Exploring Oral Manifestations and Dental Management of Coffin-Siris Syndrome

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ABSTRACT

Coffin-Siris syndrome (CSS) is a rare genetic disorder characterized by intellectual disability, developmental delays and distinctive physical features including hypoplasia of the fifth fingernails and toenails (Figures 1A & 1B), coarse facies, cardiac defects, hirsutism and sensory abnormalities. While an autosomal dominant inheritance pattern has been identified, most of the fewer than 200 affected individuals acquired the condition spontaneously. Because the syndrome is sporadic, manifests a spectrum of symptoms, and is highly phenotypically variable, definitive diagnosis is challenging and should include genetic counseling. The paucity of literature regarding dental management emphasizes the importance of identifying both clinical and oral manifestations (e.g., delayed dental eruption, crowding or diastemas, dental anomalies, etc.). Customizing dental treatment requires consideration of intellectual disability, oral manifestations, and possible cardiovascular abnormalities.

(A)  (B) 

Figures 1A-B (A) Generalized absence of nails and undeveloped phalanx on the fifth digits. (B) Hypoplasia and deformation of toes.[1]

SYNDROME OVERVIEW

Albeit the etiology of CSS has yet to be fully established, mutations in the genes of BRG-1-associated factor (BAF) chromatin-remodeling complex, including BAF250A, BAF250B, BAF200, BAF47, BRG1 and BAF57, could potentially be the causes of the syndrome (Figure 2).[2]

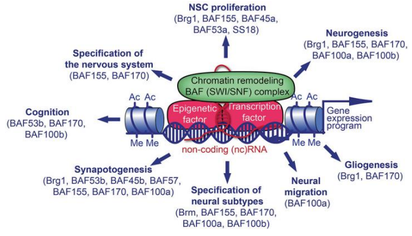


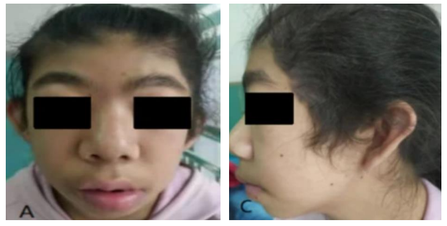
Figure 2. The BAF complex and its subunits, which influence a variety of processes involved in neural development.[2]

Currently, the diagnosis of CSS is dependent upon identification of clinical findings. Diagnostic criteria involves the following manifestations:

- Developmental delays
- Coarse facial features (Figures 3A-B)
- Hypertrichosis
- Hypoplasia or absence of fifth-digit nails, and/or
- Hypoplasia of the fingers and toes, typically, the fifth-digit[3]

CASE REPORT: MEDICAL HISTORY

A 10-year-old female presented to the Stony Brook Children's Hospital reporting a medical history remarkable for CSS, autism spectrum disorder and 48 months post-therapy for pH + b-cell acute lymphoblastic leukemia (ALL). Chemotherapeutics administered during antineoplastic therapy included imatinib mesylate (tyrosine-kinase inhibitor), methotrexate and mercaptopurine (6-MP). The mother and physician noted dental abnormalities.

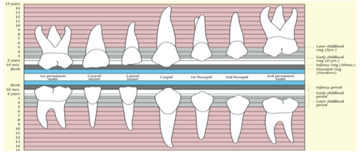


Figures 3A-B. Clinical photos of a 12-year-old female patient diagnosed with CSS obtained from Tao et al. **(A)** Photos exhibit coarse face, hair thinning, thick eyebrows and wide nose tip. **(B)** Image captured from the left lateral aspect displays micrognathia and several nevi.[4]

CASE REPORT: DENTAL HISTORY

A 10-year-old female diagnosed with CSS was referred to the Stony Brook University Dental Care Center Department of Orthodontics and Pediatric Dentistry. Accompanied by her mother, the patient presented for initial examination with chief complaint of "her teeth coming in look funny." The mother reported no tooth pain, no decay and no dental problems. Oral hygiene via toothbrush is once daily, mainly before bedtime, with assistance from the mother. The mother noted the patient exhibits nocturnal bruxism. The patient seldom consumes sweet foods, but frequently drinks soda and unsweetened iced tea. Last recall exam and prophylaxis occurred in October 2023 with an outside provider at the established dental home. Clinical examination revealed over-retained primary dentition #E and #F labial to partially erupted, rotated teeth #8 and #9. No decay, discoloration, nor abnormal tooth morphology were observed. Patient demonstrated Frankl 2 behavior and thus no radiographs were obtained. Treatment plan for next visit consists of desensitization, radiographs, prophylaxis and topical fluoride application. In addition, consideration of calcification of permanent dentition in light of chemotherapeutics should be factored into the chief complaint (Figure 4).[5] Antineoplastic treatment could lead to tooth and root morphology abnormalities,[6,7] and sequelae render a heightened risk to caries development during and after treatment.[8]

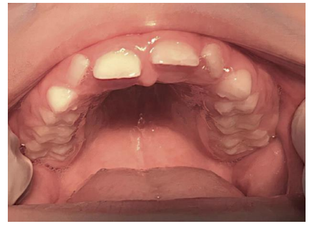
Figure 4. Calcification schedule for permanent dentition.[5]



LITERATURE REVIEW

Patients with CSS typically demonstrate clinical findings which present within the head and neck region exhibiting coarse facial features, thickened skin and abnormal facial angulation, broad nose with a depressed nasal bridge, thick lip vermillion, malformed and/or malpositioned ears, ptosis, widened mouth and a short philtrum. Hypertrichosis and hirsutism are also commonly seen along with thick eyebrows and elongated eyelashes. Of equal importance is the identification of intraoral findings in patients with the syndrome. Figueira et al. reported oral manifestations in an eight-year-old female patient with CSS including gingival hyperplasia along the maxillary arch, gingivitis due to the presence of plaque biofilm, arched palate, dolichocephalic facial type, labial hypotonia and mild enamel hypomineralization (Figure 5).[1] Houb-dine et al. published similar findings, but also noted bilateral cleft lip and palate, delayed eruption of permanent teeth and taurodontism in an eight-year-old male patient with CSS.[9]

Figure 5. Gingival hyperplasia, gingivitis and arched palate present in the maxillary arch. Clinical image obtained from Figueira et al.[1]



CONCLUSIONS

Management of CSS involves multidisciplinary care, addressing the specific needs of each individual. Early interventive programs, special education, physical therapy, and occupational therapy can optimize developmental outcomes. Treatment may target specific medical issues such as congenital heart defects or other associated abnormalities. Due to the complexity of the syndrome and the variability in its presentation, each case requires personalized medical management and support. Ongoing research into the underlying genetic causes and clinical management of CSS continues to improve understanding and care for individuals affected by this condition. Medical and dental practitioners should consider sequelae from antineoplastic therapy in cases of morphologic tooth anomalies.

REFERENCES & DISCLOSURES

Please scan the QR code to access references

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