The Hon. Greg Hunt MP

Minister for Health

MEDIA RELEASE

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$3 million for rare childhood brain disorders

The Liberal National Government is providing $3 million to close the loop from genetic diagnosis to clinical treatment for rare childhood brain disorders known as Leukodystrophies.

The research is being led by a consortium headed by Associate Professor Richard Leventer from the Melbourne Royal Children's Hospital and Professor Ernst Wolvetang from the University of Queensland.

Professor Leventer was part of the team that discovered Massimo Damiani's rare variant of Leukodystrophy.

Baby Massimo was diagnosed in 2009 with a variant of Leukodystrophy – a rare genetic condition characterised by degeneration of the white matter (myelin) of the brain.

In a world-first, an Australian-led research team, pioneering a new "trio whole genome" sequencing approach were able to establish that Massimo's condition was caused by mutations in a gene not previously associated with disease.

Today will be the one-year anniversary of Massimo's passing, who died at the age of nine.

Following Massimo's death, his parents Stephen and Sally have continued their efforts, determined to honour Massimo's legacy and complete the mission that bears his name.

In his honour, I am delighted to be able to announce our Government will provide $3 million in funding over the next three years for modern genomics with the hope of discovering novel treatments for rare childhood brain disorders, known as Leukodystrophies.

This will allow researchers to understand disease mechanisms faster and test potential therapies in potentially life-changing timeframes.

This research begins with patient recruitment for genomic diagnosis, moving to disease modelling and pre-clinical testing then full circle back to the patient with the promise of human clinical trials for novel treatments.

Leukodystrophies affect the central nervous system and causes loss of normal brain functions. This leads to progressive and severe intellectual and physical disabilities.

There are currently no cures for most Leukodystrophies, life expectancy is typically months to several years at best, and quality of life can be extremely poor.

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Until recently about half of cases presenting with these debilitating conditions remained unsolved.

This funding will support research into faster genomic diagnostics for both known and unresolved cases of Leukodystrophies, and develop adaptable, repeatable and scalable disease modelling and translational capabilities for the clinical treatment of rare childhood brain disorders.

Building on the success of the approach to diagnose Massimo's Leukodystrophy, the research aims reduce the proportion of undiagnosed Leukodystrophies to less than 10 per cent by 2020 by applying state of the art genomic technologies to provide more diagnoses for Australian children with Leukodystrophies.

A combination of patient derived stem cells and matched mouse models, will allow researchers to replicate these diseases enabling new compounds and gene therapies to be tested before they are given to the patient.

These approaches hold great promise for reducing the severity of the diseases and perhaps even curing them in the future.

It aims to reduce from decades to months the time it takes to progress from disease discovery to potential clinical treatment.

The key used to unlock the genetic cause of Massimo's mysterious illness was genomics.

Our Government recognises the power and potential of genomics and is investing $500 million over 10 years in an Australian Genomics Health Futures Mission to help save or transform the lives of Australians through research into better testing, diagnosis and treatment through genomics.

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