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# CASE REPORT

# Self-mutilation behaviour in Lesch-Nyhan syndrome

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Lesch-Nyhan syndrome (LNS), first described in 1964 by Lesch and Nyhan, is a rare X-linked genetic disorder involving (near) absence of the enzyme hypoxanthineguanine phosphoribosyl transferase (HPRT). It occurs in 1:100 000 to 380 000 live births (1, 2). The deficiency of HPRT activity leads to an excessive uric acid production resulting in neurological, renal and musculoskeletal manifestations. Death usually occurs in the second or third decade from infection or renal failure. Clinical presentation is characterized by mental retardation, choreoathetosis, spasticity, hyperuricemia and cerebral palsy. A characteristic feature of LNS is the appearance of intractable self-injurious behaviour (SIB), usually in the form of severe lip and finger biting, gouging of eyes, face scratching and head banging requiring extreme management techniques such as the application of restraints and or extraction of teeth at an early age. In this case report a unique approach of SIB in LNS is presented. J Oral Pathol Med (2005) 34: 573-5

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#### Case

A developmentally disabled 5-year-old boy with Lesch– Nyhan syndrome (LNS) was referred for evaluation of self-injurious behaviour (SIB) involving the lower lip. As a result of tonic biting, he trapped the lower lip lingually of the anterior teeth. Figure 1 reveals a severe injured central part of the lower lip. As no treatment method has a predictable result, it was decided to construct an Ercoloc-3 mm mouthguard (Erkodent®, Erich Kopp GmbH, Pfalzgrafenweiler, Germany) covering the upper jaw in order to prevent further mutilation. This mouthguard has a soft inner- and a hard outer-part.

The patient returned for evaluation 2 weeks following placement of the appliance. Positive result and healing was seen, leaving a horizontal cicatrice (Fig. 2). One month later, the boy restarted this mutilation habit which resulted in a significant bilateral soft-tissue trauma with loss of normal lip architecture. A mouthguard was now placed over the lower arch keeping this activity under control, however, sucking in of the lower lip restarted a few months later resulting in a serious swelling of the lower lip (Fig. 3).

To prevent this sucking in of the lower lip, a lipbumper was constructed replacing the lower mouthguard which resulted in a more stable condition. Orthodontic bands with buccaal tubes were adapted and cemented to the mandibular first permanent molars with a glass ionomer luting cement. The lip-bumper was inserted into the mouth and held to the buccal tubes using orthodontic elastics. This appliance was constructed in such a way that the distance between the lower arch and lower lip did not allow sucking in anymore (Figs 4 and 5).

Recall 2 months following insertion of the lip-bumper shows a continued healing of the lower lip, but with some deformation because of scarring. The appliance, worn every day, proved to be effective and periodic examination continued with success.

#### Comments

Self-injurious behaviour of patients with LNS usually starts between the ages of 1.5-2 years or shortly after the primary teeth erupt (3). In contrast to other conditions and syndromes, SIB starts sudden and violent rather than emerging gradually over time. Selfmutilation in these patients is more likely during periods of low social interaction suggesting that environmental factors contribute to the development of this behaviour (4). The injuries are topographically predictable and manifested as lip, tongue and mucosa biting, hand and finger biting, scratching with the fingernails, head banging and even aggression towards others (1). These children are unable to learn to avoid SIB which makes physical restrain necessary. Moreover, they are not insensitive to pain and therefore terrified of their own compulsive behaviour. They are usually relaxed when physical restraints are placed to that point that once the restraints are removed the patients become very agitated and often scream until replacement (1, 3). For this

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Figure 1 Severely injured lower lip.



Figure 2 A maxillar mouthguard, worn 2 weeks, results in healing, leaving a horizontal cicatrice.



Figure 3 Serious swelling of the lower lip because of sucking in.

purpose, hand and forearm bandages are often used to attach the patient to his wheelchair.

The kind of treatment chosen strongly depends on the intensity of the auto-aggressive behaviour, which is related to the activity of hypoxanthine-guanine phosphoribosyl transferase (HPRT) (5). It is an important consideration to adapt the treatment to the severity of the condition (6).



Figure 4 Lip bumper.



Figure 5 Healing of the lower lip with some deformation because of scarring.

First, numerous pharmacological trials have been administrated to improve the severe SIB with questionable effectiveness. Allopurinol has been widely used. It lowers uric acid levels to normal leading to a significant increase in life expectancy but does not affect the neurological or the behavioural aspect of the disease. Valium is one of the other common drugs given to help muscle relaxation. Medications to treat the dopaminergic dysfunction, the possible opiate and/or serotonine system dysfunction have shown variable successes (2, 3, 6).

A second approach of treatment is the extraction of teeth in both dentitions (1, 3, 5). Extractions are recommended only for severe cases and if medical problems arise (1). The overall protection of the patient should always be of concern and parents should be informed that children will find other ways to injure themselves. Lip-biting has been reported to lessen with age (3). However, it is not unusual that the patient may be aggressively mutilative for weeks to months and then, even without appliance, return to normal.

In this particular case we chose a less radical intervention by using oral appliances, however, this method has been reported to have minimal to partial success (2). The combination of a mouthguard in the

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upper jaw and a lip-bumper in the lower jaw resulted in a positive behavioural modification. This construction was necessary in the sense that the mouthguard opened the bite while the lip-bumper did not allow the lower lip to be sucked in anymore. Oral hygiene was never compromised. In periods of normal behaviour, the appliance could be easily removed, which was of great advantage.

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