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PGM1-CDG

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. PGM1-CDG. ORPHA:319646*

A rare, genetic, congenital disorder of glycosylation and glycogen storage disease characterized by a wide range of clinical manifestations, most commonly presenting with bifid uvula with or without cleft palate at birth, associated with growth delay, hepatopathy with elevated aminotransferase serum levels, myopathy (including exercise-related fatigue, exercise intolerance, muscle weakness), intermittent hypoglycemia, and dilated cardiomyopathy and/or cardiac arrest, due to decreased phosphoglucomutase 1 enzyme activity. Less common manifestations include malignant hyperthermia, rhabdomyolysis, and hypogonadotropic hypogonadism with delayed puberty.