
Research Focus Areas:

Ring chromosome 14 syndrome is a rare chromosomal abnormality where the 14th chromosome forms a ring like structure r(14). The disorder is characterized by early onset refractory epilepsy, intellectual disability, autism spectrum disorder and a number of diverse health issues. There is a heavy health burden associated with Ring14 which affects the whole the family.

One \$67,897 pilot grant is available and will be awarded to research that has the potential to lead to better understanding and better treatments to improve the quality of life for those affected by Ring Chromosome 14 and related disorders. Potential topics of interest may include but are not limited to: open source resource development (cell lines – in particular, direct differentiated neuronal cell lines as iPSCs are unstable for ring chromosomes, assays, functional studies, etc.), basic science to understand disease mechanisms relevant to Ring Chromosome 14, clinical studies to better define the national history, and translational studies. Preference may be given to novel concepts and collaborative/team approaches.