CYP11B2 gene

cytochrome P450 family 11 subfamily B member 2

Normal Function

The *CYP11B2* gene provides instructions for making an enzyme called aldosterone synthase (previously known as corticosterone methyloxidase). This enzyme is found in the adrenal glands, which are located on top of the kidneys. Aldosterone synthase is a member of the cytochrome P450 family of enzymes. These enzymes are involved in the formation and breakdown of various molecules within cells.

Aldosterone synthase helps produce a hormone called aldosterone. Aldosterone helps control 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.

Health Conditions Related to Genetic Changes

Corticosterone methyloxidase deficiency

At least 30 *CYP11B2* gene mutations that cause corticosterone methyloxidase deficiency (also known as aldosterone synthase deficiency) have been identified. These mutations 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 this disorder, which include nausea, vomiting, dehydration, low blood pressure, extreme tiredness (fatigue), and muscle weakness.

Familial hyperaldosteronism

A genetic change affecting the *CYP11B2* gene causes familial hyperaldosteronism type I, a disorder that leads to high blood pressure (hypertension). This change joins (fuses) the section of the *CYP11B2* gene that contains the instructions for making aldosterone synthase to a section of a nearby gene called *CYP11B1*. The added part of the *CYP11B1* gene contains a section called a promoter region, which normally helps start

the production of an enzyme called 11-beta-hydroxylase from the *CYP11B1* gene.

By binding to the *CYP11B1* gene's promoter region, a hormone called adrenocorticotropic hormone (ACTH) normally triggers production of the 11-beta-hydroxylase enzyme. In the fusion gene, ACTH binding abnormally triggers production of aldosterone synthase. High levels of aldosterone synthase result in excessive aldosterone production, which leads to the hypertension associated with familial hyperaldosteronism type I.

Other disorders

Normal variations (polymorphisms) of the *CYP11B2* gene have been associated with increased risk of cardiovascular problems including hypertension, stroke caused by a lack of blood flow to the brain (ischemic stroke), and a heart rhythm abnormality called atrial fibrillation in people with heart failure. Researchers suggest that differences in blood pressure control resulting from the *CYP11B2* gene variations may lead to increased risk of these cardiovascular problems.

Other Names for This Gene

- ALDOS
- aldosterone synthase
- aldosterone-synthesizing enzyme
- C11B2_HUMAN
- CPN2
- CYP11B
- CYP11BL
- CYPXIB2
- cytochrome P-450Aldo
- cytochrome P-450C18
- cytochrome P450 11B2, mitochondrial
- cytochrome P450 11B2, mitochondrial precursor
- cytochrome P450, family 11, subfamily B, polypeptide 2
- cytochrome P450, subfamily XIB (steroid 11-beta-hydroxylase), polypeptide 2
- mitochondrial cytochrome P450, family 11, subfamily B, polypeptide 2
- P-450C18
- P450aldo
- P450C18
- steroid 11-beta-monooxygenase
- steroid 11-beta/18-hydroxylase
- steroid 18-hydroxylase, aldosterone synthase, P450C18, P450aldo

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CYP11B2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1585[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CYP11B2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2; CYP11B2 (https://omim.org/entry/124080)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1585)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CYP11B2[gene])

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Genomic Location

The *CYP11B2* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chrom osome/8/).

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