

## P25 | Cytogenetics and Genomics

### *Interstitial deletion on chromosome 14q in prenatal diagnosis*

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A limited number of prenatal diagnosis (PND) cases have reported interstitial deletions of the long arm of chromosome 14 involving the 14q31-32 region. Those cases presented cardiac anomalies, urogenital anomalies, congenital diaphragmatic hernia, and mild pyelectasis. We report the PND of a 33-year-old pregnant woman, who underwent chorionic villus sampling at 12 weeks of gestation after a positive combined 1st trimester screen. The karyotype revealed a 14q interstitial deletion. Amniocentesis was performed at 18 weeks of gestation to confirm the deletion and to exclude a confined placental mosaicism and a microarray analysis was performed in order to accurately define the deletion breakpoints. Cytogenetics analysis revealed a karyotype 46,XY,del(14)(q31q32.2)dn. Microarray analysis allowed to redefined the breakpoints accurate localization and the identification of a ~21Mb deletion (arr[hg19] 14q31.1q32.31(79917376\_101568230)x1). At 18 weeks of gestation the fetus presented abnormal fetal biometric parameters (occipitofrontal diameter, cephalic perimeter and abdominal circumference) on ultrasound. After counseling the couple opted for pregnancy termination. The postmortem analysis presented decreased biometry, low weight and low fetal size, facial dysmorphism, clinodactyly, club foot, overlapping fingers and short penis. In internal habitus he presented thymus hypoplasia, bladder hypoplasia, and horseshoe kidneys. The genotype-phenotype correlation in PND pure del(14q) cases is not well established. Furthermore, to our knowledge, del(14q) had not been reported so early in the gestation yet. In this case the positive 1st trimester screen was related to the inverted ductus venosus and low PAPP-A value. The urogenital anomalies (as horseshoe kidneys) and biometry anomalies are described in the literature. However, to our knowledge, some features of the present case were not seen in other reported cases, for instance clinodactyly, club foot, overlapping fingers, thymus hypoplasia and bladder hypoplasia. Other reports described cardiac and cerebral anomalies, diaphragmatic hernia, and also UPD(14)like phenotypes, which are possibly linked to the 14q32 imprinted region. The establishment of a phenotype-genotype correlation is difficult given the size of the deletion, which includes a large number of genes in distinct regions. Nevertheless, this work contributes to a better identification of additional features associated to del(14q) that can be present in PND.