

Biochemistry team flash cards

Sphingolipids

Is essential component of membranes and plays a role in growth regulation and cell transformation.

Sphingosine is the backbone of sphingolipids

Myelin

Composed mostly of lipids and some proteins ..

Produced by

Schwann cells in **peripheral nerves**.

Oligodendrocytes **within CNS**.

Sphingolipidoses

Lysosomal lipid storage diseases ,,
When the degradation of specific sphingolipid is defected it accumulates at the organ

Usually only a single sphingolipid accumulates in the involved organs in each disease

Sphingolipidoses

Diagnosed by :

1. **measuring the enzyme activity** :
Cultured fibroblasts or peripheral leukocytes
Cultured aminocytes (Prenatal)
2. **Histological examination**
3. **DNA analysis**

Sphingolipidoses

Tay-sachs disease :

The deficient enzyme:

β -Hexosaminidase (α subunit)

The accumulated lipid

Gangliosides (Gm2)

Clinical features : Blindness, muscular weakness & seizures .

Sphingolipidoses

Gaucher disease :

The deficient enzyme:

β -glucosidase (glucocerebrosidase)

The accumulated lipid

Glucocerebrosides

Clinical features : Hepatosplenomegaly, Osteoporosis of long bones and some other features

Sphingolipidoses

Niemann-pick disease :

The deficient enzyme:

Sphingomyelinase

The accumulated lipid

Sphingomyelin

Clinical features : It has two types (A & B)

A : Fatal disease where enzyme activity is reduced

B : "Chronic" less severe and has later onset