Tay-Sachs Disease

Tay Sachs Disease is caused by a genetic mutation in the Hexosaminidase A gene, which causes accumulation of GM2 ganglioside in cells. There is an increased disease prevalence in the Eastern European Ashkenazi Jewish population. On histologic exam, neurons are ballooned with cytoplasmic vacuoles due to markedly distended lysosomes filled with gangliosides, which resemble an onion skin appearance. On physical exam, a cherry red macula can be appreciated. The disease also causes progressive neurodegeneration due to accumulation of the GM2 ganglioside and patients display progressive deterioration of mental and physical abilities at around 6 months of age.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
Tay-Sachs Disease is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be present in order for the disease to develop.

Hexosaminidase A Deficiency
Broken Hexagon-mini-As
Hexosaminidase A (also known as beta hexosaminidase A or HEXA) is an enzyme which is severely deficient in Tay-Sachs. This enzyme is responsible for breaking down GM2 ganglioside.

Increased GM2 Ganglioside
Up-arrow GM (2) Tutu car with Gangster-on-side
This is the specific type of ganglioside that accumulates in Tay-Sachs.

More Common in Ashkenazi Jewish Population
Yarmulke (Yamaka)
There is increased disease prevalence in the Eastern European Ashkenazi Jewish population.

Signs and Symptoms

Cherry Red Spot on Macula
Cherry-eyes
On fundoscopic examination, a small red spot is seen in the center of the macula of the retina. It is caused by accumulation of GM2 ganglioside in the retina and the relative transparency of the macula. This is seen in several different lipid storage diseases.

Neurodegeneration
Degenerating-nerve-arm
Progressive deterioration of the central nervous system due to accumulation of GM2 ganglioside.

Developmental Delay
Developmental-bus
Progressive deterioration of mental and physical abilities is seen at around 6 months of age due to harmful quantities of gangliosides that accumulate in the brain's nerve cells, leading to premature cell death.

Diagnosis

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Onion Skin Lysosomes

Onion-head

On histologic exam, neurons are ballooned with cytoplasmic vacuoles that constitute a markedly distended lysosome filled with gangliosides. On electron microscope, several cytoplasmic inclusions make the appearance of an onion skin.

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