VANISHING WHITE MATTER – VWM

What is Vanishing White Matter?
Vanishing White Matter (VWM) is a condition of the nervous system, that causes neurological symptoms. VWM is also called childhood ataxia with cerebral hypomyelination (CACH), and leukoencephalopathy with vanishing white matter.

What are the EIF2B1, EIF2B2, EIF2B3, EIF2B4 and EIF2B5 genes?
Mutations in the EIF2B1, EIF2B2, EIF2B3, EIF2B4, and EIF2B5 genes cause leukoencephalopathy with vanishing white matter. These genes are responsible for providing the instructions for making 5 parts of the eIF2B protein. Mutations in the eIF2B protein cause it to lose function, and make it more difficult for the body's cells to regulate protein synthesis, particularly during stressful conditions such as fever and infection.

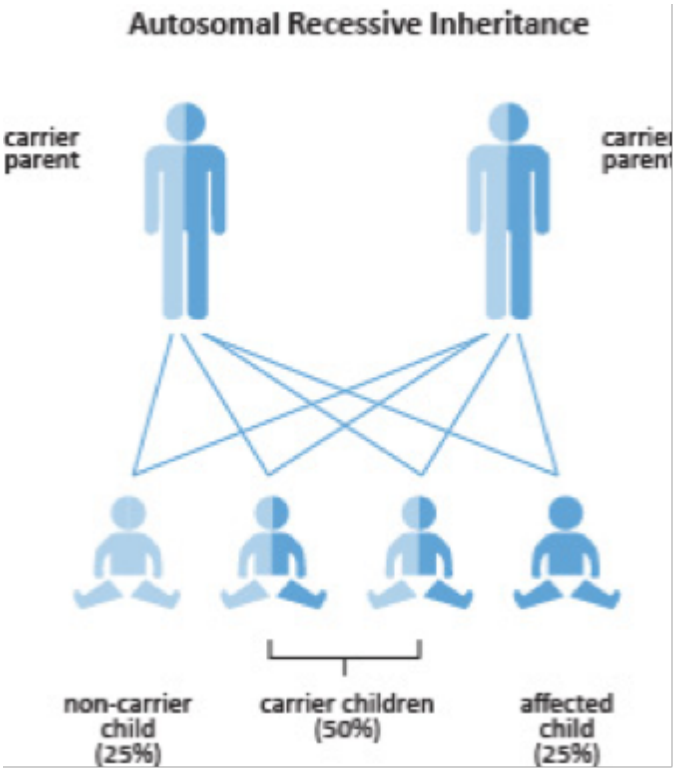
What are the symptoms of VWM?
The age of onset of VWM is highly variable, with some children showing symptoms early on, and other individuals first symptoms appearing in adulthood.
Symptoms are also variable, and can include;

- Neurological deterioration
- Episodes of fever. In VWM, this is associated with symptoms worsening, drowsiness or coma.
- Muscles spasms or involuntary contractions. Muscles are abnormally stiff and movement is restricted.
- Lethargy and abnormal drowsiness
- Coma
- Abnormal development of the ovaries in females
- Loss of muscle coordination as a result of abnormal functioning of the cerebellum (a part of the brain). This is also called cerebellar ataxia.
- Abnormalities and difficulties with the eyes.
- Seizures
- Mental impairment may be present, though generally less severe than the motor dysfunction

The motor difficulties of VWM are progressive, but the progression is often associated with fever or injuries.

How is VWM diagnosed?
VWM is diagnosed when there is a combination of symptoms in an individual, and further investigations such as a brain MRI show typical characteristics of VWM. Genetic testing for VWM will look for changes (or mutations) on both copies of the EIF2B1, EIF2B2, EIF2B3, EIF2B4, and EIF2B5 genes.

Are family members at risk of VWM?
VWM is an autosomal recessive disorder. Individuals who have one copy of the gene change on any of the five identified genes do not typically shown any signs or symptoms, and are considered as carriers of VWM. Both parents must be carriers of the condition for it to be passed on and for symptoms to be present in their child. Each pregnancy has a 1 in 4 chance of inheriting the condition.



Can VWM be treated?

There is no cure for VWM. Treatment is currently symptomatic and supportive, focussing on individual therapies for symptoms. This may include medication for seizures and the stiffness or abnormal muscle contractions that are a problem for many VWM patients.

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