Fetal isolated hydrocephalus associated with a novel missense mutation of L1CAM
Li J, Duan HL, Zhao GF, Zhu XY, Wu X
Nanjing Drum Towel Hospital, Nanjing, China

**Objective**
To provide useful information about genetic counseling or prenatal test in fetuses with isolated hydrocephalus.

**Methods**
We conducted mutation analysis of L1CAM in 13 Chinese male fetuses with isolated hydrocephalus. Genomic DNA was extracted from cord blood with QIAamp® DNA Blood Mini Kit (Qiagen, Inc., Hilden, Germany) or from the amniotic fluid cells with BioChain Amniotic Fluid Genomic DNA Kit. The 28 coding exons and intron-exon junctions of the L1CAM gene were PCR-amplified using 23 primer pairs.

**Results**
Sequencing of the L1CAM gene showed a novel P360S mutation in one case which altered a proline into serine on amino acid position 360 (p. Pro360 Ser). P360S was predicted to be deleterious by bioinformatic analysis.

**Conclusion**
L1CAM mutation was detected in 1/13 cases which implied that L1CAM mutation screening should be considered in male fetus with isolated hydrocephalus.