

Manual Therapies Promote Continence and Mobility in a Patient with Cerebellar Agenesis

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Received date: May 12, 2016; Accepted date: June 06, 2016; Published date: June 13, 2016

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Abstract

Objective: Present single case study of a 26 year old female with congenital agenesis of cerebellum and other congenital malformations who achieved fecal continence and measurable mobility improvements following a prolonged series of craniosacral therapy.

Design: Retrospective review of data based on several interviews with the patient's mother, her primary caregiver and review of the medical record.

Setting: Community based private therapy clinic in the upper Midwest.

Interventions: Craniosacral Therapy and minimal use of other therapies.

Results: Craniosacral Therapy over the course of 4 years contributed to unexpected attainment of fecal continence as well as other areas of functional improvements. The patient was 22 years of age at the time this therapy series began and had life-long neurogenic bowel and bladder dysfunction. Gains were indirectly achieved. Treatment had been initiated to address chronic pain from an older spinal surgery and sessions continued long term for health and wellness. Mobility improvements beyond pre-surgery status and fecal continence were a surprise to all parties involved.

Conclusion: Clinical significance of these outcomes stimulates curiosity as to the actual type of stimulation CST actually provides to the central nervous system and its effect upon neuroplasticity.

Keywords: Agenesis of cerebellum; Fecal incontinence; Functional mobility; Craniosacral therapy; Physical medicine modalities

Purpose

This is a case review of a 26-year-old woman with life-long incontinence who gained bowel control and measurable improvements in mobility after an extended series of Craniosacral Therapy (CST) being the primary treatment modality. Other methods were used in conjunction but minimally. Three points make this case compelling: 1) The on-going participation was requested for chronic pain and wellness, 2) the treating therapist was completely unaware of the continence status throughout the process, and 3) mobility goals far exceeded the target of pre-surgery function. The attainment of fecal continence especially surprised both the patient's family and clinic staff. Pain was quickly reduced and skills of mobility improved gradually. However, once bowel control emerged mobility improvements accelerated beyond expectations.

The purpose of the case report is to stimulate thought around the unexpected outcomes from a method that directly treats the nervous system and wonder about the neuroplasticity elements at play with such intervention.

The Medical History

History was obtained through interviews with the patient's mother. Old medical records had been lost or destroyed and not available for confirmation. During initial intake, the mother did not feel her daughter's incontinence was relevant to seeking treatment, so it was not disclosed.

The mother reported her pregnancy was uneventful though it was considered high risk. Down syndrome had been suspected based upon prenatal ultrasounds though delivery was expected to be routine. Multiple birthing complications arose including meconium aspiration, absent amniotic fluid, and cyanosis of infant with suspected anoxia. APGAR scores were 3 and 8.

Upon birth, there was the notable presence of a sub-occipital encephalocele. As mom described it, "The malformed cerebellum grew outside of the skull and grey matter was present within the encephalocele sac." Surgery to remove the encephalocele occurred within 24 hours, and the skull was reduced and sealed. Other congenital deformities were discovered including the presence of a single kidney and scattered vertebral fusions. The bladder was abnormally small and would eventually fail to grow.

Following three weeks of neonatal intensive care, with a final diagnosis of "agenesis of cerebellum with multiple congenital

anomalies,” the patient was discharged without a prognosis. No genetic syndromes were identified. Infant and childhood development and health were monitored through standard care by a local pediatrician. Her mother stated, “No one ever really knew what was going to happen or what she would be able to do”.

Early Developmental History

As an infant, the patient was able to lift her head, although due to the initial encephalocele repair, full neck range of motion was markedly restricted. Ataxia of the head and neck was expressed early, though she nursed well. Periodic MRI scans consistently revealed the absence of cerebellar tissue.

At age 8 months, the patient was enrolled in local early intervention programming, receiving occupational, physical, and speech therapies on an intensive basis. These services continued until the age of 21 since physical disabilities persisted. Lack of standing balance and intention tremors with severe ataxia interfered with gross motor skill attainment. Upper extremity skills developed to the level of crude reach with grasp and release; proximal stability was not achieved. Cursory ambulation skills did develop using a wheeled walker, but due to absence of balance and postural righting, maximal assistance navigating the walker has been a constant need. Little rotational control of the torso was ever achieved, contributing to a constant risk of falling. Falls had happened many times over the years, but there was no known head trauma or any history of seizures.

Verbal speech failed to develop due to lack of muscular control of shoulders, neck, and oral musculature. The patient did achieve the ability to vocalize with intonations to indicate wants and needs and emotional states. Due to poor motor control with finger pointing and other expressive deficits, the true measure of cognitive function and communication potential had never been fully established. The patient possessed a communication device, but her mother believed the tremors in the arms blocked the motivation to use it. Optic atrophy had been a long-term concern. Testing for ocular motor function had not been reliable.

Over the years, the family sought many interventions and advice to address the various issues. The patient’s growth was significantly impacted by the congenital spinal fusions. At age four, a cervical fusion was performed to address complications of the chronic ataxia and hypotonia. Eventually a full spinal fusion at age of 16 was necessary due to a rapidly distorting scoliosis.

The durable medical equipment the patient utilized included: bilateral ankle-foot orthosis, assisted use of a wheeled walker prior to spinal surgery, and a wheelchair for longer distances. The patient was quite petite in stature, and full body support for walking was commonly given without the use of the walker. There were no records available on the history of physical medicine, though she participated in pediatric therapies through the local school district. The mother was not able to recall what methods or goals were addressed other than, “they were always trying to help with standing and walking and make use of all the equipment available to help her function in the classroom.”

Neurogenic Bowel and Bladder Dysfunction

Pediatric urology managed the neurogenic bowel and bladder dysfunction, (NBBB). Incontinence of both was a life-long issue. Diapers as a child, and Attends undergarment as an adult, were worn

round the clock. Maximal caregiving was required for aspects of toileting care and skin hygiene.

Unsuccessful trials of medication to increase sensory awareness of fullness were conducted. Throughout her school program, therapy and nursing staff worked towards annual toileting goals using behavioral training and biorhythm schedule. A communication switch had been trialed for her to signal urges to void. Pelvic floor stimulation, sensory motor stimulation, allowance for maturation and other physical medicine regimes failed to help the patient gain any degree of continence. Mother described any fecal voiding on the toilet as, “Hit or miss. Once in a blue moon, she’d have a bowel movement on the toilet, but we had no way of knowing if it has been held and released under her control, or if it was lucky timing. There was no pattern of success to build on.” The patient would void feces randomly throughout day and night (Table 1).

1. Lack of body core awareness and lack of standing balance without assistance
2. Low speed of physical reaction times, coordination, and muscle control
3. Sensory registration and feedback
4. Poor communication of asking for assistance
5. Lack of normal developmental signs of ‘readiness’ to gain sphincter control
6. Possibly emotional resignation due to the compounding of these factors

Table 1: Known factors as a result of NBBB for this patient.

Other Factors

Chronic pain at the surgical site remained present ten years following the spinal fusion. Her persistent ataxia, coordination, and balance deficits limited her activity involvement. Her disabilities were primarily related to motor coordination and discreet control, rather than an execution of movements.

The patient lives with her parents and one sibling. She attends a sheltered workshop for adults with developmental disabilities five days a week. The cultural beliefs of the family were described by her mother: “We would never have thought of anything else except loving her as she is. She is a big part of our family. She is not that difficult to take care of and we’ve become accustomed to the routine. Our faith compelled us to raise her as we would any other child and give her the best life we could.”

The Introduction of Craniosacral Therapy

The clinical setting is a private practice in the upper mid-west of the United States that provides various therapy services. Traditional as well as health and wellness interventions are provided. CST is one of several modalities utilized to assist clients towards goals. The patient first came to our clinic at the age of 22, for a series of speech therapy. On the recommendation by this therapist, CST was suggested to address her physical symptoms. Treatment was provided by an Occupational Therapist with 32 years of experience in neurological rehabilitation, with advanced training in Upledger–CST. The patient presented with compounded symptoms (Table 2).

Extreme athetosis of head, neck and shoulder
Maximal assistance required in: standing, all transfers, any ambulation; severe ataxia of gait

Obvious pain and discomfort with body transitions, requiring maximal assistance to transition between prone, supine and sitting
Inability to comfortably recline fully into supine; requires two to three pillows under head to ease pain (assessed through facial expressions of pain)
Less than 25% control of upper extremities; could not utilize arms to support body weight or to balance; no ability to grab handles of wheeled walker and rely on for support
No midline orientation awareness or stability; lack of core strength; no balance correction; leaning heavily to either side
Inability to remain seated in a chair without armrests
Communication and cognitive expressions were rudimentary vocalizations; yes/no questions could be reliably answered with appropriate head movements

Significant keloid scar running the length of the posterior vertebral surface

Table 2: Compounded symptoms presented during initial assessment of patient.

A cursory screening was deemed the most appropriate use of resources since the mother reported providing maximal care for all activities. Minimal investigation into activities of daily life was done. Treatment commenced with goals of mobilizing tissue adhesions and reducing the impact of scarring. Long term goals and expectations from intervention were to reduce pain and return patient to pre-surgery status. CST was the primary treatment modality used to reach individual goals, though other methods were included when specific needs arose (Table 3).

% of time	Treatment Modalities incorporated	References
95%	Basic and Advanced Craniosacral Therapy (Upledger method)	Upledger, 1983, 1990, 1995; Giaquinto-Wahl [3,7,8,9]
2%	Brief use of Proprioceptive Neuromuscular Facilitation	Bass-Haugen and Mathiowetz [10]
1%	Myofascial Release to lengthen connective tissue	Barnes [20] and Duncan [21]
1%	Scar reduction and scar softening technique (Chikly method)	Chikly [15]
1%	Lymphatic drainage to enhance exchange of fluids (Chikly method)	Chikly [15]

Table 3: Distribution of treatment modalities incorporated over the four year time span of treatment

Stages of progress are summarized from daily treatment notes (Table 4). The primary goals were reached within 18 sessions. At that point the patient's mother, interpreting her daughter's communications, requested on-going sessions for the following reason: "It's been the only therapy she's ever attended where she indicates

receiving comfort. She is motivated to come and I know she feels better. Her mood is better." Bimonthly sessions were established and regularly attended. As functional improvements became evident, and then accelerated, continued intervention was appropriate.

Year One
Eighteen, one-hour weekly sessions over the course of six months. Goals that were met:
Freedom from pain with gait, transfers, and transitional movements. (Standing and walking/gait patterns remained essentially unchanged.)
Transfers onto and off treatment table improves from total lift to maximal assistance.
Moderate assistance to transition in and out of sitting and supine.
Softening of adhesions and increased tissue mobility through course of vertebral surgical scar.
Able to recline using two pillows instead of three.
Pre-surgery status reached (pain-free, improved dynamic position changes and mobility)
Year Two
Twenty-six, one-hour sessions; bi-monthly frequency. Additional goals met:
Able to comfortably recline with one pillow instead of two.
Addition of Proprioceptive Neuromuscular Facilitation added 5 minutes at the end of 25% of sessions to increase core stability
Improved dynamic sitting balance on edge of treatment table; therapist can now walk away from client sitting on table edge without patient losing of balance.
Moves in and out of supine position with minimal assistance
Year Three
Twenty-three 45-60 minute sessions; bi-monthly frequency. Goals met:
Ascends and descends treatment table with minimal assistance

Now able to lie in prone position with no pain; assumes full prone position with moderate assistance.
Can transition between prone and supine with extra time and effort, but with minimal assist or less.
Minor progress in simple standing balance while leaning against a counter top.
Year Four
Twenty-four 45 minute sessions; bi-monthly frequency. Goals met:
Ascends treatment table with slight hand-held assistance 50% faster; with greater ease
Moves into supine and/or prone position with no assistance, spotting only for safety; never loses balance sitting at edge of treatment table. She is even requesting which positions to be treated in.
Complete absence of distress with all movements, transitions, and body positions.
Tentative independent cruising around therapy table and around furniture at home, with close stand-by assistance.

Table 4: The client’s stages of subsequent progress summarized from the daily treatment notes.

Discovery of Fecal Continence

In April of 2015, approximately 4 years after intervention started, the client’s mother surprisingly announced, “My daughter has started to be able to go to the bathroom by herself. She is now using the toilet and is continent of her bowels.” During all previous treatment sessions, the patient remained fully clothed and the Attends garments felt like underwear to the therapist as the pelvic region was treated. Occasionally the mother would take her daughter to the bathroom prior to a treatment session, but no assistance from the clinic staff was ever sought. The subject of voiding and the presence of diapers were never points of conversation.

Financial Impacts

Prior to gaining continence, the typical monthly use of adult diapers (Attends) was 200 over a 16 year span. The mother reported that 30-90

diapers used monthly were specific to fecal incontinence. In the year since the fecal continence emerged, improved, and now maintained, the family has reduced their monthly average use of diapers to 100.1% or less of these are fecal related, the majority of diapers now used for urine incontinency.

This translates to a 50% reduction in the amount of diapers purchased with a significant annual cost savings (Table 5). The cost of diaper supplies are an enormous expense and for this family the estimated monthly cost ranged from \$108-130 US dollars. A national study for cost containment of fee-for-service of State Medicaid programs, reported spending a total of \$266 million annually on nine specific types of disposable incontinence supplies for the populace in the U.S. making use of this resource (Wright, 2014). The average cost per unit for the size garment our patient used ranged from \$0.40-0.60 per diaper. The effectiveness of CST reducing the cost of continence care for this patient is self-evident.

Cost allocations of incontinent products	Before	After
Total average monthly diapers used	200	100
Unit cost	\$0.54	\$0.54
Total monthly average cost of incontinence	\$108.00	\$54.00
Total annual average cost of incontinence	\$1,296.00	\$648.00
Monthly diapers for fecal soiling	30-90	≤ 1
Total monthly cost of fecal soiling	\$48.60	\$0.54
Total annual cost of fecal soiling	\$583.20	\$6.48
Annual savings for gaining fecal continence		\$576.72

Table 5: Financial impact of gaining bowel control and evidence that long term treatment reduced cost of incontinence care for this patient.

Emotional Impacts

When asked how her daughter’s independence in fecal continence has affected their lives, the mother responded, “It’s a huge impact. She’s basically independent with her bowel control now, though I still need to provide the hygiene following. It’s obvious that we’re all very happy about it. Today at adult day care she even indicated to her staff she needed to go use the restroom. Even that is a huge development. We are beyond happy and thrilled.” When asked about her daughter’s

improved mobility, the response was, “All the years of therapy never gave her this much success.”

Current Status of Patient

Traditional physical therapy was added after the client started making more spontaneous gains in mobility, but it is important to note this was after the announcement of fecal continence, and this was following four years of CST (primarily). With the physical therapy, the

client gained even greater improvement in dynamic standing balance, gait stride, and all body transitions. The physical assistance now required in mobility tasks has measurably reduced. See (Table 6) for a

summary of current mobility and self-care functional improvements since initiation of treatment 4 years prior.

Notable improvements in function	% of change
Maintains static standing balance without any assistance or arm support for 1-2 mins. Sits one edge of treatment table without side or arm support.	100%
Dynamic standing balance allows for independent cruising of furniture; 25-30 ft	100%
Gait control: midline stability and awareness; core strength has increased; can occasionally walk with only one hand held.	50%
Initiation and motivation to attempt walking alone	50%
Initiation and motivation to attempt walking alone	75%
Change body position from supine/prone and return	100%
Indicating the need to void bowels; holds bowel content during activities and sleep	100%
Indicating the need to void bladder and communicating need to get to bathroom	10%
Increased occurrence of bladder voiding on schedule with staff assistance	5%

Table 6: Summary of current mobility and self-care functional improvements with percentage of change following 4 years of treatment.

Craniosacral Therapy

Cranial therapies date back to the nineteenth century originating in A.T. Still's philosophy of osteopathic medicine [1]. W.G. Sutherland expanded Still's work in the 1920's into what is now known as Cranial Osteopathy [2]. Cranial Osteopathy emphasizes the placement and flexibility of cranial bone and sutures as etiology of dysfunction [2-7]. Upledger was an eventual student of Cranial Osteopathy in the late 1960's. Upledger and his research team went on to discover the existence and function of what is now referred to as the craniosacral system [8-11]. This system is the anatomical system of the meningeal layers and the cerebral spinal fluid, and the physiological phenomenon believed to be a semi-closed hydraulic structure within the confines of the skeletal structures [8,12].

'Craniosacral Therapy' (CST), was created to directly treat this system of fascia and fluids. Though CST embodies the essential concepts of Still and Sutherland, it is frequently confused with, or included with, cranial osteopathy as one in the same. CST has evolved into a more holistic therapy encompassing a full body work aimed at assisting the client to employ their own body's healing capabilities [9,13]. Fluid models of the physiology of the central nervous system as well as studies on the effects of gentle sustained stretch on tissues provide theoretical foundation to justify such manual techniques as CST [13-15]. Increasingly CST is being sought as a preferential treatment as evidenced in a National Institute of Health's decade-long review of the U.S. trends in the personal use of complementary health approaches [16].

The theoretical concepts of CST may be challenging to accept based on long-held anatomy and physiology doctrines. However, the evolving science of structural and energy medicine supports the basic premises of Upledger's methods [4-8,10,13,17,20]. The accepted practice of myofascial release, a common and recognized physical medicine modality, evolved from the fundamental philosophies and trainings of CST [19,20].

CST has yet to be proven through exhaustive empirical methods or quantifiable and imaging methods, though it is credited for treating a multitude of symptoms and disorders [21,22]. Two studies exist that address clinical outcomes of improved continence associated with the clinical use of CST. The first showed improved urine voiding competencies in a group of patients with Multiple Sclerosis following a trial of CST [23]. The second, a recent survey study of use of CST for autism spectrum disorders, showed 69% of the 347 participants' answers reported some level of improvement in "Bodily Functions" which included bowel control, bladder control, and awareness of inside self [24]. Other studies where changes in neurobehavioral function were inferred related to CST being the primary treatment modality include: brain injury [25], chronic pain [26,27], dementia [28], and various pediatric issues [29-31]. CST was also shown to have a favorable effect on autonomic nervous activity in patients with subjective complaints [32], positive effects on health [33], and improved quality of life [34]. These and other studies show some improvements and clinical significance but do not stand up to rigorous empirical standards. The author agrees that a high risk of bias exists in most reported low quality studies about CST. One systematic review of a small body of clinical evidence concurred with this position [35]. Nonetheless, specific effects of outcomes such as this case is noteworthy despite the fact that the best methodologies for researching CST have yet to be used.

Discussion

Agenesis of cerebellum occurring in the general population is rare, but has been described in the literature. Most have a poor prognosis for function [36,37]. In this patient's case, a formal diagnosis of agenesis was given but in reality was the consequence of a surgically excised cerebellar encephalocele. Regardless, this patient grew into adulthood without the presence, or the normal function, of a cerebellum and other parts of the neurological systems did not compensate.

Although the cerebellum accounts for approximately 10% of the brain's volume, it contains over 50% of neurons and modulates sensory processing into adaptive voluntary movements such as posture, balance, coordination, and speech [38]. Given the well-established role of the cerebellum in motor control, life time motor performance deficits appear to be common with agenesis [39]. Behavioral and functional data on living subjects with cerebellar agenesis are scarce [40]. While the term ataxia is primarily used to describe this set of symptoms and there are two major forms of cerebellar ataxia: disturbances of posture or gait or decomposition of movement [39]. Both conditions existed for our client, as did the cerebellar dysfunction of dysmetria, and dysidiadokinesia. These typical features of cerebellar dysfunction were present throughout her life, and the symptoms persist despite the amount of progress reported. Our patient did make measurable gains in voluntary control of posture, balance, and movement skills, as well as the astounding improvement in fecal continence.

Patients with neural tube defects typically present with neurogenic bowel and bladder dysfunction (NBBB) which is defined as recurrent involuntary excretion of feces and voiding of urine at inappropriate times [41]. The presenting manifestations of cerebellar dysfunction can easily be inferred as the etiology of NBBB in our patient. Persons living with toileting disability have increased care costs and caregiving dependency, and the risk of decubiti and infections are but a few factors when considering the impact NBBB has upon patients and their families. The emotional and stress tolls on the caregivers, an increase in symptoms of depression and anxiety, as well as degradation in quality of life for those with NBBB have been well substantiated [42,43].

People with NBBB limitations can, however, attain continence when the appropriate modifications to the traditional routines are made [44]. Behavioral training and operant conditioning remain the best methods for bowel training for people with developmental disabilities [45,46]. This, unfortunately, did not happen for our patient, despite 18 years of intervention with various training methods. It is suggested that self-reporting of incontinence can guide treatment planning [47], but the fecal incontinence (FI) was never brought forth as a concern in this case (at the age of 22) because the FI routine of caregiving had been adapted to by the entire family. Per mother's report, "It was just another task we needed to take care of as she needs maximal assistance in all activities. We thought she'd never use the toilet by herself."

While there is considerable literature on the causes of neurogenic bowel dysfunction, there are few studies that focus on the practical management [48,49]. Several conservative treatment modalities to treat FI include: diet, medications, pelvic floor muscle training, electrical stimulation, biofeedback, transanal irrigation, anal plugs [48-51]. Many of these avenues require minimal baseline cognitive and communication function to maximize success. There are no published studies addressing these types of therapies for adults with developmental disabilities.

It is not possible to draw a wide spread conclusion or recommendation for bowel care for all people with NBBB based upon this one case study.

Conclusion and Implications for Practice

Unexpected outcomes are common observations of Craniosacral Therapy [52,53]. The other interventions during the time of this case review was speech therapy the first two years and seasonal horseback

riding, (which had been a frequent recreational activity for over 20 years). Her speech therapy was terminated due to lack of progress and 2 years before gaining fecal continence. The majority of intervention used during this time that evoked these changes was Craniosacral Therapy.

There has been a fundamental change in our understanding of the capacity of the human nervous system to adapt and extensive research exists on the concepts of neuroplasticity [54]. Neuroplasticity refers to the lifelong capacity of the nervous system to change and rewire itself in response to the stimulation of learning and experiences [55]. Neurogenesis is the ability to create new neurons and connections between neurons throughout a lifetime [38,54,56]. [When practitioners witness changes in their patients, one curiosity is if CST somehow contributes to the phenomenon of neuroplasticity, or perhaps CST offers a unique type of somatosensory stimulation that may contribute to it. If neuroplasticity can now be measured with more sophisticated measuring tools, perhaps CST could gain more plausible investigation into its therapeutic mechanisms.

Very little empirical evidence exists that can guide treatment recommendations for long term continence issues for adults with complex disabilities. As a result many people in the world require extensive caregiving and resources in coping with incontinence associated with impaired mobility. The financial and emotional impact of gaining fecal continence and quantifiable mobility skills has been immeasurable yet easily understood, both for this patient and her family. The two existing studies of the use of CST for continence shows patients with Multiple Sclerosis gained bladder competency and some children with autism spectrum disorders improved bowel and bladder control through the experiences of receiving this treatment.

Many questions arise. Did the improvement in the patient's mobility predispose and stimulate the continence to develop? Could results of continence occur sooner if treated directly in conjunction with specific manual therapies? Could emotional depression and despair have contributed to the patient's continence and mobility status? Could this have simply been a case of under stimulation and the act of therapeutic touch was a variable? When these questions were posed to the patient's mother, she replied, "We tried everything we knew how to do. I mean everything. My daughter now has full control of her bowels, which she never had before. Plus, at the age of 26 she's starting to stand alone and walk around the house by herself. It has to be the craniosacral therapy. You have to believe me when I say we tried everything else; for years."

Implication for practice and research is that an intervention which holds potential to improve incontinence to any degree is worthy of future study, especially interventions that are as low risk and conservative as craniosacral Therapy. Furthermore, any therapy that can assist such measurable mobility skill attainment with cerebellar agenesis is also worth deeper investigation. Some existing clinical evidence of CST has indicated relatively few treatment sessions can be trialed to show initial response, but as this case suggests more time for dynamic changes to self-correct may be worth pursuing. Upledger and others have termed a phrase, "the self-correcting mechanisms of the central nervous system" as a phenomenon that arises from types of treatment such as CST [8,31,55]. Perhaps this opinion corresponds with today's views of neuroplasticity when the nervous system is given the 'right' kind of stimulation. The purpose in presenting this study is to share the anecdotal evidence, though the effectiveness of CST reducing the cost of care for this patient is self-evident.

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This article was originally published in a special issue, entitled: "**Neurodegenerative Diseases**", Edited by Tadayoshi Asaka, Hokkaido University, Japan