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"A $10 screening test could have helped my 'bubble' baby"

International Day of Immunology, April 29 / World Primary Immunodeficiency Week, April 22-29

Severe Combined Immune Deficiency (SCID) is a life-threatening condition where babies are born without a functional immune system. It gained global recognition in the 1970s due to a young boy called David Vetter, known as a 'bubble boy', who lived his short life inside sterile plastic bubbles to avoid infection. Now, a new immunodeficiency strategy for Australia and New Zealand outlines the important work that still needs to be done to address the group of illnesses that SCID belongs to.

Five days before Christmas 2020, at just 18 weeks of age, Isabelle began to turn blue and stopped breathing properly. Fortunately, she was already waiting in the Emergency Room where doctors were able to immediately assist and save her life. Identifying the underlying cause of Severe Combined Immune Deficiency (SCID), a very rare and life-threatening genetic disorder, was also a miracle. Without treatment, SCID babies are not likely to survive past their second birthday due to their inability to fight even simple infections.

"The hospital's general paediatric doctor noticed Isabelle's very low white blood cell count. Miraculously, the doctor had recently read a report on SCID, so the doctor happened to know the symptoms and immediately called the immunology specialist. Otherwise, being so rare, it could easily have not been picked up," says Louise Grant, Isabelle's Mum. "When everyone else was focusing on Isabelle's heart and lungs, these doctors fortunately looked at her immune system too."

Due to her inability to fight infection, Isabelle was immediately isolated in a positive pressure room, spent 33 days in paediatric ICU recovering from the severe lung infection she had developed, and was then in and out of hospital for the first year of her life after receiving a Stem Cell Transplant from her dad as the donor. In the absence of routine newborn screening that is available in other countries, Isabelle will now have to live with lifelong secondary consequences of SCID, such as permanent hearing loss and developmental disabilities, due to the delay in diagnosis and treatment.

"I estimate that the cost to the health system of Isabelle's medical treatment in those first few months alone would easily go into the millions. Her life-long disabilities even more. That does not even touch on the emotional cost to Isabelle and our family," says Louise. "It is incredibly frustrating that Isabelle's condition is entirely treatable if caught very early and could have been picked up on the newborn screening test which she had done at birth, as part of the Government's routine newborn screening program. It would cost less than $10 per baby to add SCID to this program."

"While Isabelle is not in a physical bubble, for the past year our whole family has been living in isolation at home to protect her from infection, rigorously disinfecting everything, until her 'new' immune system develops," explains Louise. "The impact on our lives is all consuming."

New national strategy for Primary Immunodeficiencies (PIDs)
The Australasian Society for Clinical Allergy and Immunology (ASCIA) has launched the ASCIA Immunodeficiency Strategy to improve the health and wellbeing of people living with primary immunodeficiencies and minimise the burden on individuals, carers, health services and the community. SCID is the most serious form of primary immunodeficiency disorders, a

diverse group of more than 400 potentially serious, chronic illnesses that can lead to frequent or severe infections, swellings and autoimmune problems.

Two key issues outlined in the strategy are the importance of **early diagnosis for all primary immunodeficiencies** and **newborn screening specifically for SCID**.

“Due to their rarity, delays in diagnosis of primary immunodeficiencies are common. For infants and very young children with severe PIDs, this leads to severe complications due to recurrent infections and early death, despite being curable if treated in the first few months of life,” says Dr Melanie Wong, Co-Chair of the ASCIA Immunodeficiency Strategy. “That is why we are specifically calling for newborn screening of SCID. While screening is routinely performed in New Zealand, the United States, and in some European countries, it is not yet routinely available in Australia. It is only being trialled in NSW at present.”

“SCID is fatal in the first two years of life without definitive intervention. Early diagnosis is vital to allow curative treatment such as urgent haematopoietic stem cell transplantation (HSCT), also known as bone marrow transplant (BMT),” says Dr Theresa Cole, ASCIA President Elect and ASCIA Immunodeficiency Committee Chair. “Screening is also likely to be more cost-effective for the health system than the cost of prolonged hospital and intensive care unit admissions. It should be a health priority in Australia.”

While individual PIDs are rare, taken together the overall prevalence is estimated to be 1 in 25,000 people[i]. It is estimated that 70-90% of people are still undiagnosed worldwide[ii]. PIDs affect adults as well as children, with the majority occurring in adult life. This can have a serious impact on vital organs and long-term health. Australian (Victorian) data shows that in adults the average delay from symptom onset to diagnosis is 8 years and every year of diagnostic delay costs years in life expectancy[i].

30-year-old Hayley Teasdale has common variable immunodeficiency, which is an umbrella term for a broad spectrum of PIDs. The onset of her symptoms began in her early teens, but she was not diagnosed until she was 24, which has led to chronic health issues. Hayley is now pregnant with her first child and supports improved access to tests to diagnosis for PIDs, including genomic testing.

“I kept getting repeat infections, I was always sick. It started to wear down my body to the point that I kept fainting and could not leave the house or go to university. Living in a small rural town, it went undiagnosed. It was only when I moved to the city that I was referred to an immunologist,” said Hayley. “Like most people, I don’t know if my child will be predisposed to SCID. I would love to be able to get a SCID newborn screening done for peace of mind.”

For older children and adults where curative treatment is not possible, the recent expansion of genomic technologies has the potential to transform care, informing early diagnosis and the delivery of precision medicine.

“Correct diagnosis will lead to appropriate treatment, including immunoglobulin replacement therapy (IRT), improving quality and length of life. This requires support from expert multi-disciplinary teams comprising of specialist medical, nursing and allied health professionals. With targeted resources, patients with PID can be spared unnecessary interventions, and instead utilise available medical treatments to maximise their opportunities to lead healthy and productive lives,” says Prof Jo Douglass, Co-Chair of the ASCIA Immunodeficiency Strategy.

The ASCIA Immunodeficiency Strategy also addresses the need for:

- Improved education for health professionals to recognise early warning signs of PID;
- Improved access to paediatric and adult clinical immunology/allergy specialists;
- Equitable access to funded and accredited genomic and immune testing for PID;
- Treatments to be available in rural, remote and regional centres, as well as urban areas.

The ASCIA Immunodeficiency Strategy was developed in collaboration with patient organisations, IDFA, IDFNZ, AusPIPs and HAE Australasia, and is available at www.nationalimmunodeficiencystrategy.org.au

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NOTES FOR MEDIA:

Available for interview:

- Prof Michaela Lucas (WA) - ASCIA President
- Dr Theresa Cole (VIC) - ASCIA President Elect and ASCIA Immunodeficiency Committee Chair
- Dr Melanie Wong (NSW) - Co-Chair of the ASCIA Immunodeficiency Strategy and past ASCIA President
- Prof Jo Douglass (VIC) - Co-Chair of the ASCIA Immunodeficiency Strategy and past ASCIA President
- Prof Connie Katelaris AM (NSW) – Chair, ASCIA HAE Working Party and Drug Allergy Committee and past ASCIA President
- Case studies upon request.

The Australasian Society of Clinical Immunology and Allergy (ASCIA)
The Australasian Society of Clinical Immunology and Allergy (ASCIA) is the peak professional body of clinical immunology and allergy in Australia and New Zealand. Established in 1990, ASCIA is a world leading, innovative and active professional society with strong leadership and sustainable operations.

ASCIA's purpose is to advance the science and practice of allergy and clinical immunology, by promoting the highest standard of medical practice, training, education and research, to improve the health and quality of life of people with immune system disorders, including allergy and primary immunodeficiency (PID).

ASCIA is committed to providing high quality training, education and research to improve the health and wellbeing of all people in Australia and New Zealand with PID.

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[i] Slade et al, Delayed Diagnosis and complications of Predominantly antibody Deficiencies in a cohort of Australian adults, 2018

[ii] http://www.worldpiweek.org/what-is-primary-immunodeficiency-pi/