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Severe phosphoribosylpyrophosphate synthetase superactivity

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Severe phosphoribosylpyrophosphate synthetase superactivity. ORPHA:411543

Severe phosphoribosylpyrophosphate (PRPP) synthetase superactivity is the severe and early-onset form of PRPP synthetase superactivity (see this term), an X-linked disorder of purine metabolism associated with hyperuricemia and hyperuricosuria, that is characterized by urolithiasis, gout and neurodevelopmental anomalies.