Oral and dental abnormalities in coffin siris syndrome : a new case report

Syndrome Coffin-Siris et anomalies bucco-dentaires : une nouvelle observation

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Abstract

Background : Coffin-Siris Syndrome (CSS) is a rare genetic disorder of unknown etiology. It combines digital-ungual abnormalities, facial dysmorphism, developmental and intellectual delay, and other organ-system abnormalities. Oral and dental anomalies are rarer.

Case report : 8-year-old boy with clinical diagnosis of CSS presented facial dysmorphism, sparse hair, a flat and wide nose, absence of nails on 3rd and 5th fingers of the right hand and 3rd and 4th fingers of the left hand, malformation of the feet, toes with nail hypoplasia. Oral and dental anomalies included : bilateral complete cleft lip and palate, delayed eruption of permanent teeth, presence of supernumerary tooth and taurodontism in the first permanent molars.

Conclusion : Early diagnosis of oral problems and regular follow-up in dentist are necessary to promote good oral health and improve the patient's quality of life

Key words : Coffin-Siris syndrome, oral findings, genetic abnormality, fifth digit syndrome

Résumé

Introduction : Le Syndrome de Coffin-Siris est une affection génétique rare, d'étiologie inconnue. Il associe des malformations digito-unguéales, une dysmorphie faciale, un retard staturo-pondéral, un retard mental et d'autres anomalies asscoiées. Les manifestations bucco-dentaires sont plus rares.

Présentation du cas : Un garçon âgé de 8 ans a été diagnostiqué à la naissance du Syndrome Coffin Siris avec un retard staturo-pondéral, une résistance à l'hormone de croissance, un retard mental léger et une insuffisance pancréatique. L'examen clinique du patient a montré une dysmorphie faciale, des cheveux clairsemés, un nez plat et large, une absence des ongles sur 3ème et 5ème doigts de la main droite et 3ème et 4ème doigts de la main gauche, une malformation des pieds, des orteils avec hypoplasie des ongles. En plus des caractéristiques cliniques générales, le patient présentait des anomalies bucco-dentaires incluant une fente labio-alvéolo-palatine bilatérale, un retard d'éruption des dents permanentes, la présence de dent surnuméraire et un taurodontisme au niveau des premières molaires permanentes.

Conclusion : le diagnostic précoce des anomalies bucco-dentaires et le suivi régulier chez le dentiste sont extrêmement importants pour une meilleure santé bucco-dentaire et une meilleure qualité de vie des patients porteurs du syndrome cofin siris

Mots clés : Syndrome de Coffin-Siris, anomalies bucco-dentaires, anomalie génétique

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INTRODUCTION

Coffin-Siris syndrome (OMIM #135900) also known as the fifth-digit syndrome, is a rare genetic syndrome associated with multiple congenital anomalies due to mutations in the BAF-complex or SOX gene. Subunits involved in the BAF complex or SOX genes exhibited phenotypic differences that continued to be defined despite overall well characterized. (1,2).

The first 3 cases of this syndrome were described in 1970 by Coffin and Siris (3). The clinical manifestations can be very different. The most clinical findings include the absence or hypoplasia of the distal phalanx (and nail) of the fifth fingers and/or toes, developmental disability, organ deformities, and dysmorphic facial features with sparse hair (1-4).

Other anomalies may exist, in particular oral malformations, however, oral and dental features have not been thoroughly investigated. Here, we present a new case of CSS and discuss the associated oral anomalies. The study was conducted following the CARE case report guidelines.

Clinical observation

8-year-old boy was referred by the medical genetics department for oral care. He has received clinical diagnosis of CSS 2 years after his birth with global development delay, resistance to growth hormone, mild mental retardation and pancreatic insufficiency. The pituitary MRI was normal.

Genetic testing and molecular analysis were not performed to confirm the clinical diagnosis because of the high cost of this test and the parent's lack of financial means

There was no history of consanguinity between the parents, the child was born after a first pregnancy and an uneventful delivrey.

Physical examination revealed facial dysmorphism, sparse hair, a flat and wide nose, absence of nails on 3rd and 5th fingers of the right hand and 3rd and 4th fingers of the left hand (Fig.1a), malformation of the feet, toes with nail hypoplasia (Fig.1b). We also noted a growth deficiency (P=14 kg, T=1.04m).

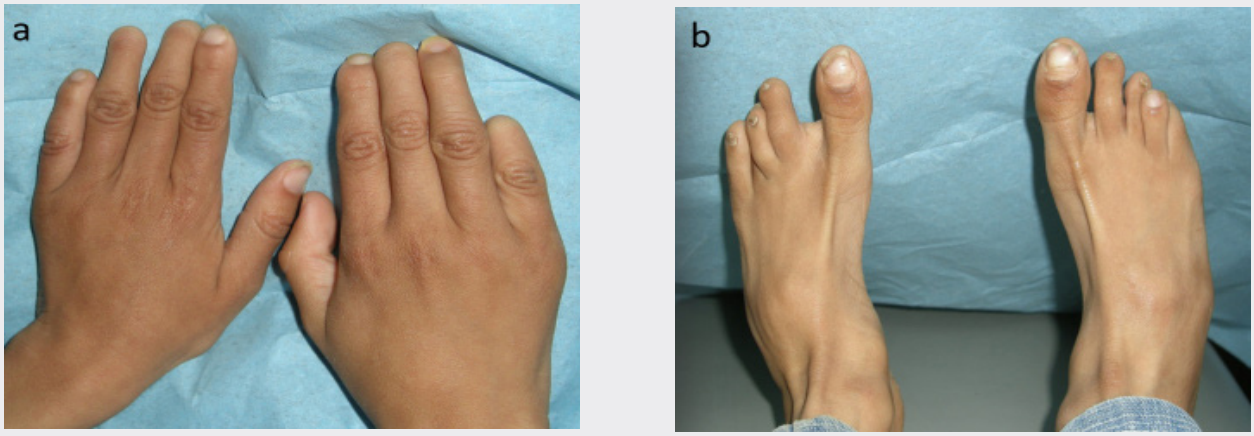

Figure 1. a. Absence of nails on 3rd and 5th fingers of the right hand and 3rd and 4th fingers of the left-hand. b. Malformations of the feet, toes with nails hypoplasia

Intra-oral examination revealed an operated bilateral complete cleft lip and palate. We noted poor oral hygiene, gingivitis with dental biofilm, caries lesions in the deciduous maxillary central incisors which presented great coronary destruction and dental abscess related to 61. Moreover, he had dento-maxillary disharmony with moderate crowding (Figure2)

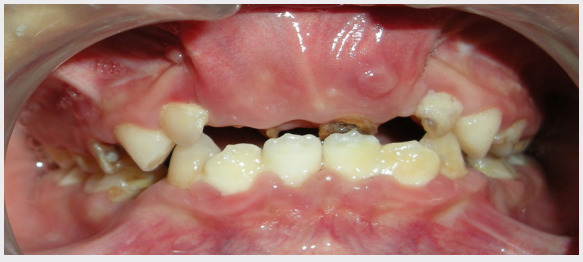

Figure 2. Intraoral Photography

Delayed eruption of permanent teeth and the presence of other dental anomalies were detected in radiographic examination : presence of supernumerary tooth (mesiodens) and taurodontism in the first permanent molars (the pulp chambers are elongated with apical displacement of the pulpal floor, and bifurcation or trifurcation of the roots) (figure 3).

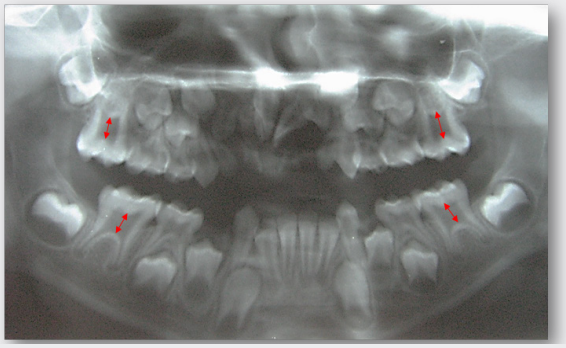

Figure 3. Panoramic X-ray showing taurodontism in the 4 first permanent molars

Treatment planning involved a multidisciplinary approach and included: instructions on oral hygiene and descaling, extraction of the mesiodens and the dilapidated teeth 61 and 71, caries care on other affected teeth and the follow-up of eruption of the permanent teeth. After dental care, orthodontic therapy (which is essentially based on patient compliance) must be reassessed because of the moderate mental delay..

DISCUSSION

Before the molecular basis was known, the diagnosis of CSS was based on clinical diagnostic criteria characterized by : aplasia or hypoplasia of the distal phalanx or the nail of the fifth fingers and/or toes, developmental delays, distinctive facial features, hirsutism mainly on the face, arms and back, associated with thinning hair (4).

Other congenital anomalies may exist, in particular malformations of the cardiac, gastrointestinal, genitourinary and/or central nervous systems (1). Other findings include feeding difficulties, slow growth, ophthalmological abnormalities and hearing impairment but these abnormalities are less constant from one patient to another (2, 5).

With the advent of large-scale genetic tests such as whole exome sequencing, more and more individuals present mutations in this pathway, and the phenotypic spectrum of CSS seems to be expanding (2).

The presented case shared many features common to all previously reported cases with CSS (developmental delay, digital anomalies and coarse facial features) and demonstrated oral and dental abnormalities include: bilateral complete cleft lip and palate, delay in eruption of permanent teeth, presence of supernumerary tooth and taurodontism in the first permanent molars.

According to the literature on the subject, Oral anomalies are rarer and non-specific, the most described are : presence of cleft palate, delayed eruption of permanent teeth, dental crowding or diastems and atypical dental defects (1, 6, 7, 8)

The cleft palate associated with Coffin Siris syndrome has been described for a long time by some authors (3, 4) and more recently :

- In 2018, Khan U. et al. (6) found for the first time a cleft palate in a boy with a rare variant of SCS (OMIM 615866)
- In 2020, Leighton Reed et al. (1) described in a 5-year-old boy with Coffin-Siris syndrome with SMARCE1 mutation palatine abnormalities which are a distinctive feature of this genotype.
- In 2022, Sofronova V et al. (7) also describes an alveolo-palatine cleft in a 3-year-old and 8-month-old boy who presents a variant of the ARID1B gene identified with the sequencing exome.

Furthermore, the literature lacks cases describing cleft lip in CSS. The delayed dentition of this patient would be linked to the global growth deficiency and the presence of mesiodens is probably related to the cleft lip and palate. According to Consolaro A. et al. (9) the presence of supernumerary teeth in cases of cleft lip and palate reaches 43.5%.

However, to the best of our knowledge, there is no publication that describes the presence of taurodontism associated with CSS. This dental anomaly is characterized by an enlarged pulp chamber, an apical displacement of the pulp floor and the absence of constriction at the cemento-enamel junction (10, 11). It most often appears as an isolated anomaly. But its association with several syndromes and abnormalities has also been reported (11). The endodontic treatment of affected teeth is considered complex and difficult due to the complexity of the dental morphology (10).

Feeding difficulties in patients with CSS would be

exacerbated if oral problems are added to the multiple congenital anomalies, therefore early management and oral care are necessary (8).

Oral management in individuals with CSS is not widely described in the literature due to the low frequency of this syndrome (1). It varies according to the needs of each individual affected and should be based on the following factors: degree of intellectual disability, oral findings with their impact on cardiovascular anomalies and influence of possible medications (8). Once the diagnosis of CSS is confirmed, dental prophylaxis, instructions on oral hygiene, dietary advice and are required.

Regular dental follow-up and multidisciplinary care are extremely important to promote good development of the child's general and oral health and improve the patient's quality of life.

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