NDIS Bill 2012 – Submission to the Senate Inquiry

Introduction

Duchenne Foundation Australia is the only National non-profit focusing on Duchenne muscular dystrophy, which is national in scope. Founded 10 years ago (formerly known as Parent Project Australia), Duchenne Foundation is, to date, a 100% voluntary organization with active directors and members in all states. Our delegations to our 4 jointly organized national conferences always exceed 300 persons as we believe in bringing the world's experts to update Australians in first-world treatment and research every 2 years.

Please visit our website to learn more about our activities. Although the site is currently below par, being reworked in preparation for our 10th anniversary year, this year, we hope to review all pages in the coming weeks. Our mission and objectives, which strive for improved quality and quantity of life for all affected families, are expounded in the "About Us" section. They are snipped below:

Duchenne Foundation's MISSION

Using National and international collaboration to improve the quality and quantity of life for those living with Duchenne and Becker muscular dystrophy and their families, through research, education and advocacy.

DF OBJECTIVES

To work together to improve the lives of Australian persons and families affected by Duchenne and Becker muscular dystrophy.

To focus on the most life-threatening forms of muscular dystrophy, Duchenne & Becker.

To support and promote Australian muscular dystrophy research.

To apply for funding from government, semi-government and private organizations for the purpose of pursuing the Company's objects.

To promote public awareness of Duchenne & Becker muscular dystrophy through media, educational and fund raising campaigns.

To network globally to identify viable research, treatments and quality care standards which will enhance the lives of persons everywhere affected by Duchenne and Becker muscular dystrophy.

To cooperate with like-minded organizations in order to advance research, treatments, equity and quality of life for persons affected by Duchenne and Becker muscular dystrophy.

All directors work for a living or care for sons with Duchenne or both. It is important to explain this fact because this submission cannot be as lengthy or as detailed as we would like – we simply do not have the time available. Yet our board and inner circle of medical advisors including the memberships of national and international neuromuscular and Duchenne organizations possess a deep knowledge of our disorder. Our initiatives lead the way in Australia because we are privileged to have access to state-of-the-art educational information and medical personnel both home and abroad. Thus the breadth of our current educational, research, clinical and awareness raising initiatives, even with a division of labour, mean that we are overstretched.

We do not mean to offend. The fact that we have been able to conference this short submission at all is testament to the very great importance we place on a workable NDIS for Australian families with Duchenne.

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What is Duchenne?

Duchenne muscular dystrophy (DMD) is the most common muscle pathology of childhood. 1 in 3,500 live born males in all races live with Duchenne. It is caused by a mutation of a gene on the X chromosome, which provides the instruction for the production of the protein Dystrophin. This protein is located inside the muscle cell membrane. When it is absent, the muscle cells collapse and die and Duchenne muscular dystrophy develops.

Symptoms of DMD usually appear between ages two and five years and may include difficulty: running, jumping, climbing and rising from the floor; frequent falls, enlarged calf muscles, toe walking and delays in language acquisition. The first muscles affected are those around the hips and upper thigh. Weakness gradually progresses to include all skeletal muscles, the muscles of respiration and the heart.

View The Duchenne Timeline for a 6 min overview of Duchenne. We distribute a wonderful verbal summary of all aspects of Duchenne, created by the world's leading neuropsychologist for Duchenne, Dr Jos Hendriksen, titled, The Psychology of Duchenne.

Duchenne is classified as a neuromuscular disorder but in fact it shares very little genetic similarities with any other disease in the group to which it was assigned in the 19th Century. With the recent genetic revolution in the late 20th century, we learned that Duchenne is not linked in cause and thus potential cure to SMA, LGMD, CM or FSHD etc. In fact Duchenne is more closely related in a genetic sense, to cardiomyopathy than any separate muscle wasting disease. To generalize about any two disorders or forms of disability as having the same requirements medically or socially is to do each complex disorder a very great disservice. This is certainly the case with Duchenne and why we believe that the international multimedia information we disseminate, the opportunities, medical treatments and research for which we advocate and finally the stewardship of our world-wide Duchenne

community gives us the mandate to be the peak body for our disorder in Australia. The existence of the Australian National Duchenne Registry which led to the Australian Neuromuscular Disorders Registry is only one example of our pro-activity because we actually do know how to advance our own separate disease to keep pace with international levels.

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Why do our stakeholders need an NDIS today?

There is a very great burden of care with all severe physical disabilities, we acknowledge. We would like to make some points to demonstrate that the burden of disability on the patient and the carer affected by Duchenne is very much universally great due to a very predictable prognosis. The pyscho-social burden on all family members is without question. The level of support required for all family members is colossal, yet within our national community the disparity of care and support can fall at both extremes and any point along a continuum for persons of any age, but alarmingly and most importantly, this disparity is more keenly felt by persons in the end stage(s) of the disease. This is very concerning indeed as a large number of our stakeholders have little to no assistance with daily tasks and as a result have little hope of reaching their potential in the short and long-term. Unlike Australia, the level of care in most developed countries is both consistent and far in excess of the level provided to a majority of our families.

Factors to consider are:

- Early or late diagnosis both result in extreme distress and guilt
- Late diagnosis, although the most common genetic disorder of childhood, impacts upon the physical wellbeing as early interventions have been missed. Subsequent affected children and carrier children may be born already as a result of late diagnosis.
- Siblings suffer psychologically and also may be relied upon for physical care and extra responsibilities
- Cyclical and anticipatory grief affect families as the degenerative symptoms progress relentlessly
- Loss of ambulation can be anywhere between 8yrs and 14 yrs
- Steroid treatment can mean our boys are very heavy and cause strain
- Slightly built boys may suffer more from recurrent chest infections and be less robust
- By early teens, a boy cannot lift a cup or toilet and bathe unassisted.
- By mid teens a youth cannot shake hands, hug, clean teeth, pick up most objects, raise his hand in class, work an elevator etc. Curvature of the spine can occur and lead to high risk and painful surgery and a lengthy recovery period.
- By early 20's a youth cannot feed himself

- Upper body strength wanes noticeably by mid teens as does heart health
- Respiratory support commences in late teens
- Significant household and transport modifications are expensive
- Gap funding of non-supplied medical equipment and mobility equipment is expensive
- The ongoing acquisition of treatment and research information helps most parents cope and hope but support must be given when families fall victim to misleading non-evidenced based 'miracle cures' such as expensive snake oil treatments. We have qualified social workers on call at our educational events and a recent alliance with counsellors at Carer's Australia
- By adolescence a boy is waking his primary carer many times each night needing to be turned or adjusted due to extreme discomfort
- Most of our boys have normal intelligence and aspire to independent living, however very few have been given the opportunity to experience this

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Further Considerations

There are many issues associated with such a slowly progressive degenerative disease. Because Duchenne is not hereditary in over 2/3rds of cases today, it cannot be prevented and often goes undiagnosed causing delays in early intervention. Eventually a combination of factors lead to diagnosis including: delayed motor milestones, gait abnormalities, difficulty running, climbing, frequent falls, toe-walking, hypertrophy of large muscle groups, and the tell-tale Gower's Manoeuvre. Note that a newborn screening trial is underway at the moment, and the lead investigator is a DF board member for WA - Klair Bayley.

Families vary in their responses, some blocking out the fact of the disability until symptoms cannot be ignored any longer - thus attempting a measure of normalcy for a time. Others are very keen to gather knowledge and begin to optimise their child immediately as a way of dealing with this tragedy for their child at a personal level. Most families fall somewhere in-between these two responses. It is fair to say however, that being relatively free of pain, and able to self-regulate for the most part to avoid tiredness and cramps, that support needed is low to medium at the early ambulatory stage. All boys require stretching and night orthotics (moon boots) to avoid contractures. Boys may require an older child's pram, manual wheelchair and now more commonly mini electric scooter. None of these devices are provided by governments. Children will benefit from extra physiotherapy sessions to support those stretches attempted by parents. Swimming lessons and regular access to a swimming pool from an early age, is not only desirable, but is highly recommended as the preferred exercise because it is non-weight bearing and therefore less likely to contribute to muscle necrosis as all other forms of exercise, for the respiratory benefit and lastly as a way to move freely suspended for mental and physical well-being.

Depending upon other factors such as behaviour and cognitive problems, there may be other early interventions required. Certainly almost mandatory steroid intervention may have various side effects most commonly behavioural ones. The child may also increasingly be

required to be involved in clinical trials of one kind or another which involves not only physical strain but emotional strain for all the family. The brain is a muscle and requires dystrophin and so in most boys there is some cognitive involvement ranging from negligible to autistic features in a small minority. Children with Duchenne are likely to have learning problems and require extra support at school and modifications so that they can keep pace with their peers. These initial difficulties disappear with age in most cases if the learning program is adapted to the student with Duchenne, but at this time when loss of ambulation is eminent, the student may begin to require extra physical support and personal care at school.

In the non-ambulatory stage - late childhood to early adolescence - behavioural problems and extreme weakness and exhaustion are exacerbated as the disease progresses, even when these milestones are delayed by corticosteroids. Weight gain is a problem as muscle dies and is replaced by fat and connective tissue and weight gain is often a side effect of steroid use as is short stature. Diet must be carefully monitored. Body image problems and fatigue mean that a power wheelchair is a welcome relief. Families must buy additional fittings on top of the base chair funding granted through the subsidy scheme. These can amount to many thousands of dollars and are not standard - head rest, pressure relieving cushion, lateral supports, backrest contoured, lumbar support, tilt & space, elevating leg rests, footplates, adductor pads, electronic or regular harness, reclining mechanism and so on, depending upon the model of the wheelchair.

Boys on the cusp of losing ambulation do not require a standing frame or tilt table as in the 19th and 20th century when institutional care was accepted. Today a standing wheelchair costing at least 3 times more than a regular motorized wheelchair, is the accepted mobility device in the first world. No state government in Australia provides a subsidy for these essential mobility devices despite the huge social cost saving; as these devices are known to prevent contractures, scoliosis, respiratory and smooth muscle problems such as continence and digestion issues etc. The benefits for independence and mental health at this very distressing juncture in the disease when one becomes officially and irrevocably confined to a wheelchair, are beyond measure. Once the wheelchair is needed, the family then need to purchase a van suiting their own budget and circumstances, in order to transport their child. Usually a family has begun to consider house modifications e.g. ramps, elevators etc. Pool chairs and beach wheelchairs to be involved in outdoor activities with one's family can cost as much as $6,000.

Throughout adolescence upper body strength declines and small tasks involving the arms are taken away - picking up objects, scratching, eating, toileting, showering, working an elevator, turning the pages of a book, turning in bed , transfers, setting up computers, etc. Parents can wake usually between 2 - 10 times a night even with the supply of pressure relieving mattresses and wheelchair cushions (government subsidized) and electric beds. For most parents the cost of a turning bed is impossible. Transfers are so difficult because unlike most disabilities, these men cannot weight-bear. Other items of equipment are required - shower chair, pool chair, hoist. Whilst these items are generally covered by the subsidy scheme, the life-saving cough assist to bring a child back from pneumonia is not covered. The cost of hospitalization for these boys and young men is then covered by Medicare - a strain on already overstretched government hospitals. Untreated boys have a 90% chance of needing spinal fusion but the risk to children who have had steroid treatment is much less. This is a long and difficult procedure needing to be done before respiratory strength is so compromised the procedure is very risky. All surgeries are risky due to the

contraindications reported when Succinylcholine and vapour anaesthetics are used, and have resulted in deaths from the anaesthetic.

Many require treatment for the certain loss of bone density as the risk is increased with steroid use, but all youths will be monitored regularly and treated for respiratory insufficiencies (sleep study and nocturnal ventilation) and dilated cardiomyopathy (ace inhibitors, beta blockers and VAT). Nutritional problems and smooth muscle problems are rife due to an inability to stretch out and to move. Certainly regular monitoring of all these factors takes place whether treatment has commenced or not.

All of these usual symptoms, and a child may have other underlying conditions too, require a daunting amount of maintenance and care giving on the part of parents, so adolescence is certainly a time when our families require as much in-home support as they can to be able to cope and to share the burden. Few families have access to night-turning services but many do not. Primary care-givers are always exhausted due to months and years of broken sleep. Respite, in-home and outside the home is welcome. Certainly regular breaks from personal care which is so physically demanding is welcome. Whilst fathers are improving and offering assistance, the mother is historically the primary carer who curtails her career to fulfil the caring role. With the physical demands over time, mothers are at a very high risk of physical, mental and emotional strains and ailments.

All family members, including siblings are at great risk of anxiety, depression etc. Youths with Duchenne almost certainly have OCD and can be anxious and depressed. The primary carer faced with at least two decades of anticipatory grief and guilt is almost certain to have recurrent mental stress on top of incredible physical, financial and social stress.

In the end stages of Duchenne - late adolescence to adulthood - the burden of care is one of compounded drudgery but also involves more equipment and medical knowledge. Non-invasive diurnal ventilation requires a second portable ventilator and a back-up ambubag. Whereas a cough assist was vital for chest infections before, we don't have any IPPV machines in Australia to save a life. Whilst cardiac meds are a boon in preserving heart function, we need access to other invasive cardiac procedures such as used in America and Europe. Sip-puff mechanisms for using devices such as chairs and computers are necessary as even the small use of the fingers throughout adolescence is gone. Jouse joy sticks(mouth operated controllers) are needed to work on a computer or activate environmental controls, and are all very expensive. Yet they prolong independence and help with the caregiver's burden of constantly completing every action for their son, moving scratching, opening, turning things on and off - every simple movement we take for granted.

Eating and swallowing difficulty means that the formerly obese body shape becomes emaciated and prone to chest infections and other complications. Nutritional supplements and peg feeding follows.

Society sees a youth with cushingoid round face from steroids, sitting in a power chair but they have no conception of the labour and devices it takes to get him to that stable point, in order, if the technology is available, for him to sit, operate a computer or wheelchair and to

breathe, eat and have some semblance of inner health, hygiene and self-actualization. The burden of care is enormous and heartbreaking. Added to this universal adversity of both the PWD and the caregiver is the enormous disparity between any two families in exactly the same familial and social circumstances. The arbitrary nature of support given between states, within states, within suburbs, means that most adult persons with advanced Duchenne can have as little as 5 hours a week of personal care and thus little to no physical respite for the primary care giver; whilst a selection of young adults can be fully self-actualized and living independently through government benefits with 24/7 care and accommodation due to either block funding or being awarded personal package of care.

The latter situation is desirable but available to so few. There are no logical differences, not in the amount of advocacy or social contribution of the primary carer, nor in ages, functional level or level of education or ability for self-determination and involvement in the community. Our anticipatory grief for our children is worsened by a feeling of being marginalized and dismissed without understanding why?

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What would a post NDIS future look like for men with Duchenne?

Duchenne Foundation, in our 10 year history, has engaged in political and medical advocacy by epetitioning, letter and national campaigns on our own initiative and for disabilities in general, government inquiries instituted by the NHMRC and the Productivity Commission, public appeals at the community level, etc. But we are 100% voluntary and have beyond offering advice and support with some equipment provision, have not had a qualified workforce to engage in personal advocacy so that our members can attempt any equity or self-determination. Organizations with large workforces are powerless to help the majority of families long-term from their own budgets or by appeals for public funding. How can we possibly hope to address each person's unmet need long-term, despite our unique and specialized knowledge, resources and international networks? One in every 3,500 males in Australia has Duchenne. Our children can succumb at any time usually anywhere between the ages of 16 and 36 with average longevity calculated to be early 20's. The majority of young men and their families are crying out for physical assistance and the opportunity for independent living before they succumb.

To give our families equitable support across geographic boundaries, city and country, the NDIS must be sustainable. There must be a guaranteed funding stream as in a levy, since the burden of care for our population is costly, although our proactive interventions and research can reduce overall social cost of the disease considerably over the person's lifespan. We are a national group, part of an international network of Duchenne organizations and we feel that the NDIS must be truly equitable and National...not just in the future, which has thus far no guaranteed funding stream, but today. It is a cruel irony that Duchenne takes away independence in almost direct alignment to the stages where a young man's (and rare woman's) peers are gaining independence as young adults. NDIS trials must help all our youths with Duchenne desperately seeking to be self-actualized and if they wish, enable them to live an independent lifestyle in the limited time they have - free from the ongoing guilt that their condition is an unbearable burden on the people they love most.

Our youths with Duchenne in the vast majority of cases have a very high level of unmet need. They do not have the time for the architects of the NDIS to test out a "NDIS methodology" on many persons whose needs are adequately met in certain arbitrary and thus discriminatory geographical zones. Our children are so severely disabled yet unsupported in the main, because of their "invisibility" due to a lack of awareness of the most common and life-limiting genetic disorder of childhood. They should have the wherewithal to survive to see their current adversity relieved and the burden of their families at least in part, alleviated.

On behalf of all our families affected by Duchenne, and all PWD who suffer with comparatively little assistance provided; we implore the government of Australia to focus on each person with a disability who is suffering today rather than on the "system" of administering that support. With PWD at the centre of the strategy we have no doubt that an immutable and sustainable funding stream must follow, to sustain adequate support throughout each person's life. We ask that we focus on keeping family strong, so that they can support and care for their sons with the access to equipment and services that will be equitable across this great nation.

We thank you for your time,

The Board

Duchenne Foundation

PO Box A83, Australind WA 6233

Ph 1300 70 60 89

info@duchennefoundation.org.au

www.duchennefoundation.org.au

Resources:

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