PURA And 5q31

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PURA syndrome

PURA syndrome

Description

PURA syndrome is a condition characterized by intellectual disability and delayed development of speech and motor skills, such as walking. Expressive language skills (vocabulary and the production of speech) are generally more severely affected than receptive language skills (the ability to understand speech), and most affected individuals are unable to speak. People with PURA syndrome may learn to walk later than their peers; many are never able to walk. In infancy, affected infants have very weak muscle tone (hypotonia) and feeding difficulties. Problems with swallowing (dysphagia) can last throughout life. In addition, affected infants can be excessively sleepy (hypersomnolent), have a low body temperature (hypothermia), and have short pauses in breathing (apnea) or episodes of abnormally slow breathing (hypoventilation). These breathing problems usually go away after age 1.Recurrent seizures (epilepsy) are also common in PURA syndrome. Seizures usually begin before age 5 with uncontrolled muscle jerks (myoclonus). Other types of seizures can develop, such as generalized tonic-clonic seizures, which involve loss of consciousness, muscle rigidity, and convulsions. In people with PURA syndrome, seizures are often

difficult to control.Other features in people with PURA syndrome can include abnormalities of the heart, eyes, urogenital tract, gastrointestinal tract, and skeleton. Some affected individuals have symptoms of a hormonal problem, such as early sexual development (precocious puberty) or low levels of vitamin D (which is a hormone).

Frequency

PURA syndrome is a rare condition affecting at least 70 individuals. It is estimated to account for fewer than 1 percent of cases of developmental delay.

Causes

PURA syndrome is caused by mutations in the PURA gene, which provides instructions for making a protein called Pur-alpha (PurÎ±). This protein has multiple roles in cells, including controlling the activity of genes (gene transcription) and aiding in the copying (replication) of DNA. The PurÎ± protein is especially important for normal brain development. PurÎ± 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.Mutations in the PURA gene are thought to lead to a reduced amount of functional PurÎ± protein. Although it is not understood how a partial loss of PurÎ± 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.

Learn more about the gene associated with PURA syndrome

PURA

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.Most cases of this condition result from new (de novo) mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) in an affected individualâ€™s parent or in early embryonic development. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

PURA-related neurodevelopmental disorder PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome

Genetic and Rare Diseases Information Center

PURA syndrome

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Catalog of Genes and Diseases from OMIM

NEURODEVELOPMENTAL DISORDER WITH NEONATAL RESPIRATORY INSUFFICIENCY, HYPOTONIA, AND FEEDING DIFFICULTIES; NEDRIHF

Scientific Articles on PubMed

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