Jordan's Syndrome

Jordan's Guardian Angels is a global nonprofit funding and facilitating research into a group of genetic mutations known as Jordan Syndrome. Jordan's Guardian Angels was founded by Cynthia and Joe Lang of Sacramento, CA, in 2015 after the diagnosis of their daughter, Jordan. Since its inception Jordan's Guardian Angels has partnered with nine research institutions including Columbia University, Seattle Children's Hospital, and UC Davis to find a treatment or a cure for Jordan's Syndrome.

Jordan Syndrome is a PPP2A related neurodevelopmental disorder on the genes PPP2R5D, PPP2R5C, and PPP2R1A, which is characterized by mild to profound neurodevelopmental delay. Affected individuals may also experience symptoms such as hypotonia (low muscle tone), macrocephaly (enlarged head size), speech impairment, developmental delay in gross motor skills, and prominent foreheads. Other common clinical signs include autism spectrum disorder, seizures, and ophthalmological abnormalities. There is wide variability in how severely individuals may be affected.

Dozens of individuals with Jordan's Syndrome have been reported in the literature to date, ranging in age from 22 months to 53 years old. A total of 293 cases have been identified in over 35 countries. It is estimated that over 250,000 individuals worldwide are affected but remain undiagnosed.

To diagnose Jordan's Syndrome, or identification of pathogenic variants in the genes PPP2R5D, PPP2R1A, and PPP2R5C, individuals must undergo molecular genetic testing. Most commonly patients are diagnosed through Whole Exome Sequencing. Whole Exome Sequencing is an expensive test not always covered by insurance, limiting access to diagnosis. Most Jordan's Syndrome cases to date are due to de novo genetic changes, meaning genetic changes that were not inherited from a parent. For parents of an infected individual the recurrence risk for future pregnancies is estimated at 1%.

After consultation with a clinical geneticist and or genetic counselor, the following management recommendations may be appropriate: referral for early intervention programs or evaluation for an individualized education plan, physical therapy for mobility, occupational therapy for fine motor skills, feeding therapy, alternative means of communication for individuals with expressive language difficulties, therapies for social or behavioral concerns, and continued consultation with a developmental pediatrician.

Jordan's Guardian Angels joins advocates in the rare disease community through events such as Rare Disease Week on Capitol Hill to advocate for legislation that would benefit the rare community at large. Jordan's Guardian Angels has joined the Epilepsy Foundation, The Rare Epilepsy Network, and a coalition of other organizations in support of Seizure Safe Schools Legislation (**H.B. 606** Ohio State Legislature), as well as the EveryLife Foundation and Rare Disease Legislative Advocates in support of the following bills:

The Access to Genetic Counselor Services Act (**H.R. 2144 /S. 1450**)
The BENEFIT Act (**H.R. 4472 /S .373**)
The Newborn Screening Saves Lives Reauthorization Act of 2021 (**H.R. 482 /S. 350**)
The Speeding Therapy Access Today Act 2021 (**H.R. 1730 /S. 670**)

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