5.17. Detection of a mosaic for a marker-chromosome 5 in prenatal diagnosis

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The clinical significance of a marker chromosome when diagnosed prenatally is always a major problem (dilemma) especially when there is a de novo situation and a mosaic with a normal cell line.

We report the case of a mosaic for a rare marker chromosome detected in prenatal diagnosis.

Amniocentesis was performed at 17 weeks of gestation in a 39 year old woman. It was a non-consanguineous couple with a normal obstetric history and a phenotypically normal daughter.

The GTG analysis revealed the karyotype 47,XY,+mar/46,XY (60% / 40%) in two different cultures. The marker was CBG positive and did not contain NOR-positive satellites. Using FISH with Chromoprobe Multiprobe System I and Octochrome (Cytocell), we have identified the chromosomal origin of the marker chromosome as derived from chromosome 5. The karyotype is 47,XY,+mar.ish der(5)(wcp5+,D5Z2+)/46,XY.

The karyotypes of the parents were normal.

The couple decided to continue the pregnancy. No ultrasound anomalies were detected at 22 weeks of gestation. A phenotypically normal boy was born at 40 weeks of gestation. Cord blood and placental chronic villi were collected for cytogenetics studies.

To our knowledge this is the first case of a chromosome 5-derived marker detected in prenatal diagnosis. A bibliographic review is presented.