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Hepatosplenomegaly and normal karyotype in a fetus with an acute megakaryoblastic leukemia

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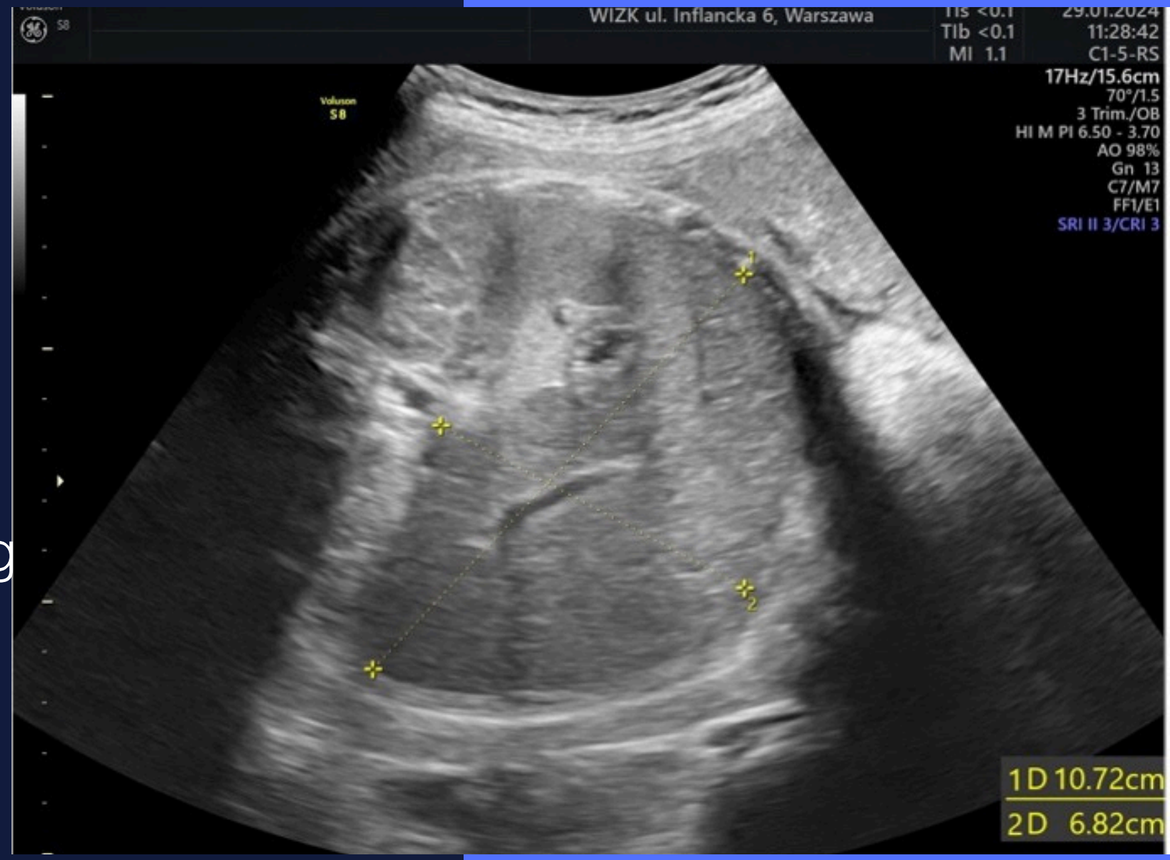
35-year-old gravida 2 para 2 was referred to the outpatient clinic at 38 weeks of pregnancy due to the gestational diabetes mellitus.

- Marked hepatomegaly with the liver measurements of 107x68x87 mm and splenomegaly with spleen dimensions of 52x30x69 mm were revealed, whereas abdominal circumference (AC) was within normal limits ~ 341 mm (58th percentile according to Hadlock).



- There were no fetal abnormalities in the previous ultrasound screening
- During the prenatal visit, the cardiotocography results were normal and the ultrasound examination revealed AGA fetus of estimated fetal weight (EFW) of 3138g
- No other sonographic abnormalities were observed

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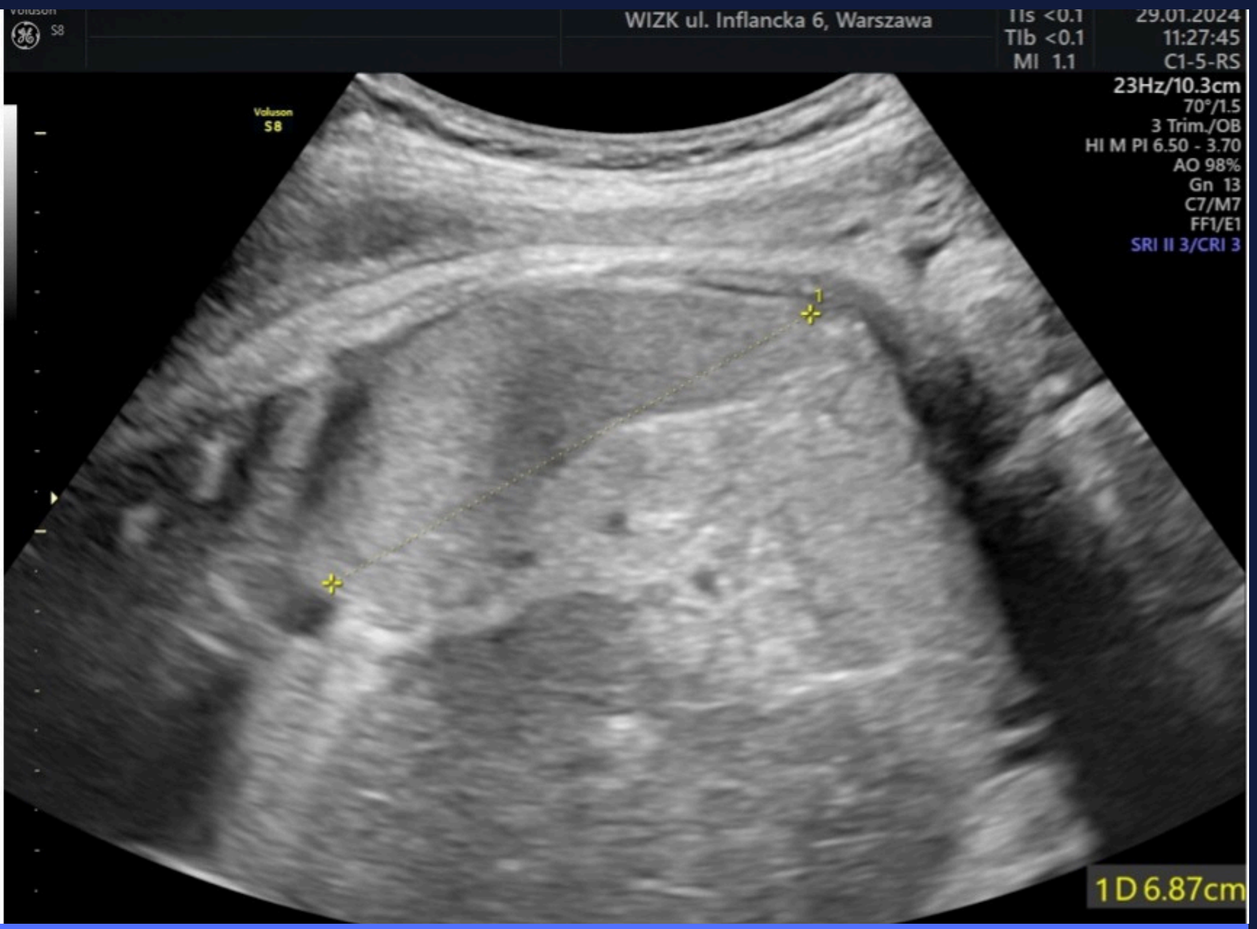

Hepatosplenomeagly diagnostic evaluation

- Infections
- Anemia
- Genetics

The patient was admitted to the ward for further diagnostic evaluation.

The most common infections – CMV, toxoplasmosis, viral hepatitis, HIV, syphilis and EBV – were ruled out by specific maternal antibodies count.

We ruled out fetal anemia by assessing MCA PSV which was within normal limits.

Due to the advanced gestational age, invasive prenatal testing was not performed.

Given the substantial hepatosplenomegaly of unknown etiology in the fetus with the possible risk of coagulation abnormalities and perinatal hemorrhage, the decision was made to proceed with a cesarean section for delivery.

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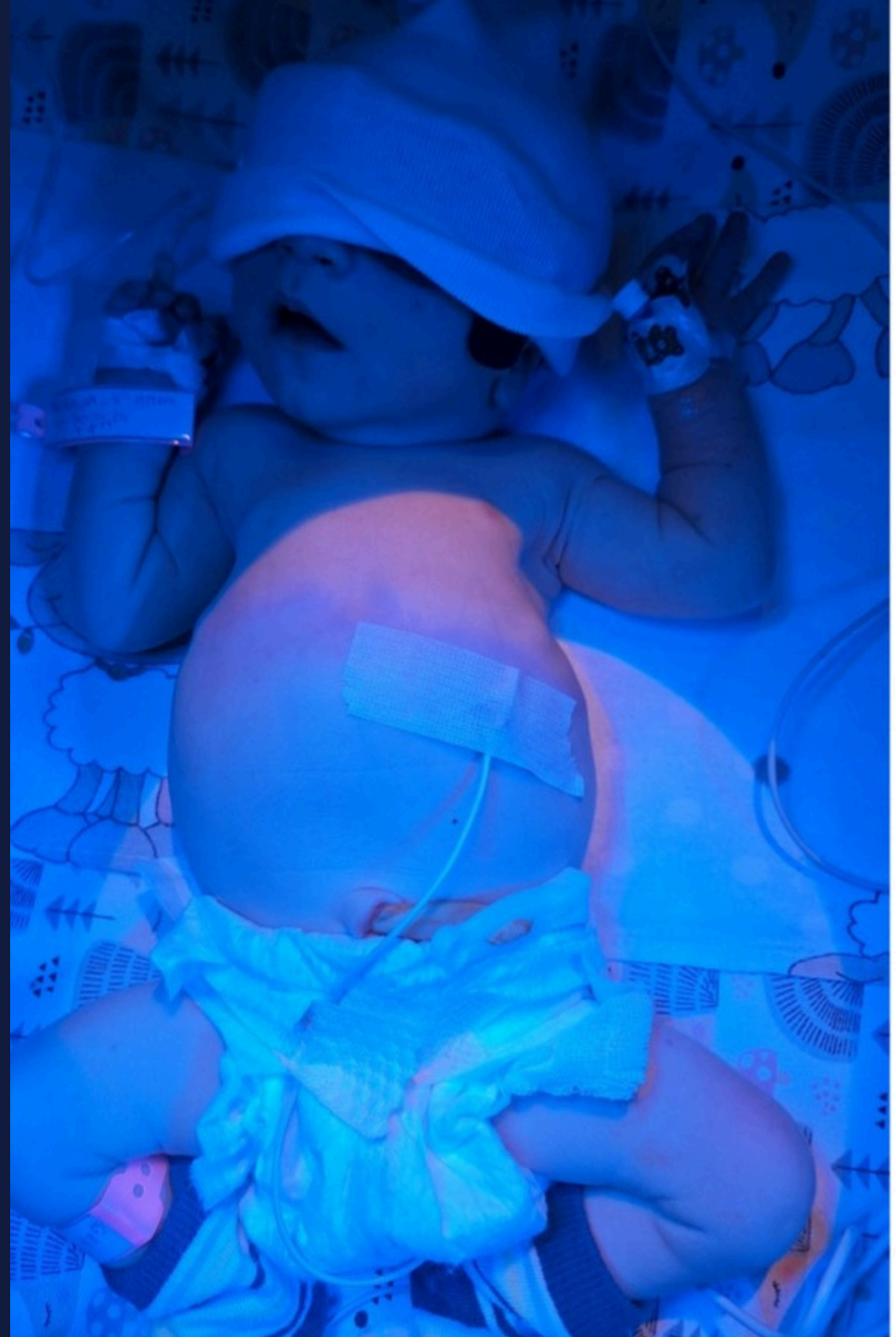
State at birth

At birth at 39 gestational weeks, the infant received an Apgar score of 10.

Since the second hour of life, the neonate began to present jaundice and petechiae on the skin. Initial clinical assessments indicated:

- hepatosplenomegaly
- thrombocytopenia (platelet count 12,000)
- elevatedhepatic enzymes
- coagulopathy
- hyperbilirubinemia

Due to the severe thrombocytopenia a transfusion of platelet concentrates was performed. Urgent transport to a specialist pediatric hospital was arranged to provide advanced medical intervention.



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Postnatal evaluation

In the days following delivery patient was hospitalized in the ICU of the pediatric tertiary care center. Although the patient maintained cardiovascular and respiratory stability, the clinical management required intensive care involving several transfusions of platelet concentrates. Postnatal karyotype analysis using the MONO WES method identified trisomies 14, 15, 19, and 21; however, these findings did not correlate with the clinical symptoms observed. A subsequent FISH test, in contrast, showed a normal male karyotype.

Given the suspicion of a myelodysplastic disorder, a trepanobiopsy was performed, which confirmed the diagnosis of megakaryoblastic leukemia.

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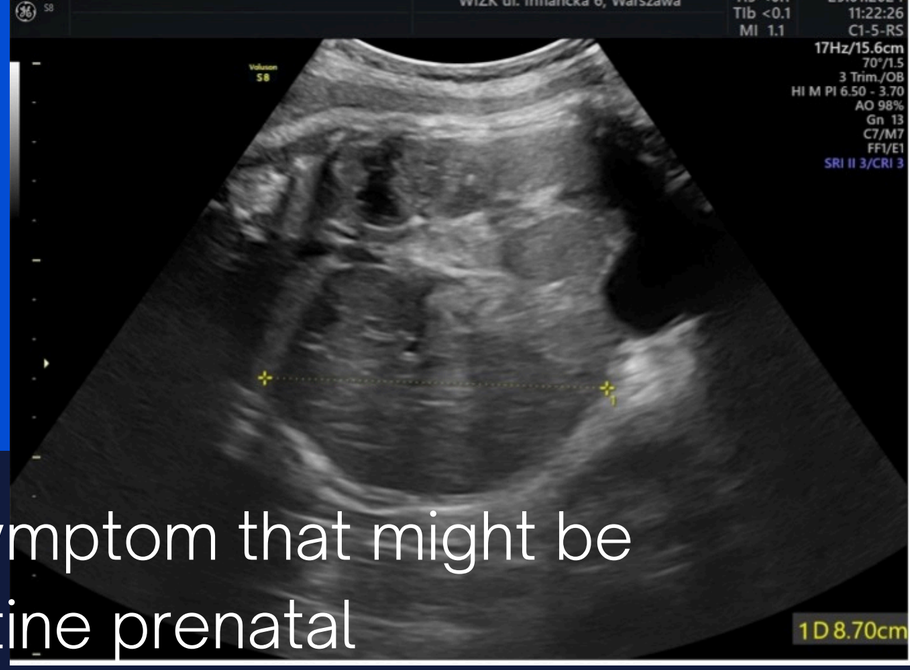
Follow-up

Chemotherapy was initiated promptly and response will soon be assessed by haematologists. The prognosis for the child remains uncertain.

Conclusion



- Fetal hepatosplenomegaly is a rare symptom that might be easily overlooked during routine prenatal screenings, especially when the abdominal circumference measurement is normal
- Prenatal leukemia is among the potential manifestations associated with Down syndrome
- In this particular case demonstrates that a normal karyotype does not exclude acute megakaryoblastic leukemia diagnosis

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