a mother and advocate for PFIC it is heartbreaking to witness lengthy delays in access to life saving treatments due to complicated approval processes.

I have also witnessed cases in Australia where a medication is finally approved for a rare disease but is not listed for PBS making the treatment unattainable due to the exorbitant cost. This is excruciating and one of my greatest fears, that we will fight for approval only to find the medication is inaccessible as it is not listed for PBS. This is due to **rare disease therapies being unable to meet the criteria for subsidy under current Pharmaceutical Benefits Advisory Committee (PBAC)/Medical Services Advisory Committee (MSAC) pathways as they were designed for the evaluation of common disease therapies.**

To solve these issues, we need to **enable all Australians to have equitable access to the best available health technology**. Unneeded delays and denying access to those with rare disease over more common conditions needs to change, every Australian life matters! Living with a rare disease is difficult enough, without having to deal with unneeded and complicated approval processes to access effective treatments.

As a solution the PFIC Network recommends the government **develop policy that supports people living with a rare disease to have timely and equitable access to new and emerging health technologies.** To implement this change the PFIC Network would like to highlight the need to;

- **Broaden the description and understanding of the principles underpinning Australian HTA processes to acknowledge the challenges associated with assessing health technologies for rare diseases.**

- **Build rare disease expertise within the Office of Health Technology Assessment (OHTA) that is responsible for analysing potential rare disease impacts.**

If PFIC patients in Australia are to gain access to these new and very promising medications that those in the USA and Europe can hopefully soon enjoy, it is crucial to not only ensure access but also **ensure funding and reimbursement pathways are fit-for-purpose and sustainable for current and new health technologies for rare diseases.** To implement these changes the PFIC Network suggests the following changes be made:

- **Build on the current processes within the OHTA to ensure all rare diseases submissions are flagged as complex and may require additional scoping and engagement to address potential challenges and uncertainties.**
- **Raise awareness among industry and rare disease organisations as to the availability of the HTA Access Point.**
- **Ensure rare disease expertise exists, or can be accessed, on all reimbursement pathways and HTA advisory bodies**.

I would like to now refer to the second term of reference*:*

Incentives to research, develop and commercialise new drugs and novel medical technologies for conditions where there is an unmet need, in particular orphan, personalised drugs and off-patent that could be repurposed and used to treat new conditions.

4-Phenylbutyrate is a medication used off label for PFIC internationally, it has been reported in peer reviewed journals to have great success in some patients with PFIC 2. To access this medication in Australia is not only extremely expensive but can also take an unnecessary length of time to gain approval for off- label access. It seems unthinkable to have a medication that can dramatically improve the quality of life of a PFIC patient that is available and affordable for other conditions in Australian but not accessible for PFIC patients.

This raises two distinct issues:

- **For rare diseases, lack of transparent and equitable pathways for repurposing existing treatments that are reimbursed for more common conditions.**

- **Lack of clear pathways and length of time for rare disease patients to access orphan drugs, new treatments and personalised medicine.**

Time is crucial in treating PFIC! Having unnecessary and complicated pathways to gain access to safe, life changing medication- means a child with PFIC suffers another hour, another day, another week, another month, another year waiting for affordable and accessible treatments.

The PFIC Network recommends implementing measures to:

Ensure people living with a rare disease have equitable access to medicines with demonstrated clinical benefit for a rare disease, including those that are already funded for another condition.

Specifically, we advocate to ensure:

- **Rare disease organisations work with the HTA Consumer Evidence and Engagement Unit to submit an application for public reimbursement of a technology eligible for assessment by the Office of Health Technology Assessment (OHTA).**

- **The Therapeutic Goods Administration (TGA) and OHTA continue to work together to develop clear processes and pathways for sponsors considering submitting applications for the repurposing of medicines already approved for use in treatment of other conditions.**

My beautiful, sparkly, vibrant 8-year daughter deserves access to best treatments available for PFIC, as do all Australian PFIC patients and all those who suffer with a rare disease in Australia.

On behalf the PFIC community in Australia, we thank you for considering our suggested changes.

With much hope,

Melanie Karakaidos
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www.pfic.org
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