

DISEASE:**Lethal hydranencephaly-diaphragmatic hernia syndrome**

NAME:	Lethal hydranencephaly-diaphragmatic hernia syndrome
DESCRIPTION:	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.
ORPHACODE:	480528
XREF(S):	<u>Orphanet</u>
ANALYTE(S):	<u>PLAT</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/2713>

RELATED CONTENT

Related Genetic Tests

- [Trombosis - Hemostasis \(gene panel\)](#)

Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

Related Analytes

- [plasminogen activator, tissue type](#)

Related Gene Panels

- [Trombosis - Hemostasis \(107 genes\) - KUL](#)

Source URL: <http://gentest.healthdata.be/disease/2713>