Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart

Description

Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart (NEDBEH) is a neurological disorder that can also affect many other body systems. This condition primarily affects neurological development, causing intellectual disability, delayed development of speech and motor skills (such as sitting and walking), or autism spectrum disorder, which is a condition that affects communication and social interaction. Some affected individuals have additional neurological features, such as weak muscle tone (hypotonia), behavioral problems, and seizures.

NEDBEH can affect development of many other parts of the body. Some affected individuals have abnormalities of brain structures, such as the tissue that connects the left and right halves of the brain (the corpus callosum), a tissue called white matter, the fluid-filled cavities (ventricles) near the center of the brain, or a structure at the back of the brain known as the cerebellar vermis. Eye abnormalities that can occur include a gap or hole in one of the structures of the eye (coloboma), underdevelopment (hypoplasia) or breakdown (atrophy) of the nerves that carry information from the eyes to the brain (optic nerves), or unusually small eyeballs (microphthalmia). These eye problems can cause vision impairment. Some affected individuals have heart defects, most commonly ventricular septal defect, which is a hole in the muscular wall (septum) that separates the right and left sides of the heart's lower chambers.

Less commonly, other systems are affected in NEDBEH, including the kidneys and inner ear. Problems with the inner ear can lead to hearing impairment (sensorineural hearing loss).

The signs and symptoms in some people with NEDBEH resemble those of another condition known as CHARGE syndrome; however, people with NEDBEH do not have changes in the gene associated with CHARGE syndrome.

Frequency

NEDBEH is a rare disorder with unknown prevalence. About twenty individuals with this condition have been described in the medical literature.

Causes

NEDBEH is caused by mutations in a gene called *RERE*. This gene provides instructions for making a protein that is critical for normal development before birth. It helps control the activity of a number of genes that are important for early development of the brain, eyes, inner ear, heart, and kidneys.

Researchers suspect that *RERE* gene mutations reduce or eliminate the function of the RERE protein. A shortage of RERE protein function likely alters the activity of several genes involved in development before birth. These changes prevent the normal development of tissues in the brain, eyes, heart, and other organs. Researchers are working to identify which genes are affected and how changes in their activity lead to the signs and symptoms of NEDBEH. It is unknown why some people with NEDBEH have only neurological problems and others also have structural abnormalities. Researchers suspect that the severity of the condition may be related to the location and type of mutation in the *RERE* gene. Additional genetic factors that have not been identified, including variations in other genes, may also help determine which body systems are affected.

Learn more about the gene associated with Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart

- RERE

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 individual'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

- NEDBEH
- RERE-related neurodevelopmental syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4310772/)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT ANOMALIES OF THE BRAIN, EYE, OR HEART; NEDBEH (https://omim.org/entry/616975)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28RERE%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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