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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Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.
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.

**References**


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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.
Teaching NeuroImages: A rare case of Jacobsen syndrome with global diffuse hypomyelination of brain
Himadri Patel, Ashutosh Kumar, Gerald Raymond, et al.

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