

EP10.59 Prenatal diagnosis of a fetus with double trisomy: 48,XXY, +21. A case report

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We describe the prenatal diagnosis and postnatal evolution of fetal patient diagnosed with concomitant Trisomy 21 and Klinefelter syndrome in 2025 at Clínica del Country, Bogotá.

A 43-year-old patient (G3P2) initially underwent combined screening, showing intermediate risk for Trisomy 21; subsequent non-invasive prenatal testing (NIPT) at 13 weeks was inconclusive. At 17 weeks, an obstetric ultrasound revealed nasal bone hypoplasia and a small phallus (micropenis). Amniocentesis confirmed a 48,XXY,+21 karyotype. Follow-up scans identified a flat facial profile, hypertelorism, low-set ears, sandal gap, and third-trimester fetal growth restriction with femoral length below the 5th percentile. The pregnancy reached 38 weeks, resulting in a male neonate weighing 2,770 grams with a clinical phenotype consistent with Down syndrome. Postnatal echocardiography ruled out congenital heart defects. A multidisciplinary management plan was initiated, involving genetics, nutrition, physiatry, speech therapy, and pediatric endocrinology. No acute surgical interventions were required, and the patient continues outpatient surveillance.

Conclusion

Double aneuploidy involving Down syndrome (T21) and Klinefelter syndrome (47,XXY) is rare, with an estimated incidence of 0.4 to 0.9 per 10,000 male births. The neonatal phenotype is typically dominated by T21 features, while Klinefelter characteristics often remain subtle until puberty or later stages of growth. Early prenatal diagnosis is essential for comprehensive parental counseling and to establish a multidisciplinary care pathway for the patient's long-term development.



Figures 1-4
2D and 3 D ultrasound
evidence of flat facial
profile

Figure 5
2D ultrasound showing
low set ears

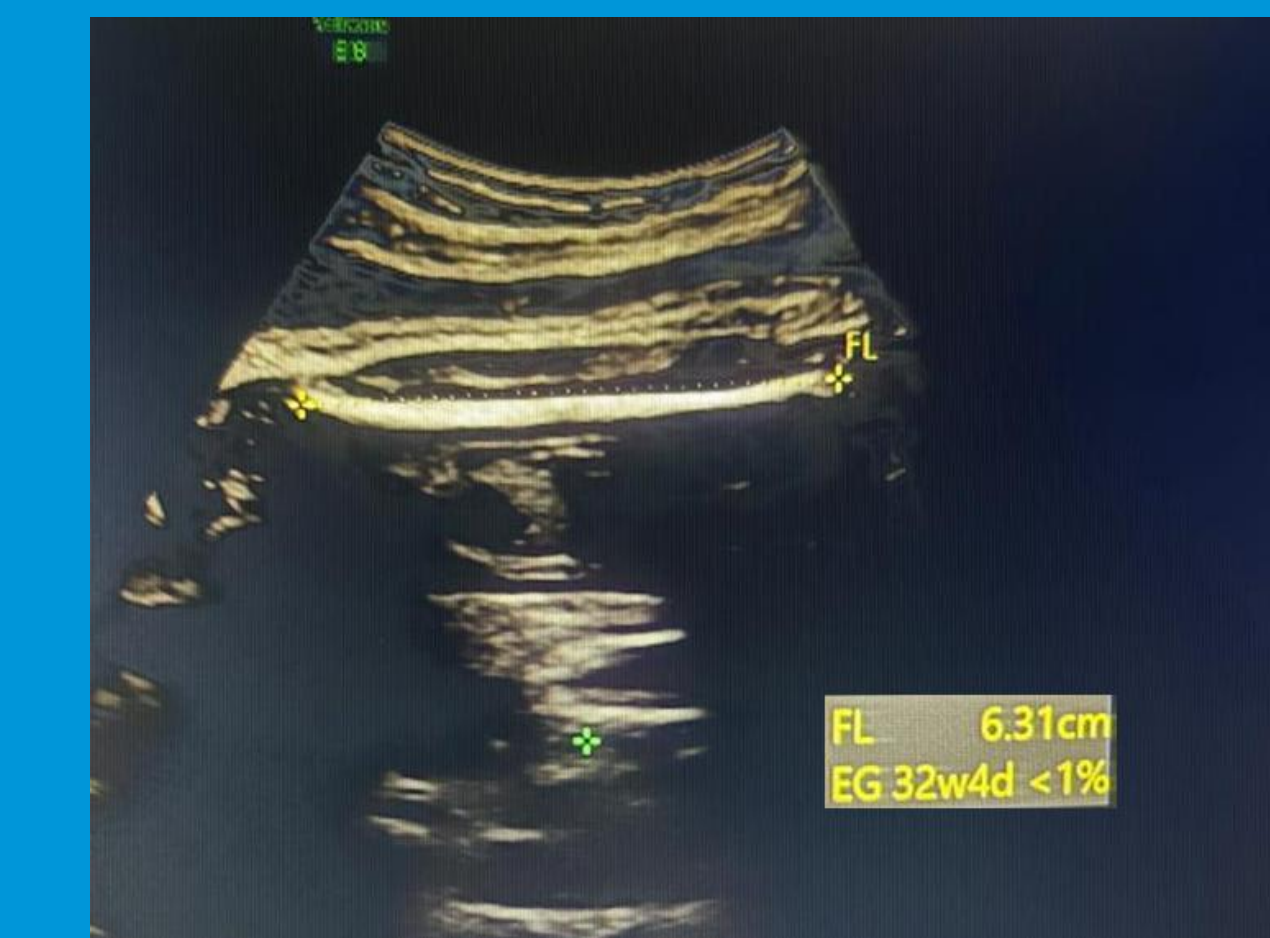


Figure 6
2D ultrasound evidence
of short long bones

Figure 7
Left ventricle
hyperechogenic foci. No
cardiopathy was associated.

