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PGM3-CDG

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. *PGM3-CDG*. ORPHA:443811

PGM3-CDG is a rare congenital disorder of glycosylation caused by mutations in the PGM3 gene and characterized by neonatal to childhood onset of recurrent bacterial and viral infections, inflammatory skin diseases, atopic dermatitis and atopic diatheses, and marked serum IgE elevation. Early neurologic impairment is evident including developmental delay, intellectual disability, ataxia, dysarthria, sensorineural hearing loss, myoclonus and seizures.