PURA Gene

Subjects: Genetics & Heredity
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purine rich element binding protein A

genes

1. Normal Function

The *PURA* gene provides instructions for making a protein called Pur-alpha (Purα), which is able to attach (bind) to DNA and RNA (a molecular cousin of DNA). This protein has multiple roles in cells, including controlling the activity of genes (gene transcription) and aiding in the copying (replication) of DNA.

The Purα protein is important for normal brain development. The protein helps direct the growth and division of nerve cells (neurons). It may also be involved in the formation or maturation of myelin, the protective substance that covers nerves and promotes the efficient transmission of nerve impulses.

2. Health Conditions Related to Genetic Changes

2.1. 5q31.3 microdeletion syndrome

5q31.3 microdeletion syndrome is caused by a chromosomal change in which a small piece of chromosome 5 is deleted in each cell. This rare condition is characterized by severely delayed or impaired development of speech and walking, weak muscle tone (hypotonia), breathing problems, recurrent seizures (epilepsy) or seizure-like episodes, and distinctive facial features. The deletion that causes this condition occurs on the long (q) arm of the chromosome at a position designated q31.3. The size of the deletion can range from several thousand to several million DNA building blocks (base pairs). The deleted region typically contains at least three genes, one of which is *PURA*.

A loss of one copy of the *PURA* gene is thought to alter normal brain development and impair the function of neurons, leading to developmental delay, hypotonia, and other neurological problems in people with 5q31.3 microdeletion syndrome. Some studies suggest that loss of another nearby gene called *NRG2* increases the severity of the signs and symptoms. It is unclear how the loss of other genes in the deleted region contributes to development of 5q31.3 microdeletion syndrome.

2.2. PURA syndrome

At least 22 *PURA* gene mutations have been found to cause *PURA* syndrome, a condition characterized by intellectual disability, delayed development of speech and walking, and epilepsy. Some of these genetic changes remove small segments of DNA from the *PURA* gene. Others change single building blocks (amino acids) in the Purα protein or lead to production of an abnormally short protein. These mutations are thought to reduce the amount of functional Purα protein. Although it is not understood how a partial loss of Purα function leads to the signs and symptoms of *PURA* syndrome, researchers suspect that it may alter normal brain development and impair the function of neurons, leading to developmental problems and seizures in people with the condition.

3. Other Names for This Gene

- MRD31
- PUR-ALPHA
- PUR1
- PURALPHA
- purine-rich single-stranded DNA-binding protein alpha
- transcriptional activator protein Pur-alpha

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