SLC25A24 Gene

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Contributor: Karina Chen

solute carrier family 25 member 24

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1. Normal Function

The *SLC25A24* gene provides instructions for producing a protein that is a member of the solute carrier (SLC) family of proteins. Proteins in the SLC family transport various compounds across the membranes surrounding the cell and its component parts. The protein produced from the *SLC25A24* gene transports molecules across the inner membrane of the mitochondria, the energy-producing centers of cells. This protein is known as an ATP-Mg/Pi carrier because it transports energy molecules called ATP that are attached (bound) to magnesium (Mg) atoms through the mitochondria inner membrane in exchange for adding or removing phosphate (P) atoms from the mitochondria. This exchange is essential for normal energy production, the formation and breakdown (metabolism) of various molecules, and protein production within cells.

2. Health Conditions Related to Genetic Changes

2.1. Gorlin-Chaudhry-Moss syndrome

At least two mutations in the *SLC25A24* gene have been found to cause Gorlin-Chaudhry-Moss syndrome. This condition, which has been found only in females, is characterized by skull abnormalities that affect the shape of the head and face, a lack of fatty tissue under the skin (lipodystrophy), excessive hair growth (hypertrichosis) on the face and body, shortened bones at the ends of the fingers and toes (short distal phalanges), and smaller-than-normal external female genital folds (hypoplasia of the labia majora).

The mutations that cause Gorlin-Chaudhry-Moss syndrome change a single protein building block (amino acid) in the ATP-Mg/Pi carrier protein. The mutations change the amino acid arginine at position 217 to either the amino acid histidine (written as Arg217His or R217H) or the amino acid cysteine (written as Arg217Cys or R217C). These mutations are thought to alter the structure of the protein, which likely decreases its ability to transport molecules across the mitochondrial inner membrane. As a result, there is an increase in mitochondrial size (mitochondria swelling), breakage of mitochondria into smaller pieces, and an overall decrease in ATP production. This increase in abnormal mitochondria and decrease in energy production can lead to cell death.

While altered cellular energy production and increased cell death are likely responsible for the features of Gorlin-Chaudhry-Moss syndrome, it is unclear how these changes lead to the specific signs and symptoms of the condition.

2.2. Other disorders

The same two *SLC25A24* gene mutations that cause Gorlin-Chaudhry-Moss syndrome (described above) have been found in individuals with a similar disorder known as Fontaine syndrome or Petty-type congenital progeroid syndrome. This syndrome has many of the same features of Gorlin-Chaudhry-Moss syndrome, including skull abnormalities, lipoatrophy, and short distal phalanges, but includes normal genital development and sparse hair, and affects boys and girls equally. Also, in contrast to Gorlin-Chaudhry-Moss syndrome, individuals with Fontaine syndrome typically do not survive past infancy.

It is unclear how the same mutations can lead to different disorders. Researchers suspect that variations in other genes involved in mitochondrial function may play a role as well as other genetic and environmental factors.

3. Other Names for This Gene

- APC1
- calcium-binding transporter
- mitochondrial ATP-Mg/Pi carrier protein 1
- mitochondrial Ca(2+)-dependent solute carrier protein 1
- SCAMC-1
- short calcium-binding mitochondrial carrier 1
- small calcium-binding mitochondrial carrier protein 1
- solute carrier family 25 (mitochondrial carrier; phosphate carrier), member 24

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