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Phosphoribosylpyrophosphate synthetase superactivity

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Phosphoribosylpyrophosphate synthetase superactivity. ORPHA:3222

Phosphoribosylpyrophosphate (PRPP) synthetase superactivity is an X-linked disorder of purine metabolism associated with hyperuricemia and hyperuricosuria, comprised of two forms: an early-onset severe form characterized by gout, urolithiasis, and neurodevelopmental anomalies (severe PRPP synthetase superactivity) and a mild late-onset form with no neurologic involvement (mild PRPP synthetase superactivity) (see these terms).