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])

References

- Ding W, Wang F, Fang Q, Zhang M, Chen J, Gu Y. Association between two geneticpolymorphisms of the renin-angiotensin-aldosterone system and diabeticnephropathy: a meta-analysis. Mol Biol Rep. 2012 Feb;39(2):1293-303. doi: 10.1007/s11033-011-0862-7. Epub 2011 May 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21607620)
- Duncan JA, Scholey JW, Miller JA. Angiotensin II type 1 receptor genepolymorphisms in humans: physiology and pathophysiology of the genotypes. CurrOpin Nephrol Hypertens. 2001 Jan;10(1):111-6. doi:10.1097/00041552-200101000-00017. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11195043)

- Gribouval O, Gonzales M, Neuhaus T, Aziza J, Bieth E, Laurent N, Bouton JM, Feuillet F, Makni S, Ben Amar H, Laube G, Delezoide AL, Bouvier R, Dijoud F, Ollagnon-Roman E, Roume J, Joubert M, Antignac C, Gubler MC. Mutations in genesin the renin-angiotensin system are associated with autosomal recessive renaltubular dysgenesis. Nat Genet. 2005 Sep;37(9):964-8. doi: 10.1038/ng1623. Epub2005 Aug 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16116425)
- Gribouval O, Moriniere V, Pawtowski A, Arrondel C, Sallinen SL, Saloranta C, Clericuzio C, Viot G, Tantau J, Blesson S, Cloarec S, Machet MC, Chitayat D, Thauvin C, Laurent N, Sampson JR, Bernstein JA, Clemenson A, Prieur F, Daniel L, Levy-Mozziconacci A, Lachlan K, Alessandri JL, Cartault F, Riviere JP, Picard N, Baumann C, Delezoide AL, Belar Ortega M, Chassaing N, Labrune P, Yu S, Firth H, Wellesley D, Bitzan M, Alfares A, Braverman N, Krogh L, Tolmie J, Gaspar H, DorayB, Majore S, Bonneau D, Triau S, Loirat C, David A, Bartholdi D, Peleg A, Brackman D, Stone R, DeBerardinis R, Corvol P, Michaud A, Antignac C, Gubler MC.Spectrum of mutations in the renin-angiotensin system genes in autosomalrecessive renal tubular dysgenesis. Hum Mutat. 2012 Feb;33(2):316-26. doi:10.1002/humu.21661. Epub 2011 Dec 22. Citation on PubMed (https://pubmed.n

cbi.nlm.nih.gov/22095942)

- Gubler MC, Antignac C. Renin-angiotensin system in kidney development: renaltubular dysgenesis. Kidney Int. 2010 Mar;77(5):400-6. doi: 10.1038/ki.2009.423. Epub 2009 Nov 18. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19924102)

- Katsuya T, Morishita R. Gene polymorphism of angiotensin II type 1 and type 2receptors. Curr Pharm Des. 2013;19(17):2996-3001. doi:10.2174/1381612811319170004. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23176211)
- Wolf G. Angiotensin II and tubular development. Nephrol Dial Transplant.2002;17 Suppl 9:48-51. doi: 10.1093/ndt/17.suppl_9.48. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12386287)

Genomic Location

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

Last updated May 1, 2013