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Prenatal diagnosis of a fetus with a ring chromosome 20 characterized by array-CGH.

Cignini P[1], Dugo N, Giorlandino C, Gauci R, Spata A, Capriglione S, Cafà EV.

Author information

Abstract

OBJECTIVE: a fetus with a ring chromosome 20 is presented.

METHODS: at 16 weeks' gestation, ultrasound examination evidenced no apparent structural malformation. Amniocentesis was performed for maternal anxiety.

RESULTS: chromosome analysis identified a ring chromosome 20 and array-CGH demonstrated that the ring including micro-deletion of the short arm in 20p13, that was extended for about 632.2 kb and a micro-deletion of the long arm in 20q13.33 region.

CONCLUSION: this is the first case of a ring chromosome 20 diagnosed prenatally. This reinforces the importance of offering amniocentesis with a-CGH to make more accurate prenatal diagnosis.

KEYWORDS: array-CGH; ring chromosome

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