Teaching NeuroImages: CLOVES syndrome

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This 17-year-old male is 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 1.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 hyperactivating mutations in \textit{PIK3CA}, belonging to the PI3K/AKT/mTOR signaling pathway\(^2\). Genetic testing on buccal swab revealed a pathogenic somatic missense mutation in \textit{PIK3CA} (NM_006218.4:c.1624G>A,p.Glu542Lys) at an alternate allele frequency of 4.5%, which was absent in blood.
### Appendix 1. Authors

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**Teaching Slides** - [http://links.lww.com/WNL/B224](http://links.lww.com/WNL/B224)
References


Figure Legend

Figure 1. Clinical and radiological findings

(A) Right hemifacial overgrowth in our patient with CLOVES syndrome. (B) Epidermal nevus on 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 the age of 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 grey-white border and polymicrogyric appearance of the cortex. Note the lipomatous overgrowth of the right face (arrowhead) (D.a).
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