Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome. ORPHA:363694

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.