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# Purine nucleoside phosphorylase deficiency

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Purine nucleoside phosphorylase deficiency. ORPHA:760*

Purine nucleoside phosphorylase (PNP) deficiency is a disorder of purine metabolism characterized by progressive immunodeficiency leading to recurrent and opportunistic infections, autoimmunity and malignancy as well as neurologic manifestations.