Adenine phosphoribosyltransferase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Adenine phosphoribosyltransferase deficiency. ORPHA:976

Adenine phosphoribosyltransferase (APRT) deficiency is a rare autosomal recessive (AR) disorder characterized by the formation and hyperexcretion of 2,8-dihydroxyadenine (2,8-DHA) in urine, causing urolithiasis and crystalline nephropathy.