Isolated hyperchlorhidrosis

Description

Isolated hyperchlorhidrosis is characterized by the excessive loss of salt (sodium chloride or NaCl) in sweat. In particular, "hyperchlorhidrosis" refers to the high levels of chloride found in sweat, although both sodium and chloride are released. Because the salt is abnormally released from the body in sweat, there are lower than normal levels of sodium in fluids inside the body (hyponatremia). Most infants with isolated hyperchlorhidrosis experience one or more episodes of dehydration with low levels of sodium in the blood (hyponatremic dehydration), which can require hospitalization. These episodes typically follow a mild illness that causes vomiting or diarrhea. Affected infants also have poor feeding and an inability to grow and gain weight at the expected rate (failure to thrive). By early childhood, though, weight and height usually catch up to normal, although the abnormal loss of salt still remains. These individuals may still experience dangerous hyponatremia when they sweat excessively, for example in warm temperatures or when exercising.

While hyperchlorhidrosis can occur as one of several features of other conditions, such as cystic fibrosis, people with isolated hyperchlorhidrosis do not have the additional signs and symptoms of these other conditions.

Frequency

Isolated hyperchlorhidrosis is a rare condition, although its prevalence is unknown. The condition has been found mostly in the Bedouin population of southern Israel.

Causes

Isolated hyperchlorhidrosis is caused by a mutation in the *CA12* gene. This gene provides instructions for making a protein called carbonic anhydrase 12 (CA 12), which belongs to a family of related proteins known as carbonic anhydrases. These proteins carry out a chemical reaction that involves the molecules carbon dioxide and water and produces a negatively charged bicarbonate molecule (bicarbonate ion) and a positively charged hydrogen atom (known as a proton). The presence of protons and bicarbonate affect the relative acidity (pH) of cells, which is important for several cellular processes, including the transport of salt into and out of cells. Researchers suggest that by regulating cellular pH in the sweat glands, CA 12 is able to control the amount of salt released in sweat.

The *CA12* gene mutation involved in isolated hyperchlorhidrosis leads to reduction of CA 12 protein function. Researchers speculate that the altered CA 12 is less able to regulate pH, and it loses its ability to control how much salt is released in sweat, leading to hyperchlorhidrosis.

Learn more about the gene associated with Isolated hyperchlorhidrosis

- CA12

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Carbonic anhydrase XII deficiency

Additional Information & Resources

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- HYPERCHLORHIDROSIS, ISOLATED; HYCHL (https://omim.org/entry/143860)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28isolated+hyperchlorhidrosis%29+OR+%28hyperchlorhidrosis%29%29+OR+%28%28hyponatremia%29+AND+%28carbonic+anhydrase+12%29+OR+%28carbonica+anhydrase+XII%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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