Diaphragmatic Hernia, Congenital

**Alternative Names**
- Diaphragmatic Hernia 1
- DIH1
- Hernia, Congenital Diaphragmatic
- HCD
- Diaphragmatic Hernia
- CDH
- Diaphragmatic Defect, Congenital
- Diaphragm, Unilateral Agenesis of
- Hemidiaphragm, Agenesis of
- Diaphragm, Complete Agenesis of

**Record Category**
- Disease phenotype

**WHO-ICD**
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations and deformations of the musculoskeletal system

**Incidence per 100,000 Live Births**
- 11-50

**OMIM Number**
- 142340

**Mode of Inheritance**
- Multifactorial; some autosomal recessive cases

**Gene Map Locus**
- 15q26.1

**Description**
Congenital diaphragmatic hernia is an abnormal opening in the diaphragm which causes part of the abdominal organs to move up into the chest cavity. The malformation occurs before birth during fetal development. Diaphragmatic hernia occurs with a frequency of 1/3,000 births with a survival rate of 50% of the children born alive. There are several types of congenital diaphragmatic hernia, which includes Bochdalek, Morgagni and central (septum transversum) diaphragmatic hernia.

Herniation through the foramen of Morgagni is rare, accounting for less than 5% of all types of congenital diaphragmatic hernia and has unique features in terms of clinical presentation and a high incidence of associated anomalies. Morgagni’s hernia can be asymptomatic, discovered accidentally during the evaluation of other non-related symptoms, or can be present with repeated attacks of chest infection or vague gastrointestinal symptoms. During infancy it can cause severe respiratory symptoms. Morgagni’s hernia is caused when normal development of the diaphragm and of the digestive tract do not occur; there is an improper fusion of structures during fetal development. In Morgagni’s hernia, an opening on the right side of the diaphragm is formed that causes the liver and intestines to move up into the chest cavity.

The rarity of Morgagni’s hernia as well as the vagueness, variability, and nonspecificity of symptoms contribute to delayed diagnosis, especially if the child is not adequately investigated. Clinical awareness of this entity among physicians caring for children needs to be emphasized.

**Molecular Genetics**
Although most cases of congenital diaphragmatic hernia are idiopathic, chromosomal abnormalities have been implicated in approximately 15% of cases. Some reports indicated either de novo deletion or unbalanced translocations involving the 15q24-q26 region, suggesting that this region is critical to normal development of the diaphragm. The myocyte-specific enhancer factor-2 (MEF2) proteins play a critical role in the control of muscle differentiation and development. Several research groups proposed MEF2A, a member of the MEF2 gene family mapping to 15q26, as a candidate gene.

**Epidemiology in the Arab World**

**Bahrain**
Ras Romani et al. (1982) described two unusual presentations of congenital posterolateral Bochdalek diaphragmatic hernia. The first case
was a two month old Bahraini male infant, who presented with vomiting of 24-hours duration. Physical examination showed a well-nourished baby, with very high pulse rate (200/min), respiratory rate (82/minute), and body temperature (102 degrees). The patient had a severely distented, tympanic, and tender abdomen. Abdominal X-rays suggested intraperitoneal perforation with diffuse peritonitis, and chest X-rays confirmed presence of a soft tissue mass in the lower posterior part of the left chest. The patient underwent laparotomy, which revealed a posterolateral diaphragmatic hernia with perforated gangrenous stomach. Partial gastrectomy was performed on the patient. The second case was that of a one-year old Bahraini female who complained of vomiting and lose motions of 24-hours duration. She later developed coughing, and became severely dyspneic and cyanotic. Chest X-ray showed pneumonitis as well as multiple cystic lesions on the left side. Barium study was not helpful. However, a left thoracotomy was performed on the patient, which confirmed the posterolateral hernia, and revealed herniation of the intraperitoneal contents into the patient’s chest. This sudden herniation was explained by Ras Romani et al. (1982) as being either due to a sudden increase in the negative intrapleural pressure sucking the abdominal contents into the chest, or due to an increase in the intra-abdominal pressure pushing the contents into the chest. The patient’s post-operative recovery was uneventful.

Egypt
Ras Romani et al (1982) described an unusual presentation of congenital posterolateral Bochdalek diaphragmatic hernia in a two month old Bahraini male infant, born to an Egyptian mother [See: Bahrain > Ras Romani et al., 1982].

Elhanalby and Abo Sikeena (2002) studied the various patterns of delayed presentation of patients with congenital diaphragmatic hernia and analyzed pitfalls in the diagnosis and treatment of these patients. Thirty-three children with congenital diaphragmatic hernia were treated between 1993 and 2000. Thirteen had a Bochdalek hernia and 2 had a Morgagni hernia. The patients had a relatively high frequency of right-sided involvement and the presence of a hernia sac. Elhanalby and Abo Sikeena (2002) concluded that: (1) late-presenting congenital diaphragmatic hernia should be included in the differential diagnosis of any child with persistent GI or respiratory problems associated with an abnormal chest X-ray film; (2) nasogastric tube placement must be considered as an early diagnostic or therapeutic intervention when the diagnosis is suspected; and (3) GI contrast studies should be a part of the diagnostic work-up of these patients.

[See also: Kuwait > Farag et al., 1994].

Jordan
[See: Kuwait > Farag et al., 1994].

Kuwait
In two separate studies, Farag et al. (1989 and 1994) described Arab families with congenital diaphragmatic defects. The first study described two male infants, born to consanguineous parents of Jordanian origin in Kuwait. The first child was born with an extensive, left sided diaphragmatic hernia, with the hernial sac containing the stomach, the intestines extending to the splenic flexure, the spleen, the left liver lobe, and the left testis. The child died following surgical repair within a day of birth. The couple's second child also died immediately after birth, following respiratory distress and acidosis. In his case, autopsy revealed left hemidiaphragmatic aplasia, with the left hepatic lobe, spleen, stomach, and most of the intestine herniated into the stomach cavity. The next pregnancy ended in a spontaneous abortion. At the time of reporting, the couple had one healthy female child, and the mother was pregnant. In their second study, Farag et al. (1994) described two affected families. The first family had a female child born to healthy, second cousin Kuwaiti parents. The child had progressive respiratory distress and an extensive left diaphragmatic hernia, and died a few hours after birth. Autopsy revealed agenesis of the left hemi-diaphragm, thoracic placement of stomach and spleen, herniation of intestinal loops and the left hepatic lobe, hypoplastic left lung, and shifted mediastinum. The parents in the second family were first cousins of Egyptian. Their child died 20 minutes after birth. In her case, autopsy revealed bilateral extensive posterolateral Bochdalek-type hernia, with herniation of the right hepatic lobe into the right hemithorax and the left hepatic lobe in the left side with the stomach, spleen, and intestinal loops. In both the families, two earlier born infants had died with a similar condition, without any necropsy being performed. Farag et al. (1994) concluded that these cases support an autosomal recessive mode of inheritance, and that high recurrence risk estimates needed to be considered when counseling Arab parents with a history of babies with extensive Bochdalek hernia or diaphragmatic agenesis.

Oman
Thakral and Sajwani (1998) reported a combination of right sided congenital diaphragmatic hernia (CDH) and esophageal atresia (EA) with tracheoesophageal fistula (TEF) in a male neonate.
The baby, born with an apgar score of six and seven at one and five minutes, respectively, presented at birth with respiratory distress and frothy secretions. There was no family history of congenital malformations. A chest radiograph showed bowel loops in the right thorax with mediastinal shift to the left, a naso-gastric tube in the upper esophageal pouch, and a distended stomach in the abdomen. The patient was managed surgically via right sided laparotomy to reduce and repair the hernia which consisted of small and large bowel (markedly distended), and the right lobe of the liver. The EA and TEF were repaired via a right thoracotomy approach. The CDH was repaired through the abdomen. A wide-based Meckel’s diverticulum was found but was left untouched. The right lung was one-third of normal size. Postoperatively, the baby was kept ventilated and paralyzed for 5 days. A contrast study on the 8th postoperative day revealed no anastomotic leak. Feeding was started and the chest tube was removed on the 9th day. The baby was discharged at 2 weeks of age when he was feeding well and was gaining weight. At 2 months of age he was seen in an outpatient clinic, and was well.

Thakral and Sajwani (1998) suggested their patient could be the first survivor with a right CDH and EA. They also related the survival of the patient to the early management before hypoxic and aspiration related complications could occur. They therefore, recommended early operation in such cases, managing both anomalies.

Sawardekar (2005) conducted a study to establish the prevalence of major congenital malformations in children born during a 10-year period in an Omani hospital in Nizwa. Of the 21,988 total births in the hospital, 15 children were born with Diaphragmatic Hernia.

Saudi Arabia
Al-Salem et al. (1990) reported three cases of infantile hypertrophic pyloric stenosis who had concomitant congenital diaphragmatic hernia.

Al-Hathal et al. (1998) conducted a retrospective analysis of all neonates with respiratory distress at birth due to congenital diaphragmatic hernia (CDH) who were treated with neither extracorporeal membrane oxygenation nor nitric oxide being used. Analysis included all neonates with CDH and respiratory distress at birth who were treated between August 1, 1992 through March 1, 1997. There were 21 patients, 11 male and 10 female. There were 17 full-term and 4 premature infants and surgery was performed in 18 infants. Al-Hathal et al. (1998) reported that one infant died during preoperative stabilization from severe pulmonary hypoplasia and pulmonary hypertension and one infant died postoperatively from the same conditions. Seventeen of 19 infants (89.5%) survived and were discharged home. Three infants (17.6%) who failed to thrive due to severe gastroesophageal reflux (GER) required fundoplication. Eleven infants (64.7%) who had sepsis proven by blood culture responded satisfactorily to appropriate antibiotics.

Al-Salem et al. (2002) reviewed 15 cases of Morgagni’s hernia from Saudi Arabia (Qatif Central Hospital) and United Arab Emirates (Tawam Hospital) that were treated for 15 years (1982-1998); comprising 11% of a total of 135 children with different types of congenital diaphragmatic hernia. The majority (12, 80%) had repeated chest infections. The fact that Morgagni’s hernia has been reported in identical twins and knowing its frequent association with other congenital anomalies, such as Down Syndrome, raises the possibility of an inherited defect. Associated anomalies were present in 10 (66.7%) patients: 4 (26.7%) had malrotation, 4 (26.7%) congenital heart disease, and 3 (20%) Down’s syndrome (Al-Salem et al., 2002).

United Arab Emirates
Nawaz et al. (2000) treated 52 children with different types of congenital diaphragmatic hernia of which 5 (9.6%) had Morgagni's hernia. There were 2 infants and 3 children including one with Down's syndrome. All suffered from repeated attacks of chest infection, and only after a chest X-ray was the diagnosis of Morgagni’s hernia suspected. In 2 cases this appeared as an opacity in the anterior mediastinum adjacent to the pericardium; diagnosis was confirmed by barium enema in one and a CT-scan in the other. The remaining 3 cases showed anterior herniation of bowel loops on chest X-ray which was bilateral in one. This bilaterality was confirmed pre-operatively by CT scan. Associated anomalies were present in all cases, including 2 with malrotation. All patients were treated surgically via a transabdominal approach. The number of cases treated in the United Arab Emirates indicates a relative high frequency of Morgagni's hernia in the country [See also: Saudi Arabia > Al-Salem et al. (2002)].

Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January 1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Sporadic conditions accounted for 26% of the cases. In their study, Al Talabani et al. (1998) observed six cases of congenital diaphragmatic hernia in families from
the United Arab Emirates. Recurrence was not reported in other members of the families. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities in the whole United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi.

References

Related CTGA Records
Down Syndrome
Pulmonary Hypoplasia
Pyloric Stenosis, Infantile Hypertrophic 1
Tracheoesophageal Fistula with or without Esophageal Atresia

External Links
http://www.lpch.org/DiseaseHealthInfo/HealthLibrary/diaphragm.html
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=2140

Contributors
Pratibha Nair: 7.8.2011
Pratibha Nair: 14.2.2008
Eiman Ibrahim: 14.4.2007
Ghazi O. Tadmouri: 10.1.2007
Pratibha Nair: 8.10.2006
Sarah Al-Haj Ali: 22.5.2005
Ghazi O. Tadmouri: 14.5.2005
Ghazi O. Tadmouri: 2.1.2005