Management of Involuntary Self-Mutilation in a Child with Lesch-Nyhan Syndrome

Lertsirivorakul J Wichajarn K** Tiamkao S*** Chawyotha P*****

Abstract

Lesch–Nyhan syndrome (LNS) is a rare X-linked recessive disorder of purine metabolism caused by deficiency of the enzyme hypoxanthine guanine phosphoribosyltransferase (HPRT). It is characterized by hyperuricemia, neurologic dysfunction, and self-mutilation. Medical and dental management in a 10-year-old boy diagnosed with LNS are described. Involuntary self-mutilation could be controlled using a soft mouth guard and repeated injections of botulinum toxin A (BTX-A) into the bilateral masseter and temporalis muscles. The findings suggest that patients with LNS require treatment from a multidisciplinary team. In addition, a soft mouth guard, repeated BTX-A injections, and pharmacological therapy could be a promising management of involuntary self-mutilation in LNS patients.

***Keywords**: Lesch–Nyhan syndrome/ Self-mutilation/ Cerebral palsy*

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Introduction

Lesch-Nyhan syndrome (LNS), also called Nyhan's syndrome and juvenile gout[1], was first described by Lesch and Nyhan in 1964.[2] It is a rare X-linked recessive disorder, occurring in 1:100,000 to 380,000 live births[3] and mainly affecting males.[4] LNS (OMIM 300322) results from a deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase (HGPRT) in purine metabolism,[1,2] caused by mutations in the hypoxanthine phosphoribosyl- transferase 1 (*HPRT1*) gene (OMIM 308000) located on the long arm of the X chromosome (Xq26.2-q26.3).[5] LNS is characterized by 3 major features, including neurologic dysfunction, uric acid overproduction, and cognitive and behavioral disturbances.[6] Affected children usually present neurological deficits before 1 year of age. The neurologic dysfunction involves the pyramidal (e.g. spasticity, hyperreflexia, and extensor plantar reflexes) and extrapyramidal (e.g. dystonia, choreoathetosis, opisthotonos, and sometimes ballismus) systems.[7] It results in global developmental delay, often leading to a misdiagnosis of cerebral palsy.[7] IQ values of LNS patients range from 25 to 101.[8] However, the explanation of how this deficiency can cause such profound neurobehavioral symptoms of LNS remains unclear.[9] The animal models demonstrated that HPRT deficiency in mice had reduced dopamine levels in the basal ganglia area and the consequences to cognitive dysfunction and neurobehavioral abnormalities.[10]

The reduced HPRT enzyme activity results in increased purine synthesis which leads to an overproduction of uric acid and consequently hyperuricemia. The excessive production of uric acid may lead to deposition of uric acid crystals, sodium urate, or calculi in the kidneys, ureters, bladder, or articular cartilages. Children with LNS may develop all the clinical signs of gout such as tophaceous gouty arthritis, uricosuria, hematuria, nephrolithiasis and have increased risk for urinary tract infections.[9-11]

A consistent symptom in all cases of LNS is the abnormal compulsive involuntary self-mutilating behavior, usually appearing about 1 year of age.[12] This behavior is generally expressed by persistent biting of the lips, tongue, fingers, shoulder and results in partial or total destruction of oral and perioral tissues. Partial or complete amputation of fingers, toes, and tongue has been also reported.[13]

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The most common cause of cognitive deficit and abnormal compulsive self-mutilating behavior is cerebral palsy. However, there are many syndromes that can also have clinical presentation like this including autism, Tourette syndrome, Cornelia de Lange syndrome, idiopathic intellectual deficit, severe psychiatric disorders, sensory neuropathies, and encephalitis.[14] LNS, in particular, should also be differentially diagnosed in an individual who presented with global delayed development and self-mutilating behavior. The prognosis of LNS is very poor. Without proper management, most LNS patients die in their childhood and survival to 20-30 years of age is rare.[12] The most common cause of death is due to infection or renal failure.[11]

Several medical and dental management methods have been employed in LNS patients. Allopurinol is effectively used to control the overproduction of hyperuricemia. It blocks the metabolism of hypoxantine and xanthine into uric acid but has no effect on neurobehavioral symptoms. To reduce complications from involuntary injuries and other deleterious behaviors, balcofen and benzodiazepine for spasticity; physical, behavioral and psychiatric therapies as well as protective equipment have been suggested.[6] However, the result on behavioral abnormalities is not satisfactory. Such problems need to be managed by a combination of pharmaceutical therapy, physical restraints and behavioral interventions. Physical interventions include bandaging, gloves, or various types of restraints that protect the body parts from continued damage. A few reports on reducing involuntary self-mutilation in LNS presented successful results of repeated injections of botulinum toxin type A (BTX-A) into muscles of mastication[15] or facial perioral muscles,[16] while another reported an ineffective outcome.[17]

Dental approaches to prevent involuntary bite injuries have included placement of a soft mouth guard or acrylic splint and different designs of intraoral appliances. While trying to maintain the dentition, vital pulpotomy and crown resections have been reported. In severe cases, orthognathic surgery to create open bite and extraction of all teeth have been suggested.[11] This report describes the characteristic of a child with LNS who received management under medical and dental collaboration and self-mutilating behavior was controlled with a soft mouth guard and repeated BTX-A injections.

Case report

A 10-year-old boy was referred to the Pediatric Dentistry Clinic, Faculty of Dentistry, Khon Kaen University (KKU), due to traumatized ulcerative tongue, lip, and fingers. He was the second child of healthy non-consanguineous parents. His mother's pregnancy and perinatal course had been unremarkable. The mother reported that at 1 month of age, he didn't raise his head. He was diagnosed with cerebral palsy at 12 months old but no treatment was provided. At 2 years old, he started banging his head and biting his lips and fingers. Although the lip biting ceased at 4 years of age after extracting all primary maxillary incisors, head banging and finger biting behaviors were presenting. Also, the mouth biting resumed at 10 years old.

Carried by his mother, he was totally dependent. He was unable to sit, stand and walk but could speak only few single words. His weight (23 kg) and height (134 cm) were below the 3rd percentile of average weight and below the 25th percentile of average height of 10-year-old Thai children. The parents reported the patient's destructive and repeated biting of lips, tongue and fingers which resulted in worse injuries. Attempting to prevent the patient's biting, chewing and drooling problems, his mother simply put a thick sponge in between his teeth, covered his mouth with a cotton mask and wrapped both of his hands with towels.

The patient exhibited uncontrolled repetitive movements, twisting, and abnormal postures which became more intense when he was tense or excited. There were contractures of his wrists, elbows, ankles and knees and generalized hyperreflexia. He presented multiple tophi (0.5-1 cm in diameter) on both ear helices (Figure 1A), red and irritated rashes on both cheeks (Figures 1A and 1B) which resulted from a smelly moist cotton mask. Chronic scars presented on both lips and thumbs (Figures 1A, 1C, and 1D). No lymphadenopathy was detected.

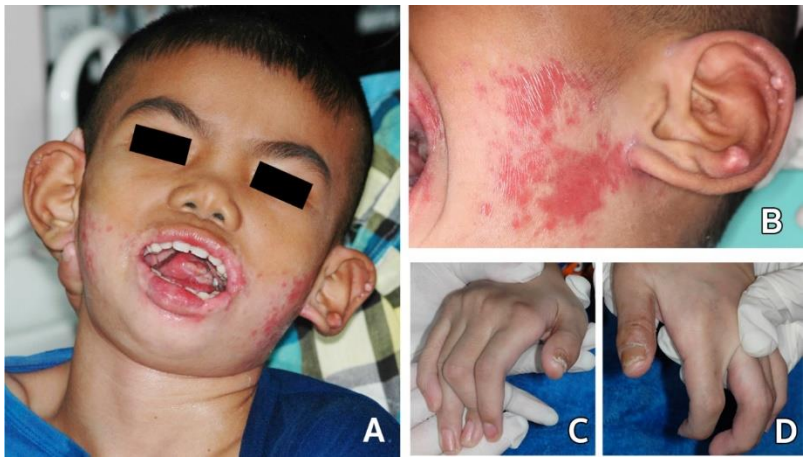

Figure 1 Clinical appearance of the patient presenting traumatized tongue and multiple tophi on both ear helices (A), rashes on the left cheek (B), and chronic scars on the right (C) and left (D) thumbs

Initial oral examination was difficult to perform, although using papoose board and mouth gag. Fresh wounds were found on the left half of the tongue with covering slough tissue (Figure 2) and deformity of the lower lip. His oral hygiene was poor with generalized gingival inflammation due to heavy deposition of plaque and calculus. The mother brushed his teeth only once daily. He was in the late mixed dentition stage which presented Class II malocclusion with generalized spacing, excessively proclined and open bite anterior incisors and partial eruption of premolars. Multiple carious lesions involved teeth 21, 22, 16, 26, 53, 63, 64, and 75. In addition, teeth 46, 54, 65, 84, and 85 retained roots.

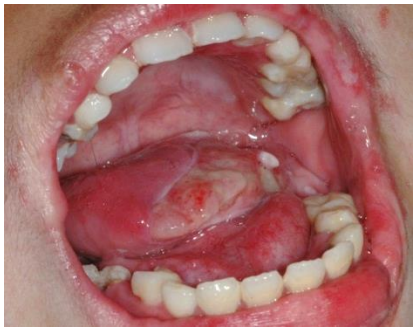

Figure 2 Intraoral feature showing the injured tongue, carious teeth, and poor oral health status with generalized heavy plaque and calculus deposition

Panoramic radiograph could not be obtained due to the patient's uncontrolled movement. Therefore, intraoral radiographs were performed by stabilizing him with a papoose board and film holding by the mother. After full mouth scaling and prophylaxis, and application of Duraphat® varnish on all remaining teeth. Extraction of teeth 46, 54, 65, 84, and 85 was accomplished under local anesthesia using stabilization with a papoose board and restraint by his mother.

Two days later, soft mouth guards were fabricated and inserted (Figure 3). Impressions of both upper and lower arches were taken with difficulty because the patient resisted mouth opening. The mother was instructed and trained how to use and clean the mouth guards. Although the patient used only the upper mouth guard, it prevented tongue biting and ameliorated a drooling problem. A few days later, the cotton mask was discarded. The mother was instructed for patient's oral hygiene care with a hands-on demonstration. He was then referred to consult with a neurologist at the Srinagarind hospital, Faculty of Medicine, KKU. He was diagnosed with dystonic cerebral palsy and 2 mg diazepam tablet at bedtime was prescribed.

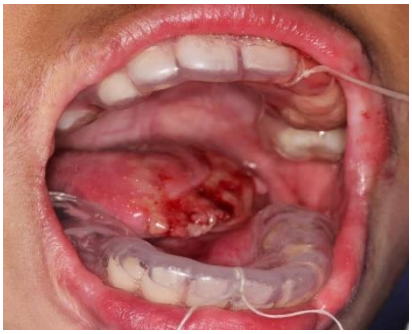

Figure 3 Soft mouth guards inserted on both upper and lower arches

At two-week follow-up, the wounds on the tongue and lip healed well (Figure 4) also the rash on both cheeks. However, a new ulceration at the mandibular left retromolar area occurred due to irritation from the distal end of the upper mouth guard. Therefore, the mouth guard was adjusted to relieve the soft tissue trauma. Dental treatment including placement of composite, prophylactic resin restorations and sealant was completed in the following appointment.

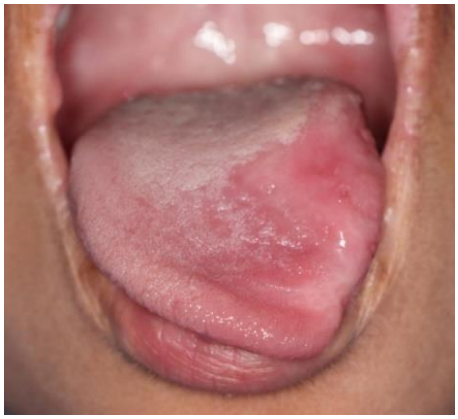

Figure 4 Appearance of the tongue and lip after using upper mouth guard

One month later, the patient returned with a complaint of lower lip biting. Oral examination revealed ulceration on lower lip and further eruption of tooth 15 which resulted in loose mouth guard. A new upper mouth guard was replaced. Two weeks later, the biting problem persisted. Therefore, a lower acrylic appliance with lip-bumper was provided. The patient was referred to the neurologist who prescribed 2 mg diazepam tablets (morning and evening) and a 5 mg diazepam tablet at bedtime. The biting problem was relieved, although the patient could not wear the lower lip bumper because of its poor retention due to heavy force of the lower lip.

Due to the continuing traumatic activities, 9 months later, the patient was referred to a geneticist for a definitive diagnosis. His karyotype was 46 XY. The complete blood count revealed normal hemoglobin level (11.6 g/dL) and normal mean corpuscular volume (86.3 fL). Serum uric acid and creatinine were13.8 (reference value = 2.7-7.0 mg/dL) and 0.8 (reference value = 0.5-1.5 mg/dL) mg/dL, respectively. The random urine uric acid to creatinine ratio was 2.66 (normal value = less than 1.0 at that age). Twenty-four urate excretion was 38.7 mg/kg. Genomic DNA was isolated from peripheral blood leukocytes. PCR amplification of 8 fragments covering *HPRT1* (NM_000194) coding region was performed and followed by direct sequencing by using oligonucleotide primers as reported previously.[18] Mutation analysis of *HPRT1* revealed c.133A > G in exon 2. This mutation changed Arginine to Glycine at position 45 (p. Arg45Gly) which confirmed the diagnosis of LNS. The treatment was initiated with allopurinol (10 mg/kg/day), baclofen (0.4 mg/kg/day) and diazepam (0.2 mg/kg/day). In addition, injections of BTX-A botulin (Dysport®) were applied in both masseter muscles 40 units each and both temporalis muscles 40 units each. The injections were repeated every 3 months.

Three months later, his serum uric acid was decreased to 3.6 mg/dl and multiple tophi on both ear helices were decreased in size. Six months after treatment, the multiple tophi disappeared (Figure 5). Self-injurious behavior, although having 80% improvement, still presented. Gabapentin was prescribed with a dose of 40 mg/kg/day because the previous study[15] reported effective treatment of self-mutilation in LNS using gabapentin. One month after initiation of gabapentin, the deteriorative behavior slightly improved. However, 3 months later, the self-injurious behavior was not improved. Therefore, gabapentin was ceased while other treatments were continued. The serum uric acid level had been controlled without new tophi occurring.

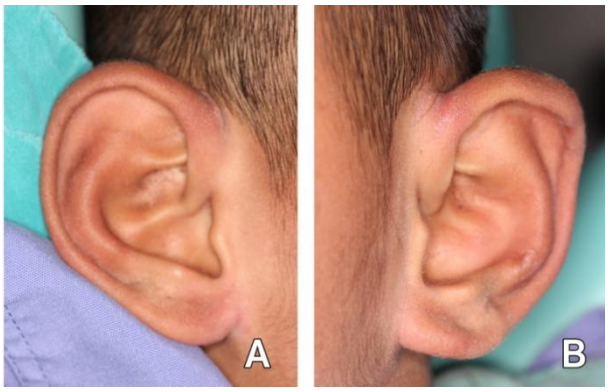

Figure 5 Appearance of the right (A) and the left (B) ear helices after treating with allopurinol

During 2-year follow-up, periodic recalls were provided for medical and dental management. Compulsive self-mutilation occurred less frequently and almost exclusively to his lips only when the mouth guard was worn out or unfitted. The traumatized lesions, however, alleviated well after applying a new mouth guard. Finally, the upper mouth guard was increased in thickness to create anterior open bite which prevented lower lip biting. With mouth guard and hand gloves combined with prescribed pharmaceuticals, the problems were under control and the patient was re-evaluated at 3-month intervals.

Discussion

Diagnosis of LNS in this patient was delayed because he lived in a remote area and the self-mutilation did not raise concern until 10 years of age. In addition, his clinical symptoms were confused with his primary diagnosis of cerebral palsy. Because of the characteristic neurologic disturbance and compulsive self-mutilation, the diagnosis of LNS was suspected. The multiple yellowish non-tender tophi presenting on both ear helices were also a possible presenting feature of the disease, secondary to gout contained uric acid crystals. Mitchell and McInnes suggested that in all males diagnosed having cerebral palsy, HGPRT deficiency must be ruled out by determining the ratio of uric acid to creatinine in a random urine specimen.[19] HPRT enzyme activity in erythrocytes and *HPRT1* mutation analysis are the gold standard for diagnosing LNS.[20] High value of uric acid to creatinine ratio in urine can be useful for diagnosing LNS, particularly if the mutation analysis is unavailable. The mutation analysis in this patient eventually revealed known pathogenic mutation which confirmed the diagnosis of LNS.[21]

Allopurinol was used to treat hyperuricemia, reduce kidney complications (nephrolithiasis, urate nephropathy), and prevent gouty arthritis and new tophi. In this case, the absence of tophi on both ear helices possibly resulted from the success of allopurinol in inhibiting uric acid production. Unfortunately, in agreement with many reports,[22-26] it has not affected neurological and behavioral problems. The subsequent dental impressions were performed more easily possibly due to the calming and muscle relaxant effect of diazepam. McManaman and Tam reported the efficacy of gabapentin for treatment of self-injurious behavior in 1999.[27] They proposed the hypothesis that brain gamma-aminobutyric acid (GABA) level is involved in self-injurious behavior. Therefore, property of gabapentin in increasing brain GABA level should improve the self-injurious behavior. However, this patient did not respond to gabapentin treatment.

The action of BTX-A both directly on the peripheral nervous system and indirectly on the central nervous system may help in reducing self-abusive behavior in LNS.[15] A successful treatment of lip biting with repeated BTX-A injections in the facial perioral muscles (zygomatic muscles, orbicularis oris muscle and levatorlabii inferioris) had been reported.[16] It is, however, useful when the injury is limited to perioral muscles. In this patient, BTX-A was administered into bilateral masseter and tempolaris muscles because they are muscles of mastication which directly control biting activity. Moreover, injecting the BTX-A in the orbicularis oris muscle or genioglossus muscle required more injection sites and is infeasible in this uncooperative patient. BTX-A is a safe treatment when administered in appropriate doses by an experienced specialist. The most common side effect is adjacent muscle weakness due to diffusion of the solution. Injection dose of BTX-A is 10-50 units per site, depending on the target muscle, with a total dose of 200 units in the masticatory system.[28]

A successful treatment of dystonia and self-mulitation with bilateral chronic stimulation of the globus pallidus internus (deep brain stimulation) in a case of LNS was also documented.[29] It was not indicated for this patient because the procedure is invasive and expensive. In addition, deep brain stimulation is effective for generalized dystonias[30] but the dystonia of this patient was segmental.

Until now, no effective management to control involuntary self-injurious behavior has been reported. Combined physical, behavioral and psychiatric management is the best management for prevention. Physical and emotional stress should be avoided to reduce further injury.

Many individuals need physical restraints to prevent self-injury. This patient still needs to wear a mouth guard and bandage both hands with towels. If the appliance was removed, the patient would become very agitated and scream. A maxillary appliance designed to raise the anterior bite exhibited favourable results in preventing self-mulitation in LNS.[31] It was not considered in this patient who needed frequently a change of appliance because of the changing oral condition due to erupting teeth which resulting in poor retention. The mouth guard in this report was, however, increased in thickness to create anterior open bite. It is inexpensive, easy to fabricate and can be cleaned easily. It also protects all soft tissue including buccal mucosa, tongue and lips better.

Conclusion

The present study suggests that LNS should be ruled out in all males diagnosed with cerebral palsy and presenting self-injurious behavior. Patients with LNS need both pharmacological and dental management to ameliorate the symptoms of disease and improve quality of life. In order to achieve these goals, a close collaboration between medical and dental personnel is required. A soft mouth guard, repeated BTX-A injections, and pharmacological therapy could be a promising management of involuntary self-mutilation in LNS patients.

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Conflict of Interest Statement The authors have no conflicts of interest to report.

Ethics Statement: Before starting the treatment, the treatment plan, procedures, potential risks and benefits was explained to the patient's mother. She agreed and signed the informed consent. This study (Reference No. HE601022) has also been reviewed and approved by the Khon Kaen University Ethics Committee for Human Research based on the Declaration of Helsinki and the ICH Good Clinical Practice Guidelines.

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การจัดการภาวะทำร้ายตนเองโดยไม่เจตนาในเด็กที่เป็นกลุ่มอาการเลส์ช–ไนแฮนซินโดรม

จินดา เลิศศิริวรกุล* กุณฑล วิชาจารย์** สมศักดิ์ เทียมเก่า*** ปณวัตร์ ชาวโยธา****

บทคัดย่อ

กลุ่มอาการเลส์ช-ไนแฮนซิน โดรม เป็น โรคที่ถ่ายทอดพันธุกรรมของยีนด้อยบน โคร โมโซมเพศที่พบได้ยาก เป็นความผิดปกติของการเผาผลาญพิวรีนที่เกิดจากขาดเอ็น ไซม์ไฮ โปแซนธีน กัวนีน ฟอสโฟไร โบซิลทรานสเฟอเรส ลักษณะเฉพาะคือ มีภาวะกรดยูริกเกินในเลือด ความผิดปกติของระบบประสาท และภาวะทำร้ายตนเอง บทความนี้บรรยายการจัดการทางการแพทย์และทางทันตกรรมในเด็กชายอายุ 10 ปี ที่ได้รับการวินิจฉัยเป็นกลุ่มอาการเลส์ช-ไนแฮนซิน โดรม ภาวะทำร้ายตนเองโดยไม่เจตนา สามารถควบคุมได้โดยใช้เฝือกป้องกันฟันชนิดนุ่มร่วมกับการฉีดโบทูลินัม ท็อกซินเอ เข้าที่กล้ามเนื้อแมสซีเตอร์และกล้ามเนื้อเทม โพราลิสทั้งสองข้าง สิ่งที่พบชี้ให้เห็นว่า ผู้ป่วยกลุ่มอาการเลส์ช-ไนแฮนซิน โดรมต้องการการรักษาจากทีมสหสาขาวิชาชีพ นอกจากนี้การใช้เฝือกป้องกันฟันชนิดนุ่มร่วมกับการฉีดโบทูลินัม ท็อกซินเอซ้ำ ๆ และการใช้ยา น่าจะเป็นการจัดการที่ให้ผลดีต่อภาวะทำร้ายตนเองโดยไม่เจตนาในผู้ป่วยเลส์ช-ไนแฮนซิน โดรม

คำไขรหัส: *เลส์ช-ไนแฮนซิน โดรม/ ภาวะทำร้ายตนเอง/ สมองพิการ*

ผู้รับผิดชอบบทความ

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