

QF-PCR on native chorionic villous for false positive and negative detection is a useful tool in fetal karyotyping by direct method

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The prenatal diagnosis (PD) on chorionic villi (CV) more reliable for the identification of fetal karyotype combines the cytotrophoblast (direct method, STC) and mesenchyme (long term culture, LTC) analyses. However, when the sample available is poor (5-10mg) the diagnosis is generally performed liking better for the direct method in order to avoid the maternal cell contamination. This condition is associated with a higher false negative risk. In our 7 years experience on 24'237 CV prenatal diagnosis, we recognised 18 cases of type V True Fetal Mosaicism (TFM) that, if they have been analysed only with STC they would have been subject to a false negative result. The abnormality found in mesenchyme of 17/18 was an aneuploidy for chromosomes 13,18,21,X,Y. In 11 of them the abnormal cell line was present in a homogeneous form, hence, possibly recognizable by QF-PCR. On these basis we performed a study to calculate in our survey the false positive and negative risk if PD would have been performed only with direct method and to evaluate in 267 poor samples (5-10mg) if QF-PCR on a minor fragment allows detection of false positive and negative cases. We evidenced 1 false negative and 2 false positive cases and in 14 instances the abnormality found in cytotrophoblast was confirmed by QF-PCR. In conclusion, QF-PCR could be an additional useful tool, which doesn't jeopardize the result of direct method, to decrease the false negative risk from 1/1136 to about 1/2941.