



Clinical Case: A 28-Year-Old G1P0 with Mild Ventriculomegaly

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Clinical Image

We present the case of a 28-year-old woman in her first pregnancy. First trimester ultrasound screening for aneuploidies (NT, β -hCG and PAPP-A) was negative. Second trimester ultrasound scan revealed a mild ventriculomegaly (posterior horn of the lateral ventricle was 11 mm). She was referred to amniocentesis for chromosomal analysis. The latter showed a lack of a region in the long arm of the chromosome 4 (46, XY, del(4)(q35 \rightarrow qter) (Figure 1), where according to UCSC genome browser the genes for FAT 1 (FAT atypical cadherin 1) [1,2], TLR3 (toll-like receptor 3) [3], CASP3 (cysteine-aspartic acid protease 3, caspase 3) [4], F11 (coagulation factor XI) [5,6], UFSF2 (ubiquitin-fold modifier 1 specific peptidase 2) [7], KLKB1 (kallikrein B1) [8] and CYP4V2 (cytochrome P450 family 4 subfamily V member 2) [9] have their loci [10]. After genetic counseling, parents underwent chromosomal analysis which revealed de Novo mutation. They decided to terminate the pregnancy in 24 weeks after informed consent. Histologic analysis revealed rocker bottom feet, camptodactyly, facial and truncal hirsutism, short neck, hypertelorism and macrocephaly.

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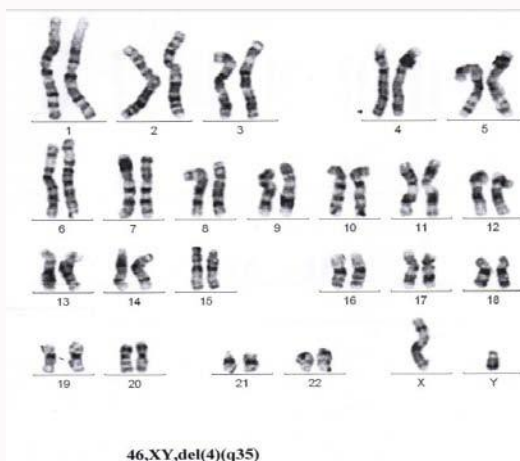


Figure 1: Chromosomal analysis of 28-year-old woman in her first pregnancy.

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