ARID1B Syndrome

https://pubmed.ncbi.nlm.nih.gov/23556151/

Clinical characteristics:

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. Congenital anomalies can include malformations of the cardiac, gastrointestinal, genitourinary, and/or central nervous systems. Other findings commonly include feeding difficulties, slow growth, ophthalmologic abnormalities, and hearing impairment.

Diagnosis/testing:

Before the molecular basis was known, the diagnosis of CSS was based solely on clinical findings (although consensus clinical diagnostic criteria have not yet been published). The diagnosis of CSS is established in a proband with suggestive findings by identification of a heterozygous pathogenic variant in one of the genes listed in Table 1.

Management:

Treatment of manifestations:

Occupational, physical, and/or speech therapies to optimize developmental outcomes. Feeding therapy, nutritional supplementation and/or gastrostomy tube placement as needed to meet nutritional needs. Routine management of ophthalmologic abnormalities and hearing loss.

Surveillance:

Yearly evaluation by a developmental pediatrician to assess developmental progress and therapeutic and educational interventions; follow up with a gastroenterologist and feeding specialists as needed to monitor feeding and weight gain. Routine follow up of ophthalmologic and/or audiologic abnormalities.

Genetic counseling:

CSS is inherited in an autosomal dominant manner; however, most affected individuals have the disorder as the result of

de novo

CSS-causing pathogenic variant. If the CSS-causing pathogenic variant has been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic testing are possible.