

Teaching NeuroImages: A rare case of Jacobsen syndrome with global diffuse hypomyelination of brain

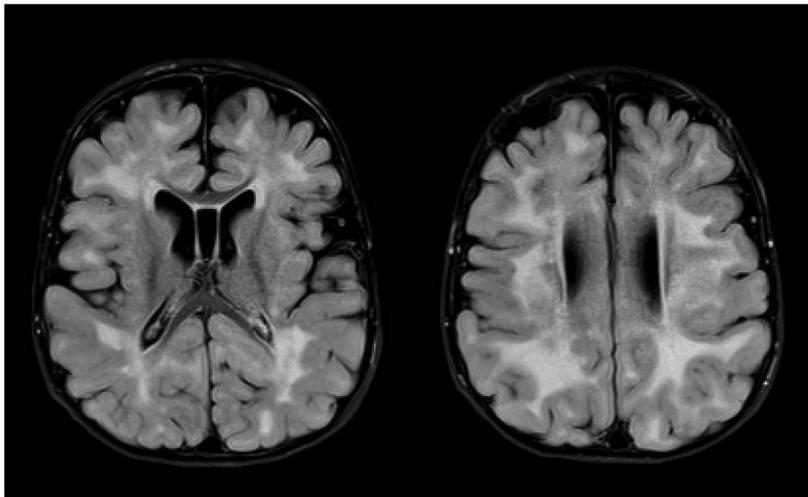
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Figure 1 MRI brain at 18 months of age



Axial fluid-attenuated inversion recovery MRI shows white matter hypomyelination of cerebral as well as deep white matter consistent with leukodystrophy.

An 8-month-old boy presented with developmental delay, diffuse hypotonia, hypoplastic left heart syndrome, undescended testes, neonatal thrombocytopenia, and unusual facies. Chromosome microarray showed an 11q23-11q24 deletion, consistent with Jacobsen syndrome (JS).

Initial head MRI at 18 months showed severe diffuse hypomyelination of cerebral white matter believed to be consistent with leukodystrophy (figure 1). Repeat MRI at 3 years of age showed less extensive hypomyelination (figure 2). The child, while still delayed, had made some developmental progression.

While delayed myelination has been reported, there are few reports of neuroimages. Awareness of the MRI presentation of JS is crucial to prevent unnecessary evaluation in this condition.^{1,2}

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Disclosure

H. Patel and A. Kumar report no disclosures relevant to the manuscript. G. Raymond is a consultant for the Department of Health and Human Services, Minoryx (Barcelona), Bluebird

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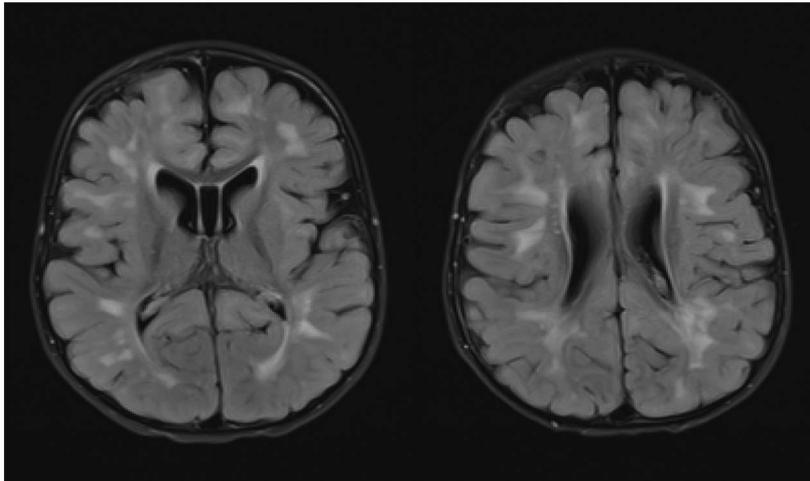
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Figure 2 MRI brain at 3 years of age



Axial fluid-attenuated inversion recovery MRI shows improved degree of white matter myelination.

bio (Cambridge, MA), and Viking Therapeutics (San Diego, CA), and previously served as a consultant for Shire and Vertex (Boston, MA). G. Mainali reports no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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2. Yu F, Carter JE, Bazan C III. A case of Jacobsen syndrome with multifocal white matter lesions. *Clin Imaging* 2016;40:705–706.

Appendix Authors

| Name | Location | Role | Contribution |
|--------------------------|-----------------------------------|--------|---|
| Himadri Patel | Penn State College of Medicine | Author | Conceptualized the case report; drafted and revised the manuscript for intellectual content |
| Ashutosh Kumar | Penn State Hershey Medical Center | Author | Conceptualized the case report; drafted and revised the manuscript for intellectual content |
| Gerald V. Raymond | Penn State Hershey Medical Center | Author | Conceptualized the case report; revised the manuscript for intellectual content |
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