Opis choroby *

Definicja

A rare disorder with multisystemic involvement and glomerulopathy characterized by progressive steroid-resistant nephrotic syndrome typically associated with focal segmental glomerulosclerosis, as well as primary adrenal insufficiency with adrenal calcifications. Age of onset and disease course are variable, with some cases presenting as severe fetal hydrops, while most patients present in infancy or early childhood and progress to end-stage renal disease within a few years. Additional features include ichthyosis, primary hypothyroidism, hypogonadism, immunodeficiency, and neurological manifestations (such as cognitive impairment, ataxia, sensorineural hearing loss, or seizures).

Dane

Klasyfikacja
Choroba

Synonimy
Primary adrenal insufficiency-steroid-resistant nephrotic syndrome due to SGPL1 deficiency
Pierwotny zespół nerczycowy oporny na sterydy z powodu niedoboru SGPL1

Kod ORPHA
506334

Kod OMIM
617575

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet