AUSTRALIAN POMPE ASSOCIATION

Media Release

Our campaign for Australian Pompe Newborn Screening

Pompe Diseases

Pompe disease is very rare, thought to be 1 in 40,000 live births (Infant onset 1 in 138,000. Juvenile and adult onset 1 in 57,000) Pompe disease is normally split into three types, Infant onset, juvenile onset or adult onset.

They are all dependant on the amount of enzyme naturally produced by the patient. Babies born with Pompe disease may produce very little or no enzyme and if so will experience rapid progressive muscle damage, muscle weakness, Cardiomyopathy and premature death and may die within weeks of birth if they are not given immediate enzyme replacement therapy.

Pompe disease is caused by a shortage of acid alpha-glucosidase whereby the lysosomal degradation of glycogen is hampered and glycogen accumulates inside the lysosomes. The lysosomes become unusually large and fuse to form large lysosomal spaces that eventually occupy a substantial part of the total cellular volume. The ensuing cellular pathology, predominantly manifesting in muscle cells as loss of architecture, ultimately leads to skeletal muscle weakness and clinical symptoms. (Pompe Disease Martina Baethmann UniMed)

Diagnosis and testing

Due to Pompe being so rare very few doctors in Australia have even seen another patient and it may require multiple referrals from doctor to specialists before Pompe can be diagnosed. Testing for Pompe is now relatively simple and can be undertaken in a few days. However, with the number of diseases with very similar symptoms
testing can often be tragically delayed particularly when the patient is outside of the metropolitan areas. The experts in Pompe disease note that the tests used in newborn screening have a potential to produce false positive test results and recommend that GAA DBS assay testing or genetic testing be undertaken to give a final diagnosis.

Incidence and tragedies

In the last 5 years the Australian Pompe's Association is aware of 5 infants passing away, in the first few months of life, having been eventually diagnosed with Pompe. Newborn screening of Pompe would have enabled these babies to have started treatment within days of birth rather than the typical three - six months once symptoms are more apparent.

Why is Pompe different?

Pompe disease is one rare disease that can be treated and the treatment is available subject to approval under the federal government life saving drugs program.This treatment will save the lives of these babies and spare parents and family from unnecessary heartbreak.

With an incidence of 1 in 138,000 and an average birth rate of 308,065 per year (ABS data for 2013) three Pompe babies will be born every year in Australia they and their parents need our help to ensure that they have the best chance of life.

This incidence of birth would also suggest that 10 babies have passed away in Australia in the last 5 years without ever being diagnosed prior to post mortem.

Experience Overseas and reports in Australia

Screening for Pompe is becoming more common with New York State currently screening for Pompe along with 47 other disorders.

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New born screening is also undertaken in Taiwan and a study (pre 2008) of 132,532 babies found 4 babies with Pompe and that due to newborn screening their diagnosis was achieved 3-6 months earlier with a much better outcome for the baby.

A 2012 Queensland report on Lysosomal disorders reviewed the incidence and advantages of new born screening and found significant benefits in testing please see Health Policy Advisory Committee on Technology
link to report below.

As Pompe can manifest at any time and continues to be very hard to diagnose in later life, the Australian Pompe's Association would strongly recommend that the families of babies born with Pompe who are asymptomatic be advised of their Pompe diagnosis for continued monitoring.

Raymond Saich
President
Australian Pompe's Association

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References

Health Policy Advisory Committee on Technology
https://www.health.qld.gov.au/healthpact/docs/briefs/WP115.pdf
Neonatal screening for lysosomal storage disorders

New York State New Born Screening Program
http://www.wadsworth.org/newborn-screening/pompe-disease-gaa

Pediatrics. 2009 Dec;124(6):e1116-25. doi: 10.1542/peds.2008-3667.
Pompe disease in infants: improving the prognosis by newborn screening and early treatment.
http://www.ncbi.nlm.nih.gov/pubmed/18519449

BioMarin
https://www.bmrn.com/pipeline/igf2-gaa-for-pompe-disease.php
The incidence is one in 40,000 births. There are two main forms of Pompe disease: adult onset with an incidence of one in 57,000 births and infantile onset with an incidence of one in 138,000 births

Genzyme
https://www.pompe.com/en/healthcare-professionals/genetics-epidemiology/incidence-prevalence.aspx

Ethnic Distribution
The estimated incidence of 1 in 40,000 reflects a worldwide average. However, several studies suggest that incidence rates may vary rates among populations, and reported estimates range from 1 in 14,000 to 1 in 300,000, depending on geographic area or ethnic group examined.[3]
In infants, the disease appears to be more common among African-Americans and in southern China and Taiwan,[3] while adults with Pompe disease may have a comparatively high incidence in the Netherlands.[1] In addition, some of the specific GAA gene mutations have been identified as more common within certain groups.

Pompe disease diagnosis and management guideline
ACMG Work Group on Management of Pompe Disease:, Priya S. Kishnani, MD,1 Robert D. Steiner
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3110959/
Incidence data are limited with reports ranging from 1 in 14,000 to 1 in 300,000 depending upon ethnicity or the geographic area studied.2 The infantile form has an apparent higher incidence among African-Americans and Chinese2 whereas the late-onset adult form has a higher incidence in The Netherlands.8 In the Netherlands, the incidence of the infantile-onset form is 1/183,000. The combined incidence of all forms of Pompe disease is estimated to be 1:40,000.2,8,9

The Advisory Committee on Heritable Disorders in Newborns and Children
http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/nominatecondition/reviews/pompe.pdf

Study	Design	Subjects	Estimated Prevalence	Comments
Ausems, 1999	Cross-sectional	3,043 anonymous dried blood spots in the Netherlands screened for 3 mutations	Infantile: 1/138,000 (95% CI: 43,169- 1/536,482) Late-onset: 1/57,000 (95% CI: 1/27,734- 1/28,255) Combined: 1/40,000 (95% CI: 17,622-1/100,073)	No clinical correlation. Only three mutations were included. Very wide confidence interval. This study suggests that late-onset Pompe disease is 2 to 3 times more common than infantile-onset disease