4H syndrome

What is 4H syndrome?

4H syndrome is short for hypomyelination, hypogonadotropic hypogonadism and hypodontia. Hypomyelination means that there is lack of myelin in the central nervous system. In hypogonadotropic hypogonadism, normal puberty development is absent because the central nervous system is not able to initiate it properly. Hypodontia means that not all teeth are present.

4H syndrome is most probably inherited in an autosomal recessive manner. This means that parents are healthy, but carry each one defective copy of the responsible gene. If a child inherits two defective copies of this gene, it will be affected. What causes 4H syndrome?

4H syndrome is a genetic disorder. Its genetic basis is however still not known. How is 4H syndrome diagnosed?

4H syndrome is diagnosed on the basis of the clinical symptoms, especially ataxia and delayed dentition or hypodontia, in combination with the results of an MRI, which shows hypomyelination (lack of myelin in the brain) and cerebellar atrophy (volume loss of the cerebellum which is a part of the brain). What are the symptoms of 4H syndrome?

At birth and during the first year of life, a child with 4H syndrome appears normal. Symptoms usually start during the second year of life, but patients with normal early childhood and symptoms only from the second decade have been described.

Neurological symptoms include:

Late walking

Early-onset ataxia (problems with balance and fine motor skills)

Deterioration of ataxia with infections with complete or partial recovery

Slow progression of ataxia over time

Dysarthria (speech is difficult to understand)

Later, development of spasticity (abnormally stiff muscles and restricted movements)

Seizures (these are rare)

Other symptoms:

Some teeth may be already present at birth (natal teeth)

Dentition (eruption of teeth) is delayed, and the first teeth to erupt are the deciduous molars, not the incisors as usual. Upper medial incisors erupt late, often after the age of 6 years. Some of the teeth, especially of the permanent teeth, may be missing (hypodontia) or are have an unusual shape.

Normal puberty development is absent.

Short stature may develop during childhood.

Myopia (short sightedness) is common.

What is the treatment for 4H syndrome?

There is no cure for 4H syndrome; treatment is supportive. Is prenatal diagnosis possible?

As long as the genetic defect has not been identified, prenatal diagnosis is not possible. How is scientific research on 4H syndrome progressing towards improved treatment or diagnosis?

Before treatment strategies can be developed, the genetic basis of the disorder must be elucidated. Genetic family studies are necessary in as many families as possible. Other names for 4H syndrome

Ataxia, hypodontia and hypomyelination (AHH)

Ataxia, delayed dentition and hypomyelination (ADDH) [1].

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http://ulf.org/4h-syndrome/