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Case Report

Lesch-Nyhan Syndrome: Evaluation of a Modified Bite Device to Prevent Bite Injuries

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Abstract: Lesch-Nyhan syndrome (LNS) is a serious form of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency, a hereditary purine metabolism disorder. The prevalence reported in the literature is 1/380,000 to 235,000 births. Males are affected and females are heterozygous carriers. LNS patients present a combination of hypotonia, spasticity, and neurological and behavioral disorders. They also show an obsessive-compulsive self-injurious behavior with bites and injuries to the lips, tongue, cheeks and fingers. The literature offers little scientific contribution related to the management of this problem. The authors describe their experience with a 4-year-old LNS patient and present a viable solution to control and avoid bite injuries, namely a specifically modified bite. The patient was treated at the Pediatric Dentistry Department of "Sapienza" University of Rome with a modified bite with the internal surface, in contact with the teeth, realized in 2-mm-thick soft silicone, and the exterior part consisting of a transparent resin shell with front and rear shields to separate lips and cheeks from the dental arches. At a 12-month follow-up visit, compliance was excellent: the child wore the device with regularity and without discomfort, even during soft-food feeding. No intraoral bite injuries were found, with a general improvement of the young patient's quality of life.

Keywords: Lesch-Nyhan syndrome; bite injuries; bite device

1. Introduction

Lesch-Nyhan syndrome (LNS) is a severe form of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency, a hereditary purine metabolism disorder linked to mutations in the HPRT1 gene located at Xq26-27. LNS is characterized by uric acid overproduction (UAO), neurological and behavioral disorders. Manifestations of the disease begin to appear at 3–6 months of age with muscle hypotonia and difficulties in maintaining the sitting position and supporting the head. Other signs include psychomotor and mental retardation of varying degrees of severity. With growth, the patients may exhibit aggressive behavior [1–3]. After dental eruption, they may also show an obsessive-compulsive self-injurious behavior with bites and injuries to the lips, tongue, cheeks and fingers [4]. These events are usually marked by stress. The disease also affects the urinary system, with crystalluria and urinary tract obstruction, ant the blood system, with megaloblastic anemia. Clinical diagnosis is confirmed by laboratory and genetic tests. Medical therapy is usually aimed at complications in the urinary tract, while no treatments exist for neurological complications [5–7]. Spasticity and dystonia are treated with benzodiazepines and physical rehabilitation. Devices to control the hands and bites to avoid bite injuries are also recommended. From the dental point of view, a review of the literature shows very few articles related to LNS, with no scientific protocols or guidelines, and only a few case reports describing the

use of mouthguards or tooth extractions [8–13]. The Department of Pediatric Dentistry of "Sapienza" University of Rome is a reference center of national relevance for the dental treatment in children [14]. All types of therapies are carried out: caries prevention and treatment [15], orthodontics, and oral surgery in small patients [16]. Special needs patients, often referred from other structures, are also cared for in collaboration with the departments of pediatrics, rare diseases, hematology, pediatric oncology, and pediatric neuropsychiatry. Special needs patients treated at our department include patients with Down syndrome, ectodermal dysplasia, Apert syndrome, osteogenesis imperfecta, Noonan syndrome, Klinefelter syndrome, Turner syndrome, and autism [17–21]. Most special needs patients are also in treatment by the Department of Pediatrics, in particular at the Rare Diseases Department. Between 2013 and 2016, the Pediatric Dentistry UOC (Complex Operative Unit) visited and treated about 140 special need patients with various pathological conditions and needs for treatment. The authors describe their experience with a 4-year-old small patient and present a viable solution to control and avoid bite injuries, based on specifically modified bite.

2. Case

A 4-year-old small patient with LNS showed up at the Department of Pediatric Dentistry of "Sapienza" University of Rome. The first signs of the disease occurred at approximately 3 months of age, with hypotonia of the neck muscles and difficulties in maintaining the posture of the head; typical orange sand-like crystals also appeared in the urine. Later, he showed difficulties in holding objects, while hypotonia progressively worsened, affecting erect posture, gait, and speech. At about 10 months of age, a sample of cerebrospinal fluid showed an anomalous very high uric acid concentration; blood analysis confirmed this anomaly. The definitive diagnosis was confirmed at one year of age, when genetic tests showed that the mother was a carrier. The patient had no anemia, but up to about two and a half years, he suffered of kidney complications with colics and calculosis. From two and a half years, he started medical treatment addressed to the management of these complications. The therapy included daily intakes of bicarbonate, allopurinol and potassium supplements, with the aim of reducing the accumulation of uric acid. From three months of age, the patient began to present the typical bite injuries to hands and lips (Figure 1). Initially, parents controlled hand injuries with the application to the arms of guardians (Figure 2), which prevented the bending of the elbow. As a solution to intraoral injuries, parents used an extraoral elastic in an attempt to prevent biting (Figure 3). This type of solution was obviously a source of discomfort for the child and caused decubitus of the lips. For this reason, the parents came to the first visit at our department. An initial attempt to solve the problem consisted in the creation of two 2-mm-thick silicone soft bites to be applied to the two arches. Alginate dental casts of the two arches were taken and the bites were built and given to the patient. In the following days, compliance and results were monitored: the upper bite had a positive effect, with good compliance, but the patient could easily remove the lower bite as it did not have adequate retention given the reduced dental support. The case was thence only partially solved: further attempts were needed. New silicone putty dental casts were taken and a study model was realized and digitized. This virtual working model could then be reproduced in the lab with no need to take further casts from the patient, as this operation was a source of discomfort and stress for the child who presented little cooperation (Figures 4 and 5). As the literature does not offer specific guidelines, from an initial idea of the parents to add a common oral screen to the bite, and after studying the technical details with our dental laboratory, we decided to build an individual bite device with some innovative features. Our main goal was to improve the comfort and the compliance of the new device with a better retention and stability inside the oral cavity. The inner part, in contact with the dental elements, was realized in 2-mm-thick soft silicone. The external part was realized with hard transparent resin, extended to the area of the fornix, with two shields, 13-mm-high and 3-mm-thick,which removed the muscular pressure of the lower lip and cheeks. To help the parents to insert the device in the child's oral cavity and improve the overall stability of the device, we inserted a front handle in soft resin, which extended from the vestibular surface of the device up to the upper lip (Figures 6–8).

Figure 1. Bite injuries.

Figure 2. Arm guard.

Figure 3. Child with extraoral elastic.

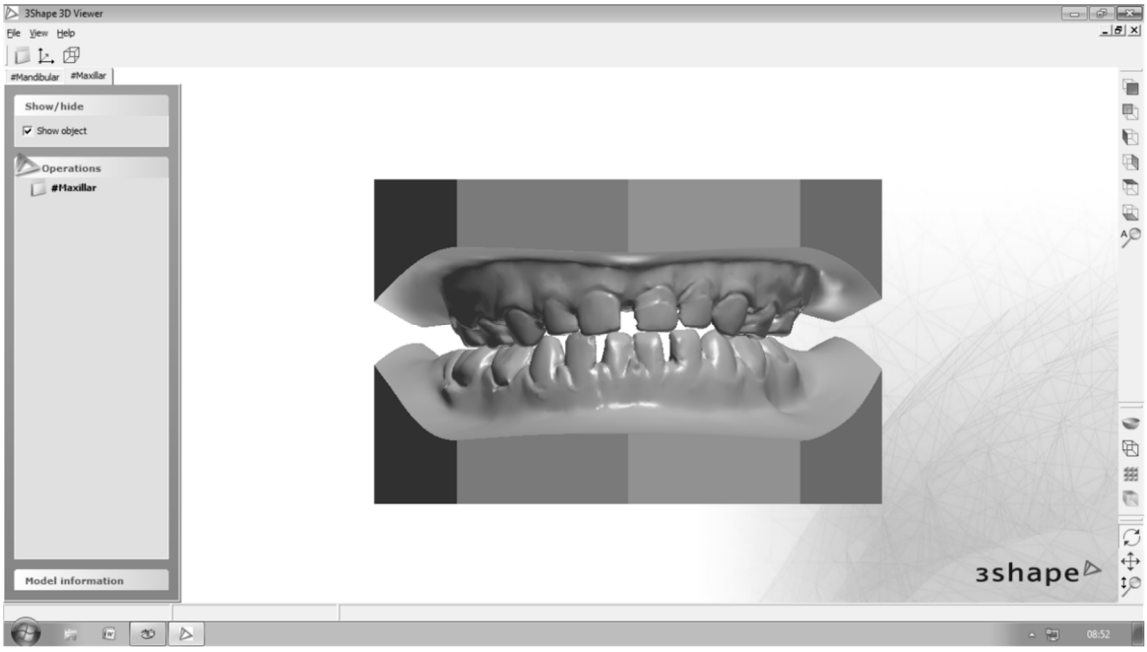

Figure 4. Front view of digital model.

Figure 5. Lower view of digital model.

Figure 6. Modified bite.

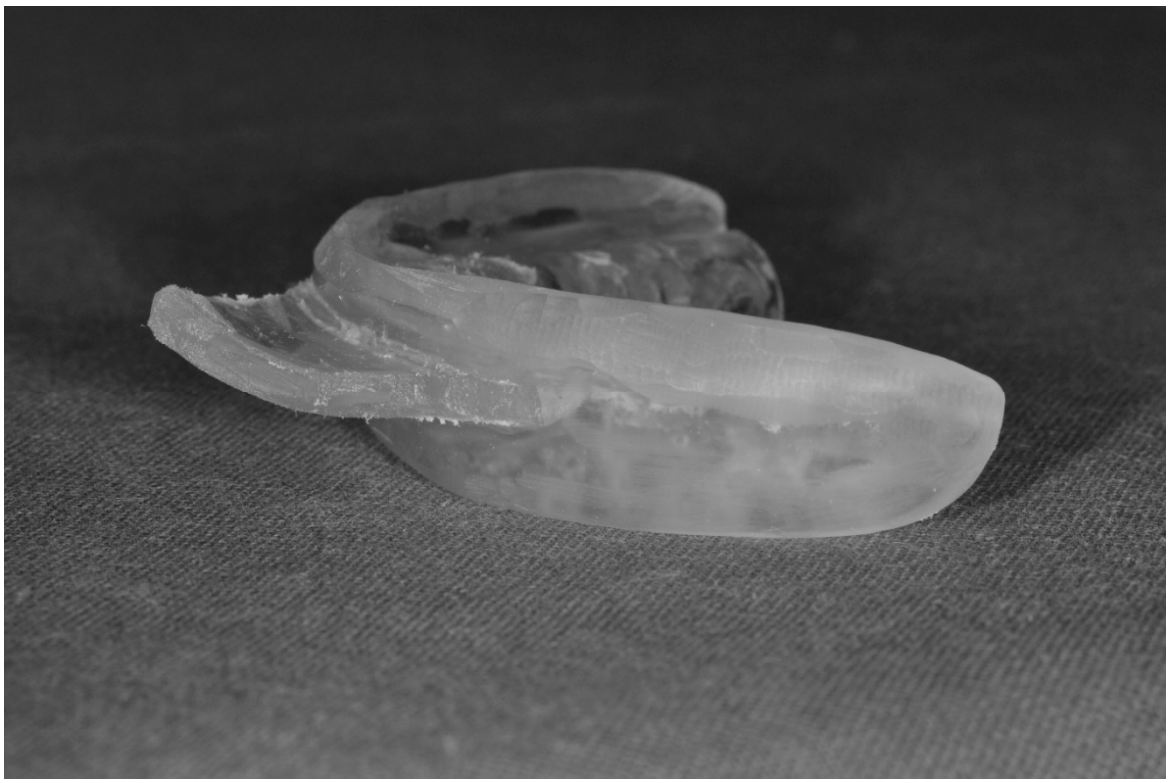

Figure 7. Modified bite.

Figure 8. Bite application.

3. Discussion

A review of previous case reports from the literature showed the usual failure of standard mouthguards and the successive resort to tooth extractions. As it is not possible to have guidelines valid for all today, it is necessary to individually evaluate the solutions. Even more difficult can be to find a solution in the pediatric patient who has poor compliance. Traditional rigid resin bites were

not accepted by our patient due to discomfort problems and a soft bite had retention problems in deciduous dentition with very small teeth. Another difficulty in these patients is taking the impression, it can be difficult because the patient tends to bite the impression itself or the dentist's hands, one must be careful and quick to remove the impression in order not to risk bites. In our case, the patient underwent regular follow up visits that highlighted the positive effect of the device. Compliance was excellent, the child wore the device regularly and without discomfort, no intraoral bite injuries were found, and the parents reported a normal night's rest, which also improved the quality of life by reducing stress and pain due to injuries. The bite was also worn during soft food feeding, if adherence was increased with a standard denture adhesive. The current 12-month follow up confirms the results obtained in the first few days of use. Of course, our experience does not represent a scientific protocol, but can be considered an example for the management of the issue. In treating this patient, we confirmed both the need for teamwork and the fact that this kind of therapies often requires long lead times. For future studies, it is important to improve the multidisciplinarity of the treatment and, after completing the current treatment programs, to get scientific value out of what is now clinical evidence.

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