CASE STUDY

Improvement in Health Outcomes for an Infant with Type 2 Gaucher Disease Undergoing Chiropractic Care: A Case Report

Lawrence Callaway, DC1 & Suzanne L. Ray, MS2

Abstract

Objective: The chiropractic care of an infant with Type 2 Gaucher disease is described with the purpose of reviewing the effectiveness of chiropractic care on aberrant somatic activity in the neuronopathic form of Gaucher disease.

Clinical features: The patient is an 8-month-old male infant who has torticollis, esotropia, hypertonia of the paraspinal musculature, hypotonia of the limbs, limited range of motion and Failure to Thrive.

Intervention and Outcomes: The patient received chiropractic adjustments utilizing an Activator Instrument to the first cervical vertebra, first thoracic vertebra, and the sacrum. After 17 visits over 10 weeks of care, improvement in torticollis and hypertonia were noted as well as increased range of motion, facial expressions, and eye control.

Keywords: Chiropractic; Subluxation; Adjustment; Type 2 Gaucher Disease; Neuronopathic Gaucher; Pediatric Torticollis

Introduction

The normal human genome is capable of providing instructions for the breakdown of sugar–lipid combinations. Mutations in the GBA gene (Gaucher disease) are known to create deficiency in the production of beta glucocerebrosidase, an enzyme necessary for this task, resulting in an accumulation of glycolipid (glucosylceramide) within the scavenging and degradative machinery (lysosomes) of the body.1

This metabolic faux pas is common enough to rank as the world’s most prevalent lysosomal storage disease, occurring at the rate of 1 per 57,000-100,000 live births.2,3 A Gaucher disease registry exists to study and support this condition, which is inherited as a recessive gene and is most prevalent among the Ashkenazi Jews.4

Clinical research currently divides Gaucher disease into three types. Type I is the most common and is referred to as the non-neuronopathic form. The patient is primarily affected in the liver, spleen, bone, and bone marrow. Type 2 (GD2), the acute neuronopathic form, constitutes only 1% of all Gaucher cases, but it includes severe central nervous system involvement and is usually fatal within the first two years of life. Type 3 does involve the CNS, however, it is more gradual in its progression.5

Knowledge of Gaucher disease is important to the chiropractor because of its skeletal manifestations, including the potential for pathological fractures.6,7 Type 1 typically presents with bone pain and can involve the spine in both adult and pediatric populations.8,9

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1. Assistant Professor, Life Chiropractic College West & Private Practice of Chiropractic, Hayward, CA
2. Professor, Life Chiropractic College West, Hayward, CA
The GD2 patient is an infant who notably displays hepatosplenomegaly and anemia along with a failure to thrive (FTT).10 Motor involvement is extensive, including severe weakness,3 hypertonia, torticollis, myoclonus, esotropia, dysphagia, and difficulty breathing. Seizures are also common.11

Currently, the most successful treatment for Gaucher disease is enzyme replacement therapy (ERT),12 although other options include substrate reduction therapy, bone marrow transplant, liver transplant, supportive care, and counselling.10 These treatment options are largely relegated to the Type I condition where clinical trials have proven more successful and where the disease course permits time for a definitive diagnosis through glucocerebrosidase assay.11,12 Consequently, this has left few options for parents carrying the recessive gene and giving birth to the Type 2 Gaucher baby.

Under such circumstances knowledge of alternative means of palliative care become particularly important, especially for the underserved communities.13-15 It is the reason this mother sought chiropractic care for her infant. We report the first chiropractic intervention for a case of GD2 in a child of Hispanic descent.

Case Report

We describe the care of an 8-month-old infant with a medical diagnosis of neuronopathic Gaucher disease. He was diagnosed by a geneticist around 3-months of age. His mother notes that he has been constantly sick and rated his discomfort as 8-10 on the Visual Analog Scale (VAS) with 10 being inconsolable. He has experienced fever, excessive pulmonary secretions, difficulty breathing, vomiting after feedings, diarrhea with creamy green stools, and has been told that he is in the Failure to Thrive (FTT) category. His mother noted that his head has been in constant extension for at least two months.

Upon examination the findings indicated chronic extension torticollis with cervical hypertonicity and myospasms. The patient was unable to flex his head at all. He had right eye esotropia and hypotonicity in all four extremities.

Subluxations were detected and recorded at C0, C1, T1-T12, costothoracic articulations, and sacroiliac region. The subluxations were determined by global and segmental decreased range of motion (ROM), visual and palpatory muscle hypertonicity, and postural deviations.

Interventions

The patient was managed with a spring-loaded instrument (SLI), Activator V, which is a hand-held device with a blunt stylus capable of delivering a high-velocity low-amplitude force (HVLA) to the targeted spinal areas. The HVLA was administered with a pediatric stylus and tensioned to the lowest setting at the areas of decreased ROM: C1 on the right (ASR), T1 on the right (PR), and at S2 (Base Posterior Sacrum) for a period of 10 weeks with a total of 17 office visits.

These adjustments were based on the size of the patient and in accord with best clinical pediatric practices and Activator Methods.16-18 A reevaluation was performed at six weeks.

A typical visit lasted 30 minutes. The patient’s mother was consulted regarding the current status of the patient. Any relevant dietary actions; patterns pertaining to sleep, defecation, and vomiting; and changes in movement were noted. Conversations with other medical providers (e.g. geneticist, gastroenterologist, and neurologist) and updated medical exam findings were shared, especially as related to FTT status.

After completion of the subjective evaluation, a physical examination was performed. This included palpation of muscle tonicity, assessment of segmental biomechanics, and determination of global ROM. The infant was inspected for nasal secretions, percussed for lung consolidations, and monitored for breathing difficulty.

Following the evaluation, treatment was administered to the spinal segments as previously described. Approximately one month into care, passive spinal stretching into flexion and extension was implemented.

Outcomes

The patient became visibly more active, had more control over his body and his torticollis substantially reduced. Cervical ROM improved as follows: passive flexion changed from 50 to 75 degrees, right and left lateral flexion from 25 to 30 degrees, and right and left rotation from 30 to 35 degrees. The infant was able to actively flex his upper extremity, initiate knee and hip flexion as well as support himself on his forearms after seven visits. His breathing improved initially as noted by less consolidation and normalization of his original barrel chest.

The re-evaluation showed right eye motor function improvement in both control and location. More facial expressiveness was present. Tongue movement was more apparent with and without stimulation. The mother estimated the torticollis reduced to a 6 out of 10 on theVAS. The General Pain Disability Index Questionnaire started at a 48 and remained at a 48. No adverse effects were documented from the chiropractic care.6

Discussions

From its inception chiropractic has concerned itself with the health of the spine, the functioning of the nervous system, and fundamental elements necessary to thrive. Although many models exist to explain the benefits of chiropractic,19 practitioners typically administer HVLA forces to the human body at points determined by diagnostic protocols and appropriate for the age and size of the patient.17 A review of the literature suggests the appeal of SLI and Activator Methods to pediatric populations.18

A recent study posits that chiropractic continues to be viewed as a favorable health solution in general and is sought by adults in proportion to available chiropractic practitioners.20

While chiropractic has largely demonstrated itself to be
effective for back \(^{21}\) and neck pain \(^{22}\) in adults, there is a growing utilization for pediatric concerns \(^{23,24}\). However, data collected by the Centers for Disease Control (CDC) on use of alternative therapies, including chiropractic, reveal that Hispanic children access it 1.5 times less than non-Hispanic children. \(^{14}\)

Parents are more likely to reach for complementary and alternative strategies where medical costs delay access to care. \(^{14}\) This would especially apply to conditions such as GD2 for which medicines are in the earliest stages of development for limited applications and where populations fall out of access economically. \(^{25}\)

Although chiropractors currently manage pediatric patients with a wide variety of conditions, there are few clinical trials documenting outcomes. \(^{24}\) Numerous case reports exist for chiropractic intervention with pediatric torticollis, both congenital and acquired, \(^{26,27}\) including one review. \(^{28}\) For those cases in which the torticollis improved, it is interesting to note that there were other gains in motor function, such as less facial distortion, less feeding difficulty, normalization of muscle tone, resolution of colic, and improved oculomotor function. \(^{26,28}\) Although the literature \(^{7}\) generally supports the use of chiropractic for pediatric torticollis, this is the first case study to report its application and benefit for torticollis in the neuronopathic form of Gaucher disease.

The objective findings that occurred during this retrospective study are consistent with several other studies on reduction of hypertonia, improved motor function, and change in overall health status. \(^{26-30}\) Although results of manipulative therapies for visceral disorders like colic remain mixed, \(^{24,29,30}\) it has been proposed that segmental dysfunction could theoretically alter spino-visceral reflexes, potentially compromising visceral functions. \(^{31,32}\) The chiropractic adjustment might affect these reflexes involving the autonomic nervous system and result in improved visceral function and overall well-being.

In general, it is the clinician’s responsibility to determine whether underlying pathology exists such that co-management is indicated. This case was previously medically diagnosed and managed to the extent of its limitations. Although there are a few case studies which support successful chiropractic intervention for FTT, \(^{33-35}\) our case was confounded by the severity of the underlying genetic Gaucher condition.

Our study shows that chiropractic care can result in the amelioration of at least some of the distress found in association with this otherwise incurable disease. The needs demonstrated by cases such as we describe compel us to conduct further research to rule out the possibility that changes observed were simply due to the natural course of a disease process or some otherwise unknown intervention. It encourages investigation of cases outside the traditional scope of neck and back utilization for chiropractic, areas for which chiropractic care might not previously have been considered, as well as to promote its application to a wider demographic. \(^{36}\)

**Conclusion**

The development of medical treatments for extremely rare conditions may of necessity lag behind those of the more common. Our findings suggest that chiropractic may provide some relief for the effects of neuronopathic Gaucher disease. More evidence is required to both confirm and expand this application.

**References**