Appendix A: Discussing Duchenne with children

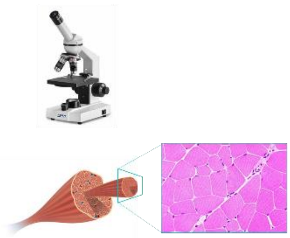
Discussing the diagnosis

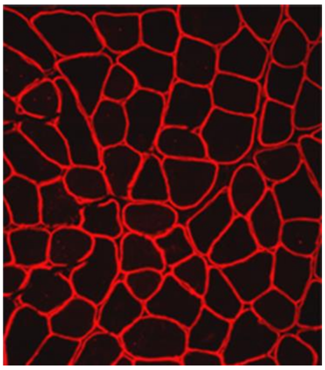
The use of comics to give information in an engaging medium should be encouraged. The PTC Comics "Mission Duchenne" are a good example.

Simple metaphors and visual tools are often beneficial. For example, younger children may be helped by the concept that dystrophin acts like a "shock absorber" to muscles (they will have seen these on cars and bikes so may understand the importance of them in preventing damage).

Explaining DMD to older children

Even complex scientific ideas can be explained visually, for example these images from Professor Straub, which show how dystrophin 'coats' the muscle fibres may be helpful for slightly older children:





Many individuals may find it difficult to process complex information presented verbally. Some clinicians might find it helpful to use visual representations to assist with explanations. There are diagrams available which show the complexity of the DMD gene in simplified form that can be used in dialogue to explain the site of mutation. This could be helpful in an older child's understanding as to why 'their Duchenne' might vary in phenotype from others affected by the condition.

Some patients may be very keen to have an in depth understanding of the genetics of the condition. If this interest is there, it should be encouraged. Invariably, things that frighten us become less scary when we know more about them.

Some useful resources to help with this include:

Types of Mutations - Parent Project Muscular Dystrophy (parentprojectmd.org)

Individuals may feel more able to discuss their condition with other patients (e.g. consider regional groups for adolescents), where ideas and concerns can be brought collectively for discussion, facilitated by clinical staff such as specialist nurses.

- *The Abilities in Me - Duchenne Muscular Dystrophy* (Gemma Keir) **ISBN-13 :** 979-8767770656
- Talking to your Children about Duchenne Muscular Dystrophy (originally on Action Duchenne website): https://media.gosh.nhs.uk/documents/Talking_to_your_child_Schonfeld_booklet.pdf
- Resources for newly diagnosed families - Duchenne UK: https://www.duchenneuk.org/support-at-diagnosis/

Appendix B: Support resources from charities

There are a number of UK-based charities that can offer support to individuals and families. It is recommended that websites are directly visited in order to check the most up to date information and support being offered by each charity. Some examples are given below:

- **Duchenne UK:** https://www.duchenneuk.org/
- **Pathfinders Neuromuscular Alliance:** https://www.pathfindersalliance.org.uk/
- **Muscular Dystrophy UK:** https://www.musculardystrophyuk.org/
- **Action Duchenne:** https://www.actionduchenne.org/
- **Duchenne Family Support Group:** https://dfsg.org.uk/dev/

Appendix C: DMD Specific Screening Tools

DMD Specific screeners

In recent years, three condition-specific screening tools have been developed to better identify neurodevelopmental, behavioural, emotional, and psychosocial difficulties associated with Duchenne muscular dystrophy (DMD). These tools aim to address limitations of generic screeners, which may miss the distinctive neurocognitive and behavioural profile linked to dystrophin deficiency in the brain. These tools are **complementary rather than competing.** Used together (or selectively), they may help move DMD care toward **proactive, brain-inclusive monitoring**, rather than relying solely on reactive referrals.

The three measures are:

1. Duchenne Muscular Dystrophy–Associated Neurodevelopmental Disorders Screener (DuMAND; Geuens et al., 2024)
2. Behavioural, Emotional, Learning, and Social questionnaire (BELS; Truba et al., 2025)
3. BIND screener (Miranda et al., 2026)

Comparison of DMD-specific screeners

Feature	BELS	BIND	DuMAND
Primary focus	Broad neurodevelopmental & psychiatric symptoms	Neurobehavioral & emotional difficulties	Neurodevelopmental disorder risk
Core domains	Behavioural, Emotional, Learning, Social	Attention, emotion regulation, social interaction, and behaviour	ADHD, autism traits, learning, emotional/behavioural regulation
Reporter	Parent/caregiver	Parent/caregiver	Parent/caregiver
Age range	~4–19 years (pediatric)	Early childhood → adolescence	Early childhood → young adulthood
Time to complete	~5–10 minutes	~5 minutes	~5–10 minutes
Output	Domain scores + total score	Domain-level risk flags	Risk stratification
Diagnostic?	No	No	No
Possible clinical use	Identify psychosocial & mental health concerns	First-line DMD-specific behaviour screener	Structured neurodevelopmental surveillance
Best used when	Routine clinic screening	Quick flagging of DMD behavioural phenotype	Developmental transitions & longitudinal monitoring

Appendix D: Generic Mental Health Screening Measures

General guidance for using screening tools in neuromuscular and mental health clinics for either mental health or neurodevelopmental concerns

These are screening/monitoring tools, not diagnostic tests. A positive screen should trigger clinical assessment, collateral history, and (where relevant) specialist referral. Measures often include 'clinical cut-off points' that may indicate a certain level of need. However, it is important to note that many measures used in regular clinical practice are not normed in the DMD population and therefore risk either over- or underestimating the level of need. Clinical judgement is still required in all situations.

It is also important to note that symptom overlap is common in neuromuscular disease (fatigue, poor sleep, pain, steroid effects, school absence, breathlessness) and can influence scores on different measures. It is important to interpret results in a clinical context and consider repeating once physical contributors are addressed/acknowledged.

PHQ-9 (depression; Kroenke et al., 2001)

Purpose: depression symptom severity (and monitoring change over time). Free to use.

Age range: widely used in adults; adolescent versions exist (PHQ-A/PHQ-9 modified for teens).

Time to administer: typically, ~2–5 minutes; some studies report an average of ~2 minutes.

Scoring/interpretation (practical): Total score between 0 and 27. Common severity cut points range from mild to severe.

Item 9 screens for suicidal thoughts: any positive response requires competent risk assessment (local policy).

When to use: routine adult mental health screening, baseline/monitoring, steroids/transition periods.

What to do next: If someone scores in the moderate range or above, or if item 9 is positive, it is recommended that clinicians assess risk, impairment, and comorbidity. A referral for formal psychological support is recommended.

GAD-7 (anxiety; Spitzer et al., 2006)

Purpose: generalised anxiety symptom severity (screening/monitoring). Free to use.

Age range: validated in adults and also used in adolescents in research/clinical contexts.

Time to complete: ~1–2 minutes.

Scoring/interpretation (practical): Total score between 0–21; common cut-off points included and overall score can be mild/moderate/severe).

What to do next: If the score is in the moderate range or above, assess functional impact (does the patient report that anxiety is impacting daily life, and if yes, how), panic/social anxiety/trauma, sleep, and physical symptom overlap.

RCADS (Revised Children's Anxiety and Depression Scale; Chorpita et al., 2000)

Purpose: symptom profiles for anxiety disorders and depression in children and young people. This questionnaire includes both parent report and self-report. Two versions- one with 47 items (RCADS-47) and one with 25 (RCADS-25). Free to use but copyrighted; check permissions for adaptations/electronic use.

Age range: typically 8–18 years.

Time to complete: RCADS-47: ~10–15 minutes. RCADS-25: ~5–10 minutes

Interpretation (practical): Subscales are used. Scores are converted to T-scores/percentiles with 'normal/elevated/clinical' style bands depending on scoring method/source.

When to use: you want more disorder-specific anxiety/depression detail than SDQ (see below) provides. Many child and adolescent services nationally will be familiar with the RCADS and it may support referrals to these services.

Strengths and Difficulties Questionnaire (SDQ; Goodman, 1997)

Purpose: The SDQ is a 25-item screening tool to assess emotional symptoms, conduct problems, hyperactivity and inattention, peer relationship problems and prosocial behaviour. This is a simple and quick measure of mental health used in nearly all child and adolescent mental health services (CAMHS). It can be completed in a waiting area and scored during or after an appointment. It is recognised to be appropriate for use in DMD and is highlighted in international guidelines (Birnkrant et al., 2018).

Age range: 2–18 years, with different versions for parent/teacher/self-report.

Time to administer: 5–10 minutes.

Interpretation (practical): Produces subscale scores and a Total Difficulties score used to flag risk bands (exact banding depends on version/scoring norms used locally). Can be used as a general first-line screener; if elevated, follow with RCADS or disorder-specific tools.

Availability: free to download/manual score; online scoring/services may cost and electronic reproduction may be restricted.

PARS-III (Personal Adjustment and Role Skills Scale- III; Hendriksen, 2009)

Purpose: This scale was originally developed to assess psychosocial adjustment in children 4–18 years of age and has been applied in children and adults with DMD and was found to be reliable and valid (Hendriksen et al., 2008). The PARS III consists of 28 items that measure psychosocial functioning in six areas: peer relations, dependency, hostility, productivity, anxiety-depression, and withdrawal.

Age range: originally developed for children (often cited ~4–18; DMD studies commonly in school-age to adolescence).

Time to administer: ~5-10 minutes

Availability/licensing: Some versions explicitly state not to reproduce without permission.

When to use: you want a chronic-illness-relevant adjustment measure rather than DSM symptom screens.

PARS-A (adult adaptation of PARS-III; Weerkamp et al., 2022)

The PARS-A is a shortened version based on the PARS-III and suitable for patients aged 16 years and older (Weerkamp et al, 2022). It contains 22-items that are suitable for adult patients and has the same structure as the PARS-III. The PARS-A will be used as a self-report measure. This questionnaire is less standard than PHQ-9/GAD-7/SDQ/RCADS in routine mental health screening.

Practical guideline stance: if your service uses it, include it as a condition-specific psychosocial adjustment measure; otherwise, for adult mental health screening, most services will prioritise PHQ-9/GAD-7 plus a functional review.

Appendix E: Screening for Autism and/or ADHD

As with many mental health screening tools, commonly used autism and ADHD screening tools were not designed for children with neuromuscular conditions. In DMD, physical disability, fatigue, treatment effects, and environmental limitations may influence questionnaire responses, potentially leading to false negatives or misleading results. Screening outcomes should therefore be interpreted cautiously and in conjunction with clinical judgement, caregiver concerns, and DMD-specific screening measures.

Some commonly used measures are listed below; however, our primary recommendation is that, if parents or clinicians suspect that a child may benefit from a formal assessment for Autism and/or ADHD, a referral to relevant neurodevelopmental assessment services should be made as soon as possible.

Autism Spectrum Quotient (AQ- 50; Baron-Cohen et al., 2001)

Purpose: self-report autistic traits (screening/quantification), not diagnostic.
Age range: commonly ≥16 years. Child version is also available.
Time: ~5–10 minutes (sometimes reported 10–15).
Scoring/interpretation (practical): Total score 0–50; higher scores are linked with more 'autistic traits'.

Autism Spectrum Quotient- 10 items (AQ-10; Allison et al., 2012)

Purpose: brief referral screener for possible autism in adults.
Age range: ≥16 years in the adult version.
Time: ~2 minutes.
Scoring/interpretation: Scored 0–10. NICE implementation materials note ≥6 as a threshold to consider referral for specialist autism assessment (context-dependent).

SNAP Rating Scale (commonly SNAP-IV; Swanson, 1992; Swanson et al., 2012)

Purpose: ADHD symptom screening via parent/teacher ratings (and some versions include ODD items).

Age range: commonly used in children/adolescents (e.g., ~8–18).

Time: often ~5–10 minutes (varies by respondent and setting).

Scoring/interpretation (practical): Items rate symptom frequency/severity on a 4-point scale. Typically summarised as Inattention (items 1–9) and Hyperactivity/Impulsivity (items 10–18); some versions add ODD items.

Adult ADHD Self-Report Scale (ASRS-v1.1)

Purpose: adult ADHD screening (Part A 6-item screener) + symptom checklist.

Age range: adults (commonly used 18+).

Time: about 5 minutes.

Appendix F: Template for Clinical Letters to be shared with school/ college/Local Authority

Below you will find a suggested template that we hope will be useful in drafting impactful letters for consideration by authorities developing Education Health and Care Plans for people with Duchenne muscular dystrophy. You can adapt this to suit the needs of your clinical set up, but the terminology we have proposed is aligned with that used by education providers – it 'speaks the right language'.
An example report shared by Dr. Alhaswani can be found with this guidance.

Keep the parts on black as part of your template – you can edit to personalize or reflect your clinical set-up if you prefer. Parts in red are to be deleted (some are instructions) or replaced as needed.

If you have any questions, please get in touch with the DMD Care UK project Education group lead – Dr Janet Hoskin: j.hoskin@uel.ac.uk

Dear xxxx

I recently met with Sami Begum and his parents on 1 July 2024 for a regular clinical review as part of my role as neuromuscular consultant in his care delivery for Duchenne muscular dystrophy (DMD).

I have separated this summary of our appointment into sections that include his medical care and physiotherapy as well as information about his psychosocial needs in order to better support holistic care for Sami.

Part 1 Clinical Health report. (Medical and Therapy)
This is your usual clinic report/emergency plans/precautions and physiotherapy report.

Part 2: Information for Education, Health and Care Plans
Please complete this Part 2 if the child is about to start school, is transitioning between primary and secondary school or between secondary and Further Education/Sixth Form, **OR** at any time the family or school request it.

Every young person or adult living with DMD who is in school or college should have an Education Health and Care Plan (EHCP). As well as experiencing severe muscle weakness, young people with DMD are at high risk of psychosocial and neuro-divergent conditions as well as emotional challenges for which they will require support from local services. **Even in the absence of a formal diagnosis of these conditions, it should be assumed that young people with DMD may experience aspects of them – it is a part of having DMD – and their care, education and support should be tailored accordingly.**

Hopes Dreams and Aspirations
As most young people with DMD can now expect to live into their fourth decade (Landfeldt et. al., 2020) it is essential that services support young people to plan for the future they want.
Add a sentence to sum up discussion with young person and family e.g. I met Sami today and he told me he loves computer games and in the future he wants to be a video games designer.

SEND Category 1: Communication and Interaction
This category refers to Speech, Language and Communication Needs for example, Speech Delay/Disorder, and Autism Spectrum Condition. You can provide specific examples under sections a, b and c below.

It has been established that Speech and Language development are often delayed in DMD (Parsons et.al., 2004; Ricotti et. al., 2015). Differences associated with Autism Spectrum Condition are often apparent even though a child with DMD may not qualify for a full diagnosis.

a. Evidence of SEND from observation/ any psycho-social assessment/school report (delete accordingly and add evidence available)
b. At clinic, Sami's parents reported that….. eg. Sami can only say three words
c. Sami would benefit from a referral to…. eg. Speech Therapy Service for a Speech and Language assessment

SEND Category 2: Cognition and Learning

This category refers to difficulties with learning. You can provide specific examples under sections a, b and c below.

Working memory and phonological processing skills are often affected in DMD which can impact literacy and numeracy and there is a high risk of dyslexia and reading difficulties (Hinton et. al., 2004; Hendriksen & Vles 2008; Hoskin, 2014). Most children with DMD have average cognitive ability and have strengths in vocabulary (Hinton et. al., 2004)

a. Evidence of SEND from observation/ any psycho-social assessment/school report (delete accordingly and add evidence available)
b. At clinic, Sami's parents reported that…… eg. He is having difficulties learning sounds/phonemes
c. Sami would benefit from a referral to …eg. An educational psychologist for a cognitive assessment

SEND Category 3: Social Emotional and Mental Health

This category relates to well-being and mental health as well as neuro-developmental differences such as ADHD. You can provide specific examples under sections a, b and c below.

Young people with DMD are at higher risk of neurodevelopmental/ mental health differences such as ADHD, OCD, Depression and Anxiety (Banihani et. al., 2015; Hendriksen et. al. 2007; Pane et al 2013). They often experience social isolation due to lack of equity of access to a range of activities.

a. Evidence of SEND from observation/ any psycho-social assessment/school report (delete accordingly and add evidence available)
b. At clinic, Sami/Sami's parents reported…..eg. Sami is having regular meltdowns at home which take a long time to get over
c. Sami would benefit from a referral to ………eg. The local CAMHS service for an assessment for ADHD

SEND Category 4: Physical Disability and Sensory Needs

This category refers to any physical needs as well as sensory difficulties a child may experience at school. This may include (depending on child/young person):

Physical abilities walking, stairs, participation in PE, safety in playground, access, fatigue, risk of falls, seating equipment if needed, support with toileting, arm function, pain and cramps ,pacing of activity, myoglobinuria, help with feeding, any assistive respiratory devices, emergency plans, explanation about any observed side effects of steroids and its impact on the young person, advice about risk of rhabdomyolysis, Risk of bone fractures (in particular if already had one). You can provide specific examples under sections a and b below.

DMD is a progressive muscle-wasting condition and provision and forward planning must be made for this (Birnkrant et. al.,2018). Please see Physiotherapy Therapy report in Section 1 of this letter for full details. Many children with DMD also appear to experience sensory difficulties such as over stimulation of sound/visuals/touch etc. (PPMD 2024). Please be aware of this in planning any support activities.

a. At clinic Sami's parents reported eg. Problems with school accessibility/ frequent falls etc (delete accordingly)
b. Sami would benefit from a referral to ….eg. a local OT to support with fine motor skills/ OT for equipment/ a Sensory Processing Service

Information about Social Care

You can provide specific examples under sections a and b below.

Education Health and Care Plans should include information about Social Care needs. Young people with DMD have additional care needs and benefit from an assessment from a local Children/ Adult Disabilities Team. They should initially qualify for Short Breaks funding and subsequently need assessing for social care support.

a. At clinic, Sami/Sami's parents reported …….. eg. Sami needs help to get up in the morning and to go to bed

b. Sami would benefit from a referral to………eg. The local Children and Disabilities team for a social care assessment

Family

You can provide specific examples under sections a and b below.

DMD impacts the whole family (Porteous et. al., 2021).

a. Parents report at clinic….eg. Sami's older sister is finding it difficult to attend school

b. Parents/ siblings would benefit from referral to…… eg. Counselling for psycho-social support

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Appendix G: Employment

Muscular Dystrophy UK's report 'Ready and Able: Removing the barriers that prevent young disabled people from finding employment'. Available at: https://www.musculardystrophyuk.org/app/uploads/2024/05/Ready-and-Able-EV-08May19-1.pdf

UK Government Access to Work scheme: https://www.gov.uk/access-to-work

Evenbreak – job board for people with disabilities: https://www.evenbreak.co.uk/

Evenbreak Career Hive: provides relevant and accessible careers advice, including career coaching by people with lived experience of being disabled employees: https://hive.evenbreak.co.uk/

Enable Works – provide training and employability programmes in the UK for people who experience barriers to employment: https://www.enable.org.uk/enable-works/

Disability Jobsite: https://www.disabilityjobsite.co.uk/

Appendix H: Sex and Intimacy

The Outsiders Trust

https://outsiders.org.uk/

Peer support and dating club for socially and physically disabled people. Referred to in Duchenne UK's family folder for adolescence and adulthood (DMD-Family-Folder-Adolescence-and-Adulthood-complete-reduced-size-pdf (cloudinary.com))

TLC Trust

https://tlc-trust.org.uk/

The TLC Trust are a committed and passionate non-profit organisation who are striving to promote sexual services for disabled people in a safe, fun and responsible way.

Scope

The disability charity, Scope, has advice sections on:

- Intimacy, disability and relationships

https://www.scope.org.uk/advice-and-support/talking-about-intimacy-sex-relationships

- Sex and disability

https://www.scope.org.uk/advice-and-support/disability-and-getting-ready-for-sex

You can register to join Scope's online community. Here you can talk about dating, sex and relationships, for example.

Disability Horizons

https://disabilityhorizons.com/category/disabled-dating-relationships-and-sex/

Has a range of blogs/posts on Relationships and Sex.

This includes a summary of the best disability dating sites, e.g.

https://disabilityhorizons.com/2024/03/disability-dating-sites-we-round-up-the-best/

SHADA: Sexual Health and Disability Alliance

https://shada.org.uk/