SOS1 Gene

Subjects: Genetics & Heredity
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SOS Ras/Rac guanine nucleotide exchange factor 1

Keywords: genes

1. Normal Function

The *SOS1* gene provides instructions for making a protein that is involved in controlling (regulating) the activation of the RAS/MAPK signaling pathway, which helps control several important cell functions. Specifically, the pathway regulates the growth and division of cells (proliferation), the process by which cells mature to carry out specific functions (differentiation), cell movement (migration), and the self-destruction of cells (apoptosis). Within the RAS/MAPK signaling pathway, the SOS1 protein regulates a protein, called Ras, that stimulates cells to grow and divide. This regulation tightly controls the growth of cells and tissues, and is especially important for proper embryonic development.

2. Health Conditions Related to Genetic Changes

2.1. Noonan syndrome

More than 55 mutations causing Noonan syndrome have been identified in the *SOS1* gene. Noonan syndrome is characterized by mildly unusual facial characteristics, short stature, heart defects, bleeding problems, skeletal malformations, and many other signs and symptoms. The *SOS1* gene mutations change single protein building blocks (amino acids) in the SOS1 protein. The resulting protein is either continuously turned on (active) or has prolonged activation, rather than promptly switching on and off in response to other cellular proteins. This increase in protein activity disrupts the regulation of the RAS/MAPK signaling pathway that controls cell functions such as growth and division. This misregulation can result in the heart defects, growth problems, skeletal abnormalities, and other features of Noonan syndrome.

2.2. Other disorders

Mutations in the *SOS1* gene can also cause hereditary gingival fibromatosis type 1. This disorder is characterized by a slowly progressive overgrowth of the tissue of the gums (gingiva). Too much of this tissue can impair teeth from emerging through the gums, which can cause difficulties in speech and chewing food. At least one mutation in the *SOS1* gene has been shown to cause hereditary gingival fibromatosis type 1. This mutation is an addition of a single building block of DNA (nucleotide). The mutation inserts the nucleotide cytosine into an area of the gene called exon 21 (written 3248_3249insC) and disrupts the gene's instructions, resulting in a shortened protein. Unlike the normal SOS1 protein, the shortened protein is permanently active because it is missing areas that regulate its activity. Instead of triggering cell growth in response to particular signals from outside the cell, the overactive protein directs cells to grow and divide constantly. It is unclear why the overgrowth of tissue is seen only in the gums.

3. Other Names for This Gene

- alternate SOS1
- GF1
- GGF1
- GINGF
- gingival fibromatosis
- gingival fibromatosis, hereditary, 1
- HGF
- son of sevenless homolog 1

- son of sevenless homolog 1 (Drosophila)
- SOS1_HUMAN

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