Fabry's Disease

Fabry’s disease is caused by a deficiency of the enzyme alpha galactosidase A, which causes accumulation of ceramide trihexoside in blood vessels and other organs. Unlike the majority of other sphingolipidoses, this disease is inherited in an X-linked recessive fashion. Symptoms include angiokeratomas, cardiovascular disease, and peripheral neuropathy. Kidney complications are especially common in Fabry’s disease. End stage renal failure typically occurs in the third decade and is a common cause of death in Fabry’s disease.



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Pathophysiology

X-linked Recessive
X-suit with Recessive-chocolate
This disease is inherited in an X-linked recessive pattern. This means boys are more likely to have disease.

Alpha-galactosidase A Deficiency
Broken Afro-galaxy-side-A
Enzyme deficient in Fabry's disease that usually breaks down ceramide trihexoside.

Increased Ceramide trihexoside
Up-arrow Sewer-bride with Tri-hexagons
Substance accumulated in this disease due to deficient enzyme.

Early Signs and Symptoms

Decreased Sweating (Hypohidrosis)
Down-arrow Antiperspirant
Patients with Fabry's disease display decreased sweating, or hypohidrosis. This presents in childhood or adolescence, and has been attributed to selective peripheral nerve damage, or to intracytoplasmic lipid deposits in the small blood vessels surrounding sweat glands.

Angiokeratoma
Angel-carrot
Angiokeratomas are benign, red scaly papules that represent enlarged vascular spaces in the top layer of the dermis. In addition, there is thickening of the overlying epidermis with hyperkeratosis. In Fabry's disease, angiokeratomas can be extensive and spread all over the body.

Episodic Peripheral Neuropathy
Purple-wavy Neuron-extremities
This is episodic pain or tingling in extremities caused by damage to nerve fibers from accumulation of ceramide trihexoside.

Late Signs and Symptoms

Cardiovascular Disease
Heart Diseased
Cardiac complications occur when ceramide trihexoside builds up in heart cells. Hypertension and cardiomyopathy are commonly observed.

Renal Failure
Dead Kidney
Kidney complications are common in Fabry's. Proteinuria is often the first sign of kidney involvement. End stage renal failure typically occurs in third decade and is common cause of death due to the disease.

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