Teaching NeuroImages: The curious case of the brainstem kink

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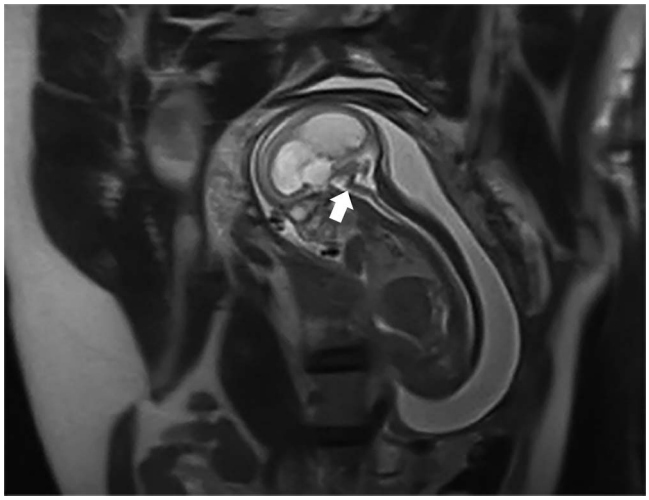
Figure 1 Prenatal imaging



Fetal half-Fourier acquisition single-shot turbo spin echo MRI demonstrates a small "kinked" brainstem (arrow) associated with severe supratentorial ventriculomegaly.

A 25-year-old woman presented for routine prenatal ultrasound, which was concerning for severe hydrocephalus. Fetal MRI at 22 weeks showed a brainstem "kink" that suggested arrest or impairment in brain maturation around 7 weeks gestation, a sign of severe neurodysgenesis (figure 1). Differential diagnoses included α-dystroglycanopathies, X-linked hydrocephalus, and tubulinopathies.[1] Postnatal imaging (figure 2) and clinical findings of hypotonia, macrocephaly, and optic nerve hypoplasia were consistent with Walker-Warburg syndrome (WWS), confirmed on sequencing of the *ISPD* gene. WWS is a severe dystroglycanopathy of autosomal recessive inheritance characterized by muscle, eye, and brain abnormalities.[2] Death typically occurs within 1 year.

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Author contributions

R. Srivastava: drafting/revising the manuscript, data acquisition, accepts responsibility for conduct of research and final approval. F.-D. Morneau-Jacob: drafting/revising the manuscript, data acquisition, accepts responsibility for conduct of research and final approval.

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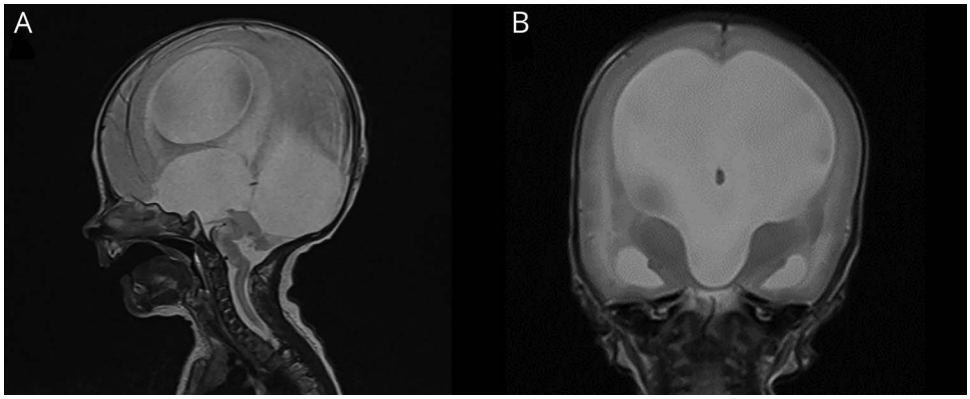
Figure 2 Postnatal imaging



T2-weighted MRI in (A) sagittal and (B) coronal cuts demonstrates multiple striking anomalies including cobblestoning lissencephaly, pontine and cerebellar hypoplasia, and a kink at the cervicomedullary junction consistent with Walker-Warburg syndrome.

References

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2. National Library of Medicine. Walker-Warburg Syndrome: Genetics Home Reference [Internet]. Bethesda: The Library; 2017. Available at: ghr.nlm.nih.gov/condition/walker-warburg-syndrome. Accessed October 17, 2018.

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