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# Lethal polymalformative syndrome, Boissel type

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Lethal polymalformative syndrome, Boissel type. ORPHA:210144*

Lethal polymalformative syndrome, Boissel type is a rare, genetic, lethal, multiple congenital anomalies/dysmorphic syndrome characterized by failure to thrive, severe developmental delay, severe postnatal microcephaly, frequent congenital cardiac defects and characteristic facial dysmorphism (including coarse face with anteverted nostrils, thin vermillion, prominent alveolar ridge and retro- or micrognathia). Additional common features include neurologic abnormalities (hyper-/hypotonia, sensorineural deafness, hydrocephalus, cerebral atrophy, seizures), as well as brachydactyly, cutis marmorata and genital anomalies.