
Research Focus Areas:

Dup15q Syndrome is a clinically identifiable neurodevelopmental disorder that occurs when a key portion of chromosome 15 is duplicated at the 12.1-13.2 region. The syndrome, a leading genetic causes of autism, also leads to hypotonia, GI issues, epilepsy, motor and speech delays and intellectual disability. There are currently no approved therapeutics targeted at Dup15q syndrome despite a significant unmet medical need. We are soliciting applications for **one \$47,158 grant** that supports work towards a potential therapeutic.

While we are keeping the opportunity broad in scope, preference will be given to awards that focus on three areas:

- A formalized Natural History study that will support clinical trials and the creation of key outcome measures that can be leveraged in clinical trial design
- Model systems that explore any aspect of the pathophysiology of the syndrome to include, but not limited to, identification/confirmation of the role of the duplicated genes, genotype/phenotype correlation, biomarkers
- Exploration of novel approaches to therapeutic development; ASOs, siRNA, small molecule inhibitors of the enzyme, PROTACs