The importance of follow-up dental findings to diagnose Sauk syndrome: a case report in a child

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Abstract
This case report presents a rare case of a 10-year-old Brazilian boy who was referred to a dental clinic of a Public Institution when he was 7-years-old because of early loss of teeth and delayed teeth eruption. Clinical features included delay in cognitive development and growth, under-weight, short stature, microcephaly, ocular hypertelorism, small but prominent ears, depressed nasal bridge, hypoplastic maxilla and prognathism. He had no nail, skin or hair abnormalities. Radiographic examination demonstrated delayed bone age, microcephaly, minimal scoliosis of the spine, taurodont mandibular and maxillary first permanent molars, diminished root formation in other mandibular and maxillary teeth, microdontia and hypodontia. Based on these dental findings after a follow up of two years, the medical history and complementary examinations, the condition of this patient was diagnosed as Sauk syndrome.

Key-words: Sauk syndrome, child, microdontia, taurodontism, hypodontia

Introduction
Originally described in 1973 (Sauk and Delaney, 1973), Sauk Syndrome (SS) is a recessive autosomal condition, with no detected numerical and/or structural alterations in the chromosomes (Sauk and Witkop, 1990). Of the four cases reported in the literature, three were male and presented normal G band karyotypes (Sauk and Delaney, 1973; Sauk and Witkop, 1990; Gardner and Girgis, 1977).

Among the chief clinical manifestations of the condition are microcephaly, low birth weight and proportionate small stature (Gardner and Girgis, 1977). The pathognomonic symptoms of the condition are alterations in dental development, such as taurodontism, microdontia and short-rooted teeth (Sauk and Delaney, 1973; Gardner and Girgis, 1977; Sauk and Witkop, 1990). Furthermore, teeth may present with mobility and exfoliate spontaneously (Sauk and Witkop, 1990). Radiographic findings include taurodont molars which may contain pulpal calcifications, short rooted anterior teeth which may undergo external resorption, or are associated with radiolucent areas in the absence of a history of trauma (Sauk and Witkop, 1990).

The principal SS effects (Sauk and Witkop, 1990) are a delay in cognitive development, premature loss of teeth, delay in the eruption of permanent teeth, malocclusion and diastemae. SS can only be conclusively diagnosed once the early mixed dentition is established (Sauk and Witkop, 1990). However, if this condition is suspected, a follow-up is fundamental so that alterations in dental development which are manifestations of this syndrome may be identified.

The aim of this paper is to report the case of a male child in whom Sauk syndrome was diagnosed as consequence of dental findings gathered over a two-year period of follow-up, as well as to describe other manifestations which have not previously been associated with SS.

Case Report
A 7-year-old male Caucasian patient was referred to the paediatric dental clinic at the School of Dentistry of a public university in Rio de Janeiro. His complaint was of premature loss of some primary teeth and a delay in the eruption of the permanent successors. He was accompanied by his guardian.

During initial consultation it was noted that the patient was the first-born child of non-consanguineous parents, both of small stature, and that he had a healthy sister. During his gestation the mother suffered hypertension
Figure 1. Frontal view of the patient showed a Class III Angle’s molar relation; bilateral posterior cross bites, the absence of the upper six anterior primary teeth and the upper primary molars. Plaque accumulation and brown extrinsic pigmentation in the 73, 72, 71, 81, 82 and 83 and carious lesions on the distal surface of 71 and 81.

Table 1. Clinical manifestations of the proband

- Delayed growth and cognitive development *
- Low weight*
- Short stature*
- Microcephaly*
- Hypertelorism
- Small but prominent ears
- Hypoplastic nares
- Short philtrum
- Lack of sealing lips
- Maxillary hypoplastic and mandibular prognathism
- Depressed nasal bridge
- No hair, skin, nails and sweats anomalies
- Pes valgus
- Thorax-lombar scoliosis
- Campodactyly of five finger of the hands

* Features associated with SS previously described in the literature.

Figure 2. Panoramic radiography showing 73 and 83 with conservative endodontic therapy, agenesis of 15, 25, 35, 31, 44, 45; size and crown form alteration of 23, delay of development of root formation of 13, 12, 11, 21, 22, 23, 24; and suspect of taurodontism of 16, 26, 36 and 46.

crises, for which she was not given any treatment. There were complications during childbirth, with the detection of a previously unidentified congenital cardiac malformation in the mother.

The patient was born at term by caesarean section, weighing 2,550g. He was 48cm long with a 32cm skull diameter hence, he was small for his gestational age. The patient's medical history revealed some episodes of infection during childhood including pneumonia at one month, seven months and two years of age and a urinary infection at five years.

Clinical examination revealed microstomia, a thin upper lip, a hypoplastic maxilla, deep palate, bilateral posterior cross bite and mandibular prognathism. Dentally, his first permanent molars were present and appeared healthy, but the primary mandibular anterior teeth displayed extreme mobility (Figure 1). There was also dental caries in 71 and 81 and restorations in 83, 82, 72 and 73. Asked about the reason for the absence of the remaining teeth, the guardian reported that ‘the teeth which were decayed, had become mobile, and because the child experienced pain the teeth were taken out by a dentist’.

The panoramic radiograph confirmed the clinical findings (Figure 2). As well as pulpal therapy in teeth 73 and 83, there was absence of some primary and permanent teeth, alteration in the form of the upper left canine, absence of radicular formation in succeeding permanent teeth and taurodont first permanent molars.

As a consequence of his medical history and the radiographic findings, the patient was referred to the genetics sector of the same educational institution. The evaluation performed by the geneticist verified a series of clinical alterations (Table 1). Some of these characteristics can be seen in Figures 3A, B and C. However, the patient’s band G karyotype test, as well as his ophthalmology examination, were normal.

In order to complete the diagnostic investigation, further tests were requested (Table 2). Based on their results, and taking into consideration the clinical and the radiological evidence, a provisional SS diagnosis was suggested. This diagnosis was confirmed after a two-year follow-up period, which started with the radiographic confirmation (Figure 4) of the alterations in dental development (Figures 5 A and B).

During the follow-up period, the patient was initially given dental treatment in the ambulatory clinic, but afterwards the visits had to be moved to the hospital, owing to difficulty in controlling the patient’s behaviour. To this end, and before the actual procedure, a surgical risk assessment was requested. The preventive-restorative dental treatment was performed with no problems. Because of the necessity of restoring form, function and aesthetic aspects to the patient’s teeth, a second phase of the dental treatment was planned. This second stage will include, besides the removal of the embedded teeth, the placing of implant retained prostheses once growth is complete.
Table 2. Complementary examinations and their results

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<th>Hormonal tests</th>
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<td>DHEAS</td>
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<td>17-Hydroxyprogesterone</td>
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<td>Cortisol</td>
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<td>Thyrode: T3, T4, T4 free, TSH</td>
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<td>GH hormone: basal and stimulated</td>
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<td>Echo-dopplercardiography - normal</td>
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<td>Abdominal ultrasound imaging - normal</td>
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<td>Audiometric tests - normal: Type C Tympanogram tracing; bilaterally absence of stapedial reflex</td>
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<td>Spirometry – low pulmonary function capacity</td>
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<td>Bone age – low: compared to 3 years-old child</td>
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<td>Simple X-Ray of cranium - microcephaly and hypoplastic posterior corpus callosum</td>
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<td>Spinal radiography – minimal right scoliosis of the spine</td>
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Figures 3A, B and C. Some clinical features of the proband.
Figure 4. Panoramic radiography after two years of follow-up, showed taurodontism of 16, 26, 36 and 46; short crown and short root formation in 13, 12, 11, 21, 22, 23, 33 and 43; lack of root formation of 17, 14, 24, 25, 27, 37, 34 and 47, agenesis (oligodontia) of 15, 35, 31, 44, 45 and crown form alteration of 41 and 25. Carious lesions in 26 and 36.

Figure 5A. Frontal view of oral conditions of the patient after two years of follow-up.

Figure 5B. Occlusal view of the lower permanent incisor teeth. Note the alteration of the crown in relation to the size and the form of 41.

In order to assess the genetic background of the patient, the medical and dental histories of the patient’s mother and sister were evaluated. It was observed that the mother had a phenotype similar to the proband’s, as well as proportionate small stature and delayed cognitive development (Figures 6 A, B and C). No alteration was detected in the sister. Under radiological examination, no dental alteration, of the type associated with SS was verified in either mother or sister (Figures 7 A and B), although microdontia was diagnosed in the mother’s upper lateral incisors. It was also
ascertained that the patient's mother and sister were in need of dental treatment.

Discussion

In the aetiopathogenesis of SS, both genetic and environmental factors may be present. In pathological conditions with a small number of cases described in the literature, it is difficult to establish an inheritance pattern. For SS, an autosomal recessive inheritance pattern has been postulated (Sauk and Witkop, 1990). Nevertheless, on the basis of this report, in which the mother presented some characteristics similar to that of the patient, another kind of inheritance is possible, such as autosomal dominant inheritance (variable penetrance and expressiveness) and x-linked (recessive) chromosome (oligosomatic carrier). However, the familial history in the cases of this syndrome, together with the tracing of the outcomes for the cases already published and molecular studies, will help to establish a more precise definition of the inheritance pattern of Sauk syndrome.

SS is characterised by alterations in dental development, which are determined after the age of seven (Sauk and Witkop, 1990). Therefore, two stipulations have to be met for the taurodont condition to be confirmed (Constant and Grine, 2001): not only must the eruption of the permanent first molars have occurred, but also the closure of at least two-thirds of their radicular apices. Furthermore, it is necessary to diagnose the reduced size of the teeth and of their roots (Neville et al., 1995). In the present report, the diagnosis was provisionally established on the basis of dental characteristics. Nevertheless, a two-year follow-up period was necessary for the dental developmental features, such as taurodontism and short-rooted teeth, to be conclusively diagnosed. Unlike the cases reported in the literature (Sauk and Delaney, 1973; Gardner and Girgis, 1977; Sauk and Witkop, 1990), other teeth also showed much reduced root formation, in addition to malformations.

The detailed clinical investigation was fundamental to the establishment of the final diagnosis. The following conditions were considered in the differential diagnosis: fibrous dysplasia (Akintoye et al., 2003), osteodysplastic primordial nanism type II (Hall et al., 2004), otodontal syndrome (Gregory-Evans, 2002), Seckel syndrome (Favre et al., 2002), Witkop syndrome (Hodges and Harley, 1999), Sensenbrenner syndrome (Amar et al., 1997), cranioectodermal dysplasia (Zanoli et al., 2001), tri-codentoosseous syndrome (Houlston et al., 1994) among others (OMIM, 2005).

There are no recommendations in the literature about specific precautions concerning the dental care for people presenting with SS. Thus, the dental treatment was initially performed at the ambulatory clinic. However, owing to difficulty in controlling the patient's behaviour (McDonald and Avery, 2001), the first stage of the treatment was in hospital under general anaesthesia. One of the advantages of this approach was to provide the treatment in a single clinical session (AAPD, 2004). Another alternative for the execution of the treatment would have been conscious sedation (AAPD, 2004), but the patient's limited respiratory capacity contraindicated its use.

By contrast with the previously reported cases (Sauk and Delaney, 1973; Gardner and Girgis, 1977; Sauk and Witkop, 1990), this patient with SS was extremely aggressive. In addition, there was a delay in the eruption of the majority of the permanent teeth, a much reduced degree of root
formation and, consequently, some teeth remained unerupted. Such alterations constitute additional clinical findings, which have not previously been associated with SS. Their presence reinforces the possibility of a differentiated phenotypic expression. Furthermore, the dental alterations detected in this case have not previously been associated with SS. These newly verified data include oligodontia, modifications in disturbance, namely, type B. Out of the twenty-three ectodermal dysplasia, expressing phenotypic variability. This ectodermal alterations may characterise the rare type B (Neville et al., 1995) or even the total absence of these in every tooth, with the exception of the lower permanent molars and permanent incisors.

However, the presence of teeth with taurodontism, teeth with minimal root formation (or even the complete absence of roots), and oligodontia in the same patient, appears to be an anthropological contradiction (Sauk and Delaney, 1973; Gardner and Girgis, 1977). According to Schalk-Van der Weide et al. (1993), such a contraindication arises as the result of ectodermal alterations, which configure instability in the dental development.

Freire-Maia and Pinheiro (1994) emphasise that the concomitant manifestation of ectodermal alterations are characteristic of an ectodermal dysplasia. Two alterations of ectodermal structures occur. One of them necessarily involves one of the classic signs, i.e. hair or skin or teeth or glands; and the second involves any other ectodermal disturbance, namely, type B. Out of the twenty-three described alterations, only one represents the type B.

In the present report, the patient displays two ectodermal alterations: one of dental origin (oligodontia, taurodontia and microodontia) (Neville et al., 1995), and the other of neurological provenance (microcephaly) (Moore and Reid, 1983). According to Freire-Maia and Pinheiro (1994) such ectodermal alterations may characterise the rare type B ectodermal dysplasia, expressing phenotypic variability. This needs to be considered because the first SS case reports were described 21 years before the classification proposed by Freire-Maia and Pinheiro (1994). Thus, further, multidisciplinary investigations are necessary to elucidate this condition.

It is evident from the foregoing that dental signs are of importance in arriving at a diagnosis for this syndrome.

References
OMIM: Online Mendelian Inheritance in Man™. Center for Medical Genetics, John Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD) www.ncbi.nlm.nih.gov/omim), 2005

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