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This list of common questions about SCN2A-Related Disorders (SRD) will help you discuss genetic test results, risk factors and potential treatments with your provider.

Questions

- What is my child's variant, type of variant (missense, truncation, deletion, duplication), and is the functional consequence of this variant known or could you predict it (Gain of Function, Loss of Function)?
- Can you tell me more about my child's phenotype based on the SRD variant?
- Is the variant pathogenic (disease-causing)?
- Does my child have any other mutations?
- What is the inheritance of the variant? Is further testing necessary (parental testing, whole exome)?
- Are there any clinical trials or research we can participate in?
- Has my child's variant been studied in the literature?
- Is there someone out there with the same variant?
- What medications or therapies are appropriate for my child?
- How do we develop a Seizure Action Plan?
- What other specialists do you recommend seeing?
- Do you feel comfortable managing or can you collaborate with an SRD specialist?

Comments / Notes

QUESTIONS

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TO ASK YOUR



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