EP06.05. It is possible to diagnose Joubert syndrome with prenatal sonographic “molar tooth sign”

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Introduction
The characteristic imaging of Joubert syndrome is “molar tooth sign”, which consists of the thickened and elongated superior cerebellar peduncle and the midbrain, however sometimes it is similar to Dandy-Walker malformation or isolated vermian hypoplasia. We report three cases of typical “molar tooth sign” at 21, 25 and 27 weeks by prenatal 2D and 3D ultrasound.

Case report
The three couples were healthy and had no family history of Joubert syndrome. The first two cases were diagnosed Joubert syndrome after birth by clinical features and MRI, one of which was diagnosed Dandy-Walker malformation prenatally for the typical hypoplastic vermis and cisterna magna, associated with mild ventriculomegaly. The result of whole genome and exome sequencing showed that the baby had pathogenic CEP290 gene mutation that inherited from both of parents, which supported the diagnosis of Joubert syndrome 5. The baby had been hospitalizing since birth for pneumonia. For the second fetus, there was no evidence showing other structural anomalies or chromosomal abnormalities prenatally except vermian hypoplasia. However, she could not walk and speak well with one eyelid ptosis when she was 3 years old. The third couple chose termination of pregnancy for the fetus with bilateral autosomal recessive polycystic kidney disease (ARPKD) and oligohydramnios besides hypoplastic vermis.

Conclusion
It is possible to detect the “molar tooth sign” and diagnose Joubert syndrome by prenatal ultrasound, especially in cases associated with other malformations such as polycystic kidney, encephalocele, post-axial polydactyly and so on.