

Message from the Women's & Children's Hospital Foundation:

Every year, one in seven babies begins life with seizures, developmental delays, or even the risk of early death because of a fatal change in their genes that disrupts brain development. Despite major advances in identifying the genes involved, most of these children still face a lifetime without effective treatments.

A breakthrough genetic technology called antisense oligonucleotides (ASOs) is transforming the way we treat previously untreatable genetic conditions. Already shown to be safe and effective in other diseases, ASOs have the potential to change the future for children living with rare and devastating brain disorders.

At present, children with conditions like PCDH19 clustering epilepsy (a severe form of epilepsy that mainly affects girls) and TIMMDC1 neurodegenerative disorder (a rare and life-limiting disease) have few, if any, treatment options. Families are left with uncertainty, distress, and a lack of therapies that can truly change their child's outcome.

The Foundation is incredibly proud to support Professors Jozef Gecz and Paul Thomas and their multidisciplinary team (Drs N. Smith, R. Sharma, L. Jolly, S.M. Tasheva & S. Montazaribarforoushi), who are working to change the future for children with devastating genetic brain disorders.

Their research will not only create real treatment options for children living with PCDH19 clustering epilepsy and TIMMDC1 disorder, but also to open the door for therapies that could transform the lives of countless others by:

- ***Develop and test*** new ASO therapies in models that reflect how these diseases affect children.
- ***Identify safe and effective doses***, giving children the best possible chance of benefit.

Build a treatment pipeline that can be applied to many other rare childhood brain disorders in years to come.