



Dear JdVS Foundation Supporters,

It is hard to believe that 2023 is coming to a close. The JdVS Foundation and the JdVS Community are growing and thriving because of your generous contributions and commitment to helping put JdVS on the map! We are deeply grateful for your commitment to the JdVS community and wish you and your family a healthy and happy New Year. Over the past year we have been hard at work. Here are some of the amazing ways your financial support has helped us grow!

- We have connected with 37 more JdVS families and have a growing community of over 190 JdVS individuals. We have identified JdVS individuals in 6 out of 7 continents ranging in age from 4 months - 77 years old!
- We hosted our First JdVS Family and Medical Professionals Summit in Charlotte, NC this past July. We had a total of 22 families attend. Your generous gifts supported this event and made it a truly spectacular weekend filled with connections, mutual understanding and support. Families were delighted to meet Dr. de Vries and Dr. Boles as well as hear from Dr. Lachman and Dr. Wojcik. Check out our YouTube channel for videos!
https://www.youtube.com/channel/UCAOVfY5ZUILPPWwfK4Hu_RQ
- We have partnered with Dr. de Vries at Radboud University in Nijmegen, Netherlands and have committed to funding a 3 year Natural History Study and the development of a JdVS Center for Excellence to gain greater insight into the course of the syndrome overtime. This is an important step to developing data driven interventions and potential therapeutics. We have a goal of raising \$50,000 by April 1st. All money raised from now until Feb. 1st will be matched by an anonymous donor (up to \$15,000)
- Once again, this year the JdVS Foundation was awarded a Rare Is Horizon Therapeutics Grant which was used to support the cost of hosting our first Family Conference
- Dr. Curry and Dr. Monica Wojcik published an article in The American Journal of Medical Genetics on their work collecting data and expanding the clinical and phenotypic spectrum of Jansen de Vries Syndrome.



Jansen de Vries
Syndrome Foundation

- The JdVS Foundation had the privilege of being invited to speak at the New England Regional Genetics Group's 46th Annual Educational Conference where we presented alongside Dr. Catherine Nowak, Clinical Director of the Center for Genetics and Metabolism at Mass General Hospital in Boston, MA. Kara joined Dr. Nowak to educate future and current genetics professionals about JdVS!
- We have created Virtual Meet-Up spaces for JdVS Champions, JdVS Caregivers and JdVS Siblings allowing our community to connect with each other, no matter the distance!
- We continue to support our community through the JdVS Uplift grant where families can apply for financial assistance to be used towards academic, athletic, extracurricular, artistic and other enrichment activities that support the lives, interests and passions of individuals diagnosed with JdVS.
- The Lachman lab is continuing its work on the underlying molecular basis of JdVS using induced pluripotent stem (iPS) cell technology, which allows them to make neurons from patients. They are currently preparing a manuscript describing their RNA sequencing and proteomics findings; molecular techniques that provide a window into neuronal pathways that are dysregulated as a result of exons 5 and 6 PPM1D mutations found in JdVS.

THANK YOU for allowing us to continue doing this work and supporting the JdVS Community! Should you feel inclined to support us once again this year, a tax deductible donation can be made by clicking the link below!

[Donate HERE](#)

With sincerest appreciation and thanks,

Kara Kilroy, President

Rachel Horne, Secretary

Marinda du Toit, Treasurer