A case of trisomy 15 detected in paraffin-embedded villous tissue

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Objective

To emphasize an additional diagnostic value of placental examination in detecting chromosomal abnormalities using fetal DNA from a formalin fixed, paraffin-embedded and microdissected villous tissue material in the event of stillborn fetus without prior prenatal screening for chromosomal abnormalities.

Methods

Case report.

Results

Placenta of stillborn MC-DA twins at 23 weeks of gestation with severe Twin Reverse Arterial Perfusion Syndrome (TRAP) was examined after delivery. Microscopic placental examination revealed histologic features that characterize placenta with trisomy: scalloping of villi, trophoblastic inclusions, cytotrophoblast giant cells, mild villous edema. This findings were indication for further genetic investigation. DNA was extracted from the formalin fixed, paraffin embedded and microdissected villous tissue. QF-PCR method using fluorescence-labelled primers referring amplification of chromosome-specific polymorphic microsatellite markers was performed. An abnormal quantity of chromosome 15 was detected.

Conclusion

DNA testing from paraffin-embedded villous material can be a useful tool in detecting trisomy in pregnancies without previous screening for chromosomal abnormalities.