DHH Gene

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Desert Hedgehog

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1. Normal Function

The *DHH* gene provides instructions for making a member of the hedgehog protein family. Hedgehog proteins are important for early development in many parts of the body. The protein produced from the *DHH* gene is believed to be involved in male sexual development and in the formation of the perineurium, the protective membrane around each bundle of fibers within a nerve.

2. Health Conditions Related to Genetic Changes

2.1 Swyer Syndrome

DHH gene mutations have been identified in a small number of people with Swyer syndrome, a condition affecting sexual development also known as 46,XY complete gonadal dysgenesis or 46,XY pure gonadal dysgenesis. Affected individuals have two mutated copies of the *DHH* gene in each cell.

People usually have 46 chromosomes in each cell. Two of the 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics. Girls and women typically have two X chromosomes (46,XX karyotype), and boys and men ordinarily have one X chromosome and one Y chromosome (46,XY karyotype).

Mutations in the *DHH* gene in people with Swyer syndrome affect the process of sexual differentiation, preventing affected individuals with a 46,XY karyotype from developing male gonads (testes) and causing them to develop female reproductive structures (a uterus and fallopian tubes).

2.2 Other disorders

DHH gene mutations have been identified in people with 46,XY disorder of sex development, also known as partial gonadal dysgenesis. These individuals have one mutated *DHH* gene in each cell. They may have external genitalia that do not look clearly male or clearly female (ambiguous genitalia) or other changes in the genitals and reproductive organs.

In addition to gonadal dysgenesis, some people with *DHH* mutations also have nerve abnormalities. These abnormalities affect nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat, and sound (the peripheral nervous system). Affected individuals may experience weakness and loss of sensation in their extremities (peripheral neuropathy).

3. Other Names for This Gene

- desert hedgehog homolog (Drosophila)
- DHH_HUMAN
- HHG-3
- MGC35145

References

1. Canto P, Söderlund D, Reyes E, Méndez JP. Mutations in the desert hedgehog(DHH) gene in patients with 46,XY complete pure gonadal dysgenesis. J ClinEndocrinol Metab. 2004 Sep;89(9):4480-3. Erratum in: J Clin Endocrinol Metab.2004 Nov;89(11):5453.
2. Canto P, Vilchis F, Söderlund D, Reyes E, Méndez JP. A heterozygous mutationin the desert hedgehog gene in patients with mixed gonadal dysgenesis. Mol HumReprod. 2005 Nov;11(11):833-6.
3. Das DK, Sanghavi D, Gawde H, Idicula-Thomas S, Vasudevan L. Novel homozygousmutations in Desert hedgehog gene in patients with 46,XY complete gonadaldysgenesis and prediction of its structural and functional implications bycomputational methods. Eur J Med Genet. 2011 Nov-Dec;54(6):e529-34. doi:10.1016/j.ejmg.2011.04.010.
4. King TF, Conway GS. Swyer syndrome. Curr Opin Endocrinol Diabetes Obes. 2014Dec;21(6):504-10. doi: 10.1097/MED.0000000000000113. Review.
5. Mohnach L, Fechner PY, Keegan CE. Nonsyndromic Disorders of TesticularDevelopment. 2008 May 21 [updated 2016 Jun 2]. In: Adam MP, Ardinger HH, PagonRA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews®[Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Availablefrom http://www.ncbi.nlm.nih.gov/books/NBK1547/
6. Sugie K, Futamura N, Suzumura A, Tate G, Umehara F. Hereditary motor andsensory neuropathy with minifascicle formation in a patient with 46XY puregonadal dysgenesis: a new clinical entity. Ann Neurol. 2002 Mar;51(3):385-8.
7. Umehara F, Tate G, Itoh K, Osame M. Minifascicular neuropathy: a new conceptof the human disease caused by desert hedgehog gene mutation. Cell Mol Biol(Noisy-le-grand). 2002 Mar;48(2):187-9. Review.
