A 25-years-old pregnant woman achieved a first trimester screening for Down syndrome with an increase risk (MoM NT 1.18, MoM PAPP-A 0.17, MoM Hcg 4.76). In consequence, a chorionic villus sampling was ruled out at 13 weeks of gestation and concluded 47 XY, +2 (trisomy 2). Given the ongoing pregnancy without any abnormal ultrasound sign, an amniocentesis was planned at 16 weeks of gestation and the result was: 46 XY (normal karyotype). A trisomy 2 confined placental mosaicism (CPM) was suspected at this moment. The parents (normal karyotype for both) decided to continue the pregnancy. Follow-up by serial ultrasound examinations revealed intrauterine growth retardation (IUGR) without malformation. A boy of 1230 g was born by caesarean section at 34 weeks of gestation after fetal maturation. We expect the analysis of polymorphic markers of chromosome 2 on the placenta.

Placental mosaicism can be confined either to trophoblast (type I), chorionic stroma (type II) or both cell lineages (type III). Type I and II are mostly of mitotic in origin and type III is mainly meiotic. A uniparental disomy can be found in meiotic confined placental mosaicism with possible repercussion on prenatal fetal development.

CPM has been observed in over 20% of pregnancies with idiopathic IUGR. Therefore it is important to study placentas from pregnancies with idiopathic IUGR for the presence of CPM, using molecular cytogenetic methods. In fact, the detection of CPM provides important information for postnatal follow-up.