Opis choroby *

Definicja

A rare genetic disease characterized by early-onset respiratory difficulties and frequent respiratory infections, congenital heart defects, dysostosis multiplex, hepatosplenomegaly, renal involvement, hematopoietic abnormalities, facial dysmorphism (coarse facial features, large forehead, synophrys, long eyelashes, broad nasal bridge, macroglossia, short neck, and low hairline), and global developmental delay. Laboratory examination shows increased urinary excretion of glycosaminoglycans and increased plasma heparan sulfate, but no lysosomal enzyme deficiency. The disease is usually fatal in the first years of life.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mucopolysaccharidosis-like plus disease
Mucopolysaccharidosis-like plus disease

Kod ORPHA
505248

Kod OMIM
617303

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet