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Aromatic L-amino acid decarboxylase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Aromatic L-amino acid decarboxylase deficiency. ORPHA:35708

Aromatic L-amino acid decarboxylase deficiency is a very rare, severe, genetic neurometabolic disorder associated with clinical manifestations related to underproduction of serotonin and dopamine, mainly hypotonia, hypokinesia, ptosis oculogyric crises, and signs of autonomic dysfunction.