Hypoxanthine-guanine phosphoribosyltransferase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypoxanthine-guanine phosphoribosyltransferase deficiency. ORPHA:206428

Hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency is a hereditary disorder of purine metabolism associated with uric acid overproduction and a continuum spectrum of neurological manifestations depending on the degree of the enzyme deficiency.