Mucopolysaccharidosis IV. type

- ***synonyms:*** dysostosis enchondralis epimetaphysaria, m. Morquio-Brailsford [1]
- accumulation of **keratan sulfate** and **chondroitin sulfate**
- significant skeletal impairment (dwarfism with shortened torso, thoracolumbar kyphosis, genua valga)
- limbs noticeably long compared to the short spine, the patient rests his hands on his thighs [2]
- intellect unaffected [3]
- OMIM 253010 (https://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=253010) OMIM 25300 (https://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=253000) OMIM 252300 (https://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=252300)

Links

Bibliography

1. SOSNA, A. – VAVŘÍK, P. – KRBEC, M.. *Základy ortopedie.* 1. edition. Praha : Triton, 2001. ISBN 80-7254-202-8.
2. POVÝŠIL, Ctibor – ŠTEINER, Ivo, et al. *Speciální patologie.* 2.. edition. Praha. 2007. vol. 297-299. ISBN 978-80-7262-494-2.
3. DUNGL,. *Ortopedie.* 1. edition. Praha : Grada Publishing, 2005. ISBN 80-247-0550-8.

Related articles

- Mucopolysacharidosis
- 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/)