

# JORDAN'S SYNDROME

## PPP2R5D CARE BEFORE THE CURE GUIDEBOOK



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.

## LIVING WITH JORDAN'S SYNDROME

### INDIVIDUALS WITH JORDAN'S SYNDROME LIVE WITH:



#### Hypotonia or low muscle tone

It is a condition that makes it difficult and tiring to even sit down for an extended period of time. Every activity and movement feels like a workout. It also affects other aspects like speech and feeding. You can't change muscle tone, but you can get your core and muscles stronger to compensate. Hypotonia or low muscle tone impact internal muscles as well as the external ones. This has direct implications on the gastrointestinal (GI) tract, creating a variety of additional daily challenges.

#### Language difficulties and delays

...as well as apraxia of speech and dystonia - all fancy words that mean acquiring speech is difficult. While receptive language is unaffected, expressive language is a struggle. In some cases, communication is very limited, creating lots of frustration.

#### Complicated sensory profiles

Most of our children seek sensory input (such as lights, sounds, sensory play) but could be averse to other sensations (such as touch).

#### A spectrum of learning difficulties

While some might attend mainstream schools, most require some level of specialized instruction. No matter the setting and the level of support required, families urge educators to not lower their expectations and always push the limits.



#### Global developmental delays

On average, the walking milestone is achieved by 4 years of age. Typically developing children are walking before 18 months of age. This delays our children from exploring their environment and having a higher sense of independence.

#### Social interaction difficulties

Some of the Jordan's Syndrome population has been diagnosed with Autism. Parents often share about their child's super ability to create a strong connection with strangers in minutes. You just have to take a minute to understand, accept, and listen (even if it's with your heart at times).



#### Vision difficulties

The challenges include nystagmus, strabismus, amblyopia, and near sightedness to name a few. Glasses and corrective surgeries are required in many cases.

#### Fine motor skill difficulties

This hinders a lot of activities that many of us take for granted such as self-dressing, putting on shoes, self-feeding, and writing. Many require help even at a later age from a parent or a care giver. Some continue to require support into adulthood.

#### Difficulty at birth

Many end up in the intensive care unit. The exact reason for this is not known but the low muscle tone is suspected to play a role.

#### Sleep disorders

These disorders reveal in many ways including trouble falling asleep and trouble staying asleep. This creates a new level of exhaustion for the families year after year.



#### Feeding challenges that vary

Some of the population has required a feeding tube for the first few years of life. The reasons for needing a feeding tube include high risk of aspiration and failure to thrive. Many require feeding therapy to tolerate different textures and to strengthen mouth and jaw muscles.

#### Focus Challenges

While not many of the individuals with Jordan's Syndrome have an official ADHD diagnosis, maintaining focus on one activity till completion is generally challenging. Families use different interventions in the hopes of providing the support needed, including medications and regulated amounts of caffeine.

#### Seizures varying in types and onset age

For some, seizures happened right at birth. For others, they started in the teenage years. About 40% of the Jordan's Syndrome population has experienced seizures at least once. Families have used a wide variety of interventions to control the seizures, including medications and diet changes.



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Graphic pictured above is available for download at:  
[jordanguardianangels.org/resource-center](http://jordanguardianangels.org/resource-center)

## COMMON FEATURES OF JORDAN'S SYNDROME

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

## 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.

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

## 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 \(jordansguardianangels.org\)](https://www.jordansguardianangels.org)

Season 1, Episode 1: Living with Jordan's Syndrome

Season 1, Episode 6: Bringing Speech and Language to Life

## AMBASSADOR PROGRAM

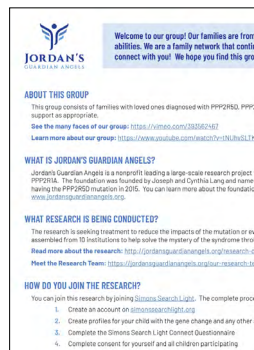
To best support our global community, Jordan's Guardian Angels Ambassadors span across regions and languages including Spanish, French, German and Italian. You can connect with your local community and access local resources as well as participate in virtual and in person regional events. Email [connect@jordansguardianangels.org](mailto:connect@jordansguardianangels.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](https://jordansguardianangels.org/resource-center)

**Website:** [jordansguardianangels.com](https://jordansguardianangels.com)

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