Ribose-5-P isomerase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ribose-5-P isomerase deficiency. ORPHA:440706

Ribose-5-P isomerase deficiency is an extremely rare, hereditary, disorder of pentose phosphate metabolism characterized by progressive leukoencephalopathy and a highly increased ribitol and D-arabitol levels in the brain and body fluids. Clinical presentation includes psychomotor delay, epilepsy, and childhood-onset slow neurological regression with ataxia, spasticity, optic atrophy and sensorimotor neuropathy.