A case of Joubert syndrome
Hamdi A, Jaziri D, Abid C, Znaigui I, Boudaya F, Achour E, Chelli D
First Department of Obstetrics and Gynecology, Maternity Center Rabta Tunis, Tunis, Tunisia

Objective
To describe the prenatal imaging findings in fetuses with Joubert syndrome.

Methods
Case report.

Results
A G3P0 woman with two previous miscarriages presented to our unit at 24 weeks. The anomaly scan revealed moderate bilateral ventriculomegaly, short femur and bilateral club foot. The mother opted for termination of pregnancy. Neonatal autopsy showed hypertelorism, bilateral aniridia, retrognathia and cleft palate, occipital meningocele and polydactyly. In a macroscopic level retinitis pigmentosa, hypoplastic lung, multicystic kidney and a molar tooth in the mid-brain.

Conclusion
The diagnosis of Joubert syndrome can be suspected prenatally on the basis of ultrasound findings or MRI. However the diagnostic confirmation is provided by the examination of clinical pathologist.