

# Jansen de Vries Syndrome

Other names: PPM1D, IDDGIP

## FOR PATIENTS, FAMILIES & EDUCATORS

Over the past several years, geneticists have discovered pathogenic variants in the PPM1D gene, now known as Jansen de Vries Syndrome (JdVS). It can be described as a nonsense alteration that results in a shortened protein. This mutation causes a wide range of overlapping clinical features including neurodevelopment issues, anxiety, unique facial features, short and wide hands/feet, growth hormone deficiency and a variety of other health issues. It is important to note that not every individual exhibits all of these symptoms. Therapies and early interventions have been proven to be very helpful in ensuring those diagnosed can be the best version of themselves. Family members, therapists, and teachers play an extremely important role in recognizing the needs of these students and learning how to help them succeed. Family members of recently diagnosed children have also found their children to be extremely social, loving and outgoing. In other words, joyous and beautiful people to be around! Welcome to our community, we're so glad you found us!

What is an IEP? An IEP is an Individualized Education Plan designed specifically for a child who has unique needs based on a diagnosed disability. The IEP plan spells out specific accommodations and related services a child needs to be successful within the school environment.

For more information on how to advocate for your child's academic and educational needs visit <https://www.wrightslaw.com/>

<b>Clinical Presentation/Diagnostic Category</b>	<b>Recommended Academic Accommodations and Services within Individualized Education Plans (IEP)</b>
ADHD	distraction free environment, cueing for attention, testing/assessments in private location, privacy board when working, clarification of directions, movement breaks, specialized seating (such as seat cushion, balance ball or wobble chair), support of aide within the classroom
Autism	Applied Behavior Analysis (ABA) both in home and at school -discrete trial learning -visuals incorporated throughout the home and school environment -social pragmatics group



	-support of aide within the classroom
Anxiety	-school psychologist or counselor on school TEAM -social emotional learning supports within school (such as Zones of Regulation or similar programming)
Speech	Speech and Language therapy in school
Occupational Therapy	Occupational therapy within school
Physical Therapy	Physical therapy within school
Vision	Teacher for the Visually Impaired (TVI) on school treatment TEAM high contrast materials with large font distraction free materials/worksheets
Reading/Writing	additional academic support within school
Math	additional academic support within school
Health	Individualized Healthcare Plan (IHP) for feeding, cyclical vomiting or constipation related issues

