Ornithine Transcarbamylase Deficiency (OTC)

Ornithine transcarbamylase deficiency is caused by mutations in the OTC gene. This gene encodes for the enzyme ornithine transcarbamylase, which carries out a key step in the urea cycle. The liver alters toxic ammonia and converts it to urea, a much safer is converted into urea, a much more neutral compound. If the enzyme is partially or wholly inactivated, damaging levels of ammonia will tend to build up in the body. The condition is much more common in males than females. Symptoms often occur within the first few days of life. They include poor feeding, muscle weakness, lethargy, seizures, and hyperventilation. Severe hypothermia and brain damage result if prompt treatment is not started. Dialysis and nitrogen scavenger compounds, such as sodium benzoate, can be used to remove ammonia from the body. Even when ammonia levels appear to be under control, a crisis can appear in which they become elevated again. Low protein diets are needed throughout life. Infants may even require a liver transplant. A late-onset form of the disease can commence later in life, sometimes triggered by injuries, operations, or starting a high protein diet. Typical symptoms include mental problems, headaches, and vomiting.

The incidence of the disease is roughly 1 in 70,000 births, occurring in roughly 4,300 patients in the USA. There does not seem to be huge differences in its occurrence among different ethnic groups. The faulty gene resides on the X chromosome, also known as an X-linked disease. Unlike females, any male with the faulty gene will have the disease since males only have a single X chromosome. The severe version of the disease is very rare in females, since they would need two faulty genes, which is highly unlikely. Females with one faulty gene normally act as carriers with no symptoms, however 15% of them will show some symptoms during their lifetime. As the disease is linked to the X chromosome, affected fathers cannot pass it on to their sons. Their daughters of affected fathers will normally receive the faulty gene.. Mothers with the faulty gene, whether they are asymptomatic or not, have a 50% chance of passing it on to each child.

Sources

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