Minerva Ginecol. 2003 Jun;55(3):253-7.

[Prenatal diagnosis of congenital diaphragmatic hernia: an update]

[Article in Italian]

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Congenital diaphragmatic hernia (CDH) has an incidence of approximately 1:4000 live births. Most frequently the diaphragmatic defect is a left and posterolateral (Bochdalek) one. Prenatal diagnosis is made at ultrasonography; the relevant sonographic features will be described in the paper. Cystic adenomatoid malformation of the lung (CAML), pulmonary sequestration, bronchogenic cysts, pulmonary hypoplasia/agenesia need to be considered in differential diagnosis. In some cases, diagnosis of CDH is not possible "in utero": in such cases, herniation of abdominal viscera into the thorax takes place presumably just at delivery through a small diaphragmatic defect. CDH may be associated with intrauterine growth retardation (IUGR), chromosomal abnormalities (3%) and/or other malformations (10-50%): such as Central Nervous System, digestive, cardiac and urogenital anomalies. Therefore, search of associated malformations and amniocentesis with analysis of fetal karyotype are mandatory, whenever a CDH is diagnosed. CDH is still at present characterised by a high mortality (reportedly, about 45%). Many prognostic factors have been correlated to postnatal outcome of CDH: some of them are valuable prenatally by ultrasonography. However, the role of sonography in the prediction of neonatal outcome is still controversial: in particular, although many ultrasonographic parameters have been proposed, prenatal evaluation of pulmonary hypoplasia (a crucial factor related to postnatal survival) has not proved to be very accurate so far. Nevertheless, it is undisputable that prenatal diagnosis itself represents a crucial prognostic factor for CDH, since it allows birth of the affected fetuses in 3d level Perinatologic Centres provided with a Neonatal Intensive Care Unit and Neonatal Surgery.

PMID: 14581871 [PubMed - indexed for MEDLINE]