AMHR2 gene

anti-Mullerian hormone receptor type 2

Normal Function

The *AMHR2* gene provides instructions for making the anti-Müllerian hormone (AMH) receptor type 2, which is involved in male sex differentiation. The AMH receptor type 2 is found on the surface of Müllerian duct cells. The Müllerian duct, found in both male and female fetuses, is the precursor to the female reproductive organs. During development of male fetuses, cells of the testes release a protein called the AMH protein. The AMH protein attaches (binds) to the AMH receptor type 2, which signals self-destruction (apoptosis) of the Müllerian duct cells. As a result, the Müllerian duct breaks down (regresses) in males. In females, who do not produce the AMH protein during fetal development, the Müllerian duct becomes the uterus and fallopian tubes.

Health Conditions Related to Genetic Changes

Persistent Müllerian duct syndrome

Persistent Müllerian duct syndrome type 2, a disorder of sexual development that affects males, is caused by mutations in the *AMHR2* gene. Males with this condition have female reproductive organs in addition to normal male reproductive organs. At least 24 mutations in the *AMHR2* gene have been identified in people with persistent Müllerian duct syndrome type 2. Most mutations change single protein building blocks (amino acids) in the AMH receptor type 2 protein. Other mutations result in a premature stop signal that leads to an abnormally short protein. Still other mutations delete regions of DNA from the *AMHR2* gene, which changes the instructions for the protein. The most common mutation, a deletion of 27 DNA building blocks (nucleotides), occurs in about half of affected individuals with an *AMHR2* gene mutation.

Mutations in the *AMHR2* gene lead to an abnormal protein that is stuck inside the Müllerian duct cells and not found on the surface. Therefore, the cells cannot receive the signal for apoptosis. As a result, the Müllerian duct persists and becomes a uterus and fallopian tubes. Because the AMH receptor type 2 is not involved in the formation of male reproductive organs, affected males also have male reproductive organs.

Other Names for This Gene

- AMH type II receptor

- AMHR
- AMHR2_HUMAN
- anti-Muellerian hormone type II receptor
- anti-Muellerian hormone type-2 receptor
- anti-Mullerian hormone receptor type II
- anti-Mullerian hormone receptor, type II
- MIS type II receptor
- MISR2
- MISRII
- MRII
- Mullerian inhibiting substance type II receptor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMHR2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=269[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28AMHR2%5BTIAB%5D%29+OR+%28%28AMH+type+II+receptor%5BTIAB%5D%29+OR+%28AMHR%5BTIAB%5D%29+OR+%28MIS+type+II+receptor%5BTIAB%5D%29+OR+%28MISR2%5BTIAB%5D%29+OR+%28MISRII%5BTIAB%5D%29+OR+%28MRII%5BTIAB%5D%29+OR+%28Mullerian+inhibiting+substance+type+II+receptor%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ANTI-MULLERIAN HORMONE TYPE II RECEPTOR; AMHR2 (https://omim.org/entry/600956)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/269)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AMHR2[gene])

References

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- Imbeaud S, Belville C, Messika-Zeitoun L, Rey R, di Clemente N, Josso N,Picard JY. A 27 base-pair deletion of the anti-mullerian type II receptor gene isthe most common cause of the persistent mullerian duct syndrome. Hum Mol Genet.1996 Sep;5(9):1269-77. doi: 10.1093/hmg/5.9.1269. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8872466)
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Genomic Location

The *AMHR2* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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