SNP ARRAYS CAN HELP IN PRENATAL GENETIC COUNSELING: A CASE REPORT

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Introduction: SNP arrays detect genetic gains and losses but also loss of heterozygosity (LOH). They are used in first line for prenatal diagnosis in cases of increased nuchal translucency (TN), one major or several minor ultrasound anomalies or severe intrauterine growth retardation (IUGR). **Case:** A 32 years old woman with a 12 weeks gestation. In first trimester ultrasound TN 6.3mm, fetal hydrops and a reverse ductus venous were observed. Therefore the patient underwent an invasive procedure for a microarray study (Affymetrix 750K CytoScan). This analysis identified a duplication and a deletion: arr [hg19] 3q26.32q29 (178,372,618-197,851,444) x3,11q23.3q25 (119,461,090-134,937,416) x1, associated with 3q29 microduplication Syndrome and 11q deletion Syndrome / Jacobsen Syndrome. It was reported to the couple that decided to interrupt the pregnancy. Suspecting an inherited imbalanced translocation, fetal karyotype was analyzed: 46,XY,der(11)t(3;11)(q26.3;q23.3). In order to determine the risk of recurrence in future pregnancies and discard a cryptic rearrangement parents karyotypes and FISH with LSI BCL6 probe (3q27) dual color break apart on parent metaphases were performed. The results were normal, thus excluding a germ line mosaic. The anomaly was considered *de novo*. Affymetrix platform did not observe a LOH on chromosome 3 so it can be assumed that the anomaly was caused during gametogenesis of one parent. **Conclusions**: Microarrays and cytogenetics help to identify and characterize fetal anomalies. With the detection of LOH we can identify the origin of the alteration allowing us to offer a better genetic counseling to establish the risk of recurrence in future pregnancies.