Introduction
This study reports the feasibility of non-invasive prenatal screening (NIPS) combined with invasive detection in identifying fetal with rare autosomal aneuploidies mosaicism.

Methods
NIPS was performed followed by amniocentesis. Then, the amniotic fluid was analyzed with karyotyping, fluorescence in situ hybridization (FISH), and chromosomal microarray (CMA). Meanwhile, detailed morphology of the fetus was carefully evaluated with Ultrasound and MRI.

Results
NIPS suggested a triploid of chr 8. However, FISH and CMA on cultured amniotic fluid cell were negative. Karyotype study was 47,XY,+8[1]/46,XX[72]. Also, some trisomy 8 mosaicism structure malformations were detected using Ultrasound and MRI. Finally, FISH on cord blood cells confirmed that the karyotype of 47,XY,+8 accounted for approximately 10%.

Conclusion
Rare aneuploidies mosaicism should be included when performing NIPS for chromosome abnormalities to prevent congenital diseases. If rare aneuploidies mosaicism was detected in NIPS, subsequent genetic tests is necessary to confirm the diagnosis. FISH for uncultured amniotic fluid cells or cord blood cells may be excellent methods for screening for rare and low-level aneuploidies mosaicism in prenatal diagnosis. Additionally, our study presents a detailed strategy for diagnosis of rare chromosomal abnormalities by a combination of a series of genetic methods.