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Dopa-responsive dystonia due to sepiapterin reductase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dopa-responsive dystonia due to sepiapterin reductase deficiency. ORPHA:70594

Dopa-responsive dystonia (DRD) due to sepiapterin reductase deficiency (SRD) is a very rare neurometabolic disorder characterized by dystonia with diurnal fluctuations, axial hypotonia, oculogyric crises, and delays in motor and cognitive development.