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Visual Diagnosis

Cerebellar Cysts and Dysplasias: More Diagnoses to Consider

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This 2.5-year-old boy presented with developmental delay from early infancy and tremulousness while reaching for objects. Examination revealed microcephaly (46 cm, −3.57 *z* score), generalized hypotonia, hyporeflexia, limb dysmetria, and gait ataxia, but no muscular weakness. On ophthalmologic assessment, he had high myopia bilaterally. Magnetic resonance imaging revealed cerebellar dysplasia and multiple cortical-subcortical cysts in the cerebellum (Fig). A next-generation whole-exome sequencing revealed a homozygous, pathogenic frameshift variant,*Chr18(GRCh37):g.6997844_6997845delNM_005559.3:c.4702_4703delp.(Leu1568Glyfs*2)Exon 33*, confirming the diagnosis of *LAMA 1*-associated Poretti–Boltshauser syndrome. The child began rehabilitation, and corrective glasses were given for myopia.

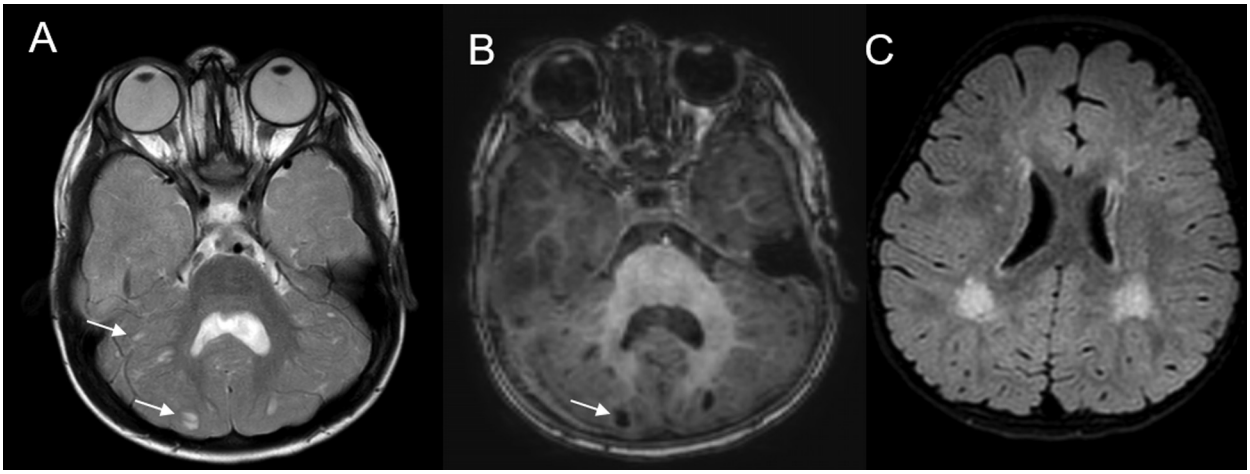

FIGURE. Axial magnetic resonance images (T2 (A), T1 (B), and fluid-attenuated inversion recovery (C)) of the posterior fossa showing cerebellar dysplasia with multiple cortical-subcortical cysts (white arrows). The fourth ventricle is dysmorphic, enlarged, and crescent shaped. In addition, bilateral peritrigonal periventricular white matter changes are also seen (FigC).

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Cerebellar dysplasias with cysts are believed to arise from genetic or acquired developmental defects of cerebellar foliation and fissure formation.[1,2] These malformations occur most commonly in patients with congenital muscular dystrophies, particularly alpha-dystroglycanopathies.[3] However, Aldinger and colleagues reported seven children with *LAMA1* mutation and similar neuroimaging findings. The reported children had ataxia, intellectual disability, and language impairment but no muscular weakness.[4] The characteristic neuroimaging changes described by these authors were cerebellar hemispheric dysplasia, cerebellar cysts, vermian hypoplasia, elevated and splayed superior cerebellar peduncles, short pons, thin isthmus, enlarged tectum, arachnoid cyst, enlarged square-shaped fourth ventricle, large globes, and cortical white matter changes.[4]

We conclude that *LAMA 1*-gene-related Poretti-Boltshauser syndrome is a cause of cerebellar dysplasia with cysts without associated muscular dystrophy.

References

1. Mickalizi A, Poretti A, Romani M, et al. Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Poretti–Boltshauser syndrome). Eur J Hum Genet. 2016;24:1262–1267.
2. Poretti A, Boltshauser E. Cerebellar dysplasia. In: Boltshauser E, Schmahmann J, eds. Cerebellar disorders in children. London: Mac Keith Press; 2012:172–176.
3. Clement E, Mercuri E, Godfrey C, et al. Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. Ann Neurol. 2008;64:573–582.
4. Aldinger KA, Mosca SJ, Tétreault M, et al. Mutations in LAMA1 cause cerebellar dysplasia and cysts with and without retinal dystrophy. Am J Hum Genet. 2014;95:227–234.