

# AGT Gene

Subjects: **Genetics & Heredity**

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angiotensinogen

genes

## 1. Normal Function

The *AGT* gene provides instructions for making a protein called angiotensinogen. This protein is part of the renin-angiotensin system, which regulates blood pressure and the balance of fluids and salts in the body. In the first step of this process, angiotensinogen is converted to angiotensin I. Through an additional step, angiotensin I is converted to angiotensin II. Angiotensin II causes blood vessels to narrow (constrict), which results in increased blood pressure. This molecule also stimulates production of the hormone aldosterone, which triggers the absorption of salt and water 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1 Renal tubular dysgenesis

At least six mutations in the *AGT* 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. Most of the mutations in the *AGT* gene that cause this disorder change single protein building blocks (amino acids) in the angiotensinogen protein. These changes occur in a region of the protein that is necessary for its conversion to angiotensin I. It is thought that the altered angiotensinogen cannot be converted, leading to 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.

## 2.2 Hypertension

MedlinePlus Genetics provides information about Hypertension

## 2.3 Other disorders

Variations in the *AGT* gene are associated with susceptibility to a form of high blood pressure (hypertension) called essential hypertension. Essential hypertension is a complex disorder associated with many genetic and environmental factors. The *AGT* gene variations associated with this condition affect single DNA building blocks (nucleotides) and likely lead to higher levels of the angiotensinogen protein.

## 3. Other Names for This Gene

- angiotensinogen (serpin peptidase inhibitor, clade A, member 8)
- angiotensinogen preproprotein
- ANGT\_HUMAN
- ANHU
- pre-angiotensinogen
- serpin A8
- SERPINA8

## References

1. 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.
2. Gribouval O, Morinière 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, Rivière 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, Triaux 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.

3. Gubler MC, Antignac C. Renin-angiotensin system in kidney development: renal tubular dysgenesis. *Kidney Int.* 2010 Mar;77(5):400-6. doi: 10.1038/ki.2009.423.
4. Watkins WS, Hunt SC, Williams GH, Tolpinrud W, Jeunemaitre X, Lalouel JM, Jorde LB. Genotype-phenotype analysis of angiotensinogen polymorphisms and essential hypertension: the importance of haplotypes. *J Hypertens.* 2010 Jan;28(1):65-75. doi: 10.1097/HJH.0b013e328332031a.
5. Wolf G. Angiotensin II and tubular development. *Nephrol Dial Transplant.* 2002;17 Suppl 9:48-51. Review.

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