## 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

Synonimy

Choroba

HUPRA syndrome Zespół HUPRA

**Kod ORPHA** 363694

Kod OMIM

**Kod ICD10** 

613845 E88.8

**Kod ICD11** 

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## \*Źródło

orphanet