Submission No 350

INQUIRY INTO HEALTH OUTCOMES AND ACCESS TO HEALTH AND HOSPITAL SERVICES IN RURAL, REGIONAL AND REMOTE NEW SOUTH WALES

Organisation: Australian Pompe Association

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Portfolio Committee No 2 Health
Parliament House
Macquarie Street
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Australian Pompe Association submission to Portfolio Committee No2 Health inquiry into and report on health outcomes and access to health and hospital services in rural, regional and remote New South Wales

Dear Committee Secretariat,

The Australian Pompe Association would like to thank the inquiry into the health outcomes and access to health and hospital services in rural, regional and remote New South Wales for the opportunity to respond to Terms of Reference a and b.

The Australian Pompe association is deeply concerned at the delays in diagnosis of Pompe disease in Pompe babies born in NSW regional and remote areas.

Pompe is a spectrum disorder which can occur at any age, from birth to the seventh decade. In babies Pompe Disease is relentless and if not treated within days of birth will often result in the baby passing away within the first year. If the incidence of Pompe is to be reduced, families be saved from the tragic impact of the loss of a child, it is vital that the disease is diagnosed within days of birth and treatment started without delay.

Pompe disease is caused by a complete or partial deficiency of the enzyme which is responsible for breaking down excess glycogen which accumulates in the muscles. If untreated the excess glycogen continues to accumulate in the muscle cells and results in progressive muscle damage, severe muscle weakness, increasing disability and premature death.

The treatment for Pompe Disease is funded under the Federal Government Life Saving drugs program but early diagnosis is critical. Without treatment from birth significant and irreversible damage is done to a young baby, in particular in relation to cardiovascular complications. Overseas evidence has shown that there is a difference in outcome for a Pompe baby who is treated at 21 days of birth compared to 5 days from birth.

Currently for us in Australia diagnosis alone takes about three months, as first the parent must see the baby is not meeting milestones and then get to see a paediatrician who will most probably refer the baby to another specialist by which time three precious months have passed. Unfortunately, it is twice this in regional and remote areas due to the extended periods required to see the succession of specialists required.

In a world where treatments to stabilise the condition are available, and earlier diagnosis can lead to better health outcomes, this is far from where NSW needs to be focussed. With potentially curative genetic treatments for these insidious conditions, currently undergoing trials overseas, NSW needs to regionalise the current system of Newborn screening testing.

The present practice is that all Newborn Screening heal prick tests taken from babies in remote, regional and metropolitan NSW are transported to Sydney and tested at the Children's Hospital at Westmead. This causes a delay of some 2-4 days in the transportation and stops NSW from achieving the critical 5-day diagnostic window recommended.

Newborn screening testing should be expanded to the NSW regional centres to ensure that tests are processed within 2 days, either by the government or private sector laboratories. The newborn screening testing laboratory at Westmead Children's is at full capacity, which limits the introduction of new diseases to the program. The transfer of Newborn screening services to the NSW regional centres would provide the much-needed increase in capacity.

The State Government's and Federal Government National Framework for Newborn screening established in May 2018 has not met up to its expectations and still only one disease in one state has been added to the newborn screening list in 18 years.

Twenty years ago, Australia lead the world in the number of diseases we proactively sought to diagnose in our newborns, as a way to ensure the best possible chance of care and a healthy life for babies at risk. In 1986 Australia added Cystic Fibrosis again a major step forward for these vulnerable children. However, in 2020, Australia only screens for 35 – 36 conditions, compared to over 60 in California. 23 USA states having already approved screening for Pompe.

Many rare diseases, especially Pompe, rely on early diagnosis to maximise the effect of treatment and reverse the damage done by the disease which is obvious at birth. Without neonatal testing in Victoria alone, three babies have been lost in the last 20 months to Pompe because the disease was not diagnosed fast enough for treatment to be initiated or was started far too late.

Pompe remains in both babies and adults a difficult disease to diagnose, once the baby has left the protection of the hospital. Once regional testing is established it would also serve to stop the diagnostic odyssey that many adult-onset patients experience, some taking years to be diagnosed, many losing years of possible treatment while struggling to find a diagnosis.

We are on the precipice of new treatments and curative genetic techniques that will rely on early diagnosis for the best outcomes for Pompe children and their families. A major expansion of our newborn screening capacity is urgently required.

Summary

Serious inequities exist in remote and regional areas of NSW where the diagnosis of critical rare diseases is being significantly delayed due to the time required to access the series of specialist required. Effective treatments are available and 2nd generation treatments and Gene Therapy cures are also currently undergoing trials overseas. All NSW parents need timely access to diagnosis of rare diseases with a treatment like Pompe.

The national newborn screening program needs to be expanded to the NSW regions to ensure that Pompe babies and other rare disease babies are tested and diagnosed while they are still within the protection of the hospital to ensure that babies are given the best chance of a normal life and families spared the needless loss of a child.

The Australian Pompe Association would once again like to thank the Standing Committee for this opportunity to contribute to the enquiry.

Yours faithfully,

The Australian Pompe Association Committee