

Muscular Dystrophy

Senator McLUCAS (Queensland) (1.25 pm)

I rise today to talk about muscular dystrophy and the effect it has on those who have been diagnosed and their families. Neuromuscular disorders are not terribly prevalent in our country. There are 60 separate disorders which have in common the progressive and irreversible wasting of muscle tissue.

They include muscular dystrophies, spinal muscular dystrophies, motor neurone diseases and peripheral neuropathies. One in 1,000 people are affected—that is, 30,000 children and adults in Australia have been diagnosed.

Today I want to specifically focus on Duchenne muscular dystrophy. Duchenne muscular dystrophy, DMD, and Becker muscular dystrophy, BMD, are progressive, degenerative muscle diseases affecting one in about 3,300 people, mainly boys. Until recently, there were no treatments available to halt the disease process or to restore muscle function.

DMD is caused by alterations in the dystrophin gene that encodes for a protein essential for muscle integrity. Advances in gene technology, though, have facilitated potential treatment options to overcome the altered gene and to produce sufficient protein to restore the integrity of the muscle cells and improve muscle function.

On average, boys lose the ability to walk by the time they are 10, have respiratory insufficiency in their teenage years and most have cardiac involvement from an early age to varying degrees. Their lifespan is shortened to their late teens or early 20s without specialist quality care. For the many boys and their parents, the diagnosis of DMD and BMD is devastating. The happy, healthy and adored little boy stops crawling, is listless and does not progress.

Concerned parents seek an answer and, unfortunately, mainly due to the rarity of the disease, diagnosis is often delayed. When diagnosis is made it usually results in the loss of employment, usually of the mums, and the juggling of myriad doctor, physiotherapist and other medical specialist visits. For those who live outside the major capital cities, this necessitates regular trips to the city, with the inherent dislocation, concern about accommodation and travel costs—large costs are associated with the regular trips—and then the effect on the family that is left at home. The financial burden on a family with a son diagnosed with DMD is estimated at \$126,000 every year.

I am indebted to members of Muscular Dystrophy Australia, MDA, Parent Project Australia, the Duchenne Foundation—and those names are sometimes interchanged—and local constituents of mine, including Ms Deb Robins, for informing me about DMD and sharing their life experiences following their diagnosis or the diagnosis of their sons. I also acknowledge Mr Laurie Stroud, who was a strong advocate and ambassador for MDA. I also know there are many other organisations right across the country that are supporting families who have been diagnosed with muscular dystrophy or Duchenne muscular dystrophy.

Parent Project Australia, which is also known as the Duchenne Foundation, is a voluntary organisation advocating for patients and families affected by DMD and BMD. They recently initiated a nationwide campaign to seek support for expanded gene sequencing of those children with DMD, who are usually boys, and to select appropriate patients to enter specific gene therapy clinical trials. Further, they are seeking government funding for the development of a national database of DMD patients that would then feed into the global database managed in Europe. I commend the former Chief Medical Officer, Professor John Horvath AO, for listening to the request and taking up their case with the relevant parties.

The creation of a national registry for DMD has been referred to the Clinical, Technical and Ethical Principles Committee of the Australian Health Ministers Advisory Council for consideration and further discussion.

I am advised that the committee is still considering the proposal. Parents tell me that this is the best way forward, that it is not 'expensive' in the scheme of things and that it will give them real hope that their sons will get the best medical support that is currently available. I urge the committee to look

favourably on this proposal and consider the value that such a register would have for these children.

Why do we want a register? For each person enrolled in the DMD registry, the most important data are the precise type and location of the particular mutation, as potential therapies for DMD are generally mutation specific. For example, the clinical trials currently proposed will use specific therapies based on whether the disease is caused by, firstly, the deletion of one or more exons in the gene sequence, which accounts for about 60 per cent of cases of DMD and 80 per cent of BMD cases; secondly, large DNA duplications and complex arrangements as occur in between five and eight per cent of cases; or, thirdly, point mutations, including premature stop codons which occur in an estimated 30 per cent of DMD cases and about 15 per cent of cases of BMD. The establishment of the register is supported naturally, by the families of children with DMD and BMD, but it is important to note that it is supported by their treating medical personnel and by the MD research community.

There are other ways that we can assist families having to work with this particular diagnosis. As mentioned earlier, after diagnosis families are confronted with managing a huge list of medical appointments, with many and various practitioners—cardiologists, physiotherapists, dieticians, geneticists, respiratory clinicians and social services. They are invariably in different locations and sometimes the order in which one sees them is important. It a crazy juggle which is stressful for families and stressful for the little boys who are being shunted round.

One solution has been found at the Royal Children’s Hospital in Melbourne. MDA has employed a neuromuscular coordinator whose role is to arrange to have development specialists available and present at the one location at the one time. The MDA November 2009 journal states:

“Parents and their children have the benefit of coming to the clinic and seeing all their specialists within the one visit, which drastically increases the emotional wellbeing of the children.”

I am sure it increases the emotional wellbeing of the parent, usually the mother, who is having to manage this list of appointments. It is a solution that I am sure could be replicated in other centres in other states. It is a simple, cheap and clearly effective solution. The presence of such a coordinator surely reduces costs not only for families but also for the health systems that are supporting those families.

I move on to another solution. There are a range of support options available at varying levels of suitability right across the country. But, rather than talk about what is not available, I would like to take this opportunity to highlight one service that is very much valued by the muscular dystrophy community. CampMDA has been operating since 1989 and has held 76 camps since its inception. It has evolved since then into a quarterly activity conducted at different locations throughout urban, regional and rural Victoria. CampMDA offers activities that are planned and resourced to reflect the needs, wants and abilities of the participants, catering for both young and not so young people. The youngest camper to date has been six years old, and the oldest 76.

MDA has value-added the experience for those with muscular dystrophy by using the opportunity to train undergraduate students who are undertaking studies in physiotherapy, nursing, occupational therapy and disability studies. As we all know, the curriculum for these courses is invariably packed with all sorts of material that a student has to know and understand. This often means that a focus on a disease such as muscular dystrophy is limited, and graduates leave their training with only a small degree of understanding of the condition.

So MDA is funding—over \$1 million to date—the placement of these students at the camps in order to improve their understanding of the range of types of muscular dystrophy and the range of responses that are appropriate. Over the years more than 2,000 students have attended, and reports indicate that the experience is positive for them in that they are gaining knowledge that is hard to access in a university or hospital placement.

The more understanding that the medical profession has of these disorders, the better the rate of diagnosis will be and the better the treatment provided will surely be.

I turn to research. In the late 1980s, MDA made a deliberate decision to work towards the establishment of a dedicated research facility to accelerate the rate of research into muscular dystrophy. In 1993, the Melbourne Neuromuscular Research Institute was established. Its work is well regarded both here in Australia and internationally. I am pleased to report that the National Health and Medical Research Council is currently funding research into Duchenne muscular dystrophy, with a grant of \$800,000. I also take this opportunity to commend the Duchenne Foundation for the work it has undertaken to bring together a conference to be held at the University of Sydney on 26 and 27 February—later this week.

The people who work so hard are often the parents and grandparents of children who have been diagnosed with DMD. Sometimes they feel very alone. Sometimes they feel as if they are not supported. I am impressed by the quality of the work that they do.

They have been rigorous, they have been unemotional when surely they could be afforded the latitude of being quite emotional about their circumstances and they have been professional. I want to pay tribute to Deb Robins in particular. She is a wonderful advocate. A woman whose son has DMD, she is a woman whom I have enormous respect for. Clearly, diagnosis of one's child with DMD or BMD is devastating for those families.

But, to quote MDA, there is hope as there is a growing number of treatments that will bring greater quality of life for these young boys and men. The potential for a register is giving families hope that Australia will be able to join the international community in seeking the right treatment for their child. Surely coordinated care is an option that other jurisdictions can consider, being a practical, cheap, simple and effective way of supporting families through the maze of medical appointments that they have to attend.

There is support for training of the medical professions so that diagnosis is quicker and treatments are best practice. There is assistance for families through respite and also the opportunity to share their experience and provide mutual support.

As I said, I commend the many groups and individuals across our country who are supporting families with children with DMD and BMD.

I thank them for all the work they are doing.

Jan McLucas

Senator for Queensland