Improving the reading skills of young people with Duchenne muscular dystrophy in preparation for adulthood

J Hoskin and A Fawcett

Abstract:

Duchenne muscular dystrophy (DMD) is a progressive genetic condition that affects both muscle and brain. Children with DMD are at risk of psycho-social difficulties such as poor academic achievement and behavioural and socio-emotional problems. This article by Janet Hoskin and Angela Fawcett, both from the University of Swansea, describes how 34 participants with DMD took part in a 36-week online literacy intervention which was delivered in partnership between home and school. The key objective was to improve reading skill. Participants were re-tested at 36 weeks for single word and text level reading, comprehension, fluency, processing and timed single word reading. Pre and post results indicated that children who followed the intervention for 36 weeks made significant improvement in their single word reading ($p = <0.0001$), timed single word reading ($p = <0.0001$) and text level reading ($p = <0.004$). They also made significant improvement in their fluency and comprehension scores. The results showed that children with DMD and related literacy difficulties benefit from a regular, structured and systematic synthetic phonics programme. With young people with DMD increasingly living into adulthood, early literacy intervention is particularly important to ensure optimum career and training opportunities.

Duchenne muscular dystrophy (DMD) is a chronic and progressive muscle wasting condition, caused by a fault on the dystrophin gene, one of the largest genes in the human genome (Emery & Muntoni 2003). It is an X-chromosome linked disease and therefore overwhelmingly affects boys, one in 3,600 – 6,000 male births (Drousiotou,
Although aspects of the condition can vary between which acts as a stabiliser in the muscle cells. Most children are diagnosed at around the age of five years (Bushby, Hill and Steel 1999) and without any intervention muscle deterioration results in lack of ambulation before they are teenagers with respiratory, cardiac and orthopaedic complications developing as they grow older. Without any treatment, the average age of death is around 19 years (Bushby et al 2010).

However, more recently with the use of interventions over the past twenty years such as daily steroids, night ventilation, cardiac monitoring and spinal surgery to address scoliosis, life expectancy has increased to an average of 27 years (Eagle et al 2002; Eagle et al 2007). Since the publication of Eagle’s research in 2007, there are now teenagers with DMD who have been using daily steroids from a much earlier age and so it might be expected that with these treatments available, young people with DMD could be living into their third decade and beyond. In Denmark, where those with DMD use steroids and ventilation in an optimal co-ordinated care programme, there are now more adults than children with the condition, and some are living into their third and fourth decades (Jeppeson et al 2003).

As well as in muscle cells, it has been suggested that the protein dystrophin is present in isoforms in the brain, and therefore it could be expected that some cognitive difficulties may arise if the protein dystrophin is missing (Muntoni, Torelli & Ferlini 2003). Some research has suggested links between genotype and phenotype, and more difficulties appear to arise where the mutation on the dystrophin gene is distal, that is, after exon33 (Bushby et al 1995; Muntoni, Torelli & Ferlini 2003; Bardoni et al 2000).
There are indeed established learning and behaviour risks associated with DMD, and the prevailing literature reports a verbal cognitive deficit, and suggested verbal developmental delay (Cotton, Voudouris & Greenwood 2001). Short term verbal memory has been reported as problematic in several studies with young people with DMD in comparison with matched controls on immediate recall of numbers sentences or stories (Hinton et al 2000; Hinton et al 2004; Billiard et al 1998) and these problems have been shown to exist across the range of intellectual ability and are not linked to more general impairments in language and memory (Hinton et al 2000; Hinton et al 2004). It has been established that the reading skills of young people with DMD appear to be compromised compared to unaffected controls and other neuromuscular conditions. (Billiard et al 1998; Billiard et al 1992; Hendriksen and Vles 2006). Hinton suggests that verbal memory span is the core cognitive deficit in DMD, thus explaining problems in learning to read, as verbal memory difficulties are known to be important in phonological manipulation and the development of early reading skills, and are associated with dyslexia and poor reading ability in the non-DMD population (Hendriksen & Vles 2006; Billiard et al 1998; Gathercole & Baddley 1990; Snowling 2000).

Nicolson and Fawcett have developed Ulmann’s Procedural Learning Difficulties model to give a ‘neural systems’ explanation for dyslexia (Nicolson & Fawcett 2007; Ulmann 2004). They suggest that due to an impaired procedural system that connects areas of the brain such as the prefrontal language system, the basal ganglia, parietal and cerebellar structures, young people with dyslexia struggle to develop skills associated with procedural learning, that is, those skills that are learned implicitly or unconsciously and enable fluency in a particular skill. However their declarative system, which depends on the medial temporal lobe structures such as the hippocampus and is linked to conscious learning and knowledge-gathering, remains intact, and often compensates for procedural weaknesses and lack of
automaticity. This may be a useful model for DMD, where procedural skills such as learning to speak and read often appear problematic or delayed (Cyrulnik et al 2007, Hendriksen & Vles 2006), whereas skills in factual understanding and abstract thinking appear relatively intact (Hinton et al 2000). In addition, neuro-imaging in both human and mouse models indicate that dystrophin is usually most abundant in the cerebellum and that as it is absent in patients with Duchenne muscular dystrophy this will cause a difference in brain structure and function (Lidov et al 1990; Cyrulnik and Hinton 2008). It is important for teachers to note that as with dyslexia, the brains of young people with DMD may work in a different way, but with appropriate intervention they can learn to read, drawing on a range of compensating skills.

As recommended in the Duchenne Standards of Care, and in guidelines for reading remediation, be it for those with dyslexia or any other reading difficulty, intervention should be as early as possible (Bushby et al 2009; Rose Report 2009; Torgeson 2001; Torgeson 2006). Limitations such as illness, absence from school, poor expectations of parents or schools can often impact on the academic aspirations of this group of young people who have life-limiting conditions, and it has been noted that the short life expectancy of young people with DMD can influence teachers, parents and the children themselves: ‘Often education is seen as a preparation for adult life, and for DMD children the whole process sometimes may seem rather meaningless.’ (Dubowitz & Leibowitz 1981). In Denmark, even though life expectancy for those with DMD is better than it has ever been, young men report regret at their lack of formal qualifications which can limit meaningful career choices (Rahbek et al 2005). It is established that difficulty learning to read can often prevent young people from making the first steps in their education towards external examinations and so for young people with Duchenne it is extremely important that now, as a range of medical interventions
are enabling them to live longer, that their learning needs are met in the same way as any other child who is struggling to read.

This project looks at the impact of a 36 week structured and systematic online literacy intervention for young people with DMD between the ages of 5 years and 12 years 2 months.

**Methods and Materials**

After gaining ethical approval from the Centre for Child Research University of Swansea, 147 letters were sent out to families through Action Duchenne’s DMD Registry. Sixty-two young people with Duchenne Muscular dystrophy underwent a series of tests for underlying verbal ability, and a variety of reading tests that are listed below. From the original sixty-two, forty seven children were identified as possible candidates for an online literacy intervention, due to standardised scores in reading below 90, or from information from schools on poor skills development. Out of the forty seven, thirty four children completed the online 36- week Decipha programme, and it is the results from these children’s tests of difference that we are presenting in this paper. As DMD is rare, and so the group is made up of children between the ages of 5 years and 12 year 2 months.

The baseline assessment battery included British Picture Vocabulary Scale BPVS II (Dunn et al 1987) which was used as a measure of verbal underlying ability. This is a test of receptive language skills, which, unlike other IQ tests, does not ask the participant to orally explain the meaning of a word or depend on memory or arithmetic skills, an important consideration given reported memory deficits in these areas in DMD.
Participants were assessed for single word reading using the Wide Range Assessment Test for Single Word Reading (WRAT 4 Wilkinson & Robertson 2006). This involves both decoding and word recognition skills, measuring the phonetic codes needed to learn reading skills and does not involve any comprehension. The one minute reading test from the Dyslexia Screening test – Junior (DST –J 6.5-11.5 years) was used to test timed single word reading for those children between the ages of 6 years 5 months and eleven years 5 months (Fawcett & Nicolson 2004). The Diagnostic Reading Assessment was used to assess text level reading for those participants over the age of 7 years (Crumpler & McCarthy 2007). This gives standardized scores for accuracy, fluency/reading rate, processing speed and comprehension. It also gives age equivalents. All tests giving a standardized score are based on a mean of 100 and a standard deviation of 15. Three non-normed tests were designed by the author to test basic phonological awareness, blending of phonemes and segmenting of phonemes (Hoskin 2008). For all Phonics tests, raw scores are presented.

Table 1 Mean standardised scores for BPVS, single word reading and reading ages at baseline

The intervention took each child one academic year to complete between 2009 - 2010. A control group is usually advocated for intervention research, but due to low incidence of DMD, and the constraints of real world research, this was not possible. Initially, children were matched for age and IQ and divided into two groups in order to cross lag the intervention and provide a control. However, due to several children having poor health and dropping out of the initial group, and the need for some of the older children to begin
intervention before starting secondary school it was not considered possible to continue with this design. Therefore through the use of pre and post standardised scores for single word reading and text level reading tests, each child acted as their own control which allowed age difference between tests to be taken into account, and gave normed scores showing how each child attained in comparison to his unaffected age group. All tests took part in quiet areas in the participants’ schools and were administered in English.

**Intervention : The Decipha Reading Programme**

The Decipha Programme was designed by the author especially for this project and is a 36 week online intervention delivered over one academic year excluding school holidays. It is based on two tenets: firstly, it is a structured synthetic phonics programme following recommendations from both the Rose Report on the Teaching of Early Reading, and that regarding the teaching of Children with Literacy Problems and Dyslexia (Rose 2006; Rose 2009). Children follow a systematic synthetic phonics programme which is delivered in short bursts little and often, in this case four sessions of twenty minutes per week. One session is delivered at home by parents. Secondly, Hinton’s study into verbal memory deficits established a shared learning profile in DMD, regardless of IQ, showing strengths in factual information and abstract thinking, and weaknesses in tests depending on intact memory skills (Hinton 2000). In addition, the author’s baseline assessments showed a similar profile where assessments that tested factual information and knowledge in the BPVS were a shared strength whereas tasks demanding intact working memory skills, such as single word and phonic tests, a shared weakness. The Decipha Programme is thus designed to play to children’s strengths in facts and knowledge whilst developing their phonological and memory weaknesses. In addition, as this group of children have established risks of ADHD, ASD and
OCD (Hendriksen & Vles 2008), and heightened behavioural and emotional difficulties (Darke et al 2006), the issue of engagement was a key consideration in the design of the intervention. Through the intervention, children became ‘Time Agents’ travelling through time and space in their personal online area, learning about different time periods, for example, meeting the Ancient Egyptians 5,000 years ago, hiding from dinosaurs 200 million years ago, in other words playing to their factual strengths, but at the same time following a structured synthetic phonics programme in order to improve memory, decoding and fluency skills. Activities were differentiated according to phonic ability so that children across the age and ability range were able to access the programme.

The programme was delivered in four sessions of twenty minutes per week, one being delivered by parents at home and three sessions delivered by a learning support assistant at school. Parents and learning support assistants were trained together at school by the author or a specialist support worker before beginning the intervention. Each session contains two structured phonic activities and one activity based in the historical context. Participants had their own password-protected dashboards where each day’s plans and resources were stored ensuring that as little preparation as possible was needed for each day. Once a week, parents and school were required to ‘blog’ each other on the child’s progress so that all parties, including the author, were aware of difficulties that the child may be experiencing or areas of achievement.

After looking at the whole group performance pre and post 36 weeks, participants were split into age groups in order to explore the optimum age for following this intervention.
Results:

Tests of difference were conducted on the pre and post test measures to identify where scores had significantly changed. Because the data was not normally distributed, a series of non-parametric Wilcoxon tests were undertaken. In accordance with literature on literacy intervention studies (Fawcett et al 2001) two factor anovas were also conducted to investigate optimum age of following the intervention. This allows effect of scores at pre-test to be examined in relation to post-test and any interactions between group and time to be examined.

Results showed significant improvement in all areas of reading tested at 36 weeks: single word reading standardised scores, text reading standardised scores, timed reading, reading fluency, reading comprehension and processing. In addition all non-standardised phonics scores improved significantly. All results can be seen in Tables 2 and 3.

Table 2 Wilcoxon non-parametric results for pre and post 36 weeks single word, text level and timed reading, fluency, processing and phonics tests

In order to investigate the effects of age on the intervention with regard to literacy scores, a further series of 2 factor anovas were undertaken with the factors of age and pre/post intervention. Participants were divided into four groups which corresponded roughly with divisions made at school: Group 1: Infants (aged 5 years – 7 years 5 months); Group 2: Lower Junior (aged 7 years 6 months – 8 years 4 months); Group 3: Mid Junior (aged 8 years 5 months – 9 years 8 months) and finally Group 4: late Juniors and early secondary
(aged 9 years 9 – 12 years 2 months). Table 3 shows the results for single word reading in age groups with effect sizes, and Figure 1 shows the single word reading scores pre and post 36 weeks intervention for each age group.

Table 3 Standardised single word reading scores in age groups pre and post 36 weeks and effect sizes*

*according to Cohen’s Criteria. Slavin & Fashiola suggest that an effect size of 0.25 is considered to have educational significance (Slavin & Fashiola 1998)

The results for the WRAT4 standardised scores show no significant effect of group at pre-test 
[F(3) = 0.12; p = .95,ns] but a significant effect of the intervention[F(1) = 25.83; p <.0001], and no significance in the interaction between group and intervention [F(3) = 1.07; p = .38, ns. This indicates that for single word reading, the intervention had a significant impact on all groups but that the difference between groups was not significant before the intervention and did not significantly change after it.

Figure 1 WRAT4 single word reading mean standardized scores pre and post 36 weeks in age groups

Discussion
From a 36 week intervention that was spread over a school year, the intervention group as a whole showed an average single word reading age increase of 21 months, from 6 years 7 months to 8 years 4 months, and a mean text level reading score improvement of 30 months, from 8 years 2 months to 10 years 8 months. This compares with Thomson’s estimate that children with reading problems, without specially designed interventions, progress by an average of 5 months in reading age over one year (Thomson 1984; 1991).

These results therefore confirm the benefits of following a structured literacy programme over an extended period for young people with Duchenne who experience reading problems. Both single word reading and text level reading standardised scores showed significant improvement over 36 weeks. As standardised scores automatically take age into account, even a stationary standardised score value indicates that a child is maintaining normal progress. A significant standardised score increase is therefore encouraging. It can be seen that, over 36 weeks, the group as a whole improved its mean standardised single word reading score from 81.4 to 88, and its mean text level reading standardised score from 85.5 to 100.3. Three participants did not improve their standardised score in either single word or text reading, two of these had other learning and behaviour issues, one of which was a diagnosis of autism.

As reading is more than simply decoding words, it is heartening to see significant improvement not just in decoding and accuracy but also in comprehension and fluency.

With regard to improvement of standardised reading scores and age, children in the early Juniors, whose baseline age ranged from 7 years 6 months to 8 years 4 months benefited most from the intervention, increasing their mean standardised single word scores from 80.89 to 91. This translates as an improvement from a reading age of 6 years 6 months to 8 years 6 months. The mean chronological age of this group at baseline was 7 years 8 months.
implying that at baseline their mean reading age was 12 months below their chronological age. However, post intervention, after an academic school year, their mean age was 8 years 8 months and so their reading age was only two months below their chronological age.

Children with DMD have been shown to be at risk of delay in speech and other developmental milestones (Cyrulnik 2007), and as it would be expected that most non-reading disabled children would learn the underlying skills for reading aged between the ages of 5 to 7 years whilst at Infants, perhaps it is not surprising that the early Junior DMD group improved the most. In addition, by the age of 7 years 6 months, these children would have benefited from at least one to two years of Phonics in a school in the UK, and this may be an indicator that children with DMD, like those with dyslexia, benefit from opportunities to ‘over-learn’ and consolidate processes such as reading and spelling. As Torgesen writes of children at risk of dyslexia:

‘If at-risk children do not receive more teaching/learning opportunities per day than other children, they will acquire reading skills more slowly ...they thus require more repetition in order to solidly establish critical word reading and comprehension skills.’ (Torgeson 2001 p 16)

Similarly, for the Infants group, their baseline mean age was 5 years 9 months and yet their mean reading age was 4 years 2 months. Post intervention their mean chronological age 6 years 9 months and their reading age was 6 years 2 months. Programmes such as the Decipha programme used in this intervention would be part of the ‘Wave 3’ in the National Strategy in the U.K., which is aimed at children with more complex neurologically based difficulties, who have consistently failed to learn to read fluently (Brooks 2006). The introduction of synthetic phonics being taught systematically at foundation level would perhaps have helped those in the younger age group and it is noted from the results that this
group began at a higher level than all of the other groups, thus perhaps having already benefited from systematic phonics instructions in nursery and reception classes.

Less improvement in standardised scores and reading age was shown in the older two groups. This may reflect difficulties associated with the progression of the disease in these particular age groups, for example beginning to lose ambulation for some children and thus increased social isolation as they fail to keep up with friends physically. However, the importance of even small increases cannot be over-estimated in a group of young people who may for the first time be engaging with reading which will serve to reduce their isolation as they grow older, as well as to add to their general quality of life.

It has also been long established that early intervention is best and that learning literacy skills when older is more challenging and difficult to improve scores and this has been explained by some early researchers (e.g. Vernon 1971) to be due to motivational factors in that children who are older have experienced more failure and frustration. In addition, a combination of less effective phonics instruction in the UK before 2006, and lack of knowledge in schools with regard to DMD may account for their lower scores. A very low starting point in reading often makes it difficult to improve, due to the need for some kind of fluency to make sense of the text which can more easily be gained once a child has a basic ‘sight vocabulary’ (Ehri, 1995; Stuart & Coltheart, 1988). This is particularly true for dyslexic learners who can often expend energy decoding words letter by letter and lose the meaning and consequently the motivation (Fawcett 2002). Stanovich has written about the ‘Matthew Effect’ in reading, based on the parable of the talents in Matthew’s Gospel, suggesting that those children who read less well than their peers, get less exposure to reading compared to unaffected children and so consequently the division between them grows (Stanovich 1986). In the same way that the servant in the parable who hides his talents has them taken away and the one who uses them has them increased in abundance, the child
with reading difficulties reads less and less and thus has very little exposure to new words or vocabulary in comparison to the unaffected child who reads more and more thus improving his ability or ‘talents’. This can result in depressed verbal IQ, as new words and ideas are not accessed through print. It can also lead to exclusion from mainstream classrooms as textbooks for various subjects become inaccessible, and in some cases of DMD a move from mainstream school to specialist provision (Billard, Gillet & Signoret 1992). We know that without intervention dyslexic children generally decline steadily in literacy relative to their peers (Singleton 2009).

The possible impact of these results should not be ignored for young people with DMD. Schools may be reticent to intervene with young people with a diagnosis of a life-limiting condition in the same way that they would with a student with dyslexia or ADHD, but evidence clearly shows that intervention as set out in the Rose Report will benefit children learning or struggling to read, in that it is a regular synthetic phonic approach (Rose 2006; Rose 2009).

Another important aspect of these results is in expectations and aspirations for the young people. The mother of a young participant who had a diagnosis of Autism described the intervention as a ‘revelation’ as she and school had never anticipated that the young person would ever learn to read. She added that it was ‘as if a light went on’, as he was able for the first time to identify letters and corresponding sounds rather than simply see ‘squiggles’ on the page. We know that parental expectations play a great role in the academic achievement of young people (Jeynes 2005), and if parents and schools are not preparing young people for adulthood due perhaps to lack of information about the disease at diagnosis then this will have a negative impact on the young person’s success at school. The use of various treatments mentioned earlier such as night time ventilation and steroids, has transformed the
life expectancy for DMD (Eagle et al 2002; Eagle et al 2007). This is not only relevant to young people with Duchenne, as strides in medical advances now mean that there are double the previously estimated children with life limiting or life threatening conditions living in the UK with a rise from 25 to 32 per 10,000 in 10 years (Fraser et al 2012).

The International Standards of Care for DMD recommend psycho-social difficulties should be treated with ‘the same effective and evidence-based interventions that are used in the general population, with a strong emphasis on prevention and early intervention....’ (Bushby et al 2010 p 89). It is essential that those working with young people with DMD are aware of the learning risks that can be present and that they understand that addressing these problems early will be most helpful to gain qualifications necessary for career opportunities and reduced social isolation. Hospitals, schools, families and the young people themselves should therefore be planning for adulthood, ensuring that the opportunities to learn skills necessary for a full and independent adult life are made available.

A potential limitation of the current study is lack of a control group due to real world constraints. However, the vast majority of tests included in this study report standardised scores which gives a participant’s score in relation to unaffected peers of the same age, and take into account the time that the intervention has taken. In addition, intervention strategies were based on recent Government advice and current research in the UK into initial and remedial reading as well as being based on new and reported information about the strengths and difficulties of young people with DMD (Rose 2006 and 2009). It was not considered ethical to expect young people struggling in literacy to follow intervention strategies that have been shown to be less effective in order to test the placebo effect, particularly in a condition such as DMD where energy levels are limited and young people are often taken
away from the classroom for hospital appointments, physiotherapy or occupational therapy. In addition observation was also given to the need for engagement due to high levels of reported emotional and behavioural risks (Darke et al 2006; Hendriksen & Vles 2008).

This is the first reading intervention that has been reported for young people with DMD it is important to show the improvement that can occur in literacy skills of young people with a complex and chronic condition. If further research confirms the feasibility and effectiveness of intervention with this group and others with life threatening conditions, it could have the potential to transform the school experience of these children.

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Table 1

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<th>Mean baseline BPVS standardised score n=34 (sd)</th>
<th>Mean baseline WRAT 4 standardised score n=34 (sd)</th>
<th>Mean baseline reading age n=34 (sd)</th>
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Table 2

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Table 3

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<tr>
<td>Late junior/secondary</td>
<td>9</td>
<td>82.1 (15.8)</td>
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<td>E.S</td>
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