CASE STUDY

Chiropractic Management of an Infant with Paroxysmal Tonic Upgaze Syndrome, Congenital Torticollis & Upper Cervical Subluxation Following Birth Trauma: A Case Report

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ABSTRACT

Objective: To describe the use of upper cervical subluxation-based chiropractic care in the management of a 12-month-old male child with Paroxysmal Tonic Upgaze (PTU) Syndrome, torticollis, and neurological issues secondary to birth trauma.

Intervention & Outcomes: The patient was assessed for vertebral subluxation using National Upper Cervical Chiropractic (NUCCA) and Activator protocols revealing right atlas laterality and sacral subluxation. The patient was seen for four months for a total of nine visits. After the first chiropractic correction parents noted that his sleep improved. Care continued weekly for one month and parents reported improvement in PTU and torticollis.

Conclusion: This case study suggests that the removal of vertebral subluxation can be beneficial in resolving symptoms related to PTU and congenital torticollis. Further research is warranted.

Key Words: *Upper cervical, paroxysmal tonic upgaze, congenital torticollis, NUCCA, Activator, vertebral subluxation*

Introduction

Paroxysmal Tonic Upgaze Syndrome (PTU) is a distinctive non-epileptic disorder of abnormal ocular movements with onset during infancy that can occur in otherwise neurologically normal subjects.[1] PTU was first described in 1988 by Ovrier and Billson.[1] The condition presents in infancy characterized by episodes of conjugated upward eye deviation with neck flexion, down beating saccades in attempted downgaze, preserved horizontal eye movements, and normal consciousness, which are often associated with an impaired ability to coordinate movements.[2]

These manifestations are frequently exacerbated by fatigue, intercurrent infection or vaccination and relieved by sleep. In some reports, this condition is associated with other neurological symptoms such as ataxia, psychomotor retardation, intellectual disability, nystagmus, amblyopia or strabismus. Additionally, some patients may exhibit developmental delay and ataxia associated with PTU syndrome.

The exact pathogenic mechanism of the disorder is still unknown. Some familial cases have been reported. Generally, PTU disappears with time and spontaneous remission occurs within a few months or years without any change in psycho-motor development.[3] There have been different sources that

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suggest the child with PTU might possibly have sensory integration dysfunction, language disorders or delays, vertigo and loss of muscle tone. Genetically, it has been occasionally associated with mutations in CACNA1A, a gene that encodes the ion-conducting pore and voltage-sensing a subunit of the neuronal CaV2.1 (P/Q-type) calcium channel and that has been linked to familial hemiplegic migraine (FHM).[4]

Congenital Muscular Torticollis (CMT) in Latin means "twisted neck" and was first defined by Tubby in 1912 as "A deformity, either congenital or acquired, characterized by lateral inclination of the head to the shoulder, with torsion of the neck and deviation of the face.'' The term congenital muscular torticollis has been used by various investigators to denote a neck deformity primarily involving shortening of the sternocleidomastoid muscle that is detected at birth or shortly after birth. Although a combination of various theories has been proposed, the true etiology of torticollis remains uncertain. Among them is birth trauma, which proposed that congenitally shortened sternocleidomastoid muscle was torn at birth with the formation of a hematoma, which then underwent fibrous contracture.[5]

Congenital muscular torticollis (CMT) is the third most common musculoskeletal impairment in infants next to hip dysplasia and clubfoot. The reported incidence of CMT is 0.4%-2.0%; however, the incidence may be higher; Stellwagen et al found that 16% of newborns had torticollis. CMT is a result of shortening or excessive contraction of the sternocleidomastoid muscle, with limited range of motion in both rotation and lateral flexion in the neck and an imbalance in muscle function around the neck. Due to the positional preference, there is a high risk that infants with CMT will develop deformational plagiocephaly if they spend most of their time in the same position,[6] Researchers hypothesize that congenital postural torticollis is caused by anomalous fetal skull positioning present in a footling or breech presentation and following difficult labors and deliveries. Congenital muscular torticollis is due to birth trauma with resultant endomysial fibrosis and shortening of the SCM muscles.[7]

Case Report

History

The patient is a 13-month year old male presented at the office for assessment and management of PTU, congenital torticollis, and infantile spasms/tremors. After inconclusive MRI and various neurological examinations, parents sought alternative care. Mother described pregnancy and delivery as difficult. Inclusive of two weeks of nonproductive labor prior to delivery. The baby was laying on his back and presented "face" first, breaking his mother's coccyx during the delivery process.

At one month of age the child received vaccinations, the mother began to notice the child had muscular spasms in the left shoulder with right head deviation. Also, Moro Reflex was present at night without stimulation. At three months an MRI was performed, then an electroencephalography in order to rule out epilepsy. Both tests were inconclusive. Three weeks following these symptoms, the child began to hit the back of his head aggressively. The child also began to show the signs and symptoms of Benign Paroxysmal Tonic Upward Gaze Syndrome (BPTU). Once noticing these sequences of symptoms and realizing that these symptoms were not going away, the mother took the child to the pediatrician. Once referred to a neurologist, the child was diagnosed with benign BPTU and congenital torticollis.

After receiving the child's diagnosis, the pediatrician believed the infantile jerky movements to be seizures, leading the doctor to have the child screened for a 24-hour electroencephalography test. An MRI revealed no neurological issues but he was monitored regularly by neurologist.

Chiropractic Examination

A physical examination was performed on the child, which consisted of the child's height, weight, vital signs, spinal range of motion, motion and static palpation, and neurological tests. A prone leg check showed a short right leg ¼ inches. Joint fixations with biomechanical alterations of the surrounding areas were noted with hypomobility, and a hard end feel at C1. Palpation of the right side of the body objectively showed pain, spasm, and change relative to the left side of cervical paraspinals. Palpation of the left side of the body showed pain and spasm within the shoulder with right head deviation.

Palpation and motion showed that the child had a left shoulder pain/sensitivity with touch and muscular spasm of the left sternocleidomastoid. During motion of the right atlas the patient's eyes began an upgaze deviation of to the left. Palpation and motion revealed a left sacral ubluxation.

Cervical ranges of motion was performed on the patient, showing a cervical flexion of 60 degrees, cervical extension of 45 degrees, cervical left lateral flexion of 55 degrees, all three tests were notable with no pain during active motion. Cervical right lateral flexion of 35 degrees pain was noted with active motion of right lateral flexion. No ataxia or nystagmus was observed. During an episode, conjugate upward deviation of the eyes and neck flexion occurred. Upon observation, the child began crawling on the floor with his left knee in crawl position and the right leg extended, as if the child was trying to walk and crawl at the same time.

Neurologic and ophthalmologic examinations produced normal results during the periods between episodes. The pupils were normoisochoric, and reacted normally. During observation, the child had jerky infantile movements. Vestibular ocular reflexes were present. The patient's consciousness was clear. The child's gait was abnormal. However, the child showed a positive Moro reflex; after seeing this test performed on the child, the mother noted that the child would perform same reaction before sleeping. The patient was diagnosed with vertebral subluxations at levels C1 and sacrum as well as infantile spasms, injury to the shoulder and upper arm, and disorder of the eyes.

Chiropractic Care

Based on the findings in the physical examination, it was determined that the goal of care would be to reduce vertebral

subluxations and restore motion. The patent was placed on a 2 times per week schedule, and was seen a total of 30 visits in a 4 month period. The patient was adjusted with Activator (contact specific, high velocity, low amplitude) technique. Adjustments were administered on a chiropractic table. In addition periodic exams were performed every 10 visits to evaluate the effectiveness of treatment.

Cofounders Warren C. Lee, DC, and Arlan W. Fuhr, DC, discovered the Activator Methods Chiropractic Technique (ACMT). The Activator Adjusting Instrument (AAI), derived originally from a dental impactor.[8] The Activator Adjusting Instrument (AAI) is a low force, moving stylus-type of mechanical instrument. The AAI is powered by the fixed potential energy of a spring that propels a 116-g hammer into a 30-g stylus. The spring is compressed manually by squeezing a sliding handle located on the shank of the instrument, and at a predetermined point is activated, propelling the hammer into the stylus. An 80-durometer rubber tip is attached to the end of the stylus and reduces the impulse force shock delivered to the spine slightly when the instrument is activated.[8]

The Activator Assessment protocol prescribes a series of prone leg-length observations and provocative tests to evaluate the function of the joints from the feet progressively upward to the cervical spine. It is believed that dysfunction of more caudal segments must be cleared before more rostral structures can be properly evaluated. The protocol has both theoretical and empirical roots. Initially derived from the leg-check concepts of Van Rumpt, the Derefield and other various isolation, pressure, and stress tests have largely evolved from the clinical experience of Activator practitioners.[8]

The ACMT involves two forms of tests that are used to determine vertebral subluxations at different levels. These tests are called stress and pressure tests. Stress tests are applied by the doctor's forefinger or thumb to accentuate the suspected dysfunction or subluxation, as indicated by leg-length inequality. The force is applied in the direction of subluxation. If no change in apparent leg length is observed, the target area is considered free of dysfunction.[8]

On the other hand, "pressure tests" involve gentle digital force applied to the suspected subluxation in a direction of correction. This vector is applied to temporarily "reduce" the positional misalignment or dynamic dyskinesia of a vertebral joint. With a pressure test, the leg-length inequality is expected to balance.[8] "Isolation tests" are maneuvers performed actively by the patient for stimulating subtle muscular changes in the body, perhaps via mechanoreceptors in muscles, diarthrodial joints, ligaments, or tendons associated with the axial and appendicular skeleton.

In the presence of articular dysfunction, specific movements in combinations of rotation, flexion-extension, and abduction-adduction are hypothesized to provoke specific neuromuscular irritations and contractions, which in turn appear to manifest in leg-length changes in a consistent manner. The reaction of the initially shorter leg in position 1 (designated the "PD leg" for the "pelvic deficiency" thought to produce the functional short-leg phenomenon) is believed to indicate the presence or absence of subluxations somewhere in the body.[8]

The patient's right cervical subluxation was adjusted supine contacting the lamina- pedicle junction of the involved segment using the Activator instrument applied over the pad of the index finger of the doctor. The line of correction was lateral-medial.

The sacral adjustment was administered prone and required the segmental contact point located at the 2nd sacral tubercle. The line of correction was superior to inferior.

Results

The patient was seen for 30 visits over a 4-month period. The patient was compliant with care. Following the first reassessment of care, the mother noticed the child was sleeping better and left shoulder and left sternocleidomastoid spasms (congenital torticollis) had diminished. The mother also noticed that the PTU had diminished. By the next reassessment it was noticeable that the sensitivity to touch of the left shoulder/trapezius was notable, however post adjustment resulted with diminishment of sensitivity. It was notable through observation that the right cervical rotation decreased; the child's head was rotated more to the left returning to a neutral position.

In addition, there was improvement overall in his coordination with the child's crawling; the child was now crawling with both knees on the ground. At the third reassessment, the mother noted that she and her husband were able to touch the child's left shoulder without him flinching, he no longer hits the back of his head, he began walking and is consistent with it on a regular basis and he continues to sleep well without any muscular spasms. Measurable results of the reassessment showed that the patient's right cervical lateral flexion range of motion increased and there was no notable pain during active motion.

The patient currently remains under chiropractic care and continues to show improvements in ranges of motion, coordination, and proprioception. However, with the infantile jerky movements still appearing (now occasionally), the mother stated the child's pediatrician wanted to schedule the child for an MRI with sedation to get a further look to discover the cause of the infantile jerky movements. However, with the great results the child had with chiropractic the mother did precede with that action which revealed a low cerebellar tonsil, Chiari 0, which the doctors noted as insignificant.

Discussion

Paroxysmal tonic upward gaze (PTU) is a rare neuro-opthalmalgic disorder. This disorder is considered a distinctive non-epileptic disorder of abnormal ocular movements with onset during infancy that can occur in otherwise neurologically normal subjects.[9]

Upward deviation of the eyes occurs in various conditions. It may represent partial epileptic activity; it is sometimes also noted in postencephalitic extrapyramidal disorders, in altered level of consciousness, in brain stem disorders and in the context of retinal visual loss with some preservation of the inferior visual field.[10] PTU of childhood appears to be a

transient paroxysmal dystonia which may present an autosomal dominant pattern of inheritance, similarly to the other familial idiopathic paroxysmal nonkinesigenic dystonias.[11]

The cause for congenital muscular torticollis is unknown; many researchers believe that various causes such as birth presentation, birth trauma, and the relation of the fetus within the uterus. A congenitally shorted SCM is torn at birth with formation of a hematoma. Subsequent endomysial fibrosis results in deposition of collagen and migration of fibroblasts around individual muscle fibers that undergo atrophy.[12] Generally, treatment for congenital muscular torticollis has consisted of an active home exercise program, gentle manual stretching therapy, application of orthosis, botulinum toxin A injection, vigorous manual myotomy, and various surgical procedures.[13] Manual therapy is a widely used method for spinal pain management but there are few reports in the literature that focus on manual therapy for cervical subluxation.[14] Chiropractic treatment is non-existent in consideration of treatment for PTU and limited for congenital muscular torticollis.

A case study in 2012 reviewed a 14-month-old boy with paroxysmal conjugate upward deviation of the eyes. He was born via normal vaginal delivery at term after an uneventful pregnancy. His psychomotor development and medical history were normal. No family history of neurologic disease or other paroxysmal neurologic phenomena was reported. The paroxysmal phenomenon began at age 11 months with episodes of upward gaze lasting approximately 1 or 2 hours.[15]

The patient**'**s gait was unstable during these episodes. These episodes occurred several times during the day, and were relieved by sleep. Episodes were exacerbated by fatigue and illness.[15] Treatment for the child consisted of Levodopa treatment. The result of this treatment has been effective. One year post treatment there was no reoccurrence of the symptoms of PTU. The child's psychomotor development, neurologic examination, and ocular movements were all normal.

In addition, McWilliams and Gloar discussed a six-year-old child with congenital torticollis with a left head tilt due to a caesarian birth. Screening of a computerized tomography (CT) of the brain revealed a plagiocephaly. Visual observation showed a left facial asymmetry. Deerfield leg-length analysis15 indicated the presence of a left cervical syndrome. Palpation of the cervical spine indicated posteriority of the atlas vertebra (C1) posterior arch on the right and laterality of the C1 vertebra on the right indicating an atlas superior right posterior (ASRP) at C1 according to the Diversified chiropractic technique.[7]

The examiner also palpated a superior orientation of the spinous process of axis vertebra (C2) with a noticeable decrease in the occipitalaxial spacing.[7] The child's subluxations were corrected via the Diversified technique. The child was adjusted on the following two visits, which resulted with a dramatic decrease in left head tilt. The child underwent physical therapy as well to retrain the left sternocleidomastoid muscle 8 months later the child was examined and once again 1 year later. The child did not need to undergo surgery for her congenital torticollis that was corrected with the chiropractic care that was provided.[7]

Proposed Mechanism

When a subluxation is present it blocks communication and sensory input between the brain and the body and can lead to neurological dysfunction. This dysfunction without any correction leads to further damage. When a subluxation is present and left uncorrected it will lead to sensory input dysfunction. Kent explains with his dysafferentation model, which states that neurological dysfunction associated with the vertebral subluxation may take other forms.[16]

The intervertebral motion segment is richly endowed by nociceptive and mechanoreceptive structures. As a consequence, biomechanical dysfunction may result in an alteration in normal nociception and/or mechanoreception. Aberrated afferent input into the CNS may lead to dysponesis.[16,17]

Successfully removing the subluxation and restoring proper motion and position of the segments, the afferent sensory information is returned to normal and in turn, resolves the neurological dysfunction thus potentially reducing the symptoms of PTU and congenital torticollis.

Conclusion

The rare condition of PTU is a challenging disorder which is characterized as "eyes up, chin down" as the characteristic of paroxysmal tonic upgaze. The present analysis indicates that benign and transient paroxysmal tonic upgaze of childhood can be managed with parental reassurance.[18] Paroxysmal tonic upgaze is an entity of heterogeneous origin. Familial cases occur, apparently of both autosomal dominant and recessive inheritance.[19] It is hypothesized that PTU probably results from a localized, transient neurotransmitter dysfunction within supranuclear pathways that control vertical eye movements but the presence of ataxia as an associated finding in these patients is presumed to reflect dystonia and classify this entity within the group of inherited dystonias.[18]

Research has shown that up to 50% of patients with PTU may manifest psychomotor and language delays.[20] Though chiropractic appears to have provided improvement in this case for both PTU and congenital torticollis there are limitations within this study.

More research is warranted to determine if chiropractic care can be beneficial in resolving symptoms related to PTU and congenital torticollis.

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