ATRX Gene

Subjects: Genetics & Heredity

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ATRX, chromatin remodeler

genes

1. Normal Function

The *ATRX* gene provides instructions for making a protein that plays an essential role in normal development. Although the specific function of the ATRX protein is unknown, studies suggest that it helps regulate the activity (expression) of other genes through a process known as chromatin remodeling. Chromatin is the complex of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development. When DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

The ATRX protein appears to regulate the expression of two genes, *HBA1* and *HBA2*, that are necessary for the production of hemoglobin. Hemoglobin is the protein in red blood cells that carries oxygen to cells throughout the body. Other genes regulated by the ATRX protein have not been identified.

2. Health Conditions Related to Genetic Changes

2.1. Alpha thalassemia X-linked intellectual disability syndrome

More than 125 mutations in the *ATRX* gene have been identified in people with alpha thalassemia X-linked intellectual disability syndrome. The most common mutations change single protein building blocks (amino acids) in the ATRX protein. Other mutations insert or delete genetic material in the *ATRX* gene or alter how the gene's instructions are used to make the protein.

Mutations may destabilize the ATRX protein or affect its interactions with other proteins. These changes prevent the ATRX protein from effectively regulating gene expression. Reduced activity of the *HBA1* and *HBA2* genes causes a blood disorder called alpha thalassemia. Abnormal expression of additional genes likely causes developmental delay, distinctive facial features, and the other signs and symptoms of alpha thalassemia X-linked intellectual disability syndrome.

2.2. Other disorders

Some gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes, which are not inherited, are called somatic mutations. Somatic mutations of the *ATRX* gene have been found in some people with blood and bone marrow disorders. In particular, researchers have found somatic *ATRX* gene mutations in some cases of myelodysplastic syndrome (MDS), which is a bone marrow disorder that usually occurs in older males.

Somatic *ATRX* gene mutations do not cause MDS; instead, they occur as the condition progresses. Like inherited mutations in the *ATRX* gene, somatic mutations reduce the activity of the *HBA1* and *HBA2* genes and cause alpha thalassemia. When alpha thalassemia occurs in people with MDS, the combination of disorders is often referred to as alpha thalassemia myelodysplastic syndrome (ATMDS).

3. Other Names for This Gene

- alpha thalassemia/mental retardation syndrome X-linked
- alpha thalassemia/mental retardation syndrome X-linked (RAD54 homolog, S. cerevisiae)
- ATR2
- ATRX_HUMAN
- DNA dependent ATPase and helicase
- helicase 2, X-linked
- MGC2094
- MRXHF1
- RAD54
- RAD54L
- SFM1
- SHS
- transcriptional regulator ATRX
- X-linked nuclear protein
- XH2
- XNP
- Zinc finger helicase
- ZNF-HX

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