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Partial trisomy 20p

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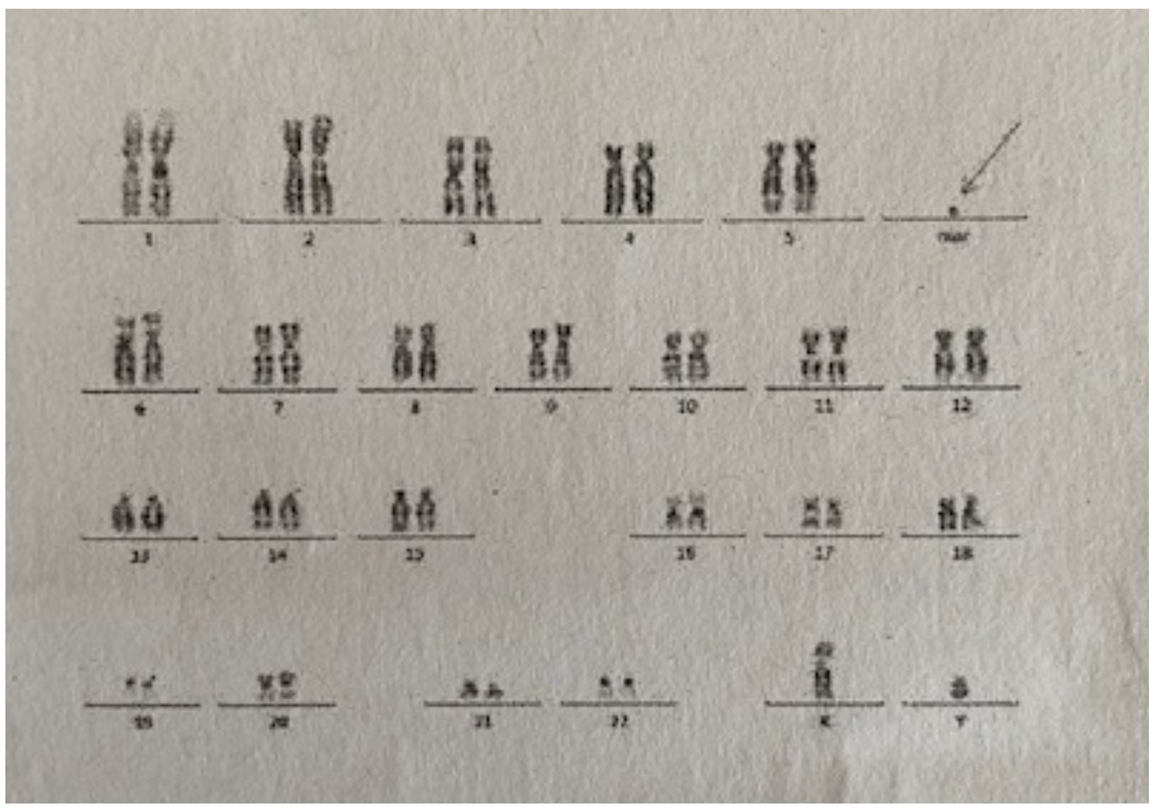
Introduction

Trisomy 20p is a chromosomal abnormality resulting from the duplication of a fraction of the short arm of chromosome 20. Although it may occur de novo, most reported cases arise from a reciprocal translocation or a parental inversion. The prognosis is variable, depending on the size and location of the duplication.

Case Report

A 40-year-old primigravida without risk factors had a 1/37 risk for Down´s Syndrome in a second-trimester serum screening. An invasive test was offered and an amniocentesis was performed. Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR) was normal, karyotype showed an excess of genetic material. A Comparative Genome Hybridization (CGH) array identified a gained region, on the short arm of chromosome 20:



arr[GRCh37] 20p11.21p11.1(23835471_26268347)x3

A parental segregation study was performed and the parents were not carriers. It was classified as a variant of unknown significance (VUS) and genetic counseling was given. At 29 weeks of gestation a short fetal femur was observed and achondroplasia and hypochondroplasia genetical studies were performed in amniotic fluid with normal results. At 33 weeks an intrauterine growth restriction (IUGR) was diagnosed and at 37 weeks a cesarean section was executed because of breech presentation. A masculine 2070gr fetus was born. Nowadays he is 10-month-old and his neurological development is normal.

Conclusion

Collaboration between geneticists and obstetricians play a vital role in order to give counseling to families when a genetic disorder as partial trisomy 20p is diagnosed.