

# A Rare Case of Fetal Hydrothorax treated through Amniocentesis and Thoracentesis

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## Abstract

We present the rare case of a 27-year-old Caucasian female which was admitted in our service at 27 weeks of gestation after being discovered with left fetal hydrothorax during routine prenatal ultrasound. Samples of maternal blood and amniotic fluid (through amniocentesis) were obtained in order to establish the cause (immune or non-immune) and fetal thoracentesis was performed in order to allow for lung re-expansion. Analysis of the amniotic and pleural fluids through QF-PCR and amniocyte cultures established the diagnosis of Down syndrome and the decision was taken to perform cesarean section at 33 weeks of gestation following two weeks treatment with dexamethasone.

**Key words:** Hydrothorax, Amniocentesis, Thoracentesis, Down Syndrome

## Introduction

Fetal hydrothorax refers to a collection of fluid within the left fetal thoracic cavity as a result of leakage or generalized fluid retention from a variety of causes which can be organized into one of the following diagnostic categories: cardiovascular (21.7%), idiopathic (17.8%), hematologic (10.4%), syndromic (4.4%), chromosomal (13.4%), lymphatic dysplasia (5.7%), infections (6.7%), thoracic (6%) and others (urinary tract malformations, extra-thoracic tumors, placental, gastrointestinal) [1]. The condition mandates urgent referral to a maternal fetal medicine specialist for extensive testing because some situations must be considered prenatal emergencies after 18 weeks and one should not wait for complete results before initiating minimally invasive diagnostic procedures and treatment [2].

## Case Report

A 27-year-old Caucasian female, pregnant for the first time, with negative triple test (AFP, uE3, hCG), was

referred at 27 weeks' gestation for a routine prenatal ultrasound. The couple had normal general health and was not consanguineous and there was no family history of genetic disorders.

Routine ultrasonography revealed normal amniotic fluid measurements and a single fetus with a large hydrothorax (~200ccm) collapsing the left lung and pushing down the diaphragm (Fig. 1). Investigations for determining the cause (non-immune or immune) of the hydrothorax were carried out and amniocentesis, followed by thoracentesis, were performed.

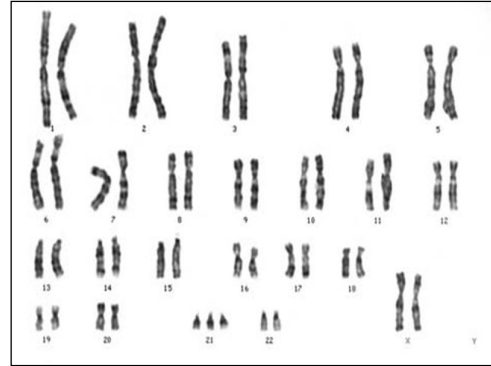
From the maternal blood acute phase titers were checked for syphilis, rubella parvovirus, toxoplasmosis, cytomegalovirus, rubella and antibody screening through indirect Coombs was performed.

The amniotic fluid (sample of 40ml) was tested for possible infection with parvovirus B19, toxoplasma, cytomegalovirus by use of PCR, while amniocyte cultures and QF-PCR were conducted in order to detect a possible genetic disorders.

Manuscript received: 24<sup>th</sup> Nov 2014  
Reviewed: 6<sup>th</sup> Dec 2014  
Author Corrected: 17<sup>th</sup> Dec 2014  
Accepted for Publication: 28<sup>th</sup> Dec 2014



**Fig 1:** 2D ultrasound showing 27 weeks fetus with hydrothorax



**Fig. 2:** Karyotype obtained from amniocyte culture showing Trisomy 21

For thoracentesis we used the same sampling kit used for amniocentesis and a sample of 70ccm pleural fluid was obtained at 28 weeks of gestation by using ultrasound guidance. Post procedural ultrasound examination revealed positive lung expansion.

Karyotype and QF-PCR from both amniotic fluid and thoracic fluid indicated the presence of Trisomy 21 (Fig. 2) and the decision was taken to perform a cesarean section at 33 weeks of gestation following a two weeks treatment with dexamethasone. Postoperative period was uneventful and the newborn developed normally, weekly ultrasound examinations showing decreasingly amounts of pleural fluid with complete resorption observed at 8 weeks after birth.

## Discussion

Despite extensive investigations, the etiology of fetal hydrothorax remains unknown in 15% to 25% of patients [3,4] while chromosomal abnormalities are the cause of fetal hydrothorax in 25% to 70% of cases [5]. Standard fetal chromosome analysis is indicated in all cases of fetal hydrothorax and early referral to a maternal–fetal specialist allows for detailed and comprehensive ultrasound examination and the early identification of any treatable causes [6]. A careful search for structural fetal anomalies or genetic syndromes, signs of fetal infection, and evidence of umbilical cord or placental anomalies may rapidly indicate the cause of fetal hydrothorax while fetal karyotyping and genetic microarray molecular testing should be conducted in all cases of unexplained hydrothorax. Cytogenetic laboratories can provide a fast but limited results within 24 to 48 hours by using QF-PCR (amniotic fluid) or more detailed results by karyotyping (amniotic fluid, fetal blood). Also, amniotic fluid should also be obtained for viral and bacterial cultures, viral PCR tests, and karyotyping [7].

Although cases of spontaneous resolution have been cited for fetal hydrothorax [8,9], we took the decision to perform amniocentesis with thoracentesis since the perinatal mortality of untreated fetal hydrothorax is still high [10] and some others have reported complete resolution after single thoracentesis [11,12,13].

## Conclusions

Even though the use of prenatal techniques (amniocentesis) for the treatment of fetal hydrothorax is still debatable with different authors reporting higher survival rates for either the conservative management [9] or the prenatal interventions [13,14, 15], we do advocate that each case of fetal hydrothorax should be investigated for genetic disorders by use of minimally invasive techniques such as chorionic villus sampling or amniocentesis and that improved access to specialized maternal–fetal medicine centers for fetal evaluation and treatment can improve outcome.

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**How to cite this article?**

Dinu-Florin Albu, Cristina-Crenguta Albu, Stefan-Dimitrie Albu, Mihai Dumitrescu. A Rare Case of Fetal Hydrothorax treated through Amniocentesis and Thoracentesis. *Int J Med Res Rev* 2015;3(1):118-120. doi:10.17511/ijmrr.2015.i1.20

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