Living with Duchenne & Becker in Australia: Supporting Families waiting for a Cure

Save Our Sons Duchenne Foundation, the peak body representing the Duchenne and Becker Muscular Dystrophy community in Australia, commissioned the McKell Institute and Equity Economics to produce a keynote report highlighting the main issues and challenges facing families of children and adults with Duchenne and Becker Muscular Dystrophy in Australia as they wait for a cure.

The report helps to better understand how Duchenne and Becker impacts families, how we can ensure that new treatments benefit Australian's sooner, and to better support those with the conditions today.

The report contains twelve recommendations for action pertaining to:

- Getting Treatment Earlier
- Getting the Care that is Needed
- Funding Future Therapies
- Improving Access to Clinical Trials

Duchenne is an X-linked genetic condition that affects 1 in 3500 boys and in very rare cases 1 in 50 million girls. It is the most common and most severe form of muscular dystrophy, with a similar milder form of the condition called Becker muscular dystrophy.

Duchenne affects every muscle through multi-systems in the body including the pelvic girdle, legs, arms, neck, hands, through to the smooth muscle affecting swallowing, digestion and elimination. The heart, lungs and diaphragm are also muscles, and the deterioration of these organs leads to progressive and inevitable respiratory and heart failure.

A boy diagnosed with Duchenne will follow a devastating trajectory of muscle loss. They will lose the precious milestones they gained as all their muscles fail them. They learn to walk, run and jump and slowly lose these abilities, most commonly requiring a mobility scooter to keep up with friends from ages 5 to 10, progressing to an electric wheelchair full-time from age 10 to 15, at which time their upper limbs become weaker leading to limited-to-no function in the arms in the late teens to early 20's, a growing dependency of respiratory support and failing cardiac function.

There is no cure or effective treatments currently available in Australia to change the natural trajectory of Duchenne. Life expectancy is in the mid-20's with young, affected Australians passing away as young as 10 years-of-age from cardiac and respiratory failure.

The key findings of the report include:

- A "cost-of-disease" study undertaken for this report shows that Duchenne in particular is associated with significant lifetime health and social care costs. It is estimated that these can total up to $2.25 Million for a child living until their mid-thirties. In addition, informal care costs total up to $630,000 in terms of reduced female participation in the workforce. On average, the financial cost of Duchenne over the lifetime of a child born today can be expected to be $1.3 Million with the cost for a child living to their mid-thirties of $2.88 Million.

- A remarkable 16.6 per cent of respondents to a survey of families completed for the report stated that the $22 Billion NDIS had worsened their situation, and a further 31.1 per cent said that the scheme had neither improved nor worsened their situation – an astonishing 47.7% in total. Delays in receiving equipment was a key issue raised by those indicating that the NDIS had worsened their situation.

- Families reported high out-of-pocket medical costs, ranging to $1800 per month. Out-of-pocket costs were much higher in NSW than in other States and Territories. Out-of-pocket costs in NSW were $430.43 per month on average, compared to $250 per month on average across the other States and Territories

- 20 per cent of families had a delay of more than 3 years between first noticing symptoms and receiving a formal diagnosis. The average age of receiving a formal diagnosis of Duchenne or Becker was 4.39 years of age, meaning that more than one child could be born into a family before the first child with Duchenne or Becker is formally diagnosed. The average delay in diagnosis was 1.09 years, with variation across State and Territories. The longest delay was in Tasmania at 3.8 years. Of interest there was a notable difference between the two largest States, with the average delay to diagnosis in NSW being 1.54 years versus an average delay in Victoria of 0.59 years.

- A high proportion of parents, being 23.5 per cent, have more than one child with Duchenne or Becker, with the majority having two children with Duchenne or Becker.

- Families consulted, on average, three professionals before they received a formal diagnosis, with 30.4 per cent seeing four or more health professionals to get a diagnosis. Most families indicated that either there was insufficient information and/or support provided around the diagnosis.

- Families see an average of 9.65 health professionals over the course of a year, thus coordination of care is critical. However only 39.4 per cent of people reported having someone help with their care coordination. Of those the majority were seen by Neuromuscular nurses which are currently funded by the Save Our Sons Duchenne Foundation.

- There are ongoing issues with the approval of clinical trials in Australia that are undermining efforts to ensure Australian children have access to the next phase of clinical trials for new gene therapies. While national reforms are underway, a review of international practice highlights that they do not go far enough in streamlining and centralising approval processes, nor do they address issues specific to gene therapies. More can be achieved to ensure that Australian children with Duchenne or Becker have the opportunity to participate in these clinical trials.

- A review of regulatory regimes in the United Kingdom, Europe and Canada found that Australia is the only jurisdiction that requires licensing of genetically modified organisms for use in clinical trials or approval by a separate gene technology regulator. This adds to approval times and hinders the ability of Australian children to gain access to important clinical trials.

- There are currently no trials of the new generation of gene therapies being undertaken in Australia, and there are concerns that Australian children will miss out on the next phase of trials due to be undertaken in 2020.

- Issues of fragmentation and inefficiency in Australia's clinical trial processes remain. In addition, for clinical trials of gene therapy for Duchenne, Australia's GMO licence approval process is far lengthier than any of the jurisdictions studied and is likely to be a major impediment to selection of Australia as a site for gene therapy trials. As stated, Australia is the only jurisdiction that requires licensing of GMOs for use in clinical trials or approval by a separate gene technology regulator.

Save Our Sons Duchenne Foundation will continue to strive to be the best possible advocates for the Duchenne and Becker community in Australia and will leave no stone unturned in our mission to enhance the quality of life for all with Duchenne and Becker and our quest to secure a cure for the conditions. We will continue to turn our ever-present and strong hope into meaningful outcomes and maximum positive impact for all whom we represent.

Please visit our website (www.saveoursons.org.au) to read a PDF version of the report, or contact us via phone on (02) 9554 6111 or e-mail at info@saveoursons.org.au and request a hard copy of the report be sent to you.

Your support is much appreciated. Thank you from the Save Our Sons team and Board.