Teaching Neuro*Images*: Prenatal MRI of muscle-eye-brain disease

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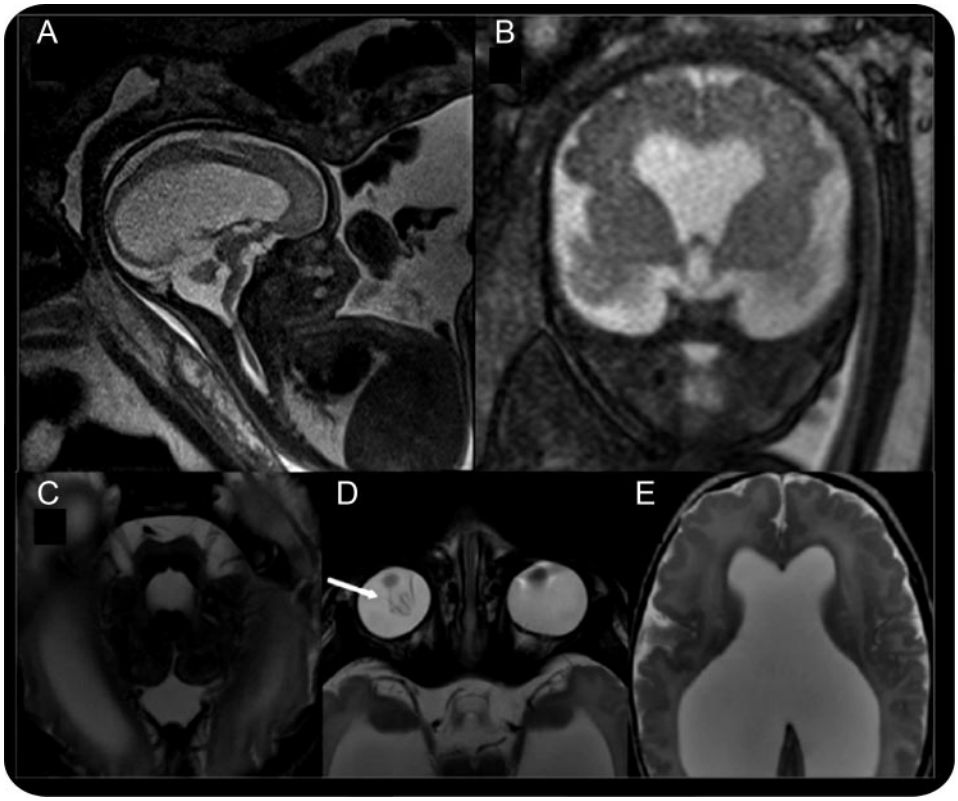
Figure Fetal (A, B) and 2-day-old (C–E) brain MRI showing malformations consistent with muscle-eye-brain disease



(A) Sagittal-T2: brainstem hypoplasia and "kinked" pontomesencephalic contour. (B) Coronal-T2: absent septum pellucidum and severe ventricular dilation. (C) Axial-T2: hypoplastic pons with an abnormal posterior indentation; dysplastic cerebellum with small cysts. (D) Axial-T2: retinal detachment (arrow); microphthalmia. (E) Axial-T2: abnormal gyral sulcal pattern with a nodular cobblestone-appearing cortex.

A 29-week fetus was noted to have dilation of the entire ventricular system on prenatal ultrasound. Fetal MRI at 33 weeks revealed brain malformations suggestive of congenital muscular dystrophy (figure, A and B).[1,2] Following normal delivery at 40 weeks, examination was remarkable for retinal detachment (figure, D) and optic nerve hypoplasia, without dysmorphic features. Sequence analysis showed a homozygous mutation in the *POMGnT1* gene consistent with muscle-eye-brain disease. Muscle-eye-brain disease is rare and presents with hypotonia, weakness, ocular abnormalities, and severe brain malformation (type II lissencephaly).[1,2] Fetal MRI can be valuable in further characterizing malformations detected by ultrasound and improving prenatal counseling and management.

REFERENCES

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