Lipid storage diseases.

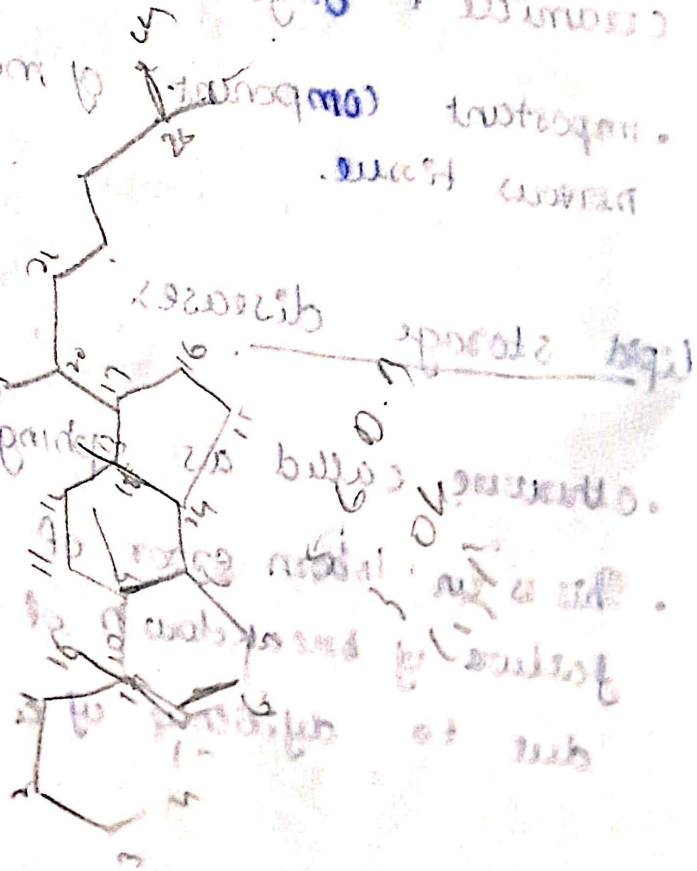
- Otherwise called as sphingolipidoses.
- This is an Inborn error of metabolism result from failure of breakdown of a particular sphingo lipid due to deficiency of a single enzyme.

- It leads to accumulation of complex lipids in CNS.
- The children affected by this disease are mentally retarded.

Impt mark:

eg: ① Niemann - pick disease.

- due to sphingomyelinase deficiency.
- sphingomyelin will accumulate.



Clinical features-

- Severe CNS damage
- mental retardation
- Hepato splenomegaly.
- cherry red spot in macula:

2). Tay - Sach's disease. { hexosaminidase A deficiency. not amina

3.) Gaucher's disease. { β glucosidase deficiency }.