Teaching NeuroImages: CLOVES Syndrome

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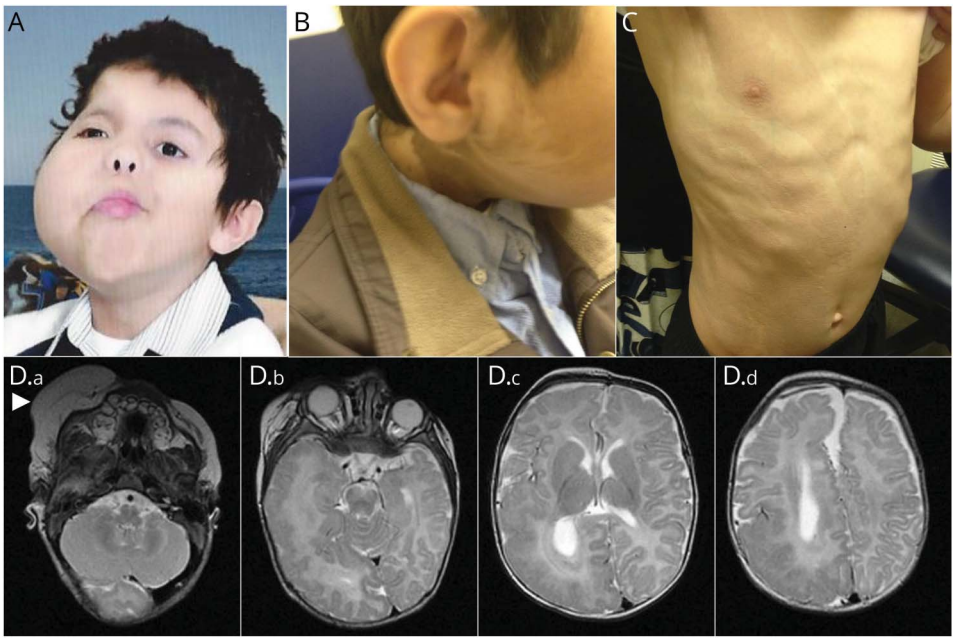
Figure Clinical and Radiologic Findings



(A) Right hemifacial overgrowth in our patient with CLOVES syndrome. (B) Epidermal nevus on the right face and neck. (C) The epidermal nevus on the trunk, characterized by hyperpigmentation and epidermal thickening, follows Blaschko lines and suggests the presence of an underlying somatic mutation. (D.a–D.d) Axial 1.5T brain MRI at age 2 months reveals right hemimegalencephaly, enlarged right ventricle, and extensive cortical dysplasia in the right temporal, parietal, and occipital lobes. There is blurring of the gray-white border and polymicrogyric appearance of the cortex. Note the lipomatous overgrowth of the right face (arrowhead) (D.a). CLOVES = congenital lipomatous overgrowth with vascular, epidermal, skeletal, and spinal anomalies.

A 17-year-old boy was diagnosed with congenital lipomatous overgrowth with vascular, epidermal, skeletal, and spinal anomalies (CLOVES) syndrome,[1] mainly affecting his right face, brain, and trunk (MIM#612918) (figure, A–C). Brain MRI revealed right hemimegalencephaly with extensive temporo-parieto-occipital cortical dysplasia (figure 1, D1-4). He developed neonatal drug-resistant seizures requiring right hemispherectomy at 15 months. He has left hemiparesis and intellectual disability. CLOVES syndrome is a segmental overgrowth syndrome associated with somatic hyper-activating mutations in *PIK3CA*, belonging to the mammalian target of rapamycin signaling pathway.[2] Genetic testing on buccal swab revealed a pathogenic somatic missense mutation in *PIK3CA* (NM_006218.4:c.1624G>A, p.Glu542Lys) at an alternate allele frequency of 4.5%, which was absent in blood.

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