ARID1B Syndrome

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Coffin-Siris syndrome (CSS) is a rare congenital malformation syndrome, caused by mutations in the ARID1B gene in over half of the cases. While the clinical characteristics of the syndrome have been increasingly described, a detailed evaluation of the epileptic phenotype in patients with ARID1B alterations and CSS has not been approached yet. We report seven patients with ARID1B-related CSS, focusing on epilepsy and its electroclinical features. The evolution of epilepsy and EEG findings of children with CSS are described and compared with patients previously reported in the literature. The patients described here reveal common features, consistent with those of patients previously described in the literature. The epilepsy phenotype of CSS due to ARID1B pathogenic variants may be described as focal epilepsy with seizures, variable in frequency, arising from motor areas, with onset in the first years of life and susceptibility to fever, and interictal perisylvian (centrotemporal) epileptiform abnormalities that are enhanced during sleep with possible evolution to an EEG pattern of continuous spike and wave during sleep (without documented developmental regression). Additional information emerging from other patients is needed to confirm this definition.