Christianson Syndrome

What syndrome? is Christianson

Christianson syndrome is a disorder that primarily affects the nervous system. This condition becomes apparent in infancy. Its characteristic features include delayed development, intellectual disability, an inability to speak, problems with balance and coordination (ataxia), and difficulty standing or walking. Individuals who do learn to walk lose the ability in childhood. Most affected children also have recurrent seizures (epilepsy), beginning between ages 1 and 2. Other features seen in many people with Christianson syndrome include a small head size (microcephaly); a long, narrow face with prominent nose, jaw, and ears; an open mouth and uncontrolled drooling; and abnormal eye movements. Affected children often have a happy demeanor with frequent smiling and spontaneous laughter.

How common is Christianson syndrome?

Christianson syndrome is a rare condition, although the exact prevalence is unknown. The condition was first described in a South African family and has since been found in people in other parts of the world.

What genes are related to Christianson syndrome?

Christianson syndrome is caused by mutations in the SLC9A6 gene, which provides instructions for making a protein called sodium/hydrogen exchanger 6 (Na+/H+ exchanger 6 or NHE6). The NHE6 protein is found in the membrane surrounding endosomes, which are compartments within cells that recycle proteins and other materials. The NHE6 protein acts as a channel to exchange positively charged atoms (ions) of sodium (Na+) with hydrogen ions (H+). By controlling the amount of hydrogen ions, the NHE6 protein helps regulate the relative acidity (pH) inside endosomes, which is important for the recycling function of these compartments. The NHE6 protein may have additional functions, such as helping to move proteins to the correct location in the cell (protein trafficking). Mutations in the SLC9A6 gene typically lead to an abnormally short NHE6 protein that is nonfunctional or that is broken down quickly in cells, resulting in the absence of functional NHE6 channels. As a result, the pH in endosomes is not properly maintained. It is unclear how unregulated endosomal pH leads to neurological problems in people with Christianson syndrome. Some studies have shown that protein trafficking by endosomes is important for learning and memory, but the role of endosomal pH or the NHE6 protein in this process has not been identified.

Related Gene(s)

Changes in this gene are associated with Christianson syndrome.

- SLC9A6

How do people inherit Christianson syndrome?

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

In X-linked recessive inheritance, a female with one mutated copy of the gene in each cell is called a carrier. She can pass on the altered gene but usually does not experience signs and symptoms of the disorder. Occasionally, however, females who carry an SLC9A6 gene mutation have mild
learning disabilities. It is unclear if these disabilities are related to the gene mutation or occur by chance.

**Where can I find information about diagnosis or management of Christianson syndrome?**

These resources address the diagnosis or management of Christianson syndrome and may include treatment providers.


You might also find information on the diagnosis or management of Christianson syndrome in Educational resources (http://ghr.nlm.nih.gov/condition/christianson-syndrome/show/Educational+resources).


To locate a healthcare provider, see How can I find a genetics professional in my area? (http://ghr.nlm.nih.gov/handbook/consult/findingprofessional) in the Handbook.

**Where can I find additional information about Christianson syndrome?**

You may find the following resources about Christianson syndrome helpful. These materials are written for the general public.

- **MedlinePlus - Health information**

- Genetic and Rare Diseases Information Center - Information about genetic conditions and rare diseases (http://rarediseases.info.nih.gov/gard/10572/christianson-syndrome/resources/1)

- Additional NIH Resources - National Institutes of Health

- **Educational resources - Information pages**
  - Disease InfoSearch: Christianson Syndrome (http://www.diseaseinfosearch.org/Christianson+Syndrome/1400)
  - Orphanet: Christianson syndrome (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=85278)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Genetic Testing Registry - Repository of genetic test information
What other names do people use for Christianson syndrome?

- Angelman-like syndrome, X-linked
- Intellectual deficit, X-linked, South African type


What if I still have specific questions about Christianson syndrome?

Ask the Genetic and Rare Diseases Information Center (http://rarediseases.info.nih.gov/GARD/).

What glossary definitions help with understanding Christianson syndrome?

acidity; ataxia; carrier; cell; channel; chromosome; disabilities; disability; endosomes; epilepsy; gene; hydrogen ions; inheritance; inherited; ions; mental retardation; microcephaly; mutation; Na; nervous system; neurological; pH; prevalence; protein; recessive; sex chromosomes; sodium; spontaneous; syndrome; X-linked recessive

You may find definitions for these and many other terms in the Genetics Home Reference Glossary (http://ghr.nlm.nih.gov/glossary).

References


The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult

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