

Prenatally detected trisomy 20 mosaicism

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Trisomy 20 is the one of the most common types of mosaic trisomy detected on prenatal diagnosis. even though few cases with congenital abnormalities have been reported in trisomy 20 mosaicism, large amount of cases has a surprisingly good prognosis.

We present a case of prenatally detected trisomy 20 mosaicism with major congenital anomaly. A 28-year-old women was referred to our hospital with abnormal amniocentesis results for genetic counseling. The result of Quad test was high risk in Down syndrome that was performed in local clinics. At 18 weeks' gestation, transabdominal amniocentesis was performed, and the result of chromosomal study was trisomy 20 mosaicism : 47,XX +20(16)/46,XX(14). At 20 weeks' of gestation, the level II ultrasonogram was performed in our hospital, and the large VSD and pyelectasia was detected. After counseling with parents, this pregnancy was terminated.