**EP06.06. Antenatal Diagnosis of Joubert Syndrome- a rare case report.**

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**Introduction**

Joubert syndrome (JS) is a rare congenital nervous system developmental disorder, was first discovered by Marie Joubert in 1969. Joubert syndrome is characterized by hypotonia, abnormal ocular movement, hyperpnea, developmental delay, hypoplasia of the cerebellar vermis and brainstem malformation. Also respiratory and renal pathology.

The most common characteristic ultrasound features of JS is the molar tooth sign (MTS) on the axial plane. There is thickened superior cerebellar peduncles (SCPs), cerebellar vermis (CV) hypoplasia/agenesis, and a deepened interpeduncular fossa.

Recurrence rate is about 25%.

We present a case of Joubert syndrome diagnosed antenatally. A woman 27 years, Gravida 4, L2 referred for routine obstetric ultrasound at 23 weeks. No consanguinity. First male child 7 years old, diagnosed postnatally with Joubert syndrome. On Ultrasound, in axial plane, vermis is hypoplastic with fourth ventricle is slightly elongated and narrowed. Also superior cerebellar peduncle appears as thickened, elongated with deepened interpeduncular fossa as “Molar Tooth” . Patient delivered full term male child with 2600gm weight.

Postnatally confirmed diagnosis by MRI.

**Conclusion**

Joubert syndrome and related disorders is a congenital anomaly with a high risk of family recurrence. On ultrasound, characteristic findings of Joubert syndrome “Molar Tooth sign” is seen.