

Opis choroby *

Definicja

Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome is a rare, genetic, mitochondrial disease characterized by early-onset progressive renal failure, manifesting with hyperuricemia, hyponatremia, hypomagnesemia, hypochloremic metabolic alkalosis, elevated BUN and polyuria, associated with systemic manifestations which include pulmonary hypertension, failure to thrive, global developmental delay, hypotonia and ventricular hypertrophy. Additional features include prematurity, elevated serum lactate, diabetes mellitus and, in some, pancytopenia.

Dane

Klasyfikacja

Choroba

Synonimy

HUPRA syndrome

Zespół HUPRA

Kod ORPHA

363694

Kod OMIM

613845

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet