DDX3X FOUNDATION

1000 N. West Street, Suite 900, Wilmington, DE 19801
ddx3x.org

DDX3X Syndrome

- DDX3X Syndrome is a rare disease caused by a mutation within the DDX3X gene. The mutation can be spontaneous meaning there is no known cause and happens randomly or inherited meaning the mutation was passed on to the child by a parent.
- The syndrome was discovered in the United States in 2014 and primarily affects girls due to its location on the X-chromosome.
- Not all individuals with DDX3X Syndrome are affected in the same ways with some cases being much more severe than others
- Common symptoms include intellectual disability, developmental delays, low muscle tone/hypotonia, difficulty with speech, epilepsy/seizures, movement disorders, abnormalities of the brain, and microcephaly.
- In order to be diagnosed, patients undergo a comprehensive blood test called Whole Exome Sequencing (WES) which looks at 20,000 genes in an individual.
- Doctors prescribe the test when they suspect there is a genetic cause for symptoms, such as missing milestones and being developmentally delayed.
- Scientists estimate that DDX3X Syndrome accounts for 1-3% of intellectual disabilities in females
- Parents of children with DDX3X Syndrome describe their children as joyful and often love water, music, and animals
- 1,013 known cases across 56 countries
- 973 Cases are female; 40 Cases are male

The DDX3X Foundation

- The first patient in the United States was diagnosed in November of 2014. The DDX3X Foundation was formed in 2015 by two moms.

- Our mission is to connect families, resources, and the medical community to advance research for a treatment or cure for DDX3X Syndrome. Our ultimate goal is to accelerate brain function in individuals affected by DDX3X Syndrome through advances in cell and gene therapy and pharmaceuticals.

- The Foundation has funded over $430,000 in research grants, raised over $1.7M, and hosted 8 Scientific Conferences.

- Our approach to finding treatments is multidisciplinary and includes research tools (animal models and iPSC lines), clinical validation (natural history study, biomarker study, nerve cell search study), and therapeutic focus (gene therapy and drug repurposing).

- Make a donation at: www.ddx3x.org/donate