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Opinion Article

Inherited Peroxisomal Disorder: Clinical and Metabolic Perspectives on Refsum Disease

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DESCRIPTION

Refsum disease is a rare inherited metabolic condition that affects the body's ability to break down a specific type of fatty acid known as phytanic acid. This fatty acid is obtained mainly through dietary sources such as dairy products, certain meats, and fish, as well as from chlorophyll-containing plant material indirectly through animal feed. In individuals with Refsum disease, the normal metabolic pathway responsible for degrading phytanic acid is impaired, leading to its accumulation in blood and tissues. This buildup gradually affects multiple organ systems, particularly the nervous system, vision, skin, and skeletal structure.

The condition is usually inherited in an autosomal recessive manner, meaning that an affected individual receives one altered gene from each parent. The genetic changes most commonly involve the PHYH gene or, less frequently, the PEX7 gene. These genes are associated with peroxisomal function, specifically the breakdown of branched-chain fatty acids. When their activity is reduced or absent, phytanic acid cannot be processed effectively and begins to accumulate over time. Symptoms typically appear during adolescence or early adulthood, although the timing can vary depending on diet and individual metabolic differences.

One of the earliest signs of the disorder is often related to vision. Individuals may develop progressive night blindness due to degeneration of the retina. As the condition advances, peripheral vision may also decline, sometimes resulting in significant visual impairment. The accumulation of phytanic acid affects retinal cells and disrupts their normal function, leading to gradual loss of photoreceptor activity. Hearing loss is another common feature and may begin subtly before becoming more noticeable over time.

Neurological symptoms are also frequently observed. These may include weakness in the limbs, reduced reflexes, difficulty with coordination, and sensory disturbances such as tingling or numbness. The peripheral nerves are particularly vulnerable to metabolic disruption, which can result in polyneuropathy. In some cases, individuals may experience difficulty with balance due to impaired nerve signaling. These neurological effects tend to progress slowly but can become more pronounced if dietary intake of phytanic acid is not controlled.

Diagnosis of Refsum disease involves a combination of clinical observation, biochemical testing, and genetic analysis. Elevated levels of phytanic acid in blood plasma are a key diagnostic indicator. Additional tests may include nerve conduction studies, ophthalmological examinations, and auditory assessments to evaluate the extent of organ involvement. Genetic testing can confirm mutations in the relevant genes and help differentiate this condition from other similar metabolic disorders.

Management of Refsum disease primarily focuses on reducing phytanic acid levels in the body. Since phytanic acid is derived from dietary intake, dietary modification plays a central role in treatment. Individuals are advised to limit or avoid foods that contain high levels of this compound. This includes certain dairy products, ruminant fats, and specific types of fish. By controlling dietary intake, the accumulation of phytanic acid can be slowed, which may help stabilize symptoms over time.

CONCLUSION

Living with Refsum disease requires continuous attention to dietary habits and medical follow-up. Early detection and consistent management can significantly influence long-term outcomes. Individuals who maintain controlled phytanic acid levels often experience slower progression of symptoms compared to those with delayed diagnosis or inconsistent dietary regulation. Research into metabolic disorders like Refsum disease continues to explore improved methods for diagnosis and management. Advances in genetic analysis have made it easier to identify affected individuals earlier in life, which can support timely dietary adjustments. Ongoing studies also aim to better understand lipid metabolism pathways and their role in human physiology.

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