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Case Report

A Unique Case of Trisomy 18 with Hypothyroidism, Chylothorax and Chylous Ascite.

Dr. Ayhan Ceri*1, Dr. Fatma Hilal Yilmaz[2]

1. Dr. Ayhan CERI, pediatrician. Dr. Ali Kemal Belviranlı Maternity and Children's Hospital, Health Ministry of Turkey. Neonatal Intensive Care Unit.

2.Dr. Fatma Hilal YILMAZ, Associate Professor and Neonatologist Dr. Ali Kemal Belviranlı Maternity and Children's Hospital, . Health Ministry of Turkey. Neonatal Intensive Care Unit.

***Correspondence to:** Dr. Ayhan Ceri, T.C. Saglik Bakanligi Dr. Ali Kemal Belviranli Kadin-Dogum ve Cocuk Hastanesi. Yeni Istanbul Cd. No:30, 42285 Horozluhan Osb/Selçuklu/Konya, Turkey.

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ABSTRACT

Background*: Trisomy 18, or Edwards syndrome, is a chromosomal anomaly characterized by multiple congenital abnormalities, including heart defects, facial dysmorphisms, and limb anomalies. While endocrine disorders are less common, they can manifest as part of the syndrome. Chylothorax, an accumulation of chyle in the pleural space, is a rare complication in Trisomy 18. Similarly, hypothyroidism is an uncommon endocrine disorder in these patients.*

Clinical Description*: We report the case of a premature infant with Trisomy 18 who presented with respiratory distress and characteristic morphological features of the syndrome, including microcephaly, low-set ears, and limb abnormalities. Further evaluation revealed a wide atrioventricular septal defect and patent ductus arteriosus. Additionally, the infant exhibited hypothyroidism with significantly elevated thyroid-stimulating hormone levels. Subsequent fluid accumulation in the abdomen and right lung area led to the diagnosis of chylothorax, confirmed by elevated triglyceride levels in the evacuated fluid.*

Management & Outcome*: The patient received medical support for cardiac failure and closure of the ductus arteriosus. Thyroid hormone replacement therapy was initiated for hypothyroidism. Despite aggressive management, including octreotide therapy for chylothorax, the patient experienced a poor clinical course. Multiple attempts at fluid evacuation were made, but the infant's condition continued to deteriorate, ultimately resulting in death on the 25th day of hospitalization.*

Conclusion*: This case underscores the complexity of managing Trisomy 18, especially when complicated by rare comorbidities such as hypothyroidism and chylothorax. It highlights the importance of considering endocrine disorders in patients with Trisomy 18 and the challenges in addressing multiple medical issues simultaneously. Further research and documentation of such cases are necessary to improve understanding and management strategies for this rare genetic syndrome.*

Keywords: *Trisomy 18, hypothyroidism, chylothorax, congenital anomalies, medical management.*

Introduction

Trisomy 18, as the name itself suggests, is an autosomal genetic disorder characterized by the presence of an extra chromosome with the presence of 3 copies of chromosome 18. Newborns with trisomy 18 often have anomalies in the heart, lung and kidney mostly. In addition, these babies are usually present at birth with intrauterine growth retardation (IUGR), microcephaly, characteristic facial appearance, low-set ears, overlapping fingers, and clubfoot deformity. In addition, in some cases, endocrine disorders such as growth-hormone deficiency, diabetes incipitus and hypothyroidism can be seen. We present a newborn with trisomy 18 who had chylous acid, chylothorax and hypothyroidism, which has not been described in the literature before.

Case Report

A 33-week-old baby born with a weight of 1910 grams from a 41-year-old mother was admitted to NICU with prematurity and respiratory distress.

It was learned that the mother did not have regular follow-up in the prenatal period. At initial examination, baby was hypotonic, hypoactive and had severe respiratory distress. On examination, there were characteristic morphological features of Trisomy 18. The patient had characteristic facial appearance, microcephaly, low-set ears, overlapping fingers and club foot deformity(Pictures 1-2). Chromosome analysis, echocardiography and ultrasonography were planned for further diagnostic tests. Abdominal and cranial ultrasonograph was reported to be normal except urachal cyst, which was consulted with pediatric surgery and advised conservative follow-up at that moment. Echocardiography revealed wide atrio-ventricular septal defect and significantly open ductus arteriosus. Genetic test confirmed the diagnosis of Trisomy 18 later.

Fıgure 1

Figure 2

Figure 3

Management and Outcome

We started medical supportive treatment for cardiac failure and ductus closure. Despite our best effort, patient had a poor clinical course during hospital stay. At the end of second week we checked thyroid function tests and thyoid stimulating hormone (TSH) level was meaningfully high as to be 26.6 (0.76-10) mIU/L while freeT4 (FT) level was 10.2 (10.35-23) pmol/L. We started the treatment with levothyroxine. Thyroid tissue ultrasonography was normal.

In the following days, gradual fluid accumulation is noted in the abdomen and right lung area and fluid evacucuated for both diagnosis and treatment. Evacuated fluid was chylous at macroscopic apperance as milky and cloudy (Picture 3) and laboratory test also confirmed that the fluid contains high level of triglycerides, that was 1411 (30-110)mg/dL.

At follow up, repetitive fluid evacuation punctures were performed and octreotide is added to the medical

treatment. However, the patient, who had always a poor clinical course, has died on 25th day of hospitalization.

Discussion and Conclusion

Trisomy 18 is caused by the presence of exta chromosome at 18 in most cases and characterised multiple system anomalies(1). Endocrinopathies are one of these system anomalies despite they are les common. As of endocrine anomalies, central diabetes incipitus, growth hormone deficiency, hypoparathyroidism, thymic and adrenal hypoplasia may be seen (2,3).

In our case the thyroid stimulating hormone level (TSH) was meaningfully higher than cut-off levels when we checked first at the around the end of second week. We presumed the case congenital hypothyroidism and started the treatment. When we searched the literature with the strings of the words "trisomy18" and "hypothyroidism", there was no report of trisomy 18 with congenital hypothyroidism. As of endocrinopathies, Nihat et al reported a trisomy case with central diabetes incipitus (2)

We searched the literature for coexistence of trisomy, chylothorax and hypothyroidism. We found only one case report of trisomy 18 with chylothorax (4). We also searched literature on the association of congenital hypothyroidism and chylothorax and found some anectodal case reports (5,6). However, we did not any report of coexistence of all trisomy 18, hypothyroidism and chylothorax. Althought it is reported that octreotide treatment may cause transient hypothyroidism in newborns(7), octreotide treatment was initiated later in our patient.

We could not see any report of trisomy with congenital hypothyroidism at our literature search, so found it worthwhile to report this very rare case of trisomy 18 with congenital hypothyroidism and chylothorax.

Key Messages:

- Trisomy 18 can present with rare endocrine disorders such as hypothyroidism, necessitating comprehensive medical evaluation in affected individuals.
- The occurrence of chylothorax in Trisomy 18, though rare, should be considered in the differential diagnosis of respiratory distress, requiring prompt diagnosis and management.
- Managing complex medical conditions in Trisomy 18 patients, including concurrent hypothyroidism and chylothorax, poses significant challenges and may necessitate multidisciplinary care and

innovative therapeutic approaches.

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