Mucopolysaccharidoses

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- MPSs: genetic disorders result from accumulation of mucopolysaccharides in tissues
- mucopolysaccharides: - a gel like substance, found in:
    1- body cells
    2- mucous secretions
    3- synovial fluids
- synthesized by connective tissue fibroblasts
- degraded within lysosomes
- Several enzymes are involved in their degradation, the lack of these enzymes leads to accumulation of mucopolysaccharides within the lysosomes
- The mucopolysaccharides that accumulate within the tissues include dermatan sulfate, heparan sulfate, keratan sulfate, and chondroitin sulfate

- there are 7 types of MPSs (MPS I to MPS VII) each resulting from the deficiency of one specific enzyme.

- **In general the MPSs**: - are progressive disorders
- characterized by involvement of many organs, including the liver, spleen, heart, and blood vessels
- Most of them are associated with:

1- coarse facial features

2- clouding of the cornea

3- joint stiffness

4- mental retardation

5- organomegaly

- affected children have short life expectancy

- Urinary excretion of the accumulated mucopolysaccharide is often increased
- treatment: enzyme and gene replacement therapy

- All of MPSs are autosomal recessive except Hunter syndrome, which is an X-linked recessive disease

1) Hurler syndrome: - MPSs type I

- caused by a deficiency of α-l-iduronidase
- associated with accumulation of dermatan and heparan sulfate
- affected children have a life expectancy of 6 to 10 years
- patients have: 1- coarse facial features
  2- clouding of cornea
  3- skeletal deformities
  4- mental retardation
  5- organomegaly
- Death is often due to cardiac complications resulting from the deposition of mucopolysaccharides in the coronary arteries and heart valves

Hurler (coarse facial features, organomegaly)

Hurler (coarse facial features, skeletal deformities, corneal clouding)

2) Hunter syndrome: - MPSs type II
- X-linked recessive disorder
- results from a deficiency of L-iduronate sulfatase
- associated with accumulation of heparan and dermatan sulfate
- patients have: 1- coarse facial features
2- progressive joints stiffness
3- organomegaly
4- mental retardation
5- No corneal clouding

Hunter

Thank you