Introduction: Our case report describes a changes on central nervous system in fetus with de novo formation of chromosomal abnormality.

Case: L.Z., 23 years old. IIg/Ilp, combined screening test in the first trimester of pregnancy was negative (risk of trisomy 21, 18 and 13 were less than 1:300) and there were found no morphological abnormalities in the fetus. But the level of nuchal translucency was pathological – 4 mm. The genetic counselling was provided, but patient refused chorionic villus sampling. The detailed morphology of the fetus by ultrasound examination at 16th week of gestation described chooroideal cysts, the other morphology was normal. In 20th week of gestation the detailed morphology normal, only the length of femur was 7th centile. Control ultrasound examination in the next 4 weeks found that the length of femur was 4th centile, nuchal fold was 6,2 mm, and there was suspicion on hypoplasia of vermis cerebelli (in caudal part of vermis was communication between cisterna magna and IV. ventricle). The MRI described megacisterna magna. The genetic counselling was provided again and patient agreed with amniocentesis, which was provided at 24th week of pregnancy.

Conclusion: Although in the first trimester combined screening was pathological level of nuchal translucency, the result on risk of trisomy 21,18 and 12 was negative. But during the pregnancy, there were pathological changes on central nervous system and the chromosomal abnormality was diagnosed. The changes on CNS dissapeared after delivery, but some new different abnormalities were found later.