Jordan University of Science and Technology

Mutations in SWI/SNF chromatin remodeling complex gene ARID1B cause Coffin-Siris syndrome.

Authors: Santen GW, Aten E, Sun Y, **Almomani R**, Gilissen C, Nielsen M, Kant SG, Snoeck IN, Peeters EA, Hilhorst-Hofstee Y, Wessels MW, den Hollander NS, Ruivenkamp CA, van Ommen GJ, Breuning MH, den

Abstract: We identified de novo truncating mutations in ARID1B in three individuals with Coffin-Siris syndrome (CSS) by exome sequencing. Array-based copy-number variation (CNV) analysis in 2,000 individuals with intellectual disability revealed deletions encompassing ARID1B in 3 subjects with phenotypes partially overlapping that of CSS. Taken together with published data, these results indicate that haploinsufficiency of the ARID1B gene, which encodes an epigenetic modifier of chromatin structure, is an important cause of CSS and is potentially a common cause of intellectual disability and speech impairment.