AMH gene

anti-Mullerian hormone

Normal Function

The *AMH* gene provides instructions for making a protein that is involved in male sex differentiation. During development of male fetuses, the AMH protein is produced and released (secreted) by cells of the testes. The secreted protein attaches (binds) to its receptor, which 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. Binding of the AMH protein to its receptor induces 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 1, a disorder of sexual development that affects males, is caused by mutations in the *AMH* gene. Males with this condition have female reproductive organs in addition to normal male reproductive organs. At least 38 mutations in the *AMH* gene have been identified in people with persistent Müllerian duct syndrome type 1. Most mutations change single protein building blocks (amino acids) in the AMH 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 *AMH* gene, which changes the instructions for the protein.

The mutated AMH protein cannot be released from the cells of the testes or cannot bind to the receptor on the Müllerian duct cells. As a result, the Müllerian duct cells never receive the signal for apoptosis. The Müllerian duct persists and becomes a uterus and fallopian tubes. Because the AMH protein is not involved in the formation of male reproductive organs, affected males also have male reproductive organs.

Other Names for This Gene

- anti-Muellerian hormone
- MIF
- MIS

- muellerian-inhibiting factor
- muellerian-inhibiting substance
- Mullerian inhibiting factor
- Mullerian inhibiting substance

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=268[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMH%5BTIAB%5D%29+OR+%28anti-Mullerian+hormone%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ANTI-MULLERIAN HORMONE; AMH (https://omim.org/entry/600957)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/268)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AMH[gene])

References

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Genomic Location

The *AMH* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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