This guidebook was developed by the Jordan's Guardian Angels team to help you learn important information about individuals with Jordan's Syndrome and how to best support them. The content found within is meant for informational and educational purposes only and does not substitute professional medical advice or consultations with healthcare professionals.

REFERENCES
Reports from the online community: PPP2R5D-Related Neurodevelopmental Disorder - GeneReviews® - NCBI Bookshelf (nih.gov)

DEFINITION
Jordan's Syndrome is a neurodevelopmental disorder caused by variants, or mutations, in genes that encode parts of a protein complex called Protein Phosphatase 2A (PP2A). These genes include PPP2R5D, PPP2R5C, and PPP2R1A. This document will focus on the PPP2R5D gene mutation.

DIAGNOSIS
Jordan's Syndrome is diagnosed through genetic testing, more commonly through Whole Exome Sequencing.

INHERITANCE
Jordan's Syndrome is typically de novo, aka non inherited.

FAMILY PLANNING
When Jordan's Syndrome is de novo, the recurrence risk to siblings is estimated to be less than 1%

Individuals with Jordan's Syndrome have a 50% chance of passing it on to their children.

COMMON FEATURES
The impact of Jordan's Syndrome varies in occurrence and severity between individuals. The table on the following page is a summary of the main symptoms that have been reported by the Jordan's Syndrome community. Please note these symptoms might or might not be present for everyone with the diagnosis.
## COMMON FEATURES OF JORDAN’S SYNDROME

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Potential Impact</th>
<th>Recommended Evaluation</th>
<th>Recommended Intervention</th>
</tr>
</thead>
<tbody>
<tr>
<td>Macrocephaly</td>
<td>Larger head size</td>
<td>MRI if needed</td>
<td>None</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>Trouble with: • Mobility • Speech • Low endurance due to reduced muscle tone</td>
<td>Physical Therapy</td>
<td>• Physical Therapy • Orthotics</td>
</tr>
<tr>
<td>Speech Delays</td>
<td>Receptive language exceeds expressive language</td>
<td>Speech Therapy</td>
<td></td>
</tr>
<tr>
<td>Vision Difficulties</td>
<td>Varying challenges including: • Nystagmus • Astigmatism • Amblyopia</td>
<td>Ophthalmology</td>
<td>• Glasses • Orthoptics • Vision Therapy (as applicable)</td>
</tr>
<tr>
<td>Feeding Challenges</td>
<td>Trouble chewing and swallowing</td>
<td>Speech Therapy or Occupational Therapy</td>
<td>• Feeding Therapy • Possible need for a feeding tube</td>
</tr>
<tr>
<td>Gastrointestinal Challenges</td>
<td>Constipation and/or diarrhea</td>
<td>Gastrointestinal Specialist</td>
<td>Specialized Diet (as applicable)</td>
</tr>
<tr>
<td>Seizures</td>
<td>Varying types</td>
<td>Neurology (EEG)</td>
<td>Treatment and Diet changes (as applicable)</td>
</tr>
<tr>
<td>Global Developmental Delay</td>
<td>Delay in achieving developmental milestones</td>
<td>Developmental Pediatrics</td>
<td>Regular and frequent therapies</td>
</tr>
<tr>
<td>Sleep Challenges</td>
<td>Trouble falling asleep or staying asleep</td>
<td>• Neurology (EEG) • Ear Nose Throat Specialist (sleep study)</td>
<td>• Increased day time activity • Treatment</td>
</tr>
<tr>
<td>Learning Disabilities</td>
<td>Challenges at school</td>
<td>Special Education Team</td>
<td>• Special accommodations • Hypotonia and communication support</td>
</tr>
<tr>
<td>Challenging Behaviors</td>
<td>•Mood swings • Self-injurious</td>
<td>Psychologist and Psychiatrist</td>
<td>• Environmental supports • Treatment</td>
</tr>
<tr>
<td>Chronic pain</td>
<td>Muscle and joint aches</td>
<td>Primary Physician and Physical Therapist</td>
<td>Physical therapy</td>
</tr>
<tr>
<td>Oral Structural Differences</td>
<td>• High vaulted hard palate • Narrow jaw • More frequent tooth decay</td>
<td>Dentist or Orthodontics</td>
<td>• Dental treatments • Potential orthodontics treatment</td>
</tr>
<tr>
<td>Spine Structural Differences</td>
<td>Scoliosis</td>
<td>• Orthopedist • Spinal Specialist</td>
<td>• Brace • Physical Therapy • Treatment</td>
</tr>
<tr>
<td>Heart Condition</td>
<td>Various Challenges</td>
<td>Cardiologist (ECG)</td>
<td>Regular cardiologist monitoring</td>
</tr>
<tr>
<td>Sensory Challenges</td>
<td>• Autism • Sensory Processing Disorder</td>
<td>• Psychologist • Primary Physician • Developmental Pediatrician</td>
<td>• Treatment • Occupational Therapy • Physical Therapy</td>
</tr>
</tbody>
</table>

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**www.jordansguardianangels.org**

**@JordansGuardianAngels**

**@JordansGAngels**

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RECOMMENDED INTERVENTIONS FREQUENCY

Maximizing on therapies especially through early development has been proven beneficial.

Ten to twelve (10-12) hours of therapies a week have been recommended. Families are encouraged to access therapies without sacrificing their mental health and the quality of life of the entire family. We recommend that you connect with local disability groups to get a better understanding of available resources.

DEVELOPMENT DELAYS MANAGEMENT

Per Gene Reviews Article: PPP2R5D-Related Neurodevelopmental Disorder - GeneReviews® - NCBI Bookshelf (nih.gov)

Consultation with a developmental pediatrician is recommended to ensure the involvement of appropriate community, state, and educational agencies and to support parents in maximizing quality of life.

The following information represents typical management recommendations for individuals with developmental delay in the United States; standard recommendations may vary from country to country.

Ages 0-3 years: Referral to an early intervention program is recommended for access to occupational, physical, speech, and feeding therapy. In the US, early intervention is a federally funded program available in all states.

Ages 3-5 years: In the US, developmental preschool through the local public school district is recommended. Before placement, an evaluation is made to determine needed services and therapies and an individualized education plan (IEP) is developed.

Ages 5-21 years:

- In the US, an IEP based on the individual’s level of function should be by the local public school district. Affected children are permitted to remain in the public school district until age 21.
- Discussion of transition plans including financial, vocation/employment, and medical arrangements should begin at age 12 years. Developmental pediatricians can provide assistance with transition to adulthood.

Early Intervention Contact Information by State | CDC
State Waivers List | Medicaid

SPECIAL ACCOMMODATIONS

Communication: Speech delays are common across individuals with Jordan’s Syndrome. Alternative ways of communicating such as sign language, pictures, and a communication device are recommended to support general communication and promote speech development.

Assistive Technology: The field of assistive technology has come a long way and can expand abilities. Some examples include communication devices, communication buttons, speech to text, typing, and read/write programs.

Motor Support: While most individuals with PPP2R5D gene mutation are able to walk, support such as a stander or a walker might be required until that milestone is reached. Stamina and endurance might continue to be an issue. Some families use adaptive strollers or wheelchairs when a lot of walking is required such as an outing to the zoo.

Special Diet: Some families have been successful in improving seizure activity through the Keto diet. Others have reported improved overall health and cognition through the gluten free and dairy free diet especially when food sensitivities were detected.
ALTERNATIVE THERAPIES
These are interventions that our families have tried and recommended.

<table>
<thead>
<tr>
<th>For Motor Skills</th>
<th>For Speech Development</th>
<th>For Brain Development</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td>Horse Riding Therapy</td>
<td>Listening Therapy</td>
<td>Book: Neuroplasticity books by Norman Doidge</td>
<td>Reiki (energy)</td>
</tr>
<tr>
<td>Aqua Therapy</td>
<td>Prompt- oral motor</td>
<td>Neurofeedback Therapy</td>
<td>Applied Behavioral Analysis (ABA) Therapy</td>
</tr>
<tr>
<td>Terasuit (Intensive PT)</td>
<td>Z vibe vibrating device- oral motor</td>
<td>Anat Daniel Method (ABM)</td>
<td>Metha casting (scoliosis)</td>
</tr>
<tr>
<td>MNRI- reflex integration</td>
<td>Class/Book: More Than Words</td>
<td>Hyperbaric Oxygen Chamber</td>
<td>Chiropractor</td>
</tr>
<tr>
<td>Conductive Education</td>
<td>Gemini Software</td>
<td>Voita Therapy</td>
<td>Acupressure/Acupuncture/Brushing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Music and Art Therapy</td>
</tr>
</tbody>
</table>

RELEVANT RARE REALITY PODCAST EPISODES
A Rare Reality is a podcast by Jordan’s Guardian Angels with varying bi-weekly topics. The following episodes contain important information about Jordan’s Syndrome and how to support our children with their disabilities. These podcasts can also be found on JGA’s YouTube channel with translation enabled.

A Rare Reality – Jordan’s Guardian Angels (jordansguardianangels.org)
Season 1, Episode 1: Living with Jordan’s Syndrome
Season 1, Episode 6: Bringing Speech and Language to Life

AMBASSADOR PROGRAM
To best support our global community, Jordan’s Guardian Angels Ambassadors span across regions and languages including Spanish, French, German and Italian. You can connect with your local community and access local resources as well as participate in virtual and in person regional events. Email connect@jordansguardianangels.org to learn more.

USEFUL RESOURCES

Jordan’s Syndrome Children’s Book
Disponible en Español

New Families Welcome Letter

Medical Professionals Kit

These resources and more can be found at: jordansguardianangels.org/resource-center

Website: jordansguardianangels.com
Facebook: facebook.com/JordansGuardianAngels
Instagram: instagram.com/JordansGuardianAngels
Twitter: twitter.com/JordansGAngels

YouTube: youtube.com/channel/UCaFqEJ_N-EcBREVI801uGXg/featured
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