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# Lethal hydranencephaly-diaphragmatic hernia syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Lethal hydranencephaly-diaphragmatic hernia syndrome. ORPHA:480528*

Lethal hydranencephaly-diaphragmatic hernia syndrome is a rare, genetic, lethal, multiple congenital anomalies syndrome characterized by hydranencephaly and diaphragmatic hernia, as well as macrocephaly, a widely open anterior fontanel, scaphoid abdomen and hypotonia. Additionally, congenital heart defects, polyhydramnios and pulmonary hypertension have also been associated.