Welcome to our group! Our families are from across the globe and our children vary in age and abilities. We are a family network that continuously learns from each other. We are happy to connect with you! We hope you find this group as helpful for you as it was for each of our families.

About this Group

This group consists of families with loved ones diagnosed with PPP2R5D, PPP2R1A or PPP2R5C. We provide emotional and information support as appropriate.

See the many faces of our group: https://vimeo.com/393562467

Learn more about our group:
https://www.youtube.com/watch?v=tNUhvSLTK3M&app=desktop

What is Jordan's Guardian Angels?

Jordan's Guardian Angels is a nonprofit leading a large-scale research project to learn all of the impacts of PPP2R5D, PPP2R5C and PPP2R1A. The foundation was founded by Joseph and Cynthia Lang and named after their daughter, Jordan. Jordan was diagnosed with having the PPP2R5D mutation in 2015. You can learn more about the foundation at www.jordansguardianangels.org.

What Research is being conducted?

The research is seeking treatment to reduce the impacts of the mutation or even reverse it. A team of the best and the brightest has been assembled from 10 institutions to help solve the mystery of the syndrome through cutting edge technology methods.

Read more about the research: http://jordansguardianangels.org/research-details-impacts/

Meet the Research Team: https://jordansguardianangels.org/our-research-team/

How do you join the research?

You can join this research by joining Simons Search Light (www.simonssearchlight.org). The complete process is outlined below:

1. Create an account on simonssearchlight.org
2. Create profiles for your child with the gene change and any other siblings under 18 that you

wish to participate.
3. Complete the Simons Search Light Connect Questionnaire
4. Complete consent for yourself and all children participating
5. Send Simons Search Light your child's genetic testing results

Once steps 1-4 are completed, the family is then connected with Ashley for the medical history phone call and assigned baseline surveys. The genetic testing results do not hold up this process but are required before a family is considered enrolled.

Enrollment = Consent for parent and child with the genetic change, completed Simons VIP Connect Questionnaire and confirmed genetic lab results.

How do you join the families' database?

In order to organize the information within the families, we maintain an online database. You can join our database by filling the form: https://forms.gle/z5AmCC54ZCoEkXvG7

You can connect with the families by joining the private Facebook group at "PPP Families (PPP2R5D, PPP2R1A, PPP2R5C)".

How do you provide EEGs and MRIs?

If you have EEGs or MRIs, Dr Ghayda Mirzaa is collecting those as part of the research. She will be using this information to better understand brain activity especially when it comes to seizures and sleep disorders. You can email her research coordinator, Jordan Zeiger, brainresearch@seattlechildrens.org and let her know you are part of this group.

What Resources are Available?

You can access the password protected family resources page at:
https://jordansguardianangels.org/family-resources/

Please email info@jordansguardianangels.org or contact our Facebook admins to obtain the password.

Know that you are not alone! This has been a journey of hope for our families; a lot of excitement is underway. If interested, we can match you with another family based on location, language, age, and/or variation. For any questions or comments, please contact us at info@jordansguardianangels.org or by phone/whatsapp at 1-720-725-1727.

New Families Frequently Asked Questions

This document has simple answers to questions often asked by new families when they get the diagnosis. This is based on input from families and is not meant to give medical advice. For any additional questions, feel free to post on the Facebook "PPP2R5D Families" group or email info@jordansguardianangels.org.

The Science in Simple Terms

What is a genetic mutation?

A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people.

What is a mutation in PPP2R5D?

Our DNA consists of 3 billion letters. Jordan's syndrome is a misspelling on the gene PPP2R5D. It is de novo (not inherited). There is less than 1% chance of it reoccurring within the same family. An individual with the mutation has a 50% chance of passing it to their own children.

Why does the mutation happen?

The mutation can happen in the sperm, egg, or at conception. It is random. There's nothing either parent have done to cause it and could not have been prevented.

The impact

How do we expect it to affect my child? What is the long-term prognosis?

There's a spectrum as to how a mutation in PPP2R5D affects individuals. Most deal with global developmental delays that appear very early on. Other concerns include potential seizures, feeding and vision challenges. Most of the current population diagnosed are children with limited data on long term impacts. We encourage you to not expect any different from your child and to keep pushing their potential! They will surprise you in how much they can accomplish!

Will my child walk?

While our children deal with a delay in walking, most have achieved this milestone. We encourage you to find ways to support their mobility until walking is achieved such as via a walker or a stander.

Will my child talk?

There's a big spectrum when it comes to speech. Some of our children are fully nonverbal while others are fully communicative. We encourage you to increase speech interventions and introduce alternative ways of communication such as sign language, pictures, or a communication device.

Will my child be able to go to a regular school?

Our current population attend a variety of schools. Some are in a regular fully inclusive programs while others are in special education programs and/or school. No matter the placement, parent's involvement to ensure the appropriate supports are put in place has been key to their success.

What specialists should we see?

Our families have seen and/or are being followed by a variety of specials including neurology, ophthalmology, gastroenterology, urology, cardiology, immunology, genetics, etc. Most are discharged or followed periodically. We encourage you to treat the symptoms if any health problems arise like you would do with any child.

What therapies should we do and how much?

Intensive therapy has proven to be helpful to our families with over 8 hours of therapy a week. In addition to physical, occupational and speech therapy, check the link below for the recommended therapies by our families. We encourage you to get familiar with services available within your country and/or state that can cover some of these therapies. If you are in the United States, most states have a Medicaid waiver program based on disability that you can explore.

https://jordansguardianangels.org/wp-content/uploads/2019/06/Recommended-Therapies-from-Families-Discussion-march-2019.pdf

The research

What research is underway?

Jordan's Guardian Angels is leading a global research looking for a treatment and/or for a way to reverse the mutation. A team of the best in the field has been recruited to study the mutation. You can learn more about the research at www.jordansguardianangels.org.

How do we expect the research to help my child?

The mutation affects the function of the brain and not the structure. Therefore, it is believed that there is a potential for a treatment or even a cure.

How do I join the research?

You can join the research by registering at www.simonssearchlight.org. With our small numbers, we highly encourage you to participate in the research as each of our children might hold another piece of the puzzle that we are together working on solving.

Next Steps

Where do I go from here?

Talk to your doctor about the new diagnosis. Setup appointments with main specialists and ramp up on therapies as needed. Reach out to the family group with any questions as we are all learning from each other's experiences. You can get more involved by spreading awareness, supporting other families, and help fundraise. Most of all however, please know that the diagnosis doesn't change who your child is! With your support, they can conquer this world in their own way.

New Families to do list

Time of diagnosis can be the hardest! We've all been there, and we are here for you. Please give yourself the time you need to process the news and all the new information that comes with that. When you are ready, here's a checklist that can help you keep track of some to-dos to integrate you within the group, provide you with information that our families find important, and include you in the research. Don't hesitate to ask any questions you might have.

- Introduce yourself at the "PPP Families (PPP2R5D, PPP2R1A, PPP2R5C)" Page
- Read your welcome letter
- Join the PPP Families Directory https://forms.gle/eEgQT4uH67YytCoN6
- Check out Jordan's Guardian Angels' website www.jordansguardianangels.org
- Sign up for the research www.simonssearchlight.org
- If you have EEG or MRI data, contact Seattle's Children's Hospital at brainresearch@seattlechildrens.org
- Fill out the informal families' database https://forms.gle/1AvieEr3EsV53iH27
- Check out JGA families Resource Page https://jordansguardianangels.org/family-resources/