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My46 Trait Profile

Congenital stationary night blindness

Congenital stationary night blindness is a childhood eye disease resulting in difficulty seeing in darkened environments. Many affected individuals may also be nearsighted and some may have small involuntary, repetitive eye movements.

Characteristics of Congenital stationary night blindness

Congenital stationary night blindness (CSNB) is a genetic eye condition in which individuals have difficulty seeing in darkened environments. This condition is present at birth (congenital) and does not usually change over time (stationary). It is considered a rare condition, but the exact estimates of its prevalence are not known. CSNB is divided into two categories based on the normal or abnormal appearance of the back of the eye (fundus). There are currently ten genes associated with CSNB without fundus abnormalities, and three genes associated with CSNB and fundus abnormalities.

Many individuals with CSNB may also be nearsighted (myopic). Individuals with myopia do not have difficulty seeing things close up, but can have trouble seeing things in the distance, such as traffic signs and billboards. Myopia can worsen in childhood, but eventually stabilizes. Some affected individuals may also have small involuntary, repetitive eye movements known as nystagmus. Strabismus, or having eyes that do not look in the same direction, can also be present in individuals with CSNB.

The severity of the condition can vary greatly from person to person. Some individuals have a marked restriction in their ability to move around in the dark, while others may have very little limitation. The same applies to the degree of nearsightedness. Some individuals examined have a visual acuity of 20/200, which means that they can see an object clearly at 20 feet that an individual without myopia could see from 200 feet. Legal blindness is defined as less than or equal to 20/200. Others individuals examined have had a visual acuity as good as 20/30, which would means little or no shortsightedness.

Diagnosis/Testing

If an individual is from a family where other family members have CSNB, parents may be alert to early signs in their children. They may be watching for indications that their child is having more difficulty adjusting to darkened spaces as compared to other children. In other families, it may take longer to establish the diagnosis, as young children may have difficulty describing their vision concerns to their parents.

To confirm a diagnosis of CSNB, an individual needs to be examined by a medical doctor who is specialized in eye and vision care (ophthalmologist), and a visual test called an electroretinography (ERG). ERG testing evaluates cell function in the retina of the eye. Individuals with CSNB have decreased cell function and would have a characteristic ERG result (commonly reported as selective reduction in the amplitude of the b-wave).

Genetic testing can confirm the clinical diagnosis of CSNB by detecting a change or mutation in CSNB-related genes.

Management/Surveillance

Annual eye exams with refraction by an ophthalmologist at an early age are important. This may help identify and correct for myopia as early as possible. Conventional strabismus surgery is available to improve vision or an unusual

head posture. Myopia and night blindness may lead to restrictions to an affected individual's driving license or prevent the individual from driving altogether.

Mode of inheritance

CSNB may be inherited in one of three patterns of inheritance: X-linked recessive, autosomal recessive, and autosomal dominant.

X-linked recessive inheritance:

CSNB is most often inherited in an X-linked recessive pattern. The gene mutations causing this type of inheritance are found on the X chromosome. An X-linked recessive pattern means that in females, both copies of a gene (i.e., one on each X chromosome) must have a change or mutation, whereas in males, only one copy of a gene must have a mutation to be affected. A female with a mutation in one copy of a gene on the X chromosome is said to be a "carrier" for an X-linked condition, and is typically not affected.

The X-linked form can be separated into complete and incomplete CSNB based on retinal function determined by ERG. Both result in overlapping symptoms but with no fundus changes. Changes or mutations in the NYX gene cause complete X-linked CSNB. Mutations in the CACNA1F gene cause incomplete X-linked CSNB.

Autosomal recessive inheritance:

Autosomal recessive inheritance accounts for a small proportion of CSNB. This inheritance pattern means that an individual has to inherit two mutations (i.e., one from each parent) to be affected. If both parents are carriers of a mutation they have a 1 in 4 (25%) chance with each pregnancy of having a child with the condition.

Mutations in the GRM6, CABP4, CACNA2D4, SLC24A1 and TRPM1 genes cause autosomal recessive CSNB without fundus changes whereas mutations in the RDH5, SAG and GRK1 genes cause autosomal recessive CSNB with fundus changes.

Autosomal dominant inheritance:

Autosomal dominant inheritance accounts for a small proportion of CSNB. This means inheriting one mutation is enough for an individual to be affected and show signs of CSNB. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Mutations in the GNAT1, RHO and PDE6B genes cause autosomal dominant CSNB without fundus abnormalities.

Risk to family members

The risk to family members depends on the pattern of inheritance.

X-linked recessive inheritance:

If a father is affected with CSNB, his daughters will be carriers of CSNB and his sons will be unaffected. If a mother is a carrier of CSNB, each daughter has a 1 in 2 chance (i.e., 50%) of being a carrier and each son has a 1 in 2 chance (i.e., 50%) of being affected with CSNB.

Autosomal recessive inheritance:

Parents of a child with CSNB are carriers of CSNB. If a sibling of a child with CSNB is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of CSNB.

Autosomal dominant inheritance:

The risk to family members depends on whether or not the individual with CSNB has a parent affected with CSNB. If a parent also has the condition, the risk of having a child with CSNB is 50% with each pregnancy. If a parent does not have CSNB, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Genetics Home Reference: X-linked Congenital Stationary Night Blindness
http://ghr.nlm.nih.gov/condition/x-linked-congenital-stationary-night-blindness
Genetics Home Reference: Autosomal Recessive Congenital Stationary Night Blindness
http://ghr.nlm.nih.gov/condition/autosomal-recessive-congenital-stationary-night-blindness
Genetics Home Reference: Autosomal Dominant Congenital Stationary Night Blindness

http://ghr.nlm.nih.gov/condition/autosomal-dominant-congenital-stationary-night-blindness

References

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