Cost of raising a child with rare genetic disorder significant but could be reduced by earlier diagnosis

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The cost of raising a child with a rare genetic disorder was significant but the economic burden could be reduced by earlier diagnosis and

targeted treatment, a new study has found.

The research, led by the Murdoch Children's Research Institute (MCRI), supports the addition of Fragile X syndrome and chromosome 15 imprinting disorders including Angelman, Prader-Willi and Chromosome 15q duplication syndromes to the newborn bloodspot screening test (heel prick test).

The four rare disorders studied are characterized by varying degrees of intellectual disability, autism and behavioral problems. About 135 people are born with one of these syndromes each year in Australia.

MCRI researcher Dr. Emma Baker said the financial burden of caring for a child with one of these rare conditions was substantial with costs disbursed across out of home care, special education, equipment support, medications and visits to health professionals and hospitals.

But Dr. Baker said higher levels of intellectual functioning were associated with lower yearly costs, suggesting that the economic impact could be reduced by earlier diagnosis and targeted interventions.

The study, published in the *Journal of Autism and Developmental Disorders*, involved 108 Australian families recruited from disorder specific support groups and organizations.

Angelman syndrome incurred the highest yearly costs per person at $96,994, followed by Prader-Willi syndrome ($57,576), Chromosome 15q duplication syndrome ($52,130) and Fragile X syndrome ($33,221). The research found that for every point increase in child's intellectual functioning the total yearly costs were reduced by $734. Many with these syndromes are more than 30 points below the IQ of what is typically seen in developing children.

"Many infants with these syndromes are not diagnosed within the first year of life," Dr. Baker said. "For Prader-Willi syndrome, diagnosis in infancy allows for early initiation of growth hormone treatment, which has shown to improve intellectual functioning when started in the first 12 months of life, while early targeted interventions such as speech, physical, and behavioral therapies are reliant on early diagnosis."

Dr. Baker said the long-term care of patients with these syndromes also put pressure on the health sector.

"Severe seizures occur in more than 80 percent of people with Angelman syndrome and Chromosome 15q duplication syndrome and they require ongoing monitoring and treatment throughout life," she said.

"For Prader-Willi syndrome, being overweight and obese is reported in 40 percent of children and adolescents, and in up to 98 percent of adults, which significantly increases the risk of chronic conditions if not prevented."

MCRI Associate Professor David Godler said early diagnoses for these syndromes was now possible using a specialized screening method called Methylation Specific-Quantitative Melt Analysis recently developed by MCRI researchers.

The one-step test can be used to screen for the four syndromes simultaneously by looking at the number of chemical modifications or marks called methylation added to affected genes, which are not present at such high or low levels in children without these disorders.

Associate Professor David Godler said given the huge costs associated with these conditions and potential savings to Governments and affected families if diagnosis was made in the first year of life, the research findings support the test being considered for addition in the newborn

bloodspot screening program.

Anu Madan's daughter, Arika, 2, was diagnosed with Angelman syndrome, at eight months of age.

To help with Arika's condition she receives speech, hydro, occupational and physio therapies, takes anti-seizure medication, and sees a team of specialists.

Ms Madan said the economic cost of raising a child with a genetic disorder had also taken a huge toll on the family.

"The NDIS covers a lot of the expenses but not everything," she said. "The medical equipment needed isn't cheap so we try to source second-hand equipment at markets where we can. On top of that we have the cost of all the specialist appointments, which easily mount up."

Ms Madan wants Angelman syndrome to be included on the newborn bloodspot screening program to spare other families the prospect of a late diagnosis and delayed treatment.

"If we had been better prepared and known what to expect things would be different," she said. We waited eight months for a diagnosis and then faced huge wait times to see specialists.

"If this disorder had been diagnosed at birth she would have received support so much sooner and her motor and cognitive skills wouldn't be as poor."

Researchers from The Royal Children's Hospital, the University of Melbourne, La Trobe University, University of Technology Sydney, Foundation for Angelman Syndrome Therapeutics, Prader Willi Syndrome Australia, Genetics of Learning Disability Service, Genetic

Services of Western Australia and Dup15q Australia also contributed to the findings.

More information: Emma K. Baker et al, The Cost of Raising Individuals with Fragile X or Chromosome 15 Imprinting Disorders in Australia, *Journal of Autism and Developmental Disorders* (2021). DOI: 10.1007/s10803-021-05193-4

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