

Coffin Siris Syndrome Genereviews

Jessica J Manson

Figure 1. [Coffin-Siris syndrome classic features]. - GeneReviews ... For clarity, excerpts of GeneReviews chapters for use in lab reports and clinic notes are a permitted use. For more information, see the GeneReviews® Copyright Notice and Usage Disclaimer. ARID1B-Related Disorder - GeneReviews® - NCBI Bookshelf May 23, 2019 · Note: The information presented in the Coffin-Siris syndrome GeneReviews chapter includes information on individuals with CSS from a variety of genetic causes including ARID1B but is not specific to individuals with a pathogenic variant in ARID1B. *Coffin-Siris syndrome - Orphanet* More than 150 cases of genetically confirmed Coffin-Siris syndrome (CSS) have been clinically reported to date. Exact prevalence and incidence are not known but the disorder is probably under-recognized. **Entry - #135900 - COFFIN-SIRIS SYNDROME 1; CSS1 - OMIM** Coffin-Siris syndrome is a multiple malformation syndrome characterized by impaired intellectual development associated with coarse facial features, hypertrichosis, sparse scalp hair, and hypoplastic or absent fifth fingernails or toenails. Coffin-Siris Syndrome - GeneReviews® - NCBI Bookshelf Apr 4, 2013 · Coffin-Siris syndrome and related disorders involving components of the BAF (mSWI/SNF) complex: historical review and recent advances using next generation sequencing. *Coffin-Siris Syndrome - PubMed* May 15, 2025 · Diagnosis/testing: The diagnosis of CSS is established in a proband with suggestive findings and a heterozygous pathogenic variant in one of the following 14 known genes identified by molecular genetic testing: ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, CMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, or SOX11. **COFFIN-SIRIS SYNDROME - Wiley Online Library** Oct 30, 2020 · Coffin-Siris syndrome (CSS) is a rare congenital malformation syndrome. The disorder is caused by heterozygous mutations in several genes encoding components of the BRG1- and BRM-associated factor (BAF) complex, including ARID1A, ARID1B, SMARCA4, SMARCB1, SMARCE1, and SOX11. Coffin-Siris syndrome: MedlinePlus Genetics Coffin-Siris syndrome appears to follow an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Coffin-Siris syndrome - NIH Genetic Testing Registry (GTR) - NCBI Clinical resource with information about Coffin-Siris syndrome and its clinical features, available genetic tests from US and labs around the world and links to practice guidelines and authoritative resources like GeneReviews, PubMed, MedlinePlus, clinicaltrials.gov, PharmGKB Coffin-Siris syndrome (Concept Id: C0265338) - National Center ... Coffin-Siris syndrome (CSS) is classically characterized by aplasia or hypoplasia of the distal phalanx or nail of the fifth and additional digits, developmental or

cognitive delay of varying degree, distinctive facial features, hypotonia, hirsutism/hypertrichosis, and sparse scalp hair.

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