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Effective treatment of self-injurious oral trauma in Lesch–Nyhan syndrome: a case report

CASE REPORT

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Abstract – Lesch–Nyhan syndrome (LNS) is an X-linked disorder originating from deficiency of the enzyme hypoxanthine guanine phosphoribosyl transferase. It is characterized by neurological manifestations, including the dramatic symptom of compulsive self-mutilation, which results in destruction of oral and perioral tissues. Several drug trials have been administered to improve the severe self-destructive behaviour, with questionable effectiveness. Invasive treatment approaches, such as extraction of teeth and orthognathic surgery, have been suggested with variable success. A conservative treatment with an intraoral appliance serving to prevent oral and peri-oral self-injury is presented in this report. The patient was a 14-year-old boy demonstrating the typical LNS behaviour, including compulsive self-biting, significant loss of lip and tongue tissue, spasticity and involuntary movements. An acrylic maxillary appliance was designed and constructed with an occlusal plate raising the bite. The appliance was retained by two Adams' clasps on the first premolars, along with three ball clasps between the incisors. Fabrication, insertion, and maintenance were uncomplicated and non-stressful to the patient. Periodic recall over 3-year period has confirmed the effective healing of the oral lesions and a high level of tolerance of the appliance.

Lesch–Nyhan syndrome (LNS) was first described in 1964 (1). It is a rare X-linked recessive disorder (2), occurring at an incidence of 1/100 000–380 000 live births (3) and affecting almost exclusively men (4, 5).

The characteristic biochemical abnormality of LNS is the congenital deficiency of the enzyme hypoxanthine guanine phosphoribosyl transferase (HPRT) which is necessary in purine metabolism. The genetic background involves mutations of the corresponding gene, which is located on the long arm of the X chromosome (Xq26-27) (6). The deficiency of HPRT activity leads to excessive uric acid production. The excess uric acid forms urate crystals, which are deposited in peripheral organs and tissues and produce neurological, renal and musculoskeletal manifestations, such as developmental delay, growth and mental retardation, choreo-athetoid spasticity, nephrolithiasis, obstructive nephropathy and acute gouty arthritis (1, 7).

The most distinctive symptom is the compulsive self-injury behaviour (SIB). The affected children appear to be normal for the first few months of life; by about 1 year of age they develop hypertonicity and involuntary movements (8). SIB is typically expressed by persistent biting of the lips, tongue, fingers, and shoulders (9) and results in partial or total destruction of perioral tissues, especially of the lower lip. Partial or complete amputation of fingers, toes, and tongue has been also observed (10). SIB may additionally be expressed by head snapping and banging, arching the spine, even by throwing the head, arm, or leg while being wheeled through a doorway. The children are sensitive to pain, although they cannot control the SIB. Therefore, they are relieved when inhibited from hurting themselves and physical restrains are necessary (11).

Several treatment approaches have been employed to manage the various abnormalities of the syndrome. Allopurinol is a drug used to reduce uric acid. It prevents the development of renal and musculoskeletal injury and leads to a significant increase in life expectancy, by delaying renal failure, which is the common cause of death in early childhood (12, 13). However, it has no effect on the behavioural, cerebral, and neurological manifestations of the disorder (5, 8, 14). Behavioural control of SIB has been sought by benzodiazepines, neuroleptics, antidepressants, chloralhydrates, and anticonvulsive drugs (15, 16). Recent therapeutic trials also include gabapentin, which is free from side effects (17), botulinum toxin A (BTX-A), which acts on the central and peripheral nervous system (18), dopamine replacement therapy and deep brain stimulation in globus pallidus (19, 20).

According to Olson and Houlihan 2000 (16), the self-destruction behaviour should be managed by a combination of physical restraints, behavioural treatment and pharmaceutical therapy. Physical interventions include bandaging, gloves, or various types of restraints that protect the body parts from continued damage (5, 8, 21, 22).

In the case that self-mutilating behaviour does not respond to medication, surgical intervention and removal of the front, or even all, teeth has been advocated (3, 23, 24). This extreme management

approach should be viewed with caution, because, besides the severe functional consequences, some authors warn that the children may find other ways to injure themselves (25, 26). Less radical treatments, based on the use of intraoral protective appliances, have been tried as a first measure to prevent continued oral self-mutilation. These include a variety of appliances such as lip-shields (27), lip bumpers (26, 28, 29), and tongue protectors (30, 31). Various types of removable intraoral appliances aiming to avoid biting by the anterior teeth as well as occlusal splints have been reported with varying success. Occlusal bite plates (32), occlusal splints (33), splints with bite blocks to produce an anterior open bite (34), and a palatal plate to raise the anterior bite (35) are commonly reported. Silva and da Fonseca (36) used a Hawley appliance modified with lateral acrylic shields, Chen and Liu (37) constructed splints using headgear to retain and stabilize the splint for patients with severe SIB and Davila et al. (38) devised an appliance attached to bubble helmets for children who self-injure the head.

This report describes the oro-dental care provided for a child with Lesch–Nyhan syndrome who presented with self-inflicted loss of tissue of the lip and tongue. An appliance was fabricated, with the purpose of preventing the soft tissue damage, while avoiding the sacrifice of the teeth.

Case report

A 14-year-old boy, diagnosed with LNS, was referred to the Paediatric Dental Clinic of the Dental School for evaluation and treatment of SIB. He suffered from spastic hypertonia and marked gross and fine motor incoordination, which was expressed as a mixture of abnormal movements, spasticity and ataxia. He exhibited hyper-reflexia and hyperkinetic choreo-athetoid movements that were intensified when he was tensed, anxious or excited. He verbally was unable to communicate. His physical growth was almost normal, but his motor disability was extensive to the point that he had difficulty raising his head. He could not sit up right, stand or walk. He was confined to a specially designed wheelchair fitted with a seat belt to keep him upright and prevent head injury and uncontrolled movement. Laboratory tests confirmed the renal failure revealing hyperuricaemia of 52.4 mg dl^{-1}, indicative of excessive purine production. Renal ultrasound examination showed kidneys of normal size but with bilateral nephrocalcinosis. He was under treatment with allopurinol (10 mg kg^{-1} per 24 h), to control the level of serum creatinine and serum uric acid.

At presentation child's arms were restrained with wraps to prevent the biting of the fingers. Extraoral and intraoral examination revealed missing tissues from the left part of the lower lip (Fig. 1a) and the left part of the tongue (Figs 1b and 2). The surrounding tissues of tongue and lip were inflamed. He never appeared to bite his buccal mucosa, which showed no scarring. He resisted opening the mouth for examination by clenching his teeth and tightening his lips, probably because the inflamed lesions of the lip were painful when stressed. His oral hygiene was poor; however, all teeth were caries-free.

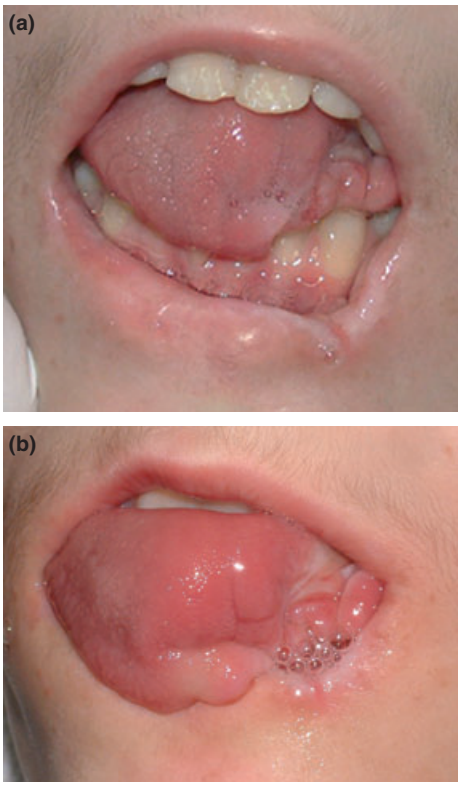

Fig. 1. (a) Missing tissues from lower lip attributed to self-injury behaviour (SIB). (b) Missing tissues from tongue attributed to SIB.

Although the SIB was extreme, it was decided to pursue at first a conservative solution. In co-operation with the Department of Orthodontics, a removable acrylic dental appliance was designed. It consisted of a posterior bite plate aiming to prevent traumatic injury of the tongue and lip and alleviate the boy's oral problems. This conservative approach was well accepted by the parents, who were unwilling to accept the extraction of anterior teeth as a treatment option.

Irreversible hydrocolloid impressions of the maxillary and mandibular arches were obtained. The procedure of impression making was hindered by the patient's resistance to open the mouth. He finally endured the making of maxillary impression, but could not cooperate for the impression of the mandibular (antagonistic) teeth. To obtain the mandibular impression, a sectional impression technique was employed. Two sectional impressions were made, one for each side of the mouth, using partial stock trays (39). The impressions were afterwards combined and poured to provide the mandibular cast (Fig. 3). The casts were mounted and the maxillary acrylic appliance was designed and constructed. A posterior occlusal bite plate was placed bilaterally on the occlusal surfaces of the posterior maxillary teeth, producing a bite-raising of about 6–8 mm of the anterior teeth. The acrylic occlusal surface of the bite plate occluded only on the occlusal surfaces of the mandibular first permanent molars. It gradually declined towards the anterior teeth, to prohibit the intercuspation of the premolars, canines and incisors. Thus, the entrapment of

the lip and tongue was not feasible (Fig. 3). The bite plate of the appliance was smooth, with no indentations, so that the mandible was able to move freely. The appliance was retained by two Adams' clasps, acting on the first premolars and by three ball clasps between the incisors (Fig. 4). During the delivery session the appliance was inserted, and checked for fit and stability (Fig. 2). The parents were instructed and trained about insertion, removal and cleaning of the appliance. To maintain good oral hygiene and plaque control and because the patient had severe motor deficiencies, instructions were given to parents to apply a brushing technique consisting of gentle horizontal strokes on the labial, lingual, and occlusal surfaces of all teeth (40). The appliance was cleaned by regular brushing followed by soaking in a denture cleanser solution (41).

The patient was re-examined after 2 weeks. Biting of the lips and tongue had ceased immediately after the insertion of the appliance and initial healing of the lesion was noted. Within 4 weeks postinsertion, the lesions had resolved completely, leaving a scar of fibrous tissue. On the 3-month recall, the lip (Fig. 5a) and tongue (Fig. 5b) had regained normal colour and texture. After 7 months, occlusal wear of the acrylic occlusal surfaces was observed and small fractures were noted and repaired. During the 3-year follow-up period, the SIB has disappeared and the patient has shown no sign of compulsive self-mutilation. The appliance is presently continuously worn.

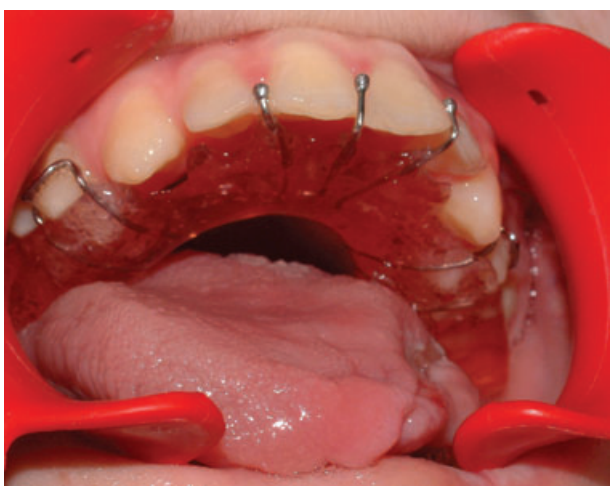

Fig. 2. The bite raising appliance intraorally. Tongue trauma is evident.

Fig. 3. The acrylic appliance on the working casts. The mandibular cast was obtained with the sectional impression technique.

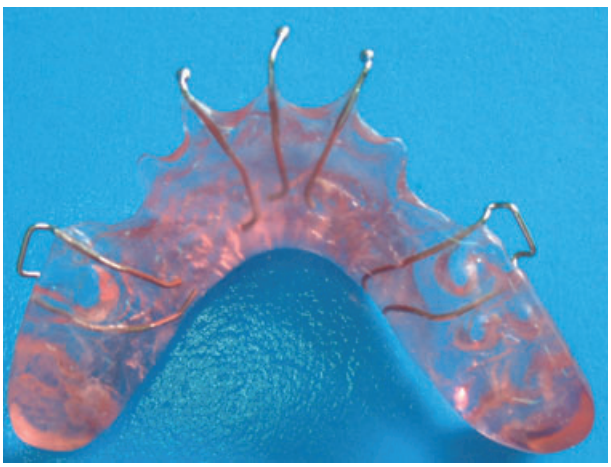

Fig. 4. The bite raising appliance.

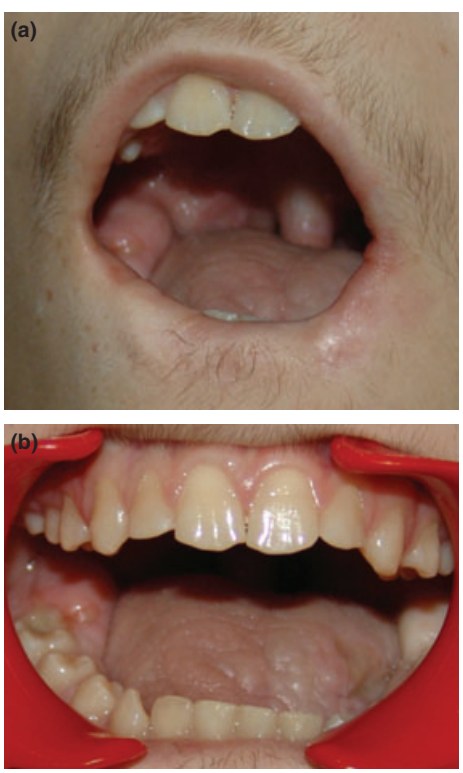

Fig. 5. (a) Complete wound healing of the lower lip. (b) Complete wound healing of the tongue.

Discussion

Oral SIB is a difficult problem for the clinician to manage (42). Extracting some or all teeth may be an effective means to confront the severe medical problems arising by the persistent self-destruction; still, it is a greatly invasive approach and introduces a major oral disability. In the case presented, in accordance with the parents' wish, the most conservative course of action was successfully followed.

Published literature suggests that an intraoral appliance can restrict self-injurious biting and protect the tissues of individuals with SIB who use their teeth to inflict damage on oral and peri-oral structures, or even on other parts of the body. Although the appliance does not treat the source of the problem, it is an effective means of controlling self-mutilation (33). The child presented in this report, accommodated to the appliance immediately and uneventfully. For the first 3 months, the patient wore the appliance continuously; until the lesions of the lower lip and tongue were fully healed. For the next 7 months, as the patient was under adult supervision and there was no SIB relapse, the parents were advised to have the appliance in use for about 12 h a day. After that period, the daily use of the appliance was gradually shortened. The patient wore the appliance during sleeping time as well as when he was not under adult supervision. The periodic examination has confirmed the effective resolution of the oral lesion and a high level of tolerance of the appliance.

The appliance was designed and fabricated in accordance with the objectives of fabricating trauma-preventing intraoral devices, as listed by Hanson et al. (1975) (43): It deflects traumatized tissues away from the occlusal table, promotes healing of injured tissues and permits normal mandibular movement and daily oral care. Additionally, it is stable and resistant to fracture, easily fabricated, comfortable, and not posing a risk to the patient.

The appliance was constructed of autopolymerizing acrylic resin. It covered the palate and extended over the occlusal surfaces of the posterior maxillary teeth. Its successful outcome was attributed to the placement of extra acrylic resin that opened the bite. Thus it protected the soft tissues from damage by breaking the self-mutilation habit and allowing inflammatory oedema to subside, thereby promoting healing. Fenton (44) argued that appliances of this type are not helpful in managing LNS, because the children can easily remove them and continue their self-mutilation. Therefore, care was taken that the appliance would not be easily removed by the child; its steady retention was accomplished by the combination of Adams' clasps, on the premolars, and ball clasps, between the incisors.

The incorporation of a cast metal framework, to improve the mechanical strength and resistance to fracture, was also considered. However, the fabrication of a cast metal element would complicate the procedure and increase construction time and cost. Therefore, it was decided not to include a metal framework in the design. The outcome justified this decision, as no mechanical complications were encountered. Probably, the appliance was not subjected to heavy loads, because the SIB ceased immediately after its insertion. No major fractures were noted and only small cracks were observed on the acrylic resin of the occlusal bite planes.

The construction procedure must be as simple and short as possible, because of the limited cooperation ability of these patients. For the patient presented in the present report, a major difficulty was encountered during the impression procedure, because of his spasticity and refusal to open the mouth. In such cases, the recording of the oral tissues needs special care and experience. Alternative impression methods may also prove useful to overcome the problems of impression making. The sectional impression technique, which was employed in the present case for obtaining the mandibular impression, is commonly used for patients with limited mouth opening attributed to scleroderma, burns or radiotherapy (45).

In conclusion, the simple intraoral appliance presented, was effective in preventing the damage of the oral and peri-oral structures. It provided a conservative solution for SIB and an alternative to the highly intrusive and substantially undesirable option of extracting the teeth. Additionally, once tissue recording and study models construction is accomplished, the appliance is easily designed and constructed and can be customized for each individual case. It can therefore be recommended as the treatment of choice for improving the quality of life of these excruciate special patients.

References

1. Lesch M, Nyhan WL. A familial disorder of uric acid metabolism and central nervous system function. Am J Med 1964;36:561–70.
2. Fernald CD. The Lesch–Nyhan Syndrome: cerebral palsy, mental retardation, and self mutilation. J Pediatr Psychol 1976;1:51–5.
3. Shoptaw JT, Reznik JI. Lesch–Nyhan Syndrome: report of three cases in one family. ASDC J Dent Child 1978;45:403–7.
4. Nyhan WL. The Lesch–Nyhan Syndrome. Annu Rev Med 1973;24:41–60.
5. Nyhan WL. Behavior in the Lesch–Nyhan Syndrome. J Autism Child Schizophr 1976;6:235–52.
6. Nyhan WL. Inherited hyperuricemic disorders. Contrib Nephrol 2005;147:22–34.
7. Bundick J. Lesch–Nyhan syndrome. ASDC J Dent Child 1969;36:277–80.
8. Christie R, Bay C, Kaufman IA, Bakay B, Borden M, Nyhan WL. Lesch- Nyhan disease: clinical experience with nineteen patients. Dev Med Child Neurol 1982;24:293–306.
9. Baumeister AA, Frye GD. The biochemical basis of the behavioral disorder in the Lesch–Nyhan Syndrome. Neurosci Biobehav Rev 1985;9:169–78.
10. Wurtele SK, King AC, Drabman RS. Treatment package to reduce SIB in a Lesch–Nyhan patient. J Ment Defic Res 1984;28:227–34.
11. Anderson LT, Ernst M. Self-injury in Lesch–Nyhan disease. J Autism Dev Disord 1994;24:67–81.
12. Balis ME, Krakoff IH, Berman PH, Dancis J. Urinary metabolites in congenital Hyperuricosuria. Science 1967;156:1122–3.
13. Sweetman L, Nyhan WL. Excretion of hypoxanthine and xanthine in a genetic disease of purine metabolism. Nature 1967;215:859–60.

14. Saemundsen SR, Roberts MW. Oral self-injurious behaviour in the developmentally disabled: review and a case. ASDC J Dent Child 1997;64:205–9.
15. Buitelaar JK. Self-injurious behaviour in retarded children: clinical phenomena and biological mechanisms. Acta Paedopsychiatr 1993;56:105–11.
16. Olson L, Houlihan D. A review of behavioral treatments used for Lesch–Nyhan Syndrome. Behav Modif 2000;24:202–22.
17. McManaman J, Tam DA. Gabapentin for self-injurious behavior in Lesch–Nyhan syndrome. Pediatr Neurol 1999;20:381–2.
18. Dabrowski E, Smathers SA, Ralstrom CS, Nigro MA, Leleszi JP. Botulinum toxin as a novel treatment for self-mutilation in Lesch–Nyhan syndrome. Dev Med Child Neurol 2005;47:636–9.
19. Pralong E, Pollo C, Coubes P, Bloch J, Roulet E, Tetreault MH et al. Electrophysiological characteristics of limbic and motor globus pallidus internus (GPI) neurons in two cases of Lesch–Nyhan syndrome. Neurophysiol Clin 2005;35:168–73.
20. Taira T, Kobayashi T, Hori T. Disappearance of self-mutilating behavior in a patient with Lesch–Nyhan syndrome after bilateral chronic stimulation of the globus pallidus internus. Case report. J Neurosurg 2003;98:414–6.
21. Dahlin PA, Van Buskirk NE, Novotny RW, Hollis IR, George J. Self-biting with multiple finger amputations following spinal cord injury. Paraplegia 1985;23:306–18.
22. Ball TS, Datta PC, Rios M, Constantine C. Flexible arm splints in the control of a Lesch–Nyhan victim's finger biting and a profoundly retarded client's finger sucking. J Autism Dev Disord 1985;15:177–84.
23. Cudzinowski L, Perreault J. The Lesch–Nyhan syndrome: report of a case. ASDC J Dent Child 1979;46:143–4.
24. Dicks JL. Lesch–Nyhan syndrome: a treatment planning dilemma. Pediatr Dent 1982;4:127–30.
25. Cusumano FJ, Penna KJ, Panossian G. Prevention of selfmutilation in patients with Lesch–Nyhan syndrome: review of literature. ASDC J Dent Child 2001;68:175–8.
26. Cauwels RG, Martens LC. Self-mutilation behaviour in Lesch–Nyhan syndrome. J Oral Pathol Med 2005;34:573–5.
27. Willette JC. Lip-chewing: another treatment option. Spec Care Dentist 1992;12:174–6.
28. Jasmin JR. Semi-fixed appliance to treat injurious lip habit: report of a case. ASDC J Dent Child 1985;52:188–90.
29. Nurko C, Errington BD, Ben Taylor W, Henry R. Lip biting in a patient with Chiari type II malformation: case report. Pediatr Dent 1999;21:209–12.
30. Kozai K, Okamoto M, Nagasaka N. New tongue protector to prevent decubital lingual ulcers caused by tongue thrust with myoclonus. ASDC J Dent Child 1998;65:474–7.
31. Peters TE, Blair AE, Freeman RG. Prevention of self-injurious trauma in comatose patients. Oral Surg Oral Med Oral Pathol 1984;57:367–70.
32. Fabiano JA, Thines TJ, Margarone JE. Management of self-inflicted oral trauma: report of case. Spec Care Dentist 1984;4:214–5.
33. Walker RS, Rogers WA. Modified maxillary occlusal splint for prevention of cheek biting: a clinical report. J Prosthet Dent 1992;67:581–2.
34. Hallett KB. Neuropathological chewing: a dental management protocol and treatment appliances for pediatric patients. Spec Care Dentist 1994;14:61–4.
35. Fardi K, Topouzelis N, Kotsanos N. Lesch–Nyhan syndrome: a preventive approach to self-mutilation. Int J Paediatr Dent 2003;13:51–6.
36. Silva DR, da Fonseca MA. Self-injurious behavior as a challenge for the dental practice: a case report. Pediatr Dent 2003;25:62–6.
37. Chen LR, Liu JF. Successful treatment of self-inflicted oral mutilation using an acrylic splint retained by a head gear. Pediatr Dent 1996;18:408–10.
38. Davila JM, Aslani MB, Wentworth E. Oral appliance attached to a bubble helmet for prevention of self-inflicted injury. ASDC J Dent Child 1996;63:131–4.
39. Topouzelis N, Kotsiomiti E, Arhakis A. An alternative impression technique for individuals with special care needs. Spec Care Dentist 2010 (accepted for publication).
40. Tesini DA, Fenton SJ. Oral health needs of persons with physical or mental disabilities. Dent Clin North Am 1994;38:483–98.
41. Jagger DC, Harrison A. Denture cleansing-the best approach. Br Dent J 1995;178:413–7.
42. Ayer WA, Levin MP. Self-mutilating behaviors involving the oral cavity. J Oral Med 1974;29:4–7. (41)
43. Hanson GE, Ogle RG, Giron L. A tongue stent for prevention of oral trauma in the comatose patient. Crit Care Med 1975;3:200–3. (42)
44. Fenton SJ. Management of oral self-mutilation in neurologically impaired children. Spec Care Dentist 1982;2:70–3. (43)
45. Suzuki Y, Abe M, Hosoi T, Kurtz KS. Sectional collapsed denture for a partially edentulous patient with microstomia: a clinical report. J Prosthet Dent 2000;84:256–9. (44).