MSX1 gene

msh homeobox 1

Normal Function

The *MSX1* gene provides instructions for making a protein that regulates the activity of other genes. The *MSX1* gene is part of a larger family of homeobox genes, which control the formation of many body structures during early development. Specifically, this gene is critical for the normal development of the teeth and other structures in the mouth. It may also be important for the development of fingernails and toenails.

Health Conditions Related to Genetic Changes

Wolf-Hirschhorn syndrome

The *MSX1* gene is often deleted in people with Wolf-Hirschhorn syndrome. The features of this condition include a characteristic facial appearance, delayed growth and development, intellectual disability, and seizures.

A loss of the *MSX1* gene probably disrupts the regulation of several other genes, particularly genes involved in the development of the mouth and teeth. Researchers believe that a deletion of the *MSX1* gene disrupts the formation of oral structures in early development, leading to missing teeth and other dental abnormalities. A loss of the *MSX1* gene probably also causes an opening in the roof of the mouth (cleft palate) and/or a split in the upper lip (cleft lip) seen in some people with Wolf-Hirschhorn syndrome.

Other disorders

MSX1 gene variants (also called mutations) are associated with abnormalities of mouth and tooth development. For example, changes in this gene contribute to some cases of cleft lip and/or cleft palate in people who do not have Wolf-Hirschhorn syndrome. These genetic variations have been associated with isolated cleft lip and cleft palate in several different populations worldwide. *MSX1* gene variants appear to be one of many genetic and environmental factors that play a role in oral clefting.

At least six *MSX1* gene variants are responsible for oligodontia, a condition in which multiple teeth fail to develop. Some individuals with *MSX1* gene variants have a combination of oligodontia and cleft lip and/or cleft palate. Variants in the *MSX1* gene

likely reduce the amount of functional MSX1 protein within cells, which disrupts the early development of structures in the mouth.

Another variant in the *MSX1* gene has been found to cause Witkop syndrome (also known as tooth-and-nail syndrome). This rare condition is characterized by missing teeth and abnormalities of the fingernails and toenails. The *MSX1* gene variant mutation responsible for Witkop syndrome leads to the production of an abnormally short, nonfunctional version of the MSX1 protein. A loss of this protein disrupts the formation of the teeth and nails during early development.

Other Names for This Gene

- homeobox 7
- homeobox protein MSX-1
- HOX7
- HYD1
- msh homeo box 1
- MSH Homeo Box Homolog 1 (Drosophila) Gene
- msh homeobox homolog 1
- MSX1_HUMAN
- OFC5

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MSX1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4487[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MSX1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TOOTH AGENESIS, SELECTIVE, 1; STHAG1 (https://omim.org/entry/106600)
- MSH HOMEOBOX 1; MSX1 (https://omim.org/entry/142983)
- WITKOP SYNDROME (https://omim.org/entry/189500)
- OROFACIAL CLEFT 5; OFC5 (https://omim.org/entry/608874)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4487)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MSX1[gene])

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Genomic Location

The *MSX1* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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