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Mild phosphoribosylpyrophosphate synthetase superactivity

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. Mild phosphoribosylpyrophosphate synthetase superactivity. ORPHA:411536

Mild phosphoribosylpyrophosphate (PRPP) synthetase superactivity is the mild and late-onset form of PRPP synthetase superactivity (see this term), an X-linked disorder of purine metabolism associated with hyperuricemia and hyperuricosuria, leading to urolithiasis and gout. This form is not associated with any neuropathy or central nervous system (CNS) disorders.