Mucopolysaccharidosis II. type

- L-iduronosulfate sulfate defatase deficiency [1]

Diseases characterized by the following features:

- accumulation of heparan sulfate
- men only (GR inheritance)
- without corneal opacity + thoracolumbar kyphosis [2]
- OMIM: +300900 (https://omim.org/entry/309900)

heavy form (A)

- starts between the ages of 1 and 3, has faster progression and people with disabilities die by the age of 15, often due to heart failure
- macrocephalus, prominent forehead, wide nose, hypertrophic gums, malformed teeth, macroglossia
- short neck, prominent abdomen as a consequence of hepatosplenomegaly
- hearing impairment
- dementia
- cardiomegaly, coronary artery stenosis

light form (B)

- manifestations often begin at a younger school age and the disability can live to be 50 years old
- slow growth, flexion holding fingers of hands that bother while typing retinitis pigmentosa
- retinitis pigmentosa
- normal intellect
- frequent hearing loss

Links

Bibliography

1. *HYÁNEK, Josef, et al. *Dědičné metabolické poruchy.* 1.. edition. Praha : Avicenum, 1990. vol. 342. ISBN 80-201-0064-4.
2. DUNGL, P., et al. *Ortopedie.* 1. edition. Praha : Grada Publishing, 2005. ISBN 80-247-0550-8.

- HYÁNEK, Josef, et al. *Dědičné metabolické poruchy.* 1.. edition. Praha : Avicenum, 1990. vol. 342. ISBN 80-201-0064-4.

Related articles

- Mucopolysaccharidosis
- Hereditary disorders of sugar metabolism
- Achondroplasia ■ Tanatophoric dwarfism ■ Diastrophic dysplasia ■ Larsen syndrome

External links

- Handbook of Genetic Counseling/Mucopolysacharidosis (https://en.wikibooks.org/wiki/Handbook_of_Genetic_Counseling/Mucopolysacharidosis_%28MPS%29)
- National MPS Society (https://mpssociety.org/)