Hydrops Fetalis
Second Qatar National Obstetric Ultrasound Training Course
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Objectives

• Definition and underlying pathophysiology and aetiology of NIHF
• Work up for NIHF
• Appropriate evaluation when fetal hydrops is detected
• Maternal risks and obstetric complications associated with NIHF
• Prognosis of NIHF
• Management of NIHF

Disclosures

• NONE

• Have you ever seen Hydrops fetalis?

• Is it difficult to diagnose Hydrops fetalis?
Definition

- Hydrops fetalis is a Greek term that describes pathological fluid ("ὕδωρ," Greek for water) accumulation in fetal soft tissues and serous cavities.

The features are detected by ultrasound

- are defined as the presence of 2 abnormal fluid collections in the fetus.
- These include ascites, pleural effusions, pericardial effusion, and generalized skin edema (defined as skin thickness >5 mm).
- Other frequent sonographic findings include placental thickening (typically defined as a placental thickness 4 cm in the second trimester or 6 cm in the third trimester) polyhydramnios
Nonimmune hydrops fetalis (NIHF)

- Refers specifically to cases NOT caused by red cell alloimmunization.
- With the development and widespread use of Rh(D) immune globulin, the prevalence of Rh(D) alloimmunization and associated hydrops has dramatically decreased.
- As a result, NIHF now accounts for almost 90% of cases of hydrops, with the prevalence in published series reported as 1 in 1700-3000 pregnancies.

Pathogenesis

- An imbalance in the regulation of fluid movement between the vascular and interstitial spaces, with an increase in interstitial fluid production or a decrease in lymphatic return.

Aetiology

- The precise pathogenesis of NIHF depends on the underlying aetiology.
The differential diagnosis is extensive, and the success in identifying a cause partially depends on the thoroughness of efforts to establish a diagnosis.

Although older studies considered many cases to be idiopathic, more recent, larger series and a systematic review report that a cause can be found in nearly 60% of cases prenatally and in 85% when postnatal detection is included.

Cardiovascular abnormalities

- Is the most common cause of NIHF
- NIHF can result from cardiac structural abnormalities, arrhythmias, cardiomyopathy, cardiac tumors, or vascular abnormalities.
- The prognosis of NIHF due to cardiac structural abnormalities is poor, with combined fetal and infant mortality reported as 92%, largely due to the severity of the heart defects that cause in utero congestive heart failure.

Chromosomal abnormalities

- Chromosomal abnormalities, particularly Turner syndrome (45,X) and Down syndrome (trisomy 21), are also common causes of NIHF, accounting for 13% in a large systematic review.
- In prenatal series, aneuploidy is the most common cause of NIHF, particularly when identified early in gestation.
- Turner syndrome is associated with 50-80% of cases of cystic hygromas, which result from a lack of communication between the lymphatic system and venous drainage in the neck.
Fetal anemia

- Fetal anemia, which can result in immune hydrops if caused by blood group alloimmunization
- Can also lead to NIHF. Etiologies include
  1. Inherited conditions such as hemoglobinopathies
  2. Acquired conditions, such as hemolysis, fetomaternal hemorrhage, parvovirus infection, or red cell aplasia.

Fetal infection

- NIHF has been reported in association with a number of viral, bacterial, and parasitic infectious diseases, including parvovirus, cytomegalovirus, syphilis, and toxoplasmosis.
- In most series, such infections account for 5-10% of NIHF.

Fetal thoracic abnormalities

- Fetal thoracic abnormalities, including masses as well as congenital hydrothorax, can also be associated with NIHF.
- The most frequent pulmonary lesion associated with NIHF is a congenital pulmonary airway malformation (CPAM).
Complicated MC twins

• Twin-twin transfusion syndrome results from an imbalance in blood flow caused by anastomoses in the placentas of monochorionic twin. Hydrops more commonly the recipient twin is affected, likely due to hypervolemia and increased central venous pressure.
• Cases of twin-twin transfusion sequence with hydrops have a very poor prognosis without treatment
• Another complication of monochorionic twinning that may result in NIHF is twin-reversed arterial perfusion sequence.


Structural urinary and gastrointestinal abnormalities

• Structural urinary and gastrointestinal abnormalities are less common causes of NIHF. A ruptured bladder or renal collecting system may cause urinary ascites and mimic NIHF.
• Congenital nephrotic syndromes have been reported to cause NIHF due to hypoproteinemia. Surviving infants may have massive proteinuria at birth and develop renal failure in childhood.

Masliah E. et al. Pediatr Dev Pathol 2002

Structural urinary and gastrointestinal abnormalities

• Few primary abnormalities of the gastrointestinal tract have been associated with NIHF.
• Those that have been reported include diaphragmatic hernia, midgut volvulus, gastrointestinal obstruction, jejunal atresia, malrotation of the intestines, and meconium peritonitis.

Randenberg AL et al. Neonatal Netw 2010

Neoplastic diseases or fetal tumors

• Neoplastic diseases or fetal tumors can occur in utero and have been associated with NIHF.
• Relatively common in this category are lymphangiomas, hemangiomas, sacrococcygeal, mediastinal, and pharyngeal teratomas, and neuroblastomas.
• Many of these are very vascular and lead to NIHF due to high output cardiac failure.

**Placental and cord lesions**

- Placental and cord lesions that have been associated with NIHF include chorioangiomas, angiomyxoma of the cord, aneurysm of the umbilical artery, cord vein thrombosis, umbilical vein torsion, true knots, and amniotic bands.

**Skeletal Dysplasias**

- A large number of skeletal dysplasias have been associated with NIHF, including achondroplasia, achondrogenesis, osteogenesis imperfecta, osteopetrosis, thanatophoric dysplasia, short-rib polydactyly syndrome, and asphyxiating thoracic dysplasia.

**Inborn Errors of Metabolism and other genetic conditions**

- Inborn errors of metabolism and other genetic conditions are historically associated with 1-2% of cases of NIHF, which may be transient or manifest as isolated ascites.
- Inherited metabolic disorders that have been implicated as a cause of NIHF are most typically lysosomal storage diseases such as various mucopolysaccharidoses, Gaucher disease, and Niemann-Pick disease.

**Inborn errors of metabolism and other genetic conditions**

- Although such disorders are a relatively uncommon cause of NIHF, they are important because of the high recurrence risk of these mainly autosomal recessive disorders.
Inborn errors of metabolism and other genetic conditions

- A number of other syndromes have been associated with NIHF.
- Many of these are disorders associated with lymphatic dysfunction, such as Noonan and multiple pterygium syndrome, both of which frequently present with cystic hygroma. Familial recurrence in some of these cases suggests a hereditary maldevelopment of lymphatic vessels.

What is the appropriate evaluation when fetal hydrops is detected?

- The diagnostic challenge is to establish the etiology and determine the appropriate therapy (if available) and timing of delivery.
- It is especially important to rule out potentially treatable conditions, as well as genetic disorders with a risk of recurrence in future pregnancies.
- Management is guided by the presence or absence of additional anomalies.

Maternal and Obstetric Implications of Hydrops Fetalis

- Mirror Syndrome
- Polyhydramnios (maternal respiratory symptoms)
- Preterm birth.
What is the prognosis of NIHF?

• The prognosis of NIHF depends on:
  • the underlying etiology,
  • gestational age at detection and delivery,
  • Apgar scores,
  • extent of resuscitation in the delivery room,
  • and whether the newborn requires transport.

Management of NIHF

• The cornerstone of counseling and management for this condition is a thorough evaluation for the underlying etiology of the hydrops.
• Pregnancy management decisions will depend on the etiology, in particular whether there is a treatable cause and the gestational age that NIHF develops or is first identified.

Cases generally fall into 1 of 3 categories:

• 1- those amenable to fetal therapy
• 2- those with a lethal prognosis
• 3- and cases in which the etiology is idiopathic and the prognosis is likely poor but uncertain.

Cont-Management of NIHF

• It is important in counseling that the potential for maternal complications with expectant management be anticipated, including mirror syndrome.
• Serial evaluation of maternal blood pressure is therefore recommended.
What are the fetal therapy options available for NIHF?

Fetuses with NIH may be candidates for antepartum surveillance if:

• (1) the underlying etiology of the hydrops is not considered lethal,
• (2) the pregnancy has reached a viable gestational age, and
• (3) the findings from surveillance would be used to assist with timing of delivery. In such cases, deterioration of testing results or worsening of the sonographic findings of hydrops might prompt delivery.

When is the optimal timing of delivery?

• There are no management trials of delivery timing in the setting of NIHF upon which to base recommendations.
• Many hydropic fetuses succumb prior to viability. There is no evidence that elective preterm delivery will improve the outcome.
• delivery is recommended in most cases if mirror syndrome develops.

What is the optimal mode of delivery?

• Prior to delivery of the hydropic fetus, consideration should be given to whether drainage of a large effusion may improve the efficacy of neonatal resuscitative efforts.
• Rarely, effusions may be so large as to pose a risk for trauma to the infant during delivery. potential for dystocia at delivery.
• Cesarean section is considered if potentially treatable condition
• If a decision has been made not to intervene for fetal indications to provide comfort care only, vaginal delivery is preferred unless otherwise contraindicated.
**TAKE HOME MESSAGE**

- Diverse aetiology
- Positive consanguinity. Think AR
- The presence of hydrops is a poor prognostic indicator for perinatal survival
- Management and intervention are dictated by the underlying disease process and the gestational age at detection.

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