AGTR1 gene

angiotensin II receptor type 1

Normal Function

The *AGTR1* gene provides instructions for making a protein called the angiotensin II receptor type 1 (AT1 receptor). This protein is part of the renin-angiotensin system, which regulates blood pressure and the balance of fluids and salts in the body. Through a series of steps, the renin-angiotensin system produces a molecule called angiotensin II, which attaches (binds) to the AT1 receptor, stimulating chemical signaling. This signaling causes blood vessels to narrow (constrict), which results in increased blood pressure. Binding of angiotensin II to the AT1 receptor also stimulates production of the hormone aldosterone, which triggers the absorption of water and salt by the kidneys. The increased amount of fluid in the body also increases blood pressure. Proper blood pressure during fetal growth, which delivers oxygen to the developing tissues, is required for normal development of the kidneys, particularly of structures called the proximal tubules, and other tissues. In addition, angiotensin II may play a more direct role in kidney development, perhaps by affecting growth factors involved in the development of kidney structures.

Health Conditions Related to Genetic Changes

Renal tubular dysgenesis

At least four mutations in the *AGTR1* gene have been found to cause a severe kidney disorder called renal tubular dysgenesis. This condition is characterized by abnormal kidney development before birth, the inability to produce urine (anuria), and severe low blood pressure (hypotension). These problems result in a reduction of amniotic fluid (oligohydramnios), which leads to a set of birth defects known as the Potter sequence.

Renal tubular dysgenesis can be caused by mutations in both copies of any of the genes involved in the renin-angiotensin system. The *AGTR1* gene mutations that cause this disorder likely change or block the AT1 receptor's ability to stimulate signaling, which results in a nonfunctional renin-angiotensin system. Without this system, the kidneys cannot control blood pressure. Because of low blood pressure, the flow of blood is reduced (hypoperfusion), and the body does not get enough oxygen during fetal development. As a result, kidney development is impaired, leading to the features of renal tubular dysgenesis.

Hypertension

MedlinePlus Genetics provides information about Hypertension

Other disorders

Variations in the *AGTR1* gene have been reported to be associated with an increased risk of a form of high blood pressure (hypertension) called essential hypertension; heart disease; or diabetic nephropathy, a complication of diabetes that affects kidney function. These are complex disorders associated with many genetic and environmental factors. The most studied *AGTR1* gene variation associated with these conditions changes a single DNA building block (nucleotide) in the gene. This change switches the nucleotide adenine to cytosine at position 1166 in the gene (written as A1166C). It is unclear how this *AGTR1* gene variation contributes to the risk of these conditions.

Other Names for This Gene

- AG2S
- AGTR1_HUMAN
- AGTR1A
- AGTR1B
- angiotensin II receptor, type 1
- AT1
- AT1AR
- AT1B
- AT1BR
- AT1R
- AT2R1
- AT2R1A
- AT2R1B
- HAT1R
- type-1 angiotensin II receptor
- type-1B angiotensin II receptor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AGTR1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=185[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28AGTR1%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

- ANGIOTENSIN II RECEPTOR, TYPE 1; AGTR1 (https://omim.org/entry/106165)
- HYPERTENSION, ESSENTIAL (https://omim.org/entry/145500)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/185)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AGTR1[gene])

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Genomic Location

The *AGTR1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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