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Purine-Nucleoside Phosphorylase Deficiency

National Cancer Institute

Source

National Cancer Institute. *Purine-Nucleoside Phosphorylase Deficiency*. NCI Thesaurus. Code C3963.

PNP reversibly catalyzes the phosphorolysis of the purine nucleosides, (deoxy)inosine and (deoxy)guanosine, to their respective purine bases and the corresponding ribose-1-phosphate. Deficiency in this enzyme is an autosomal recessive cause of combined immunodeficiency.