CYP24A1 gene

cytochrome P450 family 24 subfamily A member 1

Normal Function

The *CYP24A1* gene provides instructions for making an enzyme called 24-hydroxylase. This enzyme helps control the amount of active vitamin D available in the body. When active, vitamin D is involved in maintaining the proper balance of several minerals in the body, including calcium and phosphate, which are essential for the normal formation of bones and teeth. One of vitamin D's major roles is to control the absorption of calcium and phosphate from the intestines into the bloodstream. Vitamin D is also involved in several processes in addition to bone and tooth formation.

The 24-hydroxylase enzyme breaks down the active form of vitamin D, called 1,25-dihydroxyvitamin D_3 or calcitriol, to an inactive form when the vitamin is no longer needed. The enzyme also breaks down 25-hydroxyvitamin D (also known as calcidiol), which is the form of vitamin D that is stored in the body.

Health Conditions Related to Genetic Changes

Idiopathic infantile hypercalcemia

More than 20 mutations in the *CYP24A1* gene have been found to cause a type of idiopathic infantile hypercalcemia called infantile hypercalcemia 1, which is characterized by high levels of calcium in the blood (hypercalcemia) and urine (hypercalciuria) and deposits of calcium in the kidneys (nephrocalcinosis). The hypercalcemia typically causes vomiting, poor feeding, and an inability to grow and gain weight at the expected rate (failure to thrive) in infancy, although some affected individuals do not develop signs and symptoms of the condition until adulthood. Features in affected adults, whether they had symptoms in infancy or not, typically include hypercalciuria, nephrocalcinosis, and kidney stones (nephrolithiasis), although they may not cause any obvious health problems.

The *CYP24A1* gene mutations that cause infantile hypercalcemia 1 reduce or eliminate the activity of the 24-hydroxylase enzyme. A shortage of this enzyme's function impairs the breakdown of calcitriol. The resulting excess of calcitriol increases calcium absorption into the bloodstream, causing hypercalcemia. Dysregulation of calcium absorption in the kidneys leads to hypercalciuria, nephrocalcinosis, and nephrolithiasis.

Other Names for This Gene

- 1,25-@dihydroxyvitamin D3 24-hydroxylase
- 1,25-dihydroxyvitamin D(3) 24-hydroxylase, mitochondrial isoform 1 precursor
- 1,25-dihydroxyvitamin D(3) 24-hydroxylase, mitochondrial isoform 2 precursor
- 24-OHase
- CP24
- CYP24
- cytochrome P450 24A1
- cytochrome P450, family 24, subfamily A, polypeptide 1
- cytochrome P450, subfamily XXIV (vitamin D 24-hydroxylase)
- cytochrome P450-CC24
- exo-mitochondrial protein
- HCAI
- HCINF1
- P450-CC24
- vitamin D 24-hydroxylase
- vitamin D(3) 24-hydroxylase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CYP24A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1591[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CYP24A1%5BTIAB%5D%29+OR+%28cytochrome+P450+family+24+subfamily+A+member+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, FAMILY 24, SUBFAMILY A, POLYPEPTIDE 1; CYP24A1 (https://omim.org/entry/126065)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1591)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CYP24A1[gene])

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Genomic Location

The *CYP24A1* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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