Corticosterone methyloxidase deficiency

Description

Corticosterone methyloxidase deficiency, also known as aldosterone synthase deficiency, is a disorder characterized by excessive amounts of sodium released in the urine (salt wasting), along with insufficient release of potassium in the urine, usually beginning in the first few weeks of life. This imbalance leads to low levels of sodium and high levels of potassium in the blood (hyponatremia and hyperkalemia, respectively). Individuals with corticosterone methyloxidase deficiency can also have high levels of acid in the blood (metabolic acidosis).

The hyponatremia, hyperkalemia, and metabolic acidosis associated with corticosterone methyloxidase deficiency can cause nausea, vomiting, dehydration, low blood pressure, extreme tiredness (fatigue), and muscle weakness. Affected infants often experience failure to thrive, which means they do not gain weight and grow at the expected rate. Severe cases of corticosterone methyloxidase deficiency can result in seizures and coma and can be life-threatening. However, affected individuals who survive infancy generally have a normal life expectancy, and the signs and symptoms of the disorder typically become milder or disappear by adulthood.

Frequency

Corticosterone methyloxidase deficiency is a rare disorder; its prevalence is unknown. Researchers have described two types of the condition: Type I is more common in the Amish population of Lancaster, Pennsylvania, while type II is more common in people of Iranian Jewish ancestry. The two types have similar signs and symptoms but can be distinguished by laboratory testing.

Causes

Mutations in the *CYP11B2* gene cause corticosterone methyloxidase deficiency. This gene provides instructions for making an enzyme called aldosterone synthase. The aldosterone synthase enzyme is found in the adrenal glands, which are located on top of the kidneys.

Aldosterone synthase helps produce a hormone called aldosterone. Aldosterone regulates blood pressure by maintaining proper salt and fluid levels in the body. The aldosterone synthase enzyme is involved in a series of three chemical reactions that

produce aldosterone from other (precursor) molecules: the conversion of 11-deoxycorticosterone to corticosterone, the conversion of corticosterone to 18-hydroxycorticosterone, and the conversion of 18-hydroxycorticosterone to aldosterone.

The *CYP11B2* gene mutations that cause corticosterone methyloxidase deficiency lead to insufficient production of aldosterone, which impairs the kidneys' ability to reabsorb salt (sodium chloride or NaCl) into the blood and release potassium in the urine. As a result, excessive amounts of salt in the form of charged atoms (ions) of sodium (Na^+) and chlorine (Cl^-) leave the body in the urine, while not enough potassium is released. The resulting imbalance of ions in the body underlies the signs and symptoms of corticosterone methyloxidase deficiency.

Learn more about the gene associated with Corticosterone methyloxidase deficiency

- CYP11B2

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

- 18-hydroxylase deficiency
- 18-oxidase deficiency
- Aldosterone deficiency
- Aldosterone deficiency due to deficiency of steroid 18-hydroxylase
- Aldosterone deficiency due to deficiency of steroid 18-oxidase
- Aldosterone synthase deficiency
- CMO deficiency
- Congenital hypoaldosteronism
- Corticosterone 18-monooxygenase deficiency
- Corticosterone methyl oxidase deficiency
- Familial hyperreninemic hypoaldosteronism
- Steroid 18-hydroxylase deficiency
- Steroid 18-oxidase deficiency
- Visser-Cost syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Corticosterone methyloxidase type 2 deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3463917/)
- Genetic Testing Registry: Corticosterone 18-monooxygenase deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268293/)

Genetic and Rare Diseases Information Center

- Hypoaldosteronism (https://rarediseases.info.nih.gov/diseases/2874/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CORTICOSTERONE METHYLOXIDASE TYPE I DEFICIENCY (https://omim.org/entry/203400)
- CORTICOSTERONE METHYLOXIDASE TYPE II DEFICIENCY (https://omim.org/entry/610600)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28corticosterone+methyloxidase+deficiency%29+OR+%28aldosterone+synthase+deficiency%29+OR+%28corticosterone+methyl+oxidase+deficiency%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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