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Self-inflicted orodental injury in a child with Leigh disease

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Summary. Leigh disease is an inherited progressive mitochondrial neurodegenerative disease that affects the neurological, respiratory and cardiovascular systems and is associated with retardation of the intellectual and physical development. This report describes the case of a 4-year-old boy with Leigh disease who presented with self-inflicted traumatic injury to the teeth, alveolar bone, lips and tongue during repeated episodes of intense orofacial spasms. Conservative management of the injury included repositioning the fractured alveolar bone, splinting the traumatized teeth and planning for a mouthguard. However, after a second incident of severe self-induced injury to the teeth and alveolar bone, extraction of the anterior teeth became inevitable to protect the child from further self-mutilation and to allow healing of the injured tissues.

Introduction

First described in 1951, Leigh disease or 'subacute necrotizing encephelomyelopathy' is an inherited metabolic degenerative disease of the central nervous system [1–3], and it is considered the most common mitochondrial metabolic disorder of infancy and childhood [4], its onset most frequently occurring between 3 months and 2 years of age [1,2]. However, the onset of the disease may be delayed to adolescence or adulthood, when it is usually preceded by a chronic stage of mild neurological symptoms [2,3]. The sex ratio of Leigh disease varies according to the age of onset, with a male: female ratio of 3 : 2 in the infantile form, compared with a ratio of 4 : 1 in the juvenile form of the disease [3].

Leigh disease is a genetic disorder. The infantile and childhood forms show an autosomal recessive mode of inheritance, whereas most juvenile and adulthood cases are sporadic [1–3]. The genetic defect involves deficiency and perturbed activity of cytochrome c oxidase (COX), with the identified gene being SURF1 [5], which results in progressive neurological degeneration [5,6]. Leigh disease could also involve deficiency in pyruvate dehydrogenase and pyruvate carboxylase resulting in perturbed metabolism of brain lipids and abnormal neurological signs [1,2]. The laboratory findings include elevated levels of alanine, pyruvate and lactate in the serum, urine and cerebrospinal fluid [2].

Generally, the onset of Leigh disease is insidious although it may be acute or subacute [3]. The course of the disease is variable and follows the rate of neurodegeneration; its evolution may take months to years [3]. The first noticeable signs are feeding difficulties, weakness in the suckling ability, dysphagia, regurgitation, loss of appetite, vomiting and irritability. Most children suffer from failure to thrive and their height is below the 10th percentile [2,3]. The neurological signs include ataxia, dystonia, rigidity, generalized or partial seizures, athetosis and choreoathetosis. As the disease progresses, the patient suffers from irregular respiration and apnoea. Involvement of the cardiovascular system is manifested by perturbation in the cardiac rhythm and cardiomyopathy. Other symptoms include nystagmus, optic atrophy, weakness of the extra ocular movements, deafness, speech delay, motor retardation and lactic acidosis [1–3]. The terminal stage of Leigh disease is marked by accelerated neurological deterioration, significant cardiac perturbation and respiratory depression leading to coma and death [2–5]. The maximum survival age

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varies in relation to the age of onset and ranges from 0·9 to 5·5 years [3].

Histological examination of the affected neurological tissues reveals multiple areas of spongy necrosis and microscopic vascular proliferation leading to breakdown of the myelin and tissue degeneration [1,3,4]. These lesions have been found in the basal ganglia (striatum, thalamus and hypothalamus), the brain stem and sometimes in the cerebellum and spinal cord [3,4]. Moreover, it has been suggested that the pathological picture of Leigh disease resembles Wernicke's disease, although haemorrhage and involvement of mamillary bodies are rare in Leigh disease [3,4].

Treatment of Leigh disease is palliative and includes supportive administration of thiamine, Vitamin B1 and lipoic acid. Oral sodium bicarbonate may be given to manage lactic acidosis. Sedatives and analgesics are used to relieve the patient from the clinical symptoms [1,2].

The present report describes the dental management of a child affected with Leigh disease who suffered from severe orodental injuries resulting from peculiar self-destructive behaviour.

Case report

A 4-year-old boy was admitted to the Capital Health Authority, Division of Paediatric Neurology at the Stollery Children's Hospital in Edmonton, Alberta, with a diagnosis of Leigh disease, respiratory distress and aspiration pneumonia. During his hospitalization, the child suffered from progressive neurological degeneration, pneumonia, sepsis and lactic acidosis; he required intubation and ventilation for a period of time in the Intensive Care Unit. Concurrently, the child demonstrated severe learning disability, violent physical movements of head and extremities, periods of intense facial spasms and teeth clenching. Intravenous antibiotics were required to treat the severe infection and sloughing of his right thumb resulting from repeated intense digit biting.

After an episode of self-destructive physical behaviour, severe self-inflicted injuries to the teeth and tissues of the oral cavity were noted by the child's paediatrician who referred him to the dental clinic for a consultation with the paediatric dentist. Global assessment of the child revealed intellectual delay with severe learning and communication disability, extreme uncooperative behaviour with strong and sudden movements of the head and extremities.

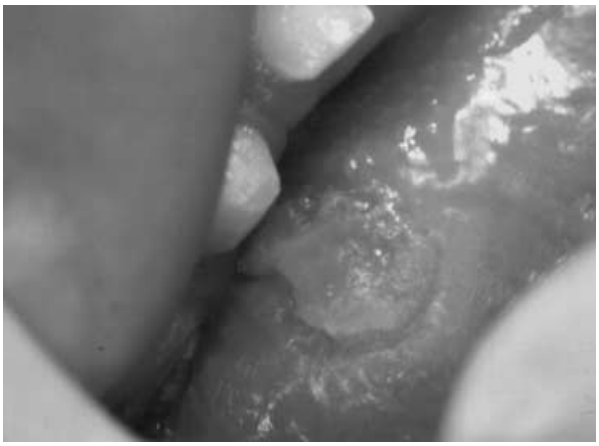

Fig. 1. Ulceration on the right border of the tongue resulting from tongue biting.

On presentation, the child's hands were restrained with plastic wrap to prevent the child from biting his fingers. Extra oral examination showed severe facial grimacing, continuous tooth clenching, drooling, repeated and intense tongue thrusting movements and inflammation with mild swelling of the upper lip. Intraoral examination showed a full complement of primary teeth, poor oral hygiene, and 2×1 cm traumatic ulceration on the lateral right border of the tongue (Fig. 1). Examination of the mandibular teeth revealed fracture of the alveolar cortical bone, laceration of the gingival tissues, severe mobility and lateral displacement in the facial direction of the mandibular central and lateral incisors (# 81, 82 and 72), and subdisplacement of the maxillary central incisors (# 51 and 61).

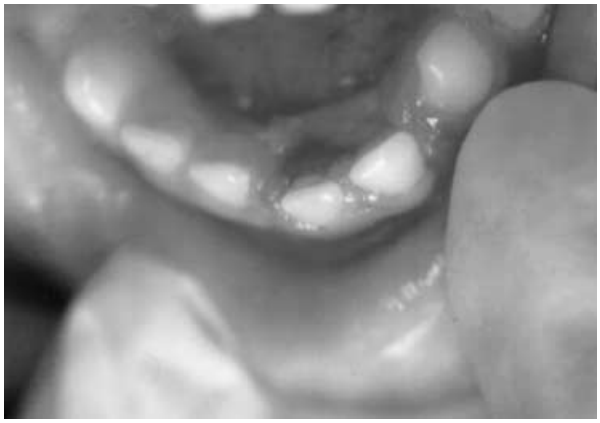

Fig. 2. Traumatic injury to the primary mandibular incisors.

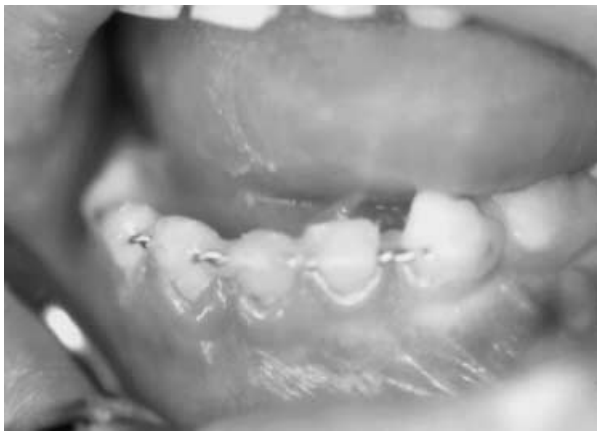

Fig. 3. Rigid composite wire splint.

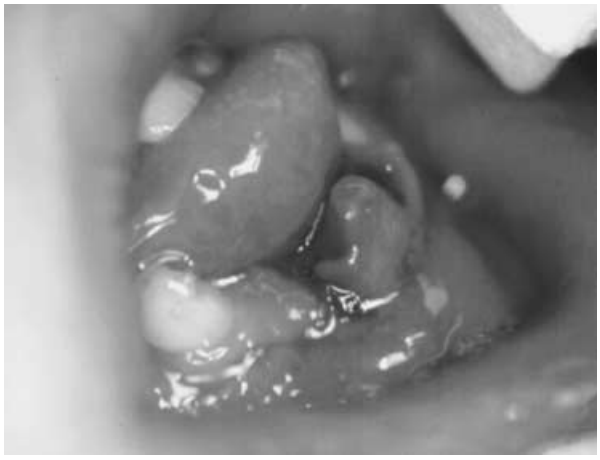

Fig. 4. Second incident of severe oro-dental self-mutilation.

Because of the child's medical history and extreme uncooperative behaviour, dental treatment was provided under general anaesthesia. Complete oral examination with intraoral radiographs was performed. Management of the traumatized teeth comprised repositioning the fractured alveolar bone and the displaced mandibular primary incisors (# 71, 72, 81 and 82), and rigid splinting of the anterior teeth (# 71, 72, 73, 81, 82 and 83) with twisted wire (0·024 inch) and composite material. To strengthen the splint, composite material was used to attach the teeth interproximally and lingually (Fig. 3). The purpose of splinting was to stabilize the displaced teeth and to obtain healing of the fractured alveolar bone. In addition, the mandibular second primary molar was restored with amalgam. Preventive measures consisted of scaling of calculus, prophylaxis and application of fluoride gel. Upper and lower alginate impressions were taken to plan for a mouthguard. Post-operative assessment was conducted 24 and 72 h postoperatively and revealed progressive healing of the injured tissues and good condition of the splint.

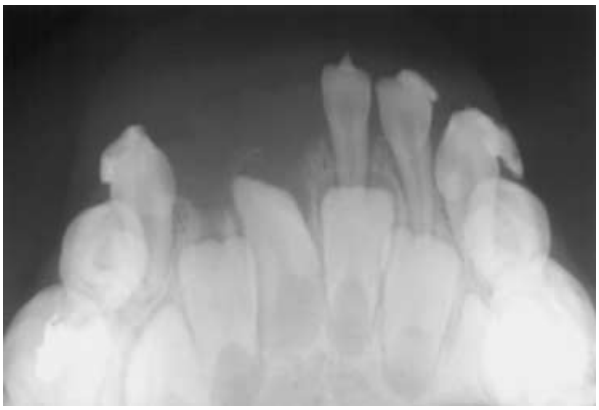

Fig. 5. Occlusal radiograph showing the extensive injury to the alveolar bone and lower incisor.

Three days later, the child was referred to the emergency dental clinic following another incident of intense spasm, tooth clenching and self-inflicted injury. This episode of violent movement and self-mutilation had occurred since the child's doses of sedatives had been reduced on the basis of compromised respiratory function. Oral examination revealed severe self-inflicted injury to the mandibular anterior segment resulting in splint fracture, severe gingival laceration and contusion, avulsion of the mandibular right central and lateral incisors (# 81 and 82), extensive fracture of the alveolar bone supporting the mandibular incisors (# 71, 72, 81 and 82), severe lateral displacement and mobility of the remaining mandibular incisors, moderate displacement and mobility of the maxillary incisors (Figs 4 and 5). Emergency management of this injury was carried out under general anaesthesia. Treatment consisted of extracting the maxillary and mandibular incisors (# 71, 72, 73, 83, 51, 52, 61, 62), repositioning the fractured bone and tight suturing. A 24 h and 7 days postoperative evaluation revealed good healing of the soft tissues and alveolar bone, and no further dento/alveolar injury was reported. The child continued repetitive tongue-thrusting movements freely with the absence of the anterior teeth, and the intensity of teeth clenching habit was much reduced.

Discussion

Self-mutilation is a repetitive learned behaviour that results in physical damage to the person. This

behaviour is compulsive, unintentional and might involve orofacial injury to the soft tissues, teeth and alveolar bone. Self-mutilation has been related to biochemical disorders, syndromes and genetic conditions such as Lesch–Nyhan syndrome [7–10], De Lange and Tourette's syndromes, learning difficulties, autism, psychological disturbances and schizophrenia [11,12]. Self-destructive behaviour may also be observed in some infectious diseases such as encephalitis [7,13], and patients with hereditary sensory and autonomic neuropathy (HSAN), which is characterized by high tolerance or insensitivity to pain [14,15]. Interestingly, self-inflicted injuries have also been noted in a child affected with Hallervorden–Spatz disease; a rare neurodegenerative condition that is characterized by progressive dystonia, rigidity and mental retardation [16]. In this case, the self-mutilating behaviour resulted in injury to the primary incisors, tongue and lips during episodes of strong orofacial muscle spasms [16].

Management of children with self-inflicted injuries is usually complicated by their lack of compliance and communication disability. There are no standard techniques to prevent or treat orofacial self-inflicted injuries. The treatment plan is established according to the special circumstances of the individual case. Sedation, behaviour modification and restraints are usually utilized to control the destructive behaviour. Orodental injuries may be managed with conservative techniques intended to protect the oral tissues, including soft/football-type mouthguard, hard resin mouthguard [9,13,16,17], acrylic splints attached to head-gear or a neck strap for retention [18] and a combination of a hard acrylic base with a silicon positioner [7]. These techniques might succeed if the appliance is well retained and tolerated by the child. In other cases, they fail because of the child's intolerance or rejection of the mouthguard, especially in patients with neuropsychiatric problems. When conservative techniques fail, extraction of the anterior teeth or full mouth clearance becomes necessary to protect the child from further injury [9,19].

In the present case, the initial conservative treatment of splinting the traumatized bone and teeth was performed on the basis of a joint decision made by the parents, paediatrician and paediatric dentist. Despite the fact that the child was sedated and his hands were restrained, he managed to self-inflict another extensive orodental injury to the anterior primary teeth and alveolar bone. Consequently, extracting the anterior teeth became inevitable to protect the child from further injury.

It is hoped that extraction of the teeth may break the self-mutilating habit, allow time for the injured tissues to heal and prevent further and more extensive injury. Similar severe self-mutilation such in cases of Lesch–Nyhan syndrome may need to be addressed with tooth extraction when other conservative measures fail [9,19]. A report by Anderson and Ernst has shown that 60% of parents would prefer tooth extraction as opposed to of a conservative modality such as mouthguard [20].

Résumé. Le syndrome de Leigh est une maladie neuro-dégénérative mitochondriale progressive, héréditaire, qui affecte les systèmes respiratoire et cardio-vasculaire. Il est associé à un retard du développement intellectuel et physique. Cet article rapporte le cas d'un garçon de 4 ans atteint de syndrome de Leigh. Celui-ci présentait une blessure traumatique auto-infligée au niveau des dents, de l'os alvéolaire, des lèvres et de la langue, survenues lors d'épisodes répétés de spasmes oro-faciaux intenses. La prise en charge des blessures a compris le repositionnement de l'os alvéolaire fracturé, la contention des dents traumatisées et la prévision d'un protège-dents. Cependant, après un second incident d'auto-mutilation sévère des dents et de l'os alvéolaire, l'extraction des dents antérieures est devenue inévitable pour protéger l'enfant d'auto-mutilations ultérieures et permettre la cicatrisation des tissus blessés.

Zusammenfassung. Morbus Leigh ist eine angeborene fortschreitende neurodegenerative mitochondriale Erkrankung, welche das Nervensystem, den respiratorischen Trakt und das kardiovaskuläre System betrifft, es kommt zu einer verlangsamten intellektuellen und physischen Entwicklung. In diesem Bericht wird der Fall eines vierjährigen Jungen mit Morbus Leigh vorgestellt, der mit selbstverursachten Verletzungen der Zähne, Alveolarknochen, der Lippe und der Zunge nach mehreren Episoden ausgedehnter orofazialer Spasmen vorgestellt wurde. Die konservative Versorgung umfasste Reposition von frakturiertem Alveolarknochen, Schienen der traumatisieren Zähne und die Planung eines Mundschutz. Nach einem weiteren selbstverursachten Trauma der Zähne und des Alveolarknochens wurde die Extraktion der Frontzähne unvermeidlich, um das

Kind vor weiterreichenden Traumatisierungen u schützen und eine Heilung zu ermöglichen.

Resumen. La enfermedad de Leigh es una enfermedad mitocondrial neuro-degenerativa hereditaria progresiva que afecta a los sistemas neurológico, respiratorio y cardiovascular, está asociada con retraso del desarrollo físico e intelectual. Este artículo describe el caso de un niño de 4 años con enfermedad de Leigh que presentaba lesiones traumáticas autoinfligidas en los dientes, hueso alveolar, labios y lengua durante episodios repetidos de intensos espasmos buco-faciales. El tratamiento conservador de las lesiones, incluyó la reducción del hueso alveolar fracturado, la ferulización de los dientes traumatizados y la planificación de un protector bucal. Sin embargo, después de un segundo incidente de lesión autoinducida severa a los dientes y el hueso alveolar, se hizo inevitable la extracción de los dientes anteriores para proteger al niño de posteriores automutilaciones y permitir la curación de los tejidos dañados.

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