CADASIL: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy

What is CADASIL?

CADASIL is an inherited condition that causes strokes. In individuals with CADASIL, the muscular walls of blood vessels do not work properly, and this causes strokes, migraines, and seizures. CADASIL can also cause damage to the white matter in the brain causing leukoencephalopathy. CADASIL is inherited and passed on through the NOTCH3 gene.

Symptoms of CADASIL

Individuals with CADASIL often have multiple strokes, which damages the brain causing dementia. Walking and facial movements, such as chewing and speaking, can also be affected. Strokes can occur in childhood but mostly begin in adulthood between the ages of 40 and 50 years. Individuals with CADASIL frequently have migraines with aura and changes to their mood. By age 65, most individuals with CADASIL have severe cognitive problems and are dependent on others for care.

How is the NOTCH3 gene inherited?

Everyone has two copies of most of their genes: one inherited from their mum and one from their dad. CADASIL occurs when one copy of the NOTCH3 genes has a change in the gene instructions. This gene change stops the NOTCH3 gene from working properly. This means CADASIL is a dominant condition because most individuals with CADASIL inherit one NOTCH3 gene change from one of their parents who is also affected. Very rarely, an individual with CADASIL has a new NOTCH3 gene change that is not inherited.

Are family members at risk of CADASIL?

Most individuals with CADASIL will have one parent who is also affected. These individuals' siblings have a 50% chance of also having inherited the NOTCH3 gene change and developing CADASIL. Individuals with CADASIL have a 50% chance of passing on the NOTCH3 gene change to each of their children.

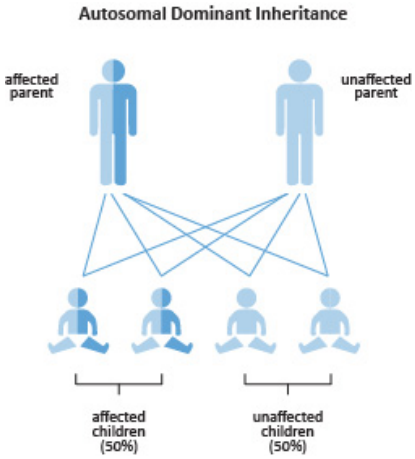

Figure 1. Diagram of autosomal dominant inheritance of NOTCH3 gene change

Source: https://www.jnetics.org/jewish_genetic_disorders/introduction_to_genetics/ accessed on 13/10/2019

What does the NOTCH3 gene do?

The NOTCH3 gene makes a protein receptor that is important in the function and survival of the smooth muscle cells that surround blood vessels. Changes in the NOTCH3 gene instructions mean the protein receptor produced does not work properly and results in the death of the smooth muscle cells surrounding blood vessels.

Treatment for CADASIL

There is no specific treatment or cure for CADASIL. Management of symptoms and yearly follow up with a neurologist who specialises in CADASIL is recommended. Support for individuals with CADASIL and their family members is recommended including genetic counselling.

Contact Us

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References

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