

Uniting Rare Genetic Variants

Web Resources

What are genetic variants?

A genetic variant is a change in the DNA sequence of a gene. Variants can be pathogenic (disease causing), benign (not disease causing), or be of uncertain significance. Sometimes a pathogenic variant is identified in a child but they are the only ones who have been reported to have it, or a variant of uncertain significance is identified and we need to learn more to find out if it is disease causing or not.

How can these resources be useful to me?

The following websites help patients and families connect with other families, healthcare providers, and researchers based on similar genetic variants and medical features.

All websites listed below are free. Inclusion on this list does not imply endorsement by Seattle Children's Hospital. Ask your child's care team if you have questions.

MyGene2

mygene2.org

You can create a "family profile" where you enter information such as your story, photos, and your child's genetic variants. This then becomes searchable so you can connect with others to share information or offer support.

GeneMatcher

genematcher.org

You can post a gene(s) of interest (for example, the gene that your child has variants in) and the site will connect you with those who post the same gene.

GenomeConnect

genomeconnect.org

An online registry designed by the Clinical Genome Resource (ClinGen). The goal of the registry is to share de-identified genetic and health information. You can then connect with other patients and families, learn about research opportunities and receive updates about your genetic test results.

To Learn More

- Genetics
206-987-2056
- Biochemical Genetics
206-987-3012
- Ask your child's healthcare provider
- seattlechildrens.org

Free Interpreter Services

- In the hospital, ask your nurse.
- From outside the hospital, call the toll-free Family Interpreting Line, 1-866-583-1527. Tell the interpreter the name or extension you need.



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Seattle Children's offers interpreter services for Deaf, hard of hearing or non-English speaking patients, family members and legal representatives free of charge. Seattle Children's will make this information available in alternate formats upon request. Call the Family Resource Center at 206-987-2201. This handout has been reviewed by clinical staff at Seattle Children's. However, your child's needs are unique. Before you act or rely upon this information, please talk with your child's healthcare provider.

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