Teaching NeuroImage: *IBA57* Mutation–Associated Infantile Cavitating Leukoencephalopathy

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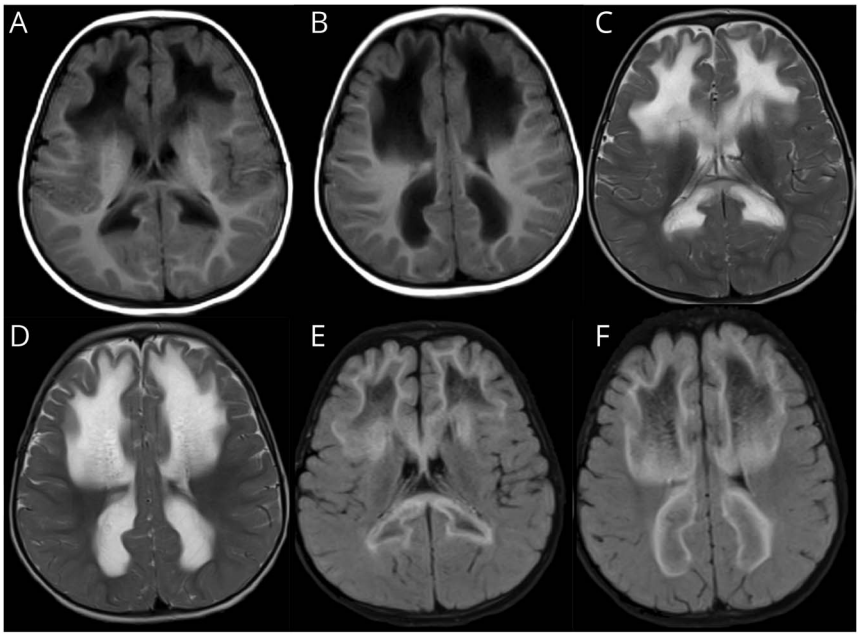
Figure 1 *IBA57* Mutation–Associated Cavitating Leukoencephalopathy



Corresponding axial T1 (A and B), T2 (C and D), and FLAIR (E and F) images show hyperintensities in frontal, parieto-occipital, and periventricular white matter with cavitating lesions suggesting a cavitating leukoencephalopathy.

A 14-month-old child, born of nonconsanguineous parentage, with normal early development till 1 year of age, presented with psychomotor regression. Examination revealed developmental age of 3 months and bipyramidal signs. Brain MRI (Figures 1 and 2) showed periventricular cavitating leukoencephalopathy (PCL). Clinical exome sequencing showed compound heterozygous, disease-associated missense variant in the *IBA57* gene.

PCL with peripheral restricted diffusion along its margins helps in the diagnosis of *IBA57* mutation–induced multiple mitochondrial dysfunction syndrome type 3.[1] PCL has also been reported with defects in complexes 3 and 4 (*LYRM7*, *APOPT1*, *COX10*, and *COX6B* variants) and *MFN2* gene mutations.[1,2]

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Teaching slides
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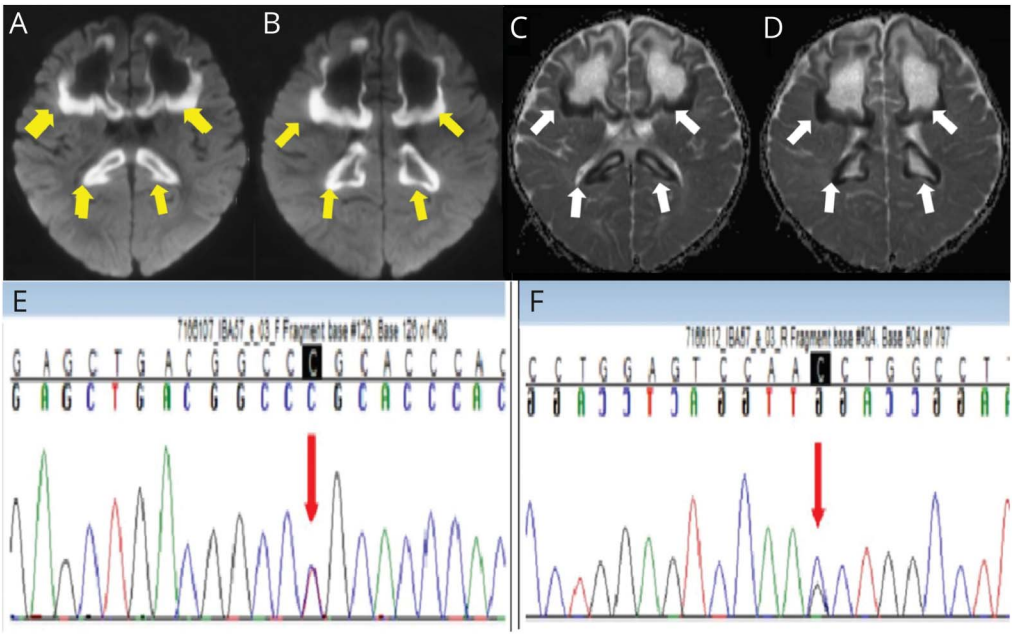
Figure 2 Diffusion-Weighted Images and Chromatogram of *IBA57* Mutation



Diffusion-weighted images (A and B) show restriction (yellow arrows) with low apparent diffusion coefficient values (C and D, white arrows) along the margins. Sanger sequence chromatogram shows variation in exon 3 of the *IBA57* gene in mother (E) (c.802C>T; p.Arg268Cys) and father (F) (c.738C>G; p.Asn246Lys) in heterozygous condition. Both are at highly conserved positions.

Disclosure

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Asish Vijayaraghavan, MD, DM	Department of Neurology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananathapuram, Kerala, India	Review and critique of the manuscript
Soumya Sundaram, MD, DM	Department of Neurology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananathapuram, Kerala, India	Review and critique of the manuscript

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