**REVIEW**

**A review of nutrition in Duchenne muscular dystrophy**

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**Keywords**

Duchenne, evidence-based dietary treatment, muscular dystrophy, nutrition therapy, paediatric dietetics.

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**Abstract**

Duchenne muscular dystrophy (DMD) is a recessive X linked genetic disorder characterised by progressive muscle weakness and reduced muscle tone. Affecting only boys, it limits life expectancy to approximately 20 years. A literature review was conducted using MEDLINE and the Cochrane Library, employing the term ‘Duchenne muscular dystrophy’. A total of 1491 articles in English were recovered. These papers were searched thematically under the headings: body composition \((n = 10)\), energy expenditure \((n = 10)\), nutrition \((n = 6)\), corticosteroid therapy \((n = 55)\) and gene therapy \((n = 199)\). Key dietetic practice points were identified relevant to nutritional management. Papers supporting these key themes were assigned a level of evidence and grade of recommendation. There is limited high-quality evidence to guide the nutritional management of boys with DMD. Currently, the majority of evidence is based on expert opinion and clinical expertise. Delayed growth, short stature, muscle wasting and increased fat mass are characteristics of DMD and impact on nutritional status and energy requirements. The early introduction of steroids has altered the natural history of the disease, but can exacerbate weight gain in a population already susceptible to obesity. Prior to commencing steroids, anticipatory guidance for weight management should be provided. Malnutrition is a feature of end stage disease requiring a multidisciplinary approach, such as texture modification and supplemental feeding. Micronutrient requirements are yet to be determined but, as a result of corticosteroid treatment, vitamin D and calcium should be supplemented. Some evidence exists supporting supplementation with creatine monohydrate to improve muscle strength. More research is needed to provide a higher quality of evidence for dietitians working within this area.

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**Background**

Duchenne muscular dystrophy (DMD) is a recessive X linked genetic disorder affecting approximately one in 3500 live male births. In DMD, the body is unable to make the protein dystrophin as a result of a large variety of mutations/deletions of the dystrophin gene, found on chromosome Xp21 (Strober, 2000). Dystrophin is essential for muscle contraction because it binds actin to the membrane bound dystroglycan complex in smooth, cardiac and skeletal muscle. The absence of dystrophin expression leads to progressive muscle weakness, with chronic degeneration of muscle and replacement of muscle with fat and endomysial fibrosis. DMD follows a predictable clinical course because the deficiency of dystrophin and inflammatory response causes muscle inflammation, necrosis and fibrosis. Characteristic progressive muscle wasting becomes noticeable in early childhood and, by their early teens, boys have become wheelchair bound. Premature death in their early twenties usually results from failure of the respiratory or cardiac musculature.

Boys with DMD present in early childhood with delayed walking, increased falls, decreased activity, difficulty climbing stairs or getting up off the floor. Unique identifiers of DMD include ‘Gower’s sign’, whereby a boy...
will use his hands to ‘walk himself up’, and pseudohyper trophy of the calf muscles, which is indicative of fibrous infiltration of the muscle. Diagnosis can be confirmed by elevated serum creatine kinase, muscle biopsy or genetic analysis. A range of other complicating features can occur, such as learning disabilities and gastrointestinal symptoms (see Supporting Information, Table S1). Medical management of the muscular dystrophies has included the use of corticosteroids; however, their precise mode of action is unclear, as is the optimal time to commence treatment. The standard initial therapeutic dose is 0.75 mg kg⁻¹ day⁻¹ for prednisone and 0.9 mg kg⁻¹ day⁻¹ for deflazacort (Moxley et al., 2005) and this is now accepted as best practice. Boys treated with steroids have improved functional capacity (Yilmaz et al., 2004), less scoliosis (Balaban et al., 2005) and better pulmonary function than those who are not treated with steroids (Pradhan et al., 2006; Daftary et al., 2007). Side effects such as excessive weight gain, cushingoid features, hirsutism, glucose intolerance, cataracts, behavioural changes and delayed puberty (Sorva & Turpeinen, 1994) can be of sufficient concern to cause their withdrawal (Balaban et al., 2005; Strober, 2006).

Daily physiotherapy combined with multidisciplinary treatment aims to reduce complications such as the development of contractures and scoliosis.

The body composition changes observed in DMD are unique, and hence nutritional management is complex. Boys with DMD can move between the spectrum of over to under nutrition within their shortened lifespan. Nutritional issues have taken a backseat in the past; however, as the life expectancy slowly inches forward with improvements in medical management, such concerns warrant immediate attention. There is no consensus available on the nutritional management, and so dietitians are forced to tread lightly through a potential minefield of issues. Here, a summary of the literature is conducted in order to provide some much needed clinical guidance for dietitians.

Methods

A literature search was conducted in MEDLINE (Ovid, from January 1950 to March 2008) and the Cochrane Library using the term ‘Duchenne muscular dystrophy’. Nutritional textbooks or chapters on management of neuromuscular disease (NMD) were examined for content and references. A total of 1690 articles were recovered, and only articles in English were included in this review (n = 1491). These papers were searched thematically under the headings: body composition (n = 10), energy expenditure (n = 10), nutrition (n = 6), corticosteroid therapy (n = 55) and gene therapy (n = 199). Reference

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**Box 1**

**Nutrition requirements**

- Monitor weight to guide energy prescription (Grade D)
- Ensure adequate intake of micronutrients as per dietary reference values (Grade D)
- Supplement Vitamin D (1000 IU daily) and Calcium (750 mg daily), especially in those receiving steroid therapy (Grade D)
- Monitor serum 25-hydroxyvitamin D (Grade D)

**Nutrition assessment and monitoring**

**Short stature is a characteristic of boys with DMD (Grade B)**

- Measure height and weight six monthly and plot on standard growth charts (Grade D)
- Upper arm length, tibial length, or knee height can be measured in the advanced stage of disease (Grade D)
- Body composition is characterised by a decreased lean body mass and increased intramuscular fat mass (Grade C)
- BMI as a screen for obesity is not accurate in boys with DMD (Grade C)
- Various tools can be used to measure body composition with DXA and MRI being accurate, appropriate and noninvasive measurement instruments (Grade C)

**Management**

- Prevent excess weight gain by providing anticipatory guidance around energy balance prior to commencement of steroid therapy (Grade D)
- Advise reduction in energy intake with caution. Follow up with regular measurement of fat free mass to monitor progress where available (Grade D)
- Dietary texture modifications may be required to accommodate eating difficulties (Grade D)
- Support patients and families in decisions regarding enteral feeding and/or gastrostomy tube placement (Grade D)
- A multidisciplinary team should be involved in the management of feeding difficulties (Grade D)

**Nutriceuticals**

- There is a limited evidence base to support the use of nutriceuticals in boys with DMD (Grade D)
- Short- and medium-term creatine monohydrate treatment improves muscle strength in people with NMD (Cochrane)
lists were examined for papers that were not identified by the search strategy. Where systematic or Cochrane reviews were available, these were utilised in preference to individual papers.

Key themes were identified from all relevant literature. Practice points were agreed between the authors as the most helpful to practitioners, and papers relevant to these key themes were assigned a level of evidence and grade of recommendation using the Oxford Centre for Evidence-based Medicine criteria (Phillips et al., 1998). These points are summarised in Box 1 with the grade of recommendation, with a scale of A being the strongest through to D being the lowest quality of evidence (see Supporting Information, Table S2). A summary of the search process is provided in Fig 1.

**Nutritional requirements**

**Energy requirements**

It is unclear whether decreased energy expenditure, either from reduced voluntary activity, or a primary defect in energy metabolism are contributing factors to obesity in DMD. Studies within this area are limited with contradictory findings. The six studies published (Gonzalez-Bermejo et al., 2005; Zanardi et al., 2003; Satomura et al., 2001; Hankard et al., 1996a,b, Okada et al., 1992, Berardinelli et al., 2001) have limitations: small sample sizes and a large age range. Only three of these studies examined energy requirements in younger children and it is unclear whether the participants were receiving steroid therapy. Energy requirements become complicated as the DMD boys become older. In particular, resting energy expenditure (REE) can be affected when gas exchange is impaired (Bodamer et al., 1997), secondary to a decrease in respiratory muscle strength. The literature is inconclusive with respect to energy requirements in the advanced stages of disease. Okada et al. (1992) measured basal metabolic rate (BMR) in 310 individuals with DMD aged 11–29 years. DMD subjects had an increased BMR compared to controls, and BMR increased with age. Gonzalez-Bermejo et al. (2005) measured REE in 20 males aged 20–33 years on mechanical ventilation. REE was significantly lower in DMD participants compared to controls. The difference in REE findings could be the result of the presence of ventilation support in the latter study. Gonzalez-Bermejo et al. (2005) also found that measured REE was 22% lower than the theoretical REE calculated using the Harris Benedict equation. Dietitians need to make an individualised ‘best guess’ to establish energy requirements using the equation: total energy expenditure = REE (measured if possible) × physical activity level (Department of Health, 1991). Individual longitudinal monitoring of body weight will provide a crude indication on the accuracy of energy prescription (Almond et al., 2007).

**Micronutrient requirements**

There has been little investigation into the micronutrient status and requirements of boys with DMD. In the advent of corticosteroid therapy, the micronutrients that require immediate attention are vitamin D and calcium. Canalis et al. (2004) describes how pharmacological doses of oral glucocorticosteroids can suppress bone formation and increase bone resorption, thereby leading to osteoporosis. The use of steroid therapy combined with decreased mobility suggests that boys with DMD are at an increased risk of fractures and poor bone health (Söderpalm et al.,

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**Figure 1** Literature search method.
Serum 25-hydroxyvitamin D \( [25(\text{OH}) \text{ D}] \) was examined in 24 boys with DMD aged 2.3–19.7 years, with the majority being treated with prednisolone, and in 24 controls (Söderpalm et al., 2007). Significantly lower levels of \( 25(\text{OH}) \text{ D} \) were found in the DMD group \((21 \pm 6 \mu \text{g} \text{L}^{-1})\) compared to controls \((30 \pm 8 \mu \text{g} \text{L}^{-1})\), as well as a lower bone mineral density and decreased bone turnover. Interestingly, food records revealed an apparent adequate intake of dietary calcium and vitamin D, implying either that interactions are increased or there is another mechanism that predisposes the boys with DMD to vitamin D insufficiency. Vitamin D metabolism is an area which would benefit from future research. Biggar et al. (2006) recommends daily calcium (750 mg) and vitamin D (1000 IU) supplementation in conjunction with corticosteroid therapy.

**Nutritional assessment and monitoring**

### Height

Short stature in boys with DMD has been documented but its aetiology is unclear. Eiholzer et al. (1988), Nagel et al. (1999) and Willig et al. (1993) determined mean the \( z \)-scores for height for their populations as \(-1.5, -1.0 \) and \(-0.936 \), respectively. McDonald et al. (1995) demonstrated that reduced stature remains a feature into adulthood. Biggar et al. (2006) demonstrated that boys treated with deflazacort were significantly shorter than boys who were not treated at age 10, 15 and 18 years. Interactions of steroids and growth require long-term follow up.

Although standing height is the preferable measurement, accurate measurements of height become difficult to obtain on approaching adolescence because of an inability to stand combined with the development of scoliosis and/or contractures. Stewart et al. (2006) suggested measuring upper arm length, tibial length or knee height in these circumstances.

### Weight

In steroid naive boys, weight is disproportionately distributed throughout the stages of DMD. McDonald et al. (1995) reported that 44% of boys aged 9–13 years were above the 90th percentile in weight, whereas, after age 17 years, 65% weighed less than the 10th percentile. Similar findings were presented by Willig et al. (1993). Of the 252 boys studied, the prevalence of obesity, defined as having a weight above the 90th percentile, reached 54% by age 13 years. Undernutrition (i.e. a weight below the 10th percentile) occurred after age 14 years, involving 54% of the boys by age 18 years. The interaction between weight and steroid treatment is significant, and requires longitudinal follow up.

**Body composition**

Body composition is characterised by a decreased lean body mass and increased intramuscular fat mass (Leroy-Willig et al., 1997). Therefore, standard anthropometric measures (i.e. skinfold thickness) are not accurate (Leroy-Willig et al., 1997; Pichiecchio et al., 2002; Mok et al., 2006a). Various body composition measures have been documented, including dual energy X-ray absorptiometry (DXA) (Palmieri et al., 1996; Douvillez et al., 2005; Söderpalm et al., 2007), bioelectrical impedance (Hankard et al., 1996a; Gonzalez-Bermejo et al., 2005; McDonald et al., 2005; Mok et al., 2006a), total body potassium (Griggs et al., 1983; Edmonds et al., 1985), magnetic resonance imaging (MRI) (Huang et al., 1994; Leroy-Willig et al., 1997; Gong et al., 2000; Berardinelli et al., 2001; Pichiecchio et al., 2002; Zanardi et al., 2003) and urinary creatinine excretion (Griggs et al., 1983; Edmonds et al., 1985; Hankard et al., 1996a; Franciotta et al., 2003). Because different methodologies and outcome measures were utilised in these studies, a meta-analysis of the outcome data was not possible. However, the studies demonstrate that DXA and MRI are accurate and noninvasive measures that would be appropriate for monitoring nutritional status in a paediatric population.

Classifying boys with DMD as being overweight or underweight by using height and weight or body mass index (BMI) without accurate body composition measures is problematic. Pessolano et al. (2003) cautioned against using BMI for the evaluation of nutritional status in boys with DMD. The researchers compared the number of boys classified as being overweight using BMI \((n = 5)\) with the number of boys classified as being overweight using the percentage of expected weight for zero muscle mass \((n = 30)\).

Stewart et al. (2006) suggest that height and weight should be measured at a minimum of every 6 months and plotted on growth charts. Standard growth charts, such as the 'UK 90' or the 'CDC 2000', can be used in clinical practice to monitor longitudinal growth and development in NMD (Almond et al., 2007). In 1988, a specific DMD weight chart was created (Griffiths and Edwards). This chart is a theoretical model that accounts for weight growth in the presence of muscle wasting. When comparing this chart with the CDC 2000 chart, the percentiles sit lower, taking into account the loss of muscle mass.

**Management**

### Obesity

Excess weight gain occurs in the early stages of the disease, with a dramatic inflexion in the observed weight
Obesity contributes to the progression of the disease by exerting extra force on already weak muscle groups, essentially decreasing mobility. In an examination of body composition of 13 males aged 9–13 years with DMD, Hankard et al. (1996a) split the group into obese DMD and nonobese DMD, sensu Griffiths & Edwards (1988). Their analysis showed that there was no difference in muscle mass between obese and nonobese participants, suggesting that obesity should be considered only as a factor increasing the handicap. Obesity also has implications for increased respiratory involvement and poorer psychosocial development, and can make activities of daily living and care at home more difficult.

Excess weight gain is exacerbated in boys treated with steroids because increased appetite contributes to an overconsumption of energy (Desilva et al., 1987) and the most frequently reported side effect (Angelini, 2007). Obesity is a major concern for parents/caregivers. A systematic review conducted by Moxley et al. (2005) determined that weight gain occurred over a prednisone dose range of 0.3–1.5 mg kg$^{-1}$day$^{-1}$. In those individuals treated for 6 months, 75–80% of patients receiving 0.75 mg kg$^{-1}$day$^{-1}$ prednisone increased their weight by ≥10% compared to 20–24% of those receiving placebo. Despite this weight gain, patients showed clinical improvement. Interestingly, the distribution of the weight gained, in relation to muscle and fat, differed between patients receiving prednisone compared to placebo. At the completion of the 18-month prednisone trial, a 36% increase in muscle mass in the 0.75 mg kg$^{-1}$day$^{-1}$ prednisone group compared to placebo-treated patients was demonstrated using urinary creatinine excretion. These findings challenge health professionals to re-consider the definition of ‘excessive weight gain’ and to consider the need for measurement of body composition as a routine part of nutrition assessment and monitoring.

There is debate about whether the type of steroid affects the severity of induced side effects. Bonifati et al. (2000) and Balaban et al. (2005) found that deflazacort was successful in reducing the amount of weight gained compared to prednisone.

Standard dietetic practice for weight management is to manipulate the energy balance equation by decreasing the intake of food energy, and by increasing activity to increase total energy expenditure. This theory of ‘eat less, do more’ should not be applied to boys with DMD. Although obesity raises considerable parental concern, caution is needed when inducing a negative energy balance because this may increase the loss of lean body mass, which does not have the potential to regenerate. In adults on various energy restricted diets, it has been shown that weight loss comprises approximately 70% fat mass and 30% lean mass (Truby et al., 2006). There is only one relatively old case study of successful weight loss in a boy with DMD and another with Becker’s muscular dystrophy (Edwards et al., 1984). These case studies demonstrated that weight loss was possible and led to an increase in mobility and self esteem without deleterious effects on total body nitrogen (i.e. a surrogate measure of lean mass). However, the authors cautioned that weight loss should be carefully monitored because of the potential loss of lean tissue that usually results during periods of negative energy balance. To achieve this weight loss in the DMD participant, energy was restricted to 2633 kJ day$^{-1}$ (54 g protein, 30 g fat, 40 g carbohydrate) from January to December 1981, raising questions about micronutrient adequacy and patient satisfaction.

Although estimating energy requirements and macronutrient composition are uncharted territory, so too is the issue of activity prescription. There is a risk of increased muscle breakdown from overuse versus increased muscle weakness from disuse (McDonald, 2002). Grange & Call (2007) discuss how muscle weakness may be attenuated by regular, low intensity exercise; however, there is a critical lack of data to support exercise prescription. The authors recommend a systematic analysis of muscle function to determine potential exercise thresholds. In 2005, Carter et al. (2005) reported anecdotal evidence of weight loss in two boys with DMD using the anti-epileptic drug, Topiramate (Topamax; Janssen-Cilag, Titusville, NJ, USA). The patients lost 26% and 41% of their initial body weight. The authors concluded that further study investigating the use of pharmaceutical weight loss agents is warranted.

The most constructive advice in the management of overweight boys with DMD is given by Griffiths & Edwards (1988): prevention of excess weight gain is preferable to severe restriction in the already obese. The modern manifestation of this advice would be to provide anticipatory guidance to boys commencing corticosteroids, and to follow up with regular monitoring of growth, in particular with respect to weight gain. However, in the already obese child, the dietitian does need to consider the devastating effects of excess weight on mobility versus the potential for muscle loss associated with inducing negative energy balance.

Malnutrition

The spectrum of over nutrition to under nutrition occurs with disease progression. As boys with DMD age, the likelihood that they will become underweight is increased. Significant muscle wasting will result in weight loss. This is accentuated by eating difficulties experienced at meal times likely leading to a reduced food intake. Iannaccone
et al. (2003) investigated malnutrition after surgical procedures and found weight loss after surgery to be associated with loss of the ability to self feed. Some possible explanations suggested for this were: a decrease in neck range of motion, or head control; lack of adequate care giving; and low cognitive function. Intervention to increase energy intake resulted in weight gain. Pane et al. (2006) reported on feeding problems experienced by individuals with DMD. These included difficulty in opening their mouth, chewing leading to an increase in meal duration, with episodic choking also reported. In a similar survey, Willig et al. (1994) found the most prominent feeding difficulty was getting food to the mouth.

Food texture modification is an important strategy to manage feeding difficulties. Modifications include cutting food into smaller pieces and changing the texture to smooth foods (Pane et al., 2006). A multidisciplinary team should be involved in the management of feeding difficulties (Stewart et al., 2006; Almond et al., 2007) with individual preference being paramount. Gonzalez-Bermejo et al. (2005) analysed dietary intake in an older group of boys (aged 22–33 years) reporting that, although they ate slowly and with difficulty, these inconveniences were largely compensated for by the pleasure of eating a normal meal.

In the literature, the incidence of gastrostomy feeding tubes in older boys with DMD ranges from nil (Gonzalez-Bermejo et al., 2005) to 50% (Hilton et al., 1993). Indications include difficulty in swallowing leading to aspiration, and undernutrition (Bushby et al., 2005). As with other conditions, multiple issues should be considered before gastrostomy placement can occur. At the stage of disease when alternate feeding options need to be considered, such patients have severe respiratory insufficiency, putting them at high risk of major complications when they are anaesthetised or sedated for such a procedure (Birnkranet al., 2006). Despite these risks, Birnkranet al. (2006) detailed successful gastrostomy placements in a 22- and 19-year-old male. The authors encourage physicians to consider whether providing enteral nutrition support should be proactively planned at earlier stages of the disease process, before the risks associated with anaesthesia in those with compromised respiratory capacity.

In the past, gastrostomy placement in the DMD population was rare (Pane et al., 2006), most likely as a result of ethical issues surrounding this procedure. Hilton et al. (1993) presents an excellent discussion on such ethical considerations associated with advanced stages of DMD. ‘The “can do” versus the “should do” ... the ability to act does not justify the action’ (Hilton et al., 1993). In the face of such confronting ethical decisions in medicine, and in this case nutrition and dietetics, the authors suggest four considerations to professionals: medical indications, patient preference, quality of life, and contextual features (i.e. social and medical support). No studies have specifically examined the effect of gastrostomy feeding in DMD on quality of life, although the nutritional benefits of enteral feeding are demonstrated by Goldstein et al. (1989).

Nutriceuticals

There are four randomised controlled trials, summarised in a recent Cochrane review (Kley et al., 2007), providing high-quality evidence for the use of dietary supplements to enhance muscle function. Because many families seek alternative or naturopathic supplements, dietitians need to be aware of their potential nutritional effects or interactions. The most promising of these is creatine monohydrate. Creatine is an amino acid found in muscle that stores energy in the form of phosphocreatine for immediate use (Strober, 2006). Several studies have supported the use of creatine for increasing athletic performance and strength in healthy populations (Grande & Graves, 2005). A recent Cochrane review (Kley et al., 2007) concluded that the available evidence from randomised trials shows that short- and medium-term creatine treatment improves muscle strength in people with NMD, and is well tolerated. In 138 participants with NMD treated with creatine, there was a significant increase in maximum voluntary contraction in the creatine group compared to placebo, with a weighted mean difference of 8.47% (95% confidence interval = 3.55–13.38). There was also an increase in lean body mass during creatine treatment compared to placebo (weighted mean difference 0.63 kg; 95% confidence interval = 0.02–1.25). The supplement doses of the included trials were 5 g day\(^{-1}\) (Escolar et al., 2005), 3 g day\(^{-1}\) (Louis et al., 2003), 100 mg kg\(^{-1}\)day\(^{-1}\) (Tarnopolsky et al., 2004) and 5 g day\(^{-1}\) for children, and 10 g day\(^{-1}\) for adults (Walter et al., 2000). Another interesting finding from these studies was the effect on bone health in boys with NMD. Creatine treatment increased bone mineral density by 3% (Louis et al., 2003), and reduced levels of N-telopeptides, a marker of bone degradation (Tarnopolsky et al., 2004). Although these results are statistically significant, the clinical relevance should be considered.

Of the studies included in the Cochrane review, all bar one (Tarnopolsky et al., 2004) was conducted in steroid naïve dystrophic males. The use of complementary therapies could possibly increase the positive effect on muscle function. Using the mdx mouse model, Payne et al. (2006) tested several compounds (creatine monohydrate, conjugated linoleic acid, α-lipoic acid and β-hydroxy-β-methylbutyrate) in combination therapy with...
prednisolone, which provided the most consistent evidence of efficacy. Glutamine has also been studied. Despite promising beginnings in nondystrophic humans (Hankard et al., 1996b, Darmoun et al., 1998) and in dystrophic males (Hankard et al., 1998, 1999; Mok et al., 2006b), a randomised controlled trial (Escolar et al., 2005) was unable to demonstrate a statistically significant effect of glutamine supplementation based on manual and quantitative measurements of muscle strength. The authors did conclude, however, that a disease-modifying effect of glutamine in younger dystrophic males cannot be excluded.

L-arginine (Chaubourt et al., 1999; Voisin et al., 2005) and green tea extract (Buetler et al., 2002; Dorchies et al., 2006) have also been investigated with some success in the mdx mouse model; however, these therapies are yet to move from ‘bench level’.

Future developments

The multidisciplinary management of boys with DMD has progressed significantly and now involves the use of steroid therapy, ventilator support, spinal management and improved therapies such as physiotherapy. As a result, the life expectancy of boys with DMD is steadily increasing. Eagle et al. (2002) concluded that the chances of survival to 25 years have increased from 0% in the 1960s to 53% in the 1990s; and this was before steroid treatment. Data on the longer-term effects of steroids are beginning to emerge (Balaban et al., 2005; Biggar et al., 2006; Angelini, 2007), although it would appear overall that the body of evidence is still catching up with the now regular use of steroids in clinical practice. Further research is required to map the natural history of DMD in steroid-treated boys. With an older DMD population emerging, it is unclear what nutritional issues will emerge in the adulthood life of DMD males. With increased body fat mass and an inability to exercise, are these young men at risk of developing a cluster of cardiovascular risk factors, such as the metabolic syndrome? Aitkens et al. (2005) and Kilmer & Zhao (2005) suggest that this might happen, which will provide a new level of complexity to dietetic management.

Future research avenues

The dystrophin gene is the largest, spanning approximately 2.5 Mb of genomic sequence, and is composed of 79 exons (Blake et al., 2002); hence, gene therapy has been hampered by the complexity, size and sheer number of mutations. Phase III clinical trials are ongoing with PTC124, an orally administered drug that promotes ribosomal read-through of nonsense mutations. The treatment will have limited reach because nonsense mutations are one of the least common genotypes. Other potential therapies include those that increase the uptake of utrophin, as well as antisense oligonucleotide-mediated exon skipping to correct out-of-frame mutations and restore truncated, yet functional dystrophins. Until a cure is found, therapeutic and supportive interventions will assist in preventing or delaying complications and preserving quality of life.

Conclusion

Reviewing the evidence base for nutrition in DMD reveals consistent limitations across all areas, with little high-quality evidence to support the key practice points highlighted. Overall, there is little research into nutrition and DMD. There are often no other completed studies to support findings in publications; small sample sizes are common because recruiting from such a sparse patient population can be difficult, randomised controlled studies are scarce, and countless projects remain at the bench stage. For these reasons, many recommendations are based on extrapolations from non DMD studies and expert opinions. Such recommendations will inevitably always be ranked as poor using evidence-based medicine criteria. However, they are not without merit. Given the obstacles that potential DMD researchers face, expert opinion based on clinical expertise and fundamental nutrition principles may comprise the strongest and most accurate evidence available to assist this neglected group.

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**Supporting Information**

Additional Supporting Information may be found in the online version of this article:

**Table S1.** Other potential issues identified in males with DMD.

**Table S2.** Key practice points and supporting evidence.

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