Presenting Author Name
Danielle Pascual

Presenting Author Category
PhD Student

Research Category
Basic Science

Abstract Title
Determining the Role of IRF2BPL in Neurological Disease

Background
De novo truncating variants in the gene IRF2BPL cause severe childhood-onset ataxia termed NEDAMSS (Neurodevelopmental disorder with abnormal movements, loss of speech and seizures). Additionally, IRF2BPL missense variants are associated with autism spectrum disorder. IRF2BPL is important for nervous system development and maintenance, but its function remains unclear.

Objective
Objective: We have generated the first Irf2bpl knockout mice and aim to characterize the behavioural outcomes and brain pathology in these mice.

Methods
We generated the Irf2bpl null allele by removal of the majority of the single exon (Δ17-651). We performed heterozygous crosses to assess viability, mass, and motor function by vertical pole test and inverted grid test across littermates of both sexes.

Results
We observed that Irf2bpl KO mice are born at lower Mendelian ratios. Although WT and HET mice did not have a significant difference in mass, the KO mice are significantly runted. Lastly, three-month-old Irf2bpl KO mice display motor defects on the vertical pole test and inverted grid test. KO mice brains are also smaller in size and mass than WT mice at 3 months of age.

Conclusion
We have generated preliminary data on Irf2bpl mice. Our data shows that the Irf2bpl KO mice display motor defects as early adults which may model NEDAMSS which may act as a preclinical model to develop therapeutics for this devastating disorder.

Authors

Name	Role	Profession
Danielle Pascual	Presenting Author	Graduate
Yina Her	Co Author	Graduate
Paul Houston	Co Author	Graduate
Robert Beattie	Co Author	Assistant Professor
Paul Marcogliese	Co Author	Assistant Professor