



POSTER – 1

PRENATAL INVESTIGATION OF A FAMILIAL PARTIAL MONOSOMY 10q

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Objective: To present the clinical, cytogenetic and molecular findings of a prenatal study of a familial partial monosomy 10q. Distal 10q deletions are rare and the majority are terminal deletions involving bands 10q25 and 10q26. Patients typically present with facial dysmorphism, postnatal growth retardation, developmental and mental retardation, genitourinary anomalies and digital anomalies.

Methods: Conventional cytogenetic analysis in metaphases obtained by chorionic villi long term cultures, multiplex ligation dependent probe amplification (MLPA), fluorescent in situ hybridization (FISH), microarray analysis.

Results: A 24-year-old gravida was referred for chorionic villus sampling at 12⁺⁶ weeks of gestation due to a previous child with facial dysmorphism, bilateral inguinal hernia, short stature and mild to moderate psychomotor delay of whom a microarray analysis was underway. His karyotype was normal but array-CGH analysis disclosed a 10q24.33-q25.1 interstitial deletion. The deletion encompasses 987Kb to 1,14Mb and includes 20 genes, in particular the COL17A gene. Fetal and parental karyotypes were normal. FISH analysis with a BAC clone located within the 10q region deleted in the phenotypically abnormal sibling showed normal results for both the mother and the fetus and a deletion in the apparently normal father.

Conclusion: In this case chorionic villi analysis as well as the application of FISH with a specific and targeted BAC clone allowed a shorter turnaround time for the prenatal investigation of the chromosomal abnormality. The authors discuss the challenges of microarray analysis application in the prenatal setting namely in cases like the one presented here where there seems to be phenotypic variability.