Smyrna Tıp Dergisi *Olgu Sunumu*

Neonatal Ascites and Liver Failure: A Case of Galactosemia
Neonatal Asit ve Karaciğer Yetmezliği: Galaktozemili Bir Olgu

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Abstract:
The most common reasons of neonatal ascites are chylous, urinary, biliary or pancreatic. Neonatal hydrops or cardiovasvular diseases can be the other reasons. Niemann-Pick disease, Gaucher and galactosemia have been reported in literature as the reasons of neonatal ascites among metabolic diseases. Galactosemia, an inborn neurometabolic disorder, results from an aberrant galactose metabolism due to the deficiency of serum galactose-1-phosphate uridyltransferase activity. It manifests in the hepatobiliary system in the form of cholestasis, prolonged icter and hepatic failure. The patient with neonatal ascites and liver failure was diagnosed as galactosemia. Following diagnosis, galactose was withdrawn from the diet (he was given lactose free formula) and the ascites diminished dramatically. The purpose of this paper is to emphasize that galactosemia can be a reason of neonatal ascites and physicians should keep in mind this for early diagnosis.
Key words: Ascites, galactosemia, neonate

Özet:
Neonatal asit en sık şilöz, üriner, bilier veya pankreatik nedenlerle oluşur. Neonatal hidrops veya kardiovasküler hastalıklar da bu duruma neden olabilir. Metabolik hastalıklardan özellikle Niemann-Pick hastalığı, Gaucher ve Galaktozemi literatürde neonatal asit sebebi olarak bildirilmiştir. Galaktozemi, galaktoz metabolizmasında yer alan serum galaktoz-1-fosfat üridil transferaz aktivite yokluğuna bağlı gelişen doğumsal nörometabolik bir hastalıktır. Neonatal dönemde hepatobilier sistem bulguları kolestaz, uzamış sarılık, karaciğer yetmezliği ve asit şeklindedir. Neonatal asit ve karaciğer yetmezliği olan hastamız galaktozemi tanısı almıştır. Tanıyı takiben, galaktozsuz diyet verilmiş (laktozsuz mama) ve asit dramatik olarak gerilemiştir. Bu yazının amacı neonatal asitin galaktozemide önemli bir ipucu olabileceğini vurgulamaktır.
Anahtar kelimeler: Asit, galaktozemi, neonatal

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Introduction

Neonatal ascites may be chylous, urinary, biliary or pancreatic. It may be secondary to neonatal hydrops, congestive heart failure or caused by the rupture of a large ovarian cyst. Chylous ascites are reported as the most common types of neonatal ascites in literature (1). On the other hand, venous anomaly of portal system can be the reason of neonatal ascites, such as ductus venous agenesis (2).

During the neonatal period, in galactosemia, attention is focused on five neonatal presentations; conjugated hyperbilirubinemia, cholestatic jaundice with otherwise good liver function, severe liver disfunction, hepatomegaly with hypotonia +/- cardiomyopathy and hepatosplenomegaly (3). Ascites is a rare presentation in the neonatal period. Early diagnosis is important because the treatment can be dramatically effective with galactose free diet (4).

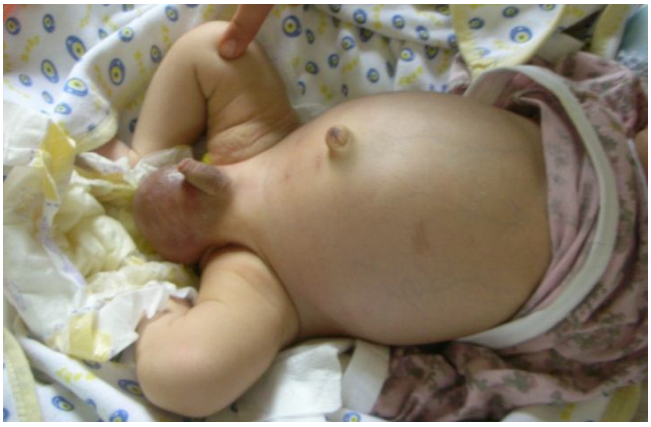
Case

Mentioned patient was born after a normal pregnancy and delivery. He was the first child and there was no history of consanguinity. The present patient became jaundiced and lethargic on the 10^{th} day of his life. He was treated after the diagnosis of sepsis and blood products were administered before admission to our clinic when he was one month old. He was mildly jaundiced, the abdomen was markedly distended with fluid, umblical and inguinal hernia and the liver was palpable about 4 cm (Figure 1).

Routine hemogram and kidney function tests were normal. SGOT: 210 U/dl, SGPT: 53 U/dl, GGT: 37 U/dl, total protein: 4,1 g/dl, albumin: 2,5 g/dl, Na: 138 mEq/L. Ultrasonography revealed hepatomegaly and diffuse ascites; in doppler portal system, blood flow was normal. Urine analysis revealed mild proteinuria, aminoaciduria and positive for reducing substances. After positive urine reducing substances, desired blood aminoacid levels were normal. Results of blood galactose-1-phosphate uridyl transferase activity was 1,23 U/gHb (N≥3 U/gHb), free galactose level was 64,3 mg/dl (N≤5 mg/dl). The

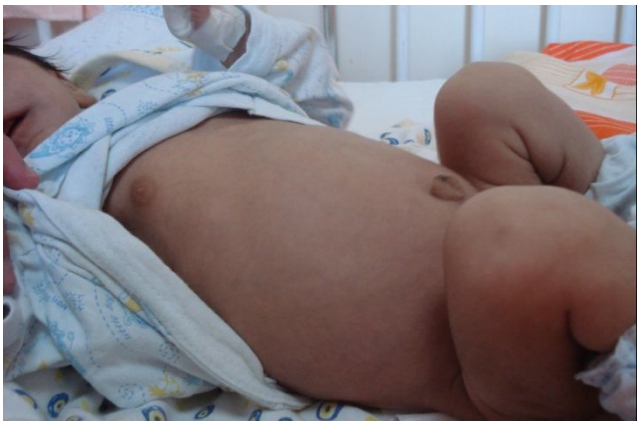
erythrocyte galactose-1-phosphate level was increased (64 mg/100 ml). Ophthalmological examination revealed bilateral cataracts. These results suggested us congenital galactosemia. The result of the GALT gene analysis was homozygous for the Q188R mutation. Magnetic resonance imaging of the brain showed cerebral white matter changes with bilateral hypomyelinization and symetrical periventricular hypersignal in T2.

Figure 1. *Abdominal distention and scrotal swelling due to ascites*



Following diagnosis, galactose was withdrawn from the diet (he was given lactose free formula) and the clinical findings became progressively normal (Figure 2).

Figure 2. *Regression of ascites, view of the abdomen on the 7th day of galactose free diet*



Discussion

Cardinal clinical features of galactosemia include marginal decrease in birth weight, prolonged neonatal jaundice, progressive hepatic dysfunction in the first few weeks of life, renal dysfunction in the form of proteinuria, aminoaciduria and acidosis, E. coli sepsis, hypoglycemia and cataracts (5). Involvement of hepatobiliary system is characterized by prolonged neonatal jaundice and cholestasis. Although progressive hepatic failure in galactosemia has been reported in the literature, mild ascites during neonatal period is a rarity in galactosemia (4,6). Ascites in the patient was attributed to galactosemia as there was no clinical or laboratory evidence for any other cause for ascites like vascular, congenital heart disease, renal pathologies and hydrops fetalis. On the 7 th day of galactose free diet, the ascites diminished dramatically.

Ascites mechanism is unclear in galactosemia. Our patient has also hypoalbuminemia; this may be a reason of ascites. However hypoalbuminemia is often found in galactosemia, ascites is unusual. The mechanism of ascites was thougth to be hepatic failure due to galactosemia. Ascites may develop 2-5 weeks after birth with continued galactose ingestion and is present in most infants who succumb to the disease. Portal hypertension is the major reason for the development of ascites. Patients with liver disease but without portal hypertension do not develop ascites. Two important factors in ascites formation thus are portal venous pressure (which directly affects hydrostatic pressure in the liver) and plasma colloid oncotic pressure. Portal venous pressure was not detected in this patient (7).

Mutation of Q188R, K285N and 5kb-deletion have been reported in severe cases. Also in this case mutation analysis result was homozygot Q188R. In a review from our country the same mutation has been detected in 57% of galactosemia cases (8). In the countries which do not include galactosemia in their newborn screening programmes immediate removal of galactose from the diet results in a full recovery from this life-threatening neonatal crisis (9).

Conclusion

We highlight a rare presentation of ascites as a feature of galactosemia in neonatal period and suggest that attention should be given to its early detection and management.

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