Teaching Neuroimage: IBA57 Mutation–Associated Infantile Cavitating Leukoencephalopathy

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A 14-month-old child, born of non-consanguineous parentage, with normal early development till 1 year of age, presented with psychomotor regression. Examination revealed developmental age of 3 months and bipyramidal signs. MRI brain (Figures 1,2) showed periventricular cavitating leukoencephalopathy (PCL). Clinical exome sequencing showed compound heterozygous likely pathogenic missense mutation in IBA57 gene.

PCL with peripheral restricted diffusion along its margins helps in 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, COX6B variants), and MFN2 gene mutations.\(^{1,2}\)

Figure 1 title: *IBA57 Mutation associated Cavitating Leukoencephalopathy*

**Legend to Figure 1:** Corresponding axial T1 (a,b), T2 (c,d) and FLAIR (e,f) images shows hyperintensities in frontal, parieto-occipital, periventricular white matter with cavitating lesions suggesting a cavitating leukoencephalopathy.
Figure 2 title: **Diffusion weighted images and chromatogram of IBA57 Mutation**

**Legend to Figure 2:** Diffusion weighted images (a,b) shows restriction (yellow arrows) with low apparent diffusion coefficient values (c,d -white arrows) along the margins. Sanger sequence chromatogram shows variation in exon 3 of *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.

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References


Appendix 1. Authors

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