CASE STUDY

Upper Cervical Chiropractic Care of a Patient with Joubert Syndrome and Hemifacial Spasm

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Abstract

Objective: To describe the upper cervical chiropractic care of a pediatric patient with Joubert syndrome and hemifacial spasms.

Clinical Features: A 3-year-old boy with Joubert Syndrome presented to the office with tachypnea, occasional apnea, hemifacial spasm, nystagmus, strabismus, delayed motor skill development and hypotonia. The mother's main concern was a hemifacial spasm that occurred three to four times a week. This was the patient's first chiropractic experience. The atlas subluxation was detected from leg length evaluation, postural analysis and radiographic measurement.

Intervention and Outcomes: The patient received orthogonal-based, upper cervical low-force (UCLF) chiropractic care based on the National Upper Cervical Chiropractic Association (NUCCA) technique protocol. After the first adjustment, the patient did not have a hemifacial spasm for nine days, which was the longest interval between spasms since birth. The spasms completely disappeared after 31 months of care. Except for slightly improved coordination, no other significant change was reported as a result of the chiropractic care.

Conclusions: The positive response observed in this case suggests that hemifacial spasm may be linked to atlas subluxation. The need for further investigation of the connection between atlas vertebra subluxation and cranial nerve dysfunction is indicated.

Key Words: *Upper cervical, subluxation, pediatric, Joubert syndrome, hemifacial spasm, Grostic, orthogonal, NUCCA, orthospinology, microvascular decompression.*

Introduction

Joubert syndrome is a rare disorder of an autosomal recessive trait, first reported by Joubert et al[1] in 1969. Clinical features of Joubert syndrome include ataxia, hypotonia, abnormal breathing, irregular jerky eye movements, and mental retardation.[1-2] Joubert syndrome is characterized by the Molar Tooth Sign on magnetic resonance imaging (MRI). This sign is the result of the hypoplasia or agenesis of the cerebellar vermis and malformed brainstem.[3-5] Other variable clinical features include a protruding tongue, hemifacial spasms, polycystic kidney, odd facial features and polydactyly.[1-2, 6-8] Although hemifacial spasm is not a classic feature of Joubert syndrome, several cases associated with multiple hemifacial spasms have been documented.[1,8] Hemifacial spasms in general are often successfully treated by microvascular decompression surgery of the facial nerve.[9-13] This case report will describe the upper cervical chiropractic care of a 3-year-old boy with Joubert syndrome and hemifacial spasms.

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Case Report

Case History

The patient was brought by his mother for chiropractic consultation two weeks before his fourth birthday. The case history revealed that he was diagnosed with Joubert syndrome soon after his birth. He was born at term by a forceps assisted delivery. The patient's discharge summary noted tachypnea, central apnea, left facial nerve palsy, asymmetry of facial structures and hypoplasia of the cerebellar vermis on MRI. What they initially diagnosed as a left facial palsy was later found to be hemifacial spasm.

At the time of initial chiropractic examination, the patient presented with tachypnea, occasional apnea, hemifacial spasm, nystagmus, strabismus, delayed motor skill development and hypotonia. The patient's mother was mainly concerned with the patient's hemifacial spasms. Three to four times a week, the patient had spasms while sleeping. The side of the face that the spasm occurred on alternated, although it was more frequently observed on the right side. The muscle spasms did not seem to cause excessive pain; however it caused enough discomfort to wake the patient up during the night. Each episode lasted for four to twelve hours. Because the spasms occurred during the night, the patient's mother had to stay awake to comfort the patient and thus resulted in sleep deprivation. The tachypnea and apnea which the patient experienced during the hemifacial spasms were more severe than those normally associated with Joubert syndrome. After each episode, the patient's face would be tight and sore for several days. The patient was on a seizure medication, Tegretol, for the hemifacial spasms but the medication produced only minimal improvements. He was receiving regular speech therapy, physical therapy, hippotherapy, occupational therapy and music therapy, none of which made any significant change in the spasms. At the time of the consultation, four days was the longest duration the patient had ever gone between episodes of nighttime spasms.

Chiropractic Assessment

Orthogonal-based, upper cervical low force (UCLF) chiropractic care is a method of adjusting the spine based on the Grostic model of analysis. In contrast to the high velocity, low amplitude (HVLA) thrusts, UCLF makes corrections to the upper cervical subluxation with minimal force through a specific vector to restore structural balance and optimum function to the body.[14] The chiropractor in charge of this case used NUCCA protocols and procedures in his management of the patient.

Postural distortions indicate the presence of the atlas subluxation in NUCCA analysis. Upper cervical theories state that the brainstem controls muscle tone, so displacement of the atlas affects the brainstem, resulting in spastic contractures of the postural muscles. This results in postural imbalance and deviation of the spinal column from the vertical axis of the body. Leg length measurement and postural analysis are used to determine if there is an atlas subluxation and also to assess the effectiveness of the adjustment at each visit.[14-16] In a supine position the patient had a functional leg length inequality with left leg appearing shorter than the right by 1/4". Postural analysis in the upright position indicated that the patient had left head tilt, neck deviation to the right and an elevated left shoulder.

In Grostic/orthogonal based techniques, the radiographic analysis of the cervical region provides the practitioner with a three-dimensional view of the atlas misalignment to derive the specific vector needed to correct that misalignment. This analysis requires three x-ray films looking at the cervical spine from the front (anterior-to-posterior), the top (superior-to-inferior), and from the side (lateral). The x-ray equipment is specifically designed and aligned for upper cervical chiropractic techniques. Procedures for specific patient placement results in x-rays from which the atlas displacement can be measured to one-fourth of a degree.[14] In this case, it was determined that the atlas vertebra had shifted to the left along the surface of the occipital condyles and rotated anteriorly at the left articulation between the occiput and the atlas (Table 1, Figure 1).

Chiropractic Care

NUCCA adjustments are described as a low-force adjustment that is delivered to the patient with a depth of excursion between 1/16" to 1/8" (Figure 2)[14] The patient received adjustments in a right recumbent position on a chiropractic table with a headpiece that gave firm support to the head. The doctor positioned himself in a precise stance, as dictated by the radiographic analysis, in order to create a specific vector. The doctor's left pisiform was placed over the patient's left transverse process of the atlas vertebrae between the left mastoid process and the ramus of the mandible, while the doctor's left wrist was grasped by his right hand for stability. The actual correction of the atlas vertebra was achieved by a maneuver called a *triceps pull adjustment.* This maneuver, developed by the National Upper Cervical Chiropractic Research Association (NUCCRA), allows practitioners to adjust the atlas vertebra without thrusting into the patient. This is done by contracting the long heads of the triceps muscles to generate just enough force to move the atlas vertebra, and does so without rotation or cavitation of the joint.[14-15] The doctor had the patient rest for ten to fifteen minutes after each adjustment.

Immediately after the first adjustment, the anterior-to-posterior and superior-to-inferior views of the cervical spine were taken again to confirm the effectiveness of the vector used in correction. Post-adjustment radiographs are a criticism of upper cervical care due to increased radiation exposure to the patient. However, a study by Rochester found that the dosage from post x-rays did not increase the total risk of thyroid cancer.[17] Additionally, radiation exposure was further reduced by an average of 65% through the use of lead foil filters designed for upper cervical radiographs.[18] Assessments of the difference between the length of the right and left leg and changes in posture were performed in pre-adjustment and post-adjustment evaluations at each visit.

Results

The patient received his first atlas adjustment on his fourth birthday. Post-adjustment evaluation revealed that the leg length inequality, head tilt, neck lateral deviation and shoulder height difference had been eliminated. The reduction of the atlas misalignment was confirmed by radiographic analysis of the post-adjustment anterior-to-posterior and superior-to-inferior views of the cervical spine (Table 1, Figure 3). The post anterior-to-posterior film revealed that the direction to which the atlas shifted along the occipital condyles was indeed right, not left. The vector of the adjustment was modified accordingly for the future adjustments. The radiographic re-examination that was conducted after six months of care confirmed the right shift of the atlas and the successful correction of the shift following the atlas adjustment (Table1, Figure 4, Figure 5).

Following the atlas adjustment, for the first time in his life, the patient did not have a facial spasm for nine consecutive days. His mother reported at the next visit, nine days later, that the boy began to sit up better after his first adjustment.

The patient received a total of seven adjustments during the first month of care. He experienced three hemifacial spasms on the right side during the one-month period, which was a significant improvement from having a spasm every 3-4 days. Regular check-ups were scheduled every one to two weeks for the following nine months to maintain the proper alignment of the atlas. In addition to the scheduled appointments, the mother brought the boy for the evaluation when he fell, hit his head or had facial spasms. His mother also noted that the patient had exaggerated coordination dysfunction when he was found to be out of alignment. For the eight months following the first adjustment, the hemifacial spasm only occurred on the right side. The frequency of hemifacial spasms fluctuated from zero to eight times a month, until it was completely resolved after 31 months of care. The patient, now nine years old, has continued to receive upper cervical care for wellness.

Discussion

Hemifacial spasm is an involuntary contraction of facial muscles caused by hyperactive dysfunction of the facial nerve.[9] Studies have shown that mechanical compression of the cranial nerve at the root entry/exit zone (REZ) of the brainstem is related to hemifacial spasm as well as trigeminal neuralgia, vertigo, glossopharyngeal neuralgia, and essential hypertension.[9-11, 17-20] The offending structure in adults is usually an artery, although veins and tumors can also be the source of the compression.[9-13, 19-24] The accepted treatment for these conditions is microvascular decompression at the REZ.[9-13, 23-25] The success of decompression surgery at the REZ for hemifacial spasm suggests that the source of nerve compression is on the peripheral part of the facial nerve. However, it is also suggested that the site of compression is actually on the nuclei in the brainstem which makes the facial nerve hyperexcitable.[23-25] This hypothesis is supported by the finding of De Ridder et al[25] that the hyperactivity dysfunction of the facial nerve, the trigeminal nerve and the glossopharyngeal nerve are more frequently found in individuals with longer nuclei of the cranial nerves. The author reported several cases where microvascular decompression of the CNS part of the vestibulocochlear nerve cured vertigo.

Hemifacial spasm in infancy is extremely rare.[26-27] Unlike the cases reported in adults, hemifacial spasm in infancy is usually due to a mass lesion such as cerebellar ganglioglioma or a hamartoma in the fourth ventricle.[27-33] In most cases, the removal of the lesion results in complete remission of the symptoms.[28-32] Following a literature search, only one case of spontaneous remission has been reported for an infantile hemifacial spasm without a space occupying lesion.[26]

Since a chiropractic adjustment of the atlas was successful in the management of hemifacial spasm in the case of a 4-year-old-boy with Joubert syndrome, it is possible that correction of the atlas subluxation had a decompression effect on either the nucleus or the peripheral part of the facial nerve directly or indirectly. The facial nucleus is located in the posterior pons facing the fourth ventricle, and its fibers emerge from the brainstem at the venterolateral aspect of the junction between the pons and medulla oblongata.[34] Although the level of the atlas vertebra is normally inferior to the pontomedullary junction, an atlas misalignment may deform the brainstem and spinal cord through the dentate ligament as proposed by Grostic.[35] This model suggests that the nuclei may become facilitated through cord deformation or microvascular ischemia. Another potential site of compression due to its proximity to the atlas transverse process is the facial nerve. The facial nerve exits from the skull at the stylomastoid foramen. According to the study of Greyling et al,[36] the average distance from the atlas transverse process to the facial nerve trunk is 14.31 mm on the left and 13.76 mm on the right. Since there are many soft tissue structures in that small area, slight atlas misalignment may put enough indirect pressure on the facial nerve trunk to cause hyperactivity. The literature has shown that chiropractic adjustments have successfully managed different cranial nerve dysfunctions including trigeminal neuralgia, Bell's palsy, vertigo, dizziness, tinnitus, hearing loss, spasmodic dysphonia and hypertension.[37-42] These cases might also be explained by the decompression effect of upper cervical adjustments.

It is not known in this case whether or not the hemifacial spasm was an isolated condition or was associated with the patient's Joubert syndrome. Although the first reported case of the Joubert syndrome presented with a hemifacial spasm,[1] it is not a typical characteristic of the syndrome described in the literature. It is possible that the brainstem malformation may have made the nuclei or facial nerve susceptible to compression and/or torsion. It is also possible that hemifacial spasm was a separate, incidental condition that was linked to the patient's history of forceps delivery. Kind et al[8] described three cases of the Joubert syndrome with hemifacial spasm, two of which had a forceps assisted birth while the third was delivered by caesarean section. As previously discussed, there is only one documented case of spontaneous remission for infantile hemifacial spasm. This child was delivered by forceps.[24] The association between the instrument assisted delivery and the hemifacial spasm might be worth further investigation.

Conclusion

This report described the use of orthogonal-based upper cervical chiropractic care of a child with hemifacial spasm. It is not known if the remission of the symptoms after 31 months of care was solely due to the chiropractic management or if it occurred because of the combination of all the therapies the patient was receiving. However, the decrease in the frequency of the episodes from three to four times a week to a complete cessation for nine days immediately following the first atlas adjustment the patient received, may suggest that correction of the atlas subluxation through UCLF techniques may have helped improve the patient's condition. The connection between atlas subluxation and cranial nerve dysfunction should be further studied.

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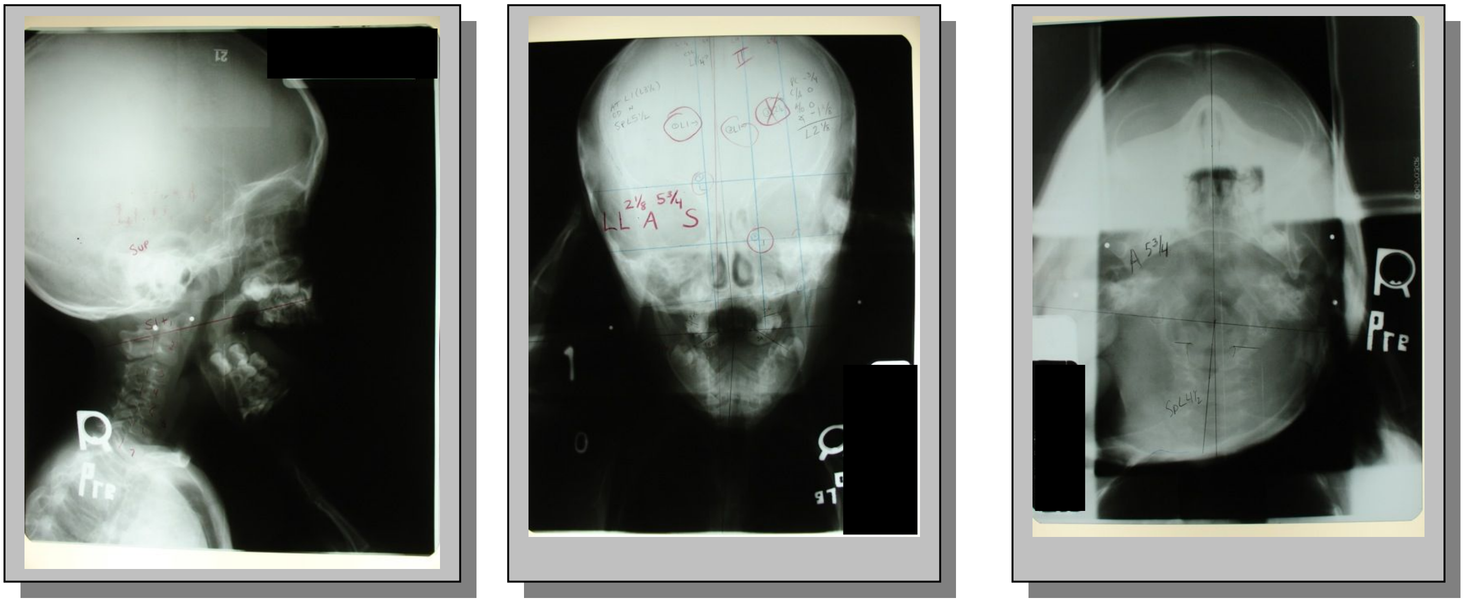
Table 1: Summary table of the radiographic measurements in degrees

	First adjustment		After six months	
Nasium	**Pre**	**Post**	**Pre**	**Post**
Atlas	Left 2 1/8	Right 1/4	Right 1 3/4	Neutral
Vertex	**Pre**	**Post**	**Pre**	**Post**
Atlas	Anterior 5 3/4	Posterior 1/2	Posterior 4	Posterior 4 1/2
Axis spinous	Left 4 1/2	Left 3	Left 4	Left 3

Figure 1: First adjustment: pre-adjustment radiographic analysis (lateral, nasium, vertex)



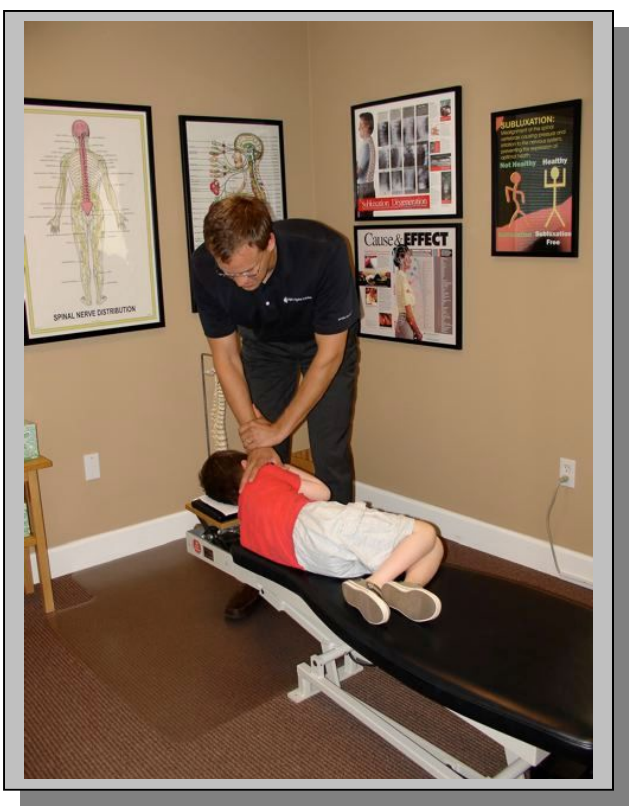


Figure 2: Demonstration of NUCCA adjustment

Figure 3: First adjustment: post-adjustment radiographic analysis (nasium, vertex)

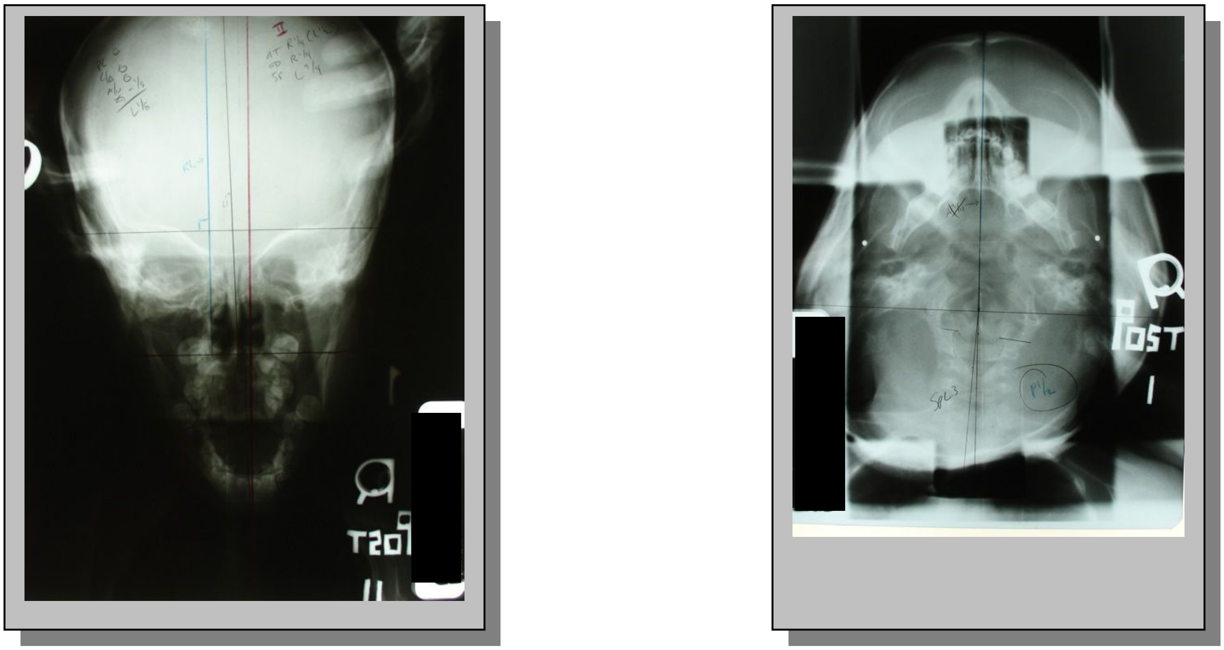


Figure 4: Six months re-examination: pre-adjustment radiographic analysis (lateral, nasium, vertex)

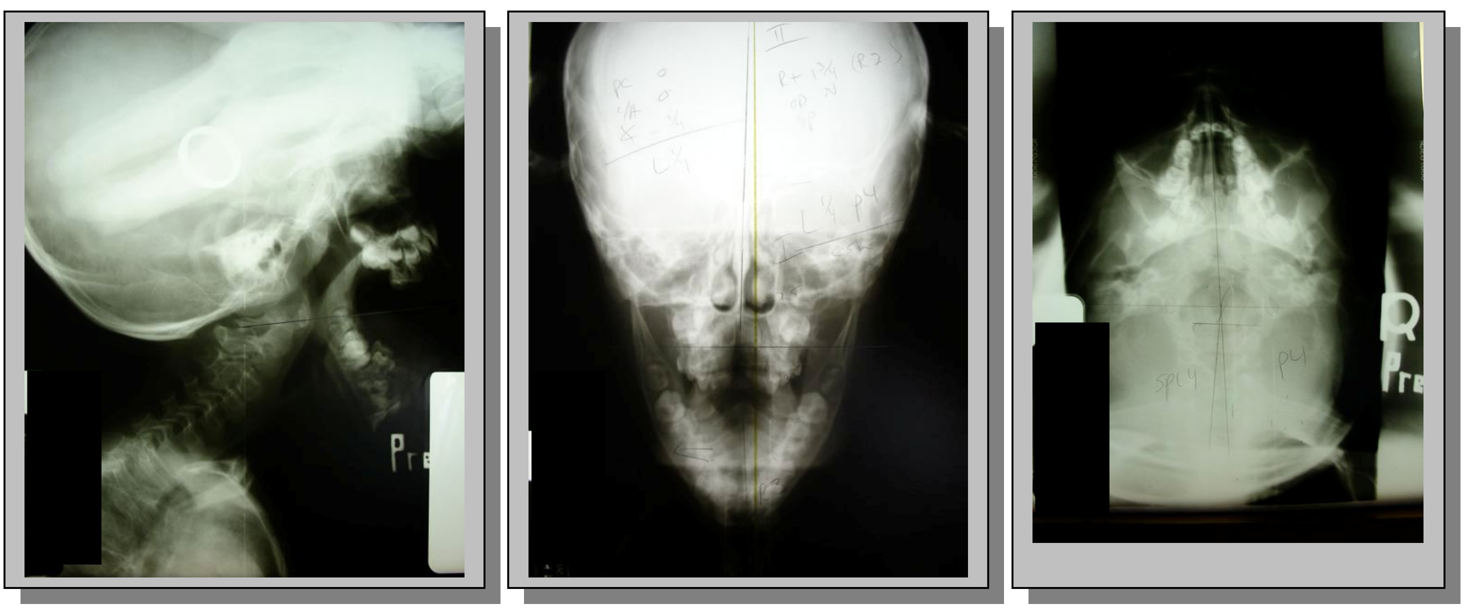


Figure 5: Six months re-examination: post-adjustment radiographic analysis (nasium, vertex)