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Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome. ORPHA:73246*

Visceral neuropathy-brain anomalies-facial dysmorphism-developmental delay syndrome is characterised by facial dysmorphology, neuropathic visceral dysmotility, neurogenic megacystis, intracerebral calcifications and developmental delay. It has been described in two siblings (brother and sister) born to consanguineous parents. The girl also had microcephaly and multicystic kidneys. The boy had a more extensive neuropathic visceral disorder, leading clinically to chronic intestinal pseudo-obstruction syndrome (CIPO).