GM1 Gangliosidosis and Oral Self-Injury: A CASE REPORT

Simon LS, Vergotine RJ, Boynton JR*

University *of Michigan School of Dentistry Ann Arbor, MI, Mott Children's Health Center, Flint, MI, Hurley Medical Center, Flint, MI*

ABSTRACT

GM1 gangliosidosis is a lysosomal storage disease that progressively destroys neurons in the brain and spinal cord. Degree of severity typically depends on the age of onset and rate of psychomotor deterioration, with Type I (infantile), being the most severe of the three types. The most notable characteristics that individuals with this condition present with include developmental arrest followed by psychomotor deterioration, dystonia, progressive feeding difficulties, and skeletal abnormalities. Oral self-injurious behavior (SIB) has been reported to be common in certain diseases, syndromes, and disorders. Biting is reported to be the most common form of SIB in patients with mental delay. Historical treatment options in patients with compulsive oral SIB have included psychological, pharmacological, intraoral devices, or surgical interventions. In this report, we present a 12-year-old male with GM1 gangliosidosis who presented with a chief concern of chronic oral self-injurious behavior, our initial treatment, and long-term comprehensive care considerations.

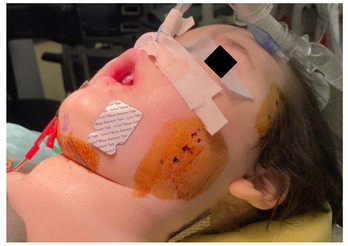
INTRODUCTION

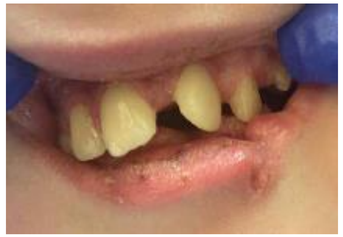
GM1 gangliosidosis, also known as β-galactosidase-1 (GLB-1) deficiency and Landing disease, is a lysosomal storage disease that progressively destroys neurons in the brain and spinal cord. A mutation in the GLB1 gene causes a decrease in the activity of β-galactosidase, the enzyme that breaks GM1 ganglioside down. The accumulation of GM1 ganglioside to toxic levels in tissues and organs, including the brain, leads to the destruction of nerve cells.

The approximate occurrence of GM1 gangliosidosis is 1 in 100,000-200,000 live births, affecting both males and females. GM1 gangliosidosis is passed down in an autosomal recessive pattern, with Type I as the most severe and most frequently reported form. Classification of the three types, based on the age at which signs and symptoms first appear, the severity, and residual enzyme activity, is a spectrum due to overlap of symptoms. Clinical defining features include developmental arrest followed by psychomotor regression, macula cherry-red spots, odontoid hypoplasia, and irregularity of endplates of vertebral bodies. Other notable features include cardiomyopathies, seizures, generalized skeletal dysplasia, clouding of cornea, swallowing/feeding difficulties, and increased oral secretions. Oral and dental features specific to GM1 gangliosidosis are inconclusive and have rarely been reported in literature. However, these patients may present with increased calculus accumulation, oral secretions, gingival hyperplasia, and possible abnormalities of teeth formation and eruption.

Oral self-injurious behavior (SIB), a deliberate, repetitive and persistent behavior, results in physical injury of the head, neck and hands. SIB is common in certain diseases, syndromes and disorders, including movement disorders of neurodegenerative origin, similar to GM1 gangliosidosis. This case report identifies chronic oral self-injurious behavior as another observed oral manifestation with GM1 gangliosidosis that has not been widely discussed in the literature.

CASE REPORT





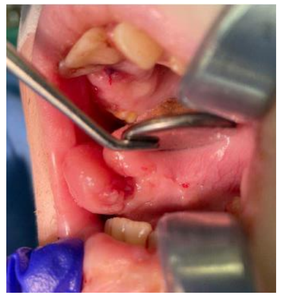
Patient A.H. 12y0m Male

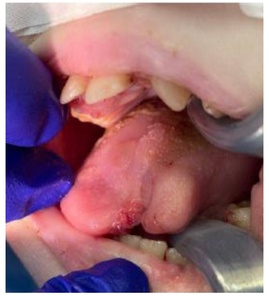
Current Medical Diagnoses:
- Late infantile GM1 gangliosidosis
- Epilepsy (ketogenic diet)
- Developmental delay
- Cerebral palsy
- Chronic respiratory insufficiency
- Obstructive sleep apnea
- Hypoxia
- G-tube dependence

Current Medications:
- Daily: Acetylcysteine, Albuterol, Baclofen, Budesonide-formoterol, Clobazam (Onfi), Cyproheptadine, Klonopin, Midazolam, Lamotrigine, and Miralax
- PRN: Tramadol, Diastat, Naloxone

Allergies: Latex, midazolam, adhesive tape silicones, Prasterone, coconut oil

CC: Injury to the point of laceration and inability to achieve hemostasis at home. EMS were called twice for bleeding >45 minutes, often multiple times per day, requiring sutures and silver nitrate.



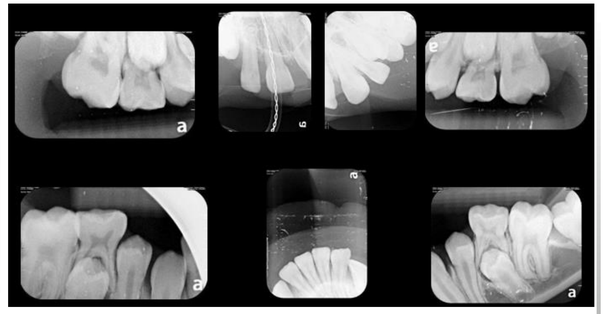


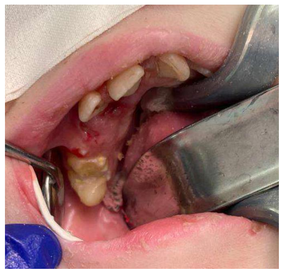
Clinical Findings:
- Lip laceration on lower left lip, extending from wet-dry line past the vermilion border with redness and scabbing

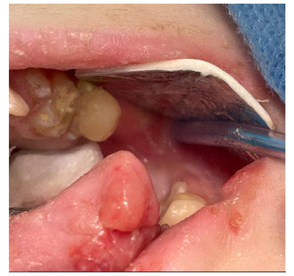
Oral & Dental Findings:
- Laceration on the left side of his tongue with erythema on the ventral surface of his tongue
- Moderate generalized gingivitis, mild calculus on molars
- Late mixed dentition – Class I molar
- 5mm overjet, 10% overbite
- Diastema between #8, #9, and #10 with distal rotation of #10
- #7, #8 Grade III mobility
- No clinical caries or existing restorations
- Low caries risk

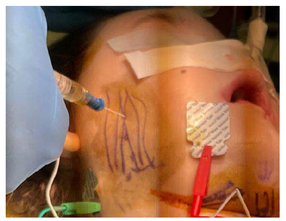
Diagnosis:
- Oromandibular dystonia, muscle spasms with traumatic wound of tongue and lower lip

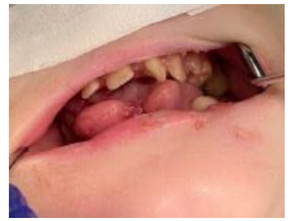
Treatment Plan:
- Dental rehabilitation under general anesthesia due to complex medical history and treatment needs
- Complete intraoral exam
- Full mouth radiographs
- Open bite with composite on maxillary first permanent molars
  - Restorations: #3-O, #14-O composite build up
- Extraction of #7, #8 due to aspiration risk; #9, #10 offending teeth
- Chemodenervation with 100 units botulinum toxin with Myoguide
  - Masseter, temporalis, anterior digastric muscles











Post-Operative:
- Two apneic episodes in the PACU, difficulties coming out of anesthesia
- PICU for observation (vomiting, seizures)
  - Acute on chronic anemia secondary to chronic lip and tongue biting
- 24 hour bedside exam revealed extraction sites healing within normal limits

DISCUSSION

Upon initial presentation to the hospital dentistry clinic at C.S. Mott Children's Hospital, Ann Arbor, MI, mother stated chief concern of chronic lip and tongue injury with profuse bleeding. Due to his dental presentation and complex medical history, A.H. had comprehensive dental treatment completed in the operating room, coordinated with oral surgery. It was determined that psychological, pharmacological and intraoral device options may not be effective for patient A.H., thus requiring a conservative surgical approach. It was discussed with mother that long-term treatment considerations may include complete edentulism if botulinum toxin injections and opening patient's bite are ineffective.

Considerations for edentulating patient A.H. in the future would include limiting additional general anesthesia procedures when possible, as patient had post-operative complications following the procedure. Maintenance of chemodenervation of muscles of mastication would require follow up injections under general anesthesia to immobilize the muscles vs. extraction of remaining dentition under general anesthesia at once. The recommendations of this patient's specialists (pulmonology, neurology, nephrology, hematology) should also be considered in determining long-term definitive treatment for this patient. Regular periodic dental exams with patient positioned in wheelchair to review oral hygiene, remove calculus, ensure retention of restorations, and evaluate effectiveness of botulinum toxin is recommended.

Patients with GM1 gangliosidosis may have variable dental presentation, depending on severity of the disease. A multidisciplinary approach should be considered to manage the symptoms that these patients present with. As clinicians involved in oral and maxillofacial care, we must be aware of how oral self-injurious behavior may precipitate bleeding issues in patients with neurodegenerative diseases. It is important to be familiar with all treatment modalities for SIB and to be cognizant of patient's medical histories to provide appropriate treatment, proper intervention, and definitive long-term care for patients.

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