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Mosaic Trisomy 9 in a Male Fetus with Severe Facial Abnormality and Genital Hypoplasia

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KEYWORDS Trisomy 9. Mosaicism. Cheilopalatognathus. Polycystic Kidney Disease. Genital Hypoplasia

ABSTRACT Fetal trisomy 9 is a rare chromosomal abnormality, which was difficult to diagnose in the prenatal period. Here is reported a case of mosaic trisomy 9 fetus, prenatally detected by ultrasound examinations in the second trimester and confirmed by karyotyping from amniocentesis and cordocentesis. The fetus presented multiple malformations, especially facial deformity, polycystic kidney disease and genital hypoplasia, which were rare in previous literature.