

## 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