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Infantile spasms-broad thumbs syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. [Infantile spasms-broad thumbs syndrome](#). ORPHA:3173*

Infantile spasms-broad thumbs syndrome is a rare neurologic disorder characterized by profound developmental delay, facial dysmorphism (i.e. microcephaly, large anterior fontanel, hypertelorism, downslanting palpebral fissures, beaked nose, micrognathia), broad thumbs and flexion and/or extension spasms. Bilateral cataracts, hypertrophic cardiomyopathy and hydrocele have also been reported. EEG shows hypsarrhythmic features and MRI may reveal partial agenesis of the corpus callosum, mild brain atrophy and/or ventriculomegaly. There have been no further descriptions in the literature since 1990.