

## Genetics 101

Genetics can be confusing and overwhelming, especially for those who do not have a background in science. Additionally, when faced with a new diagnosis it can be even more intimidating. Here is an attempt to provide a simplified genetics overview.

People inherit their chromosomes, which contain their genes, from their parents. Chromosomes come in pairs and humans have 46 chromosomes, 23 pairs. Children randomly get one of each pair of chromosomes from their mother and one of each pair from their father. The chromosomes that form the 23rd pair are called the sex chromosomes. They determine if a person is male or female.

The gene implicated in **Jansen de Vries Syndrome (JdVS)** is located on the 17th chromosome (specific location: 17q23.2) and has 6 exons. Exons are portions of the gene that code for proteins. Individuals with JdVS have variants/miscodings on the 5th and/or 6th exon of the PPM1D gene that result in a shortened (truncated) protein.

## Genetic Disorders

Genetic disorders can happen for many reasons. Genetic disorders often are described in terms of the chromosome that contains the gene. If the gene is on one of the first 22 pairs of chromosomes, called the autosomes, the genetic disorder is called an autosomal condition. If the gene is on the X chromosome, the disorder is called X-linked. **JdVS** is an autosomal dominant condition.

The PPM1D gene is affected in **JdVS**, therefore you may hear it referred to as PPM1D related disorder and your doctor will reference this gene when you review genetic testing after diagnosis. The PPM1D gene (Protein Phosphatase, Mg<sup>2+</sup>/Mn<sup>2+</sup> Dependent 1D) is a Protein Coding gene.



## JdVS and Other Single Gene Disorders

Some genetic diseases are caused by a DNA variation/mutation in one of a person's genes. For example, suppose part of a gene usually has the sequence TAC. A variant can change the sequence to TTC in some people. This change in sequence can change the way that the gene works, for example by changing the protein that is made. Think of variants as misspellings in genetic code, these misspellings thereby change the role/function of the gene. Variants can be passed down to a child from his or her parents. Or, they can happen for the first time in the sperm or egg, so that the child will have the mutation but the parents will not.

Single gene disorders can be autosomal or X-linked. Most, but not all, of the individuals identified with **JdVS** thus far have a *de novo variant*, meaning it is present for the first time in one family member as a result of a variant (or mutation) in a germ cell (egg or sperm) of one of the parents, or a variant that arises in the fertilized egg itself during early embryogenesis. Also called *de novo variant*, *new mutation*, and *new variant*. For the parents of a child with a *de novo variant*, the risk to have another child with the variant is very low; for the affected child the risk will be 50% to pass on the variant to their children.

Reference: Centers for Disease Control:  
<https://www.cdc.gov/genomics/about/basics.htm>

