HIVEP2-related intellectual disability

Description

HIVEP2-related intellectual disability is a neurological disorder characterized by moderate to severe developmental delay and intellectual disability and mild physical abnormalities (dysmorphic features). Early symptoms of the condition include weak muscle tone (hypotonia) and delayed development of motor skills, such as sitting, standing, and walking. After learning to walk, many affected individuals continue to have difficulty with this activity; their walking style (gait) is often unbalanced and wide-based. Speech is also delayed, and some people with this condition never learn to talk. Most people with *HIVEP2*-related intellectual disability also have unusual physical features, such as widely spaced eyes (hypertelorism), a broad nasal bridge, or fingers with tapered ends, although there is no characteristic pattern of such features among affected individuals. Many people with the condition exhibit neurodevelopmental disorders, such as hyperactivity, attention deficit disorder, aggression, anxiety, and autism spectrum disorder, which is a group of developmental disorders characterized by impaired communication and social interaction.

Other features of *HIVEP2*-related intellectual disability include mild abnormalities in the structure of the brain and an abnormally small brain and head size (microcephaly). Less common health problems include seizures; recurrent ear infections; and eye disorders, such as eyes that do not look in the same direction (strabismus), "lazy eye" (amblyopia), and farsightedness (hyperopia). Some people with *HIVEP2*-related intellectual disability have gastrointestinal problems, which can include backflow of acidic stomach contents into the esophagus (gastroesophageal reflux) and constipation.

Frequency

HIVEP2-related intellectual disability is a rare disorder. At least nine individuals with the condition have been described in the medical literature.

Causes

HIVEP2-related intellectual disability is caused by mutations in the *HIVEP2* gene. The protein produced from this gene is most abundant in the brain, where it controls the activity (expression) of genes involved in brain growth and development.

Mutations in the *HIVEP2* gene are thought to lead to a shortage of functional HIVEP2

protein. It is unclear how these genetic changes result in the features associated with *HIVEP2*-related intellectual disability, although researchers speculate that a shortage of the HIVEP2 protein alters the expression of several genes involved in brain growth and development. Abnormalities in the growth and development of the brain likely underlie the cognitive problems and other neurological features of *HIVEP2*-related intellectual disability. It is unclear how *HIVEP2* gene mutations contribute to the unusual physical features and health problems that can occur with this condition.

Learn more about the gene associated with HIVEP2-related intellectual disability

- HIVEP2

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

- Mental retardation, autosomal dominant 43
- MRD43

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Intellectual disability, autosomal dominant 43 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4310771/)

Genetic and Rare Diseases Information Center

- Intellectual developmental disorder, autosomal dominant 43 (https://rarediseases.info.nih.gov/diseases/13179/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22HIVEP2-related intellectual disability%22)

Catalog of Genes and Diseases from OMIM

- INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL DOMINANT 43; MRD43 (https://omim.org/entry/616977)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28HIVEP2%5BTIAB%5D%29+AND+english%5Bla%5D)

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