Lesch-Nyhan syndrome: a treatment planning dilemma

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Abstract
The characteristic self-mutilation behaviors of patients with Lesch-Nyhan syndrome are detailed. The behavioral manifestations and dental management of five cases are discussed with emphasis on the lack of viable treatment choices other than extraction. Some suggestions for categorizing the mutilative behaviors have been presented together with possible treatment choices of each group.

Lesch-Nyhan syndrome results from an inheeritable error of purine metabolism, in which characteristic self-mutilative behavior presents a major problem for the dentist. The disorder affects only males and results in overproduction and accumulation of large amounts of uric acid in body fluids, which if untreated, usually leads to renal failure and death in early childhood. Recently, it has been found that treatment of this disorder with allopurinol returns uric acid levels to normal concentrations, dramatically increasing the life expectancy of the patient. Even with normal uric acid levels, the self-mutilative and other associated behaviors of these children remain unchanged. These behaviors and the dilemma they present to the clinical dentist in planning treatment will be the main thrust of this discussion.

Clinical Manifestations
A major characteristic of the syndrome is severe mental retardation, although most of these patients appear more capable than test scores indicate. These children usually relate well to people, and seem to understand what is said to them. Most communicate adequately through affirmative or negative responses to questions, although speech is severely dysarthric. All of the patients have cerebral palsy with severe spasticity. The characteristic extensor spasms of the trunk are increased by tension or excitement.

The self-mutilative behavior is thought to begin with the eruption of teeth; patients then begin to bite themselves. This behavior continues, resulting in partial or total destruction of the perioral tissues, especially the lower lip and to a lesser extent, the upper lip. Partial or complete amputation of the fingers, nose and tongue are also common.

The characteristics of the self-mutilative behavior in Lesch-Nyhan syndrome appear to be phenotypical and differ from similar behaviors in other patients. For example, mutilation of the lips occurs in Conelia DeLange syndrome but is much less severe. Loss of tissue is rare, apparently accidental, and the behavior is easily extinguished, using aversive techniques which are not effective in Lesch-Nyhan syndrome.

The pattern of mutilative behaviors seen in children with sensory neuropathies such as congenital insensitivity to pain is also quite different. The damage is definitely accidental and these patients have been described as looking like pugilists. In addition, many autistic children and some other mentally retarded children occasionally display self-mutilative behaviors which likewise differ from those observed in Lesch-Nyhan patients. Head banging and hitting are common, with biting rarely observed; therefore, hypertrophy and callous production, not loss of tissue, usually appears in the area of trauma.

The dramatic and extremely rapid loss of tissue is a hallmark of Lesch-Nyhan syndrome. Unquestionably, these patients do perceive pain. They usually cry out when biting themselves as if against their will. They are usually relaxed when restrained and when restraints are removed they become very agitated and often scream until restraints are replaced.

Biting is not the only source of mutilation: many other actions create perioral damage. Most common is the rubbing of soft tissue between a tooth and some hard surface, a bed sheet, or even the fingers. The
damage due to trauma together with superimposed infections produces rapid tissue loss. Extension thrusts of the body, causing the head to bang against a hard surface may also cause injury to the patient, but more commonly cause injury to others. This aggression towards other people is a secondary but important facet of behavior in this syndrome.

Behavior modification measures have been disappointing in coping with the self-mutilative behaviors observed in this syndrome. Mild aversive techniques have proven generally ineffective, and more severe aversive techniques have actually increased the frequency of these maladaptive behaviors. Extinction techniques have produced mixed results although several programs show some promise and reinforce the concept that the self-mutilative behaviors have some conscious component along with their compulsive aspects. There are, however, serious attendant risks of severe mutilation associated with some extinction programs.

Chemical approaches to modification of mutilative behavior have likewise experienced mixed results. Allopurinol will lower uric acid levels to normal but does not affect the neurological or behavioral aspects of the disease. The use of hydroxytryptophan together with decaboylase inhibitor has proven effective in reducing or eliminating the self-mutilative behavior for short periods of time with certain patients. Results with this and other drugs thus far have been promising but preliminary.

Physical restraints have been the sole reliable resource for preventing self-mutilative behavior. Cloth body restraints, cloth mittens, and plastic arm splints have all been useful in reducing the frequency of injury. However, even with restraints, self-mutilation can and often does occur.

Dental Literature Review

Although Nyhan says that next to phenylketonuria, Lesch-Nyhan syndrome is the second most common inborn metabolic disorder, very little has appeared in the literature concerning the dental management of these cases.

Budnick describes two cases; the first required no dental treatment because an extreme anterior open bite prevented lip biting, and, the second utilized self-curing acrylic splints on both upper and lower arches which were successful in preventing lip biting after a splint for the lower arch only was found to be unsuccessful in this regard. These splints required modification and recementation during the mixed dentition period. Budnick also suggested that covering only the posterior teeth, creating an anterior open bite, might also be helpful.

Cudzinowski and Perreault described one case in which all primary teeth were extracted in serial fashion with local anesthetic. This approach apparently eliminated further mutilation, although no follow-up concerning the permanent dentition was described.

Shoptow and Reznik described three cases, all having anterior and posterior primary and permanent teeth serially extracted to prevent mutilation. The success of this approach can be described as mixed, since significant mutilation occurred prior to extraction.

Case Histories

1. P. K.: Noninstitutionalized, presently 21 years old — has been followed since 1976. The patient is spastic and difficult to treat. Impressions can be taken if necessary. No self-mutilation is evident to date. Restorative and preventive treatment have been accomplished.

2. B. K.: Noninstitutionalized, presently 17 years old — has been followed since 1976. This patient is the brother of P. K. There is severe spasticity and it is impossible to obtain impressions. Severe self-mutilative behavior is present and previous extractions of individual offending permanent teeth have been accomplished with local anesthesia. There is very poor cooperation and full body restraints are necessary for treatment. The patient screams constantly when in the dental clinic. In 1979, all remaining permanent teeth were extracted under general anesthesia. No further mutilation has been noted.

3. R. F.: Institutionalized, presently 13 years old — has been followed since 1970. This patient is a first cousin to P. K. and B. K. The expression of self-mutilation has varied with many remissions. Mutilation began with the primary teeth. The dentist attempted a lower “lip plumper” in 1971 which was removed in 1972 due to ineffectiveness and lack of patient cooperation. Patient cooperation and spasticity prevented any intraoral impressions after age 7. Occasionally offending teeth were extracted with local anesthesia. In 1978, all remaining permanent teeth were extracted under general anesthesia. Three months later the patient began mutilating his lower lip with his finger. Restraints have proven generally effective in preventing further mutilation.

4. J. F.: Institutionalized, presently 15 years old — has been followed since 1970. This patient is the brother of R. F. The patient presented with partial extraction of primary dentition and extensive perioral damage. Patient cooperation and spasticity have prevented any impressions. Extraction of individual offending teeth with local anesthesia was accomplished over several years. The remaining permanent teeth were extracted under general anesthesia in 1978. No further mutilation is evident to date.

5. D. C.: Institutionalized, presently 15 years old
— has been followed since 1973. The patient has an anterior open bite. Some minor cheek biting occurs when the patient is under stress. A single episode of serious mutilation occurred in 1976, when the patient was able to extract slightly loose upper central incisors on the edge of an arm restraint. The patient, who can communicate well, presently wears arm restraints. A soft mouth guard for the maxillary teeth was inserted in 1979 to prevent cheek biting. This appliance is working well and no further mutilation is evident to date.

Discussion

The dilemma of treatment planning involves (1) the relative rarity of cases, (2) the severity of treatment or nontreatment consequences, and (3) the paucity of supportive opinions in the literature. The main issue centers on the decision to extract teeth, especially permanent teeth, to prevent mutilation.

There are several factors that further complicate the choices of treatment. First, there is the problem of remissions in self-mutilative behavior; periods with no mutilative behavior may be several days or as long as a year and a half. When remission of mutilative behavior follows a decision not to extract teeth, the clinician is often given a false impression that the improvement is permanent. The re-emergence of mutilative behavior following a remission can be discouraging to the clinician who must then consider extractions again. Likewise, remission following any behavior modification program can cloud the issue and give false hopes of success. Secondly, the extraction of teeth, especially permanent teeth, to prevent further mutilation appears a dramatic and irreversible step to all professionals. If this is the treatment choice of the clinical dentist, lack of support by many professionals and outright opposition on the part of some behaviorists can be expected. The advent of behavior reduction committees, human rights committees, etc., tend to further isolate the dentist who proposes extraction of teeth as a solution to self-mutilation.

The present failure of behavior modification programs to significantly alter self-mutilating behavior in this syndrome is not surprising. Apparently there is a compulsive component to this behavior that is not readily amenable to known behavior change techniques. In addition, many of these children live in hospitals or institutions for the mentally retarded where low professional staff/patient ratios, high staff turnovers, and other manpower problems impact on the effectiveness of any behavior modification program. These manpower problems, combined with the long-term nature of the behavior (2-18 years duration), almost certainly result in periods where self-mutilation behaviors re-emerge. During these periods, however short, severe and irreparable tissue damage often occurs.

The long-term nature of the mutilative behavior and the changing dental manpower situation in most residential care facilities produces a situation in which the dentist may observe only six months to two years of behavior which may show great variances over a 10- to 12-year span. This exposure to but a small portion of the client’s behavioral spectrum, combined with lack of previous experience due to rarity of the syndrome, can result in hesitancy on the part of the dentist to recommend apparently radical treatment regimens such as full-mouth extraction.

Another aspect of the dilemma is the concept that full-mouth extraction is a radical treatment choice. Review of the literature, both dental and nondental, reveals that in cases of severe self-mutilation, ultimate loss of both teeth and perioral tissue appears inevitable. To extract teeth at some earlier phase, resulting in a teenager who is edentulous but retains most of the perioral tissues, appears the less radical treatment choice. This may still be difficult for the dentist to accept because justification for such a treatment choice in terms of positive results, may be several years away; yet the responsibility for prescribing such a treatment is immediate.

Summary

The review of cases presented in this discussion and in the literature reveal a wide variety in the expression of mutilative behaviors in children with Lesch-Nyhan syndrome. Thus it may be helpful to the clinician to divide these clients into three categories.

Category I: Children with no oral self-mutilative or biting behaviors (e.g., P. K.). No treatment other than routine preventive and restorative treatment is indicated. The fewest number of children are in this group.

Category II: Children with some oral mutilative behaviors, but those which can be controlled by restraint techniques, verbal control, and/or aided by construction of mouth guards or splints (e.g., D. C.). These patients are quite spastic, yet their spasticity and cooperation is adequate enough to take intraoral alginate impressions. Usually these patients present little biting behavior but occasionally have bouts of hitting or placing fingers or thumbs in the mouth.

There appears to be advantages to a soft splint, using mouth guard material formed on a model with an omnivac as opposed to self-cured acrylic splints described by Budnick. One advantage is the ease of construction, but most important is the adequate retention obtained without the necessity of using cements. Cementing an acrylic splint over long
periods of time produces obvious oral hygiene and
demineralization problems. Ease of construction is
very important during the mixed dentition stage. The
soft splint on the maxillary arch alone has been very
successful in preventing cheek biting in one patient
(D. C.); its use in preventing lip biting is untested.
The “lip plumper” was constructed for one patient
(R. F.) consisting of a labial myofunctional plastic-
coated arch soldered to two bands on either side of
the mandibular arch and cemented into place. Oral
hygiene quickly became a problem, even with in-
creased tooth brushing. This device became loose and
was recemented (with great difficulty) several times.
Eventually the child was able to place the scarred
lower lip between the labial plastic arch and the lower
teeth and the device was discarded as totally unsuccessful.

With this group any combination of behavior
modification techniques, chemotherapy, restraints,
or mouth guards may be appropriate treatment dur-
ing the developmental years.

Category III: Children with severe self-mutilative
behaviors involving the teeth that begin at an early age. When lip biting begins in these children, it is im-
perative that a program team be organized to closely monitor these self-mutilative behaviors. This team
would include, but not be limited to, a physician, dentist, behaviorist, social worker, direct care staff,
and/or parents. Immediate attempts should be
instituted to control the self-mutilative behavior
through behavior modification techniques and/or
chemotherapy. The dentist should attempt the con-
struction of a mouth guard/splint if cooperation
allows an intraoral impression, although this is usu-
ally not the case with Category III patients. The
most important factor is the immediate institution
of maladaptive behavior monitoring and team agree-
ment on procedures to follow if the behavior is not
corrected. Any tooth causing lip damage should be
extracted under local anesthesia when it is apparent
that other measures have failed. The difficulty is
allowing enough time to adequately evaluate the suc-
cess or failure of behavior change methods without
allowing excessive lip damage to occur. The author
suggests that the direction of the team decision
should be toward conservation of lip tissue. If tooth-
associated self-mutilative behaviors persist after the
extraction of offending teeth in spite of active and
intense behavior modification therapy, the extraction
of remaining teeth should be strongly considered. The
tigger for this decision should be the involvement
of a second tooth or group of teeth.

The extraction of only the incisor teeth has not
proven effective in preventing further lip damage in
the author’s experience. At first biting seems to
cease, but then the cuspid teeth become involved by
biting or rubbing the soft tissue between the labial
surface of the tooth and a hard surface. This produces
damage at the corner of the mouth which can be ex-
tremely disfiguring. If cuspids are removed the pro-
cess continues to the bicuspids and then to the molar
teeth. Cheek biting with the molar teeth, after
anterior teeth have been extracted, is common. Most of these children are extremely difficult to han-
dle and constantly scream during the extraction pro-
cedure. Thus, full-mouth extraction of all primary
teeth under general anesthesia is indicated. After all
primary teeth have been extracted, there is usually
a pause in mutilative behavior which allows time for
the team to plan future treatment options and ex-
plore new concepts of mutilation control, especially
in the area of chemotherapy. Unfortunately, when the
permanent teeth begin erupting, the discouraging
and disfiguring process usually begins to repeat
itself, ultimately resulting in full-mouth extraction,
preferably under general anesthesia.

The complete extraction of teeth before major
mutilation takes place still does not guarantee an
edentulous teenager with intact perioral tissues,
although this should be the limited goal with
Category III patients. Unfortunately, delays in the
decision to extract teeth have almost always resulted
in an edentulous or nearly edentulous teenager with
extreme perioral disfigurement. It is a dramatic and
difficult decision on the part of the dentist, parent,
or other professionals to extract permanent teeth in
order to prevent a greater degree of facial disfigure-
ment. At present, there appears no reasonable alter-
native for these patients.

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