Duchenne Muscular Dystrophy—A Single Case Study on the Effects of Structural Integration on an Individual with DMD

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I was tempted to title this article The Reluctant Researcher as this was my initial feeling when Peter and Lauren Brisbane brought their son, MacKinley, to my clinic in March 2006. I knew nothing at all about Duchenne Muscular Dystrophy (DMD). However, they were adamant that I could help MacKinley after hearing positive reports about structural integration from a friend so, as is often the case when our clients are insistent, I was invited to step up to a new learning experience. Initially they put me in contact with several people in the field including Helen Posselt, a world-renowned researcher and specialist paediatric physiotherapist and expert in the field of DMD. Amongst other positions she holds, she advises Montrose Access, an organization formerly known as the Crippled Children’s Society that was formed in Australia in 1932 in response to the global outbreak of poliomyelitis. Helen graciously gave her time and expertise to answer concerns and questions I had around using structural integration as an intervention and maintenance for MacKinley over the past six years. The following article describes a six-year, single case study of the effects of structural integration on an individual with Duchenne Muscular Dystrophy.

Duchenne Muscular Dystrophy

I would like to give a brief overview of this disease that is described by Tecklin (2008) in his book, Paediatric Physical Therapy, as one of the “most prevalent and disabling childhood myopathies” (p. 336). DMD is a virulent and fatal muscle-wasting genetic disease occurring early in life and was first described in 1861 by the 19th century French neurologist, Guillaume (University of Illinois, 2008). It is a genetic disorder transferred from mother to son via a compromised sex-linked X chromosome for the dystrophin gene (American Academy of Neurology, 2005). It is the largest known human gene providing instructions for making the dystrophin protein that, amongst other important functions, is necessary for cellular protection and structural support (NIH, 2010).

Dystrophin is located chiefly in muscles and is used for movement of skeletal and cardiac muscles; however, small amounts of the protein are also present in nerve cells in the brain. When the dystrophin is defective it causes the sarcolemma to tear. Calcium then flows into the skeletal muscle cells. Unable to endure the extra calcium, the cells’ mitochondria rupture. A chain reaction of cell necrosis then spreads from cell to cell through the long bundles of skeletal muscle fibers (Fleischman, 2010).

There are many different versions of dystrophin, some of which are specific to certain cell types; however, to keep things simplified, this article only addresses DMD.

Because of the way the disease is inherited, males are more likely to develop symptoms than females. The sons of females who are carriers of the disease have a 50% chance of having the disease. The daughters each have a 50% chance of being carriers (Barlow-Stewart, 2007). We find Duchenne Muscular Dystrophy occurring in approximately one out of every 3,600 live male infant births and, because this is an inherited disorder, risks often include a family history of DMD (Monckton, Hoskin, & Warren, 1982). However, it still occurs...
in people without a known family history of the condition due to the size of the gene and therefore its susceptibility to defects.

Patients usually become wheelchair-bound by the age of twelve years and die of cardiorespiratory complications in their late teens to early twenties. However, advances in the management of DMD, including treatment with corticosteroids and the use of intermittent, positive-pressure ventilation have provided improvements in function, ambulation, quality of life, and life expectancy (Yui & Kornberg, 2008), which is where manual therapies like structural integration can come in. Novel therapies still aim to provide a cure for this devastating disorder.

**Symptoms**

The symptoms typically appear before age six and may appear as early as infancy. The first noticeable symptom is developmental delay in movement, including sitting and standing independently.

The average age for walking in boys with DMD is around 18 months. There is progressive muscle weakness in the legs and pelvis because of loss of muscle mass which causes the hyper-lordotic waddling gait. Weakness also occurs in the arms, neck, and other areas, but not as severely or as early as in the lower half of the body. Muscle contractures occur in the legs, making the muscles unusable because the muscle fibers shorten and fibrosis occurs in connective tissue. Occasionally, there can also be pain in the calves and the hip flexors shorten. Contractures are especially common in non-ambulatory patients, which is why MacKinley’s case as an ambulatory teenager is so interesting. Typically, patients with DMD develop flexed, abducted, and externally rotated deformities of the hips, which eventually become fixed in position (Hosalkar, 2008).

As symptoms appear there is a steady decline in muscle strength between the ages of six and eleven years. By age ten, braces may be required for walking, and unfortunately by age twelve, most boys are confined to a wheelchair. Bones develop abnormally, causing skeletal deformities of the spine and other areas that frequently contribute to breathing disorders (Yui & Kornberg, 2008) and enlargement of the heart, occurring in almost all cases, beginning in the early teens in some, and in all after the age of 18 (Simonds, 2002).

Intellectual impairment may occur, but it is not inevitable and does not worsen as the disorder progresses. Some examples are: attention deficit hyperactivity disorder (ADHD), autistic-spectrum disorders, learning disorders such as dyslexia, and non-progressive weaknesses in specific cognitive skills (in particular short-term verbal memory) which are believed to be the result of absent or dysfunctional dystrophin in the brain (Poylsky, 2007). Few individuals with DMD live beyond their thirties, with breathing complications and cardiomyopathy being the most common causes of death.

**Management of DMD**

Mortality and quality of life for those with DMD has been improved by the introduction of the corticosteroids Prednisone and Deflazacort, which slow the progressive nature of muscle weakness in boys with DMD, with most specialists agreeing that corticosteroids should be given daily to be most effective (Sussman, 2002). The main benefits of long-term daily use of corticosteroids are improved skeletal muscle, cardiac, and pulmonary functions, and therefore delayed onset of scoliosis and contractures, and preservation of upper extremity function. However it is important to underline that the side-effects of daily corticosteroids on endocrine function such as delayed puberty, short stature, obesity, and osteoporosis are common and significant (Manzur, Kuntzer, Pike, & Swan, 2008). As you will see in the slides, MacKinley has delayed puberty and has remained the size of an eleven-year-old boy for some time now.

Educating and supporting the family around all aspects of DMD and its progression in their child is an important part of the management process (Bothwell et al., 2002). We need to remember that it is not just about the person who has DMD but also about the mental health of the whole family as often family members also suffer stress. Appropriate boundaries need to be maintained so that all family members can get their needs met.

**Case Study**

MacKinley Brisbane (Figure 1) came for treatment at Quantum Health, Brisbane, Figure 1. MacKinley Brisbane, age 9
Australia in March 2006 at the age of nine.

He presented with typical symptoms of DMD, which were first diagnosed as severe when he was as an infant. He was eventually put on daily medication of corticosteroids that brought his symptoms under control so by the time he came to me for treatment he would have looked like a pretty normal nine-year old to the uninitiated eye. However, on testing I found shortness in the lumbar dorsal spine, hamstrings, calves, plantar fasciae, and the IT band. The front line of the quadriceps, in particular the origin, was severely short with the iliopsoas and transversus abdominis lacking appropriate tone. His spine was also starting to show signs of scoliosis. The photographs taken before session one can be seen in Figure 2.

**Method (Treatment)**

Conventional treatment for DMD typically includes physical therapy to stretch and minimize contractures, breathing exercises, and massage to relieve sore muscles. I felt that a more targeted myofascial treatment plan using the ten to twelve structural integration session format, alongside movement re-education and stretching, was in order.

MacKinley first went through an initial, eleven-session structural integration series of bodywork and movement sessions. He responded favorably, which is indicated in the photographs taken afterwards. His movement became more confident and measured and his stance became more grounded, which is reflected in the anterior view up to his shoulder girdle, with an overall leveling out of his thorax (Figure 3). The arrows in the before photograph indicate some of the main tilts, rotations, and compressions that reflect the curvature of the spine. The posterior view, however, is starting to show some compensatory loss of tone in the rhomboids, and you can see the steroid treatment is starting to influence his weight (Figure 4).

Over the next three years I worked in a series of three-session treatments targeting his spine specifically with only a minimum of integration with the rest of the body. The photographs from 2009
show he has regressed in terms of his alignment, which was disappointing. I reverted back to revolving through the complete series of ten to twelve ordered structural integration sessions and by March this year (2012) he shows once again an improvement in alignment. You can see the difference in the photographs of the anterior view (Figure 5) and posterior view (Figure 6) from 2009 and 2011.

It was interesting getting back to the basics of the ordered series of structural integration which highlighted that no part of the body is separate from the other and that any influence is felt and reflected throughout the whole. We further assume that working with an individual in this way has psychophysiological and psychosocial effects that change the self, interaction with others, and ultimately how the individual sees the world around him. In MacKinley’s case we have worked together as he has progressed through his childhood years to his teenage years so, in terms of life transitions, this has been an important journey for him psychologically as much as it has been a physiological one.

Each session included movement awareness lessons that encouraged a sagittal gait pattern. An at-home stretching plan was also designed. Basic movement awareness lessons in foot placement and working with ground reaction force were assisted by heel inserts which helped MacKinley make contact with a solid surface and pitch him slightly forward, which neutralized his tendency to lean back in his structure. Because of the shortened backline, despite extensive myofascial work and stretching, it eventually (by age twelve) became difficult for him to put his heels on the ground, which created eversion of the foot and external rotation at the ankle, in particular on the less-used left foot. You can see he is a little more relaxed through his feet at age 15 because of the support given by the heel inserts (Figures 5 and 6, right). Judith Aston has always asserted that it is better to support what is rather than force a body part to a place it can’t go. This ultimately prevents other parts of the structure from going on hold in an effort to support an unnatural stance (personal communication, Aston Training, 1996).

This was the process we felt would best serve our goal of keeping MacKinley ambulatory. His dedicated parents drove two hours from the Sunshine Coast every two weeks for his treatments except for holidays where at times there were breaks of a month or so.

Results
Eventually MacKinley will, like the rest of his cohort, lose his ability to walk because of muscle contractures, weakness, and eventual scoliosis. However, our theory is that if we can keep him as aligned, flexible, and as strong as possible this in turn will facilitate ambulation, which ultimately will prolong his quality of life.

It is clear that the corticosteroid treatment MacKinley is receiving has stunted his growth significantly—you definitely wouldn’t pick him as being 15 years of age—and unfortunately has contributed to his weight gain. However, the significant payoffs are that he is out of a wheel chair and not suffering the resulting collapse through his torso with all the attending discomfort of...
musculoskeletal pain, not being able to breathe properly, and depression. If a DMD sufferer can somehow keep walking it has the effect of keeping the muscular system engaged and active, and the important associated psychological benefits of being able to fraternize with his or her school friends and feel more normal.

Although surgery, where rods are inserted in the spine to correct scoliosis, is often an option considered, MacKinley and his parents prefer to explore the manual therapy options at this stage. MacKinley had also been receiving chiropractic treatment prior to him starting structural integration sessions; however, the adjustments didn’t seem to hold for long. Once he started structural integration his chiropractic adjustments held for a longer period which his chiropractor commented on, and as the slides show he has continued to remain relatively aligned.

Currently MacKinley remains ambulatory and is the only boy with DMD in his age group in Queensland who is not in a wheelchair, and astoundingly is one of three boys in the whole of Australia in his cohort still walking. Because of the severity of his dystrophin deletions, the fact that he is still walking is significant. My case study was presented informally by Lauren Brisbane at an international medical conference held in Sydney in 2009. This drew the attention of researchers in Perth who have been working on a molecular genetic trial that hopes to restore the reading frame on the dystrophin gene to allow production of sufficient dystrophin to occur. MacKinley has been invited to be involved in this trial, once the program has satisfied the necessary ethical considerations.

Conclusion
I continue to see MacKinley and indeed now his whole family for structural integration sessions and have developed a special relationship with this amazing family who has never given up on the hope that MacKinley will be able to maximize his potential and achieve his dreams however long he is on this earth. They treat their son just like the rest of his siblings, expecting as much from him as they do from the others. This has given MacKinley a “can do” positive attitude which in my view has as much to do with his current state of being as the work that I and the other health care professionals on his team have been doing with him.

MacKinley’s dream as an avid movie goer and on-line gamer is that he hopes to attend university and ultimately gain an IT and graphics degree that will enable him to develop computer games and compose short films.

His vision is inspiring (Figure 7).

References


