INTRODUCTION
Galactosemia is a disorder of galactose metabolism that is inherited in an autosomal recessive manner. Galaktokinase (GALK), galactose-1-phosphate uridylyltransferase (GALT), and uridine diphosphate-galactose 4-epimerase (GALE) are the enzymes of galactose metabolism. The deficiency of these enzymes results in the accumulation of galactose. Classic galactosemia (CG) (OMIM 230400) is caused by a complete or profound deficiency of GALT enzyme.

Affected babies are born healthy but usually in the early days of life with breastfeeding, the symptoms occur. Most of the galactosemia patients present in neonatal period with the clinical findings of feeding intolerance, vomiting, diarrhea, jaundice, weight loss and lethargy. Hepatomegaly, excessive bleeding, cataracts, and E. coli septicaemia are the other findings. Unless it is treated, it can be life threatening with multiorgan involvement and long-term complications can develop. On the other hand, atypical manifestations of galactosemia in children and adult have been previously reported. The diagnosis is confirmed by measuring galactose-1-phosphate levels and GALT enzyme activity and molecular genetic analysis. The exclusion of lactose is important for preventing acute life-threatening events. If it is initiated immediately enough, signs resolve, and cataracts also may disappear.

Wilson disease (WD) is an autosomal recessive inherited disorder of copper metabolism that is caused by copper overload in the organs particularly in the liver and brain. Homozygous or compound heterozygous mutations in the ATP7B gene causes the disease. Impaired copper excretion results in pathological copper overloading and a wide range of symptoms occur. Hepatic, neurologic, or psychiatric symptoms, or a combination of these can be seen and vary among families. Patients can present 3 or over 50 years. The incidence is approximately 1/30000 births worldwide. Studies have shown that patients presenting primarily with liver involvement findings are younger than those presenting with neurological symptoms, but both groups may occur at an early or late age.

Herein, we present atypical manifestation of galactosemia in a patient with WD.

CASE
A 6-year-old girl was referred to our center due to elevated transaminase levels and hepatosplenomegaly.

The child, weighing 2200 g, was born to first-degree consanguineous parents after a full-term uneventful pregnancy. She was hospitalized in the neonatal period due to indirect hyperbilirubinemia, gastrointestinal bleeding, diarrhea lasting 2 weeks and elevated liver enzymes. She was discharged from the hospital for 2 days due to feeding intolerance and failure to thrive when she was 2 years old. She was examined until the age of 4, but no result could be reached, and the family did not continue to follow up.

Upon physical examination, her weight was 18 kg (10-25 percentile for age), and height was 120 cm (3-10 percentile for age). Hepatosplenomegaly was evident. Cataract was detected and psychiatric assessment revealed borderline mental capacity and cognitive and speech retardation.

Laboratory investigations revealed elevated transaminases aspartate aminotransferase (AST) 142 U/L (RR-0-35) and alanine aminotransferase (ALT) 194 U/L (RR-0-35) and low ceruloplasmin (0.03/0.04 g/L; RR:0.23-0.48) levels. No cholesterol was detected, and blood glucose was normal. Metabolic investigations showed no specific finding except trace positivity for reducing substance in urine. In the previous examinations performed in the neonatal period and at the age of 2 years in other centers, urinary reducing substance and urine sugar chromatography and GALT enzyme activity were found to be normal.

Abdominal ultrasonography revealed no pathological finding of liver and spleen. Cranial magnetic resonance imaging was normal. The copper level in the 24-hour urine was normal. Liver biopsy showed copper accumulation in hepatocytes (hepatic copper concentration was 380 mcg/d dry weight), cytoplasmic enlargement of hepatocytes and occasional glycogenated nuclei. With the finding of low ceruloplasmin levels and liver biopsy, WD was suspected in the patient. Genetic analysis revealed a homozygous c.2293G>A (p.Asp765Asn) variant in ATP7B gene (Fig.1a). Penicillamine treatment was initiated, and she had been followed up with the diagnosis of WD. In the clinical follow-up of patient, a significant mental retardation, behavioral disorder, and cataract findings which were not compatible with the diagnosis of WD were detected. Expanded genetic analysis was performed to evaluated etiology. Homozygous c.1018G>T (p.Glu340Ter) variant in GALT gene was detected (Fig.1b). Despite GALT enzyme activity was found to be normal at the age of 2 years old, GALT enzyme level was reassessed and was found to be low 0.07 mmol/mLh (RR:4-12). The patient was diagnosed with both WD and galactosemia. Galactose-free diet and appropriate doses of calcium and vitamin D supplements were initiated.

CONCLUSION
The fact that the GALT enzyme activity, which was first checked in the other center and found to be normal in this patient, may be related to erythrocyte transfusion.

Coexistence of rare genetically transmitted diseases can be seen in countries where consanguineous marriages are common, such as our country. In the presence of findings that cannot be explained by a single disease, other genetic diseases should be investigated.