

Example of a Detailed Pathway Scope for Duchenne Muscular Dystrophy (DMD)

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| **Disciplines Involved** | **Selected parameters to include and detail** | **☑** |
|  | Diagnosis of DMD |  |
|  | Clinical presentation summary including sign and symptoms |  |
| **Initial stages of diagnosis = GP / Primary care team / Paediatrician / Neurologist** | Baseline Clinical Assessment  Baseline Test (s)  Baseline Investigation (s)  Identification of Red Flags for referral to expert centre |  |
| **In-depth diagnosis = Neurology, Neuromuscular specialist, Clinical Genetics** | Specialist Clinical Assessment  Specialist Test (s)  Genetic/genomic testing  Specialist Investigation (s) |  |
|  | Diagnostic Criteria |  |
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|  | Specialist care for DMD |  |
| **Key medical specialist**  **(Co-ordinating specialist) = Neuromuscular Specialist** | * Review period e.g. at diagnosis, yearly, as indicated * Clinical Assessment * Symptomatic treatment * Prophylactic treatment * Surveillance (i.e. asymptomatic screening – age / stage, frequency * Symptomatic screening (including trigger) * Onward referrals (including trigger for referral) * Specific advice / management for pregnancy, anaesthetic, sick days * Access to Clinical Trials * Follow-up for at-risk family members |  |
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| **Core medical specialists for all patients** | Examples of indications |  |
| **Cardiology** | Cardiomyopathy, rhythm abnormalities and heart failure management including review period |  |
| **Endocrinology** | Growth, puberty, bone monitoring and adrenal insufficiency including review period |  |
| **Respirology** | Pulmonary function, vaccination, cough assist, ventilation including review period |  |
| **Orthopaedics** | Scoliosis, fractures, contractures, pain |  |
| **Gastroenterology** | Swallow, constipation, gastro-oesophageal reflux, gastroparesis, gastrostomy |  |
| **Ophthalmology** | Steroid use – monitoring for cataracts, glaucoma |  |
| **Clinical Nurse Specialist** | Role e.g. care co-ordination, point of contact, transition planning |  |
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| **Medical specialist for complex care (if required)** |  |  |
| **Orthopaedic surgery** | Scoliosis, fractures, contractures |  |
| **Pain Specialist** | Pain management |  |
| **Psychiatrist** | Moderate to severe mental health issues |  |
| **Perinatal specialist** | Maternal cardiomyopathy screening,  reproductive options |  |
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|  | **Health & Social Care Professionals (HSCPs)** |  |
| MDT team |  |  |
| **Physiotherapy** | Respiratory and neuromusculoskeletal function |  |
| **Speech & Language Therapy** | Speech, language & communication needs and / or feeding, eating, drinking & swallowing skills |  |
| **Occupational Therapy** | Activities of daily living skills, environmental adaptions, specialist equipment / aids including assistive technology |  |
| **Dietetics** | Swallow and gastrointestinal issues, nutritional status bone health, growth, weight, |  |
| **Genetic Counselling** | Recurrence risk, reproductive options, at-risk relative identification and follow-up |  |
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|  | Psychosocial Services & Supports |  |
| **Psychology** | Adjustment, anxiety, depression, neuropsychology |  |
| **Social Work** | Psychosocial, financial, housing, mobility, home care, respite & employment support |  |
| **Educational Psychology** | Signpost to national educational psychology service  Role e.g. Cognitive assessment, learning support, school placement |  |
| **Employment Support** | Signpost to national employment support services for information, advice and support |  |
| **Social Welfare Services** | Signpost to national social welfare support services for information, advice and support |  |
| **Patient Organisation (s)** | Signpost to validated relevant National, European / International patient organisations for information, advice and support |  |
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|  | Primary / Community care services |  |
| **GP** | Role e.g. vaccinations, coordination of care |  |
| **Public Health Nurse** | Role e.g. co-ordination of local services and supports |  |
| **Rehabilitation / Disability services** | Access to Neurorehabilitation / Neurodisability services and supports |  |
| **Home care services** | Access to home care |  |
| **Respite care services** | Access to respite care |  |
| **Palliative care services** | End of life care and decision-making |  |
| **Pharmacy** | Medication advice |  |
| Dentistry | Regular review and referral to Orthodontics (if required) |  |
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|  | General Outcome Measures (from which KPIs can be developed) |  |
| **Outcome measure 1** | Access to neuromuscular specialist services for males affected with DMD |  |
| **Outcome measure 2** | Access to clinical trials for gene therapy - genetic testing to assess eligibility for genotype-specific therapies e.g. Ataluren |  |
| **Outcome measure 3** | Opportunity for pre-conceptual genetic testing to clarify carrier status / reproductive risk for at-risk female relatives |  |
| **Outcome measure 4** | Cardiac assessment / follow-up of female relatives who carry the familial pathogenic variant |  |
| **PROM 1** | Family informed about national patient organisations |  |
| **PROM 2** | Family offered referral to Genetic Counselling at diagnosis |  |
| PROM 3 | Family and GP provided with patient-friendly information about DMD |  |
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|  | Core Information |  |
| Orphanet | Orphacode, Orphanet definition & link |  |
| Evidence | Recent Clinical Practice Guidelines and other key publications/evidence used to inform the care pathway |  |
| **ERNs** | Links to relevant ERN-endorsed resources on ERN-NMD (e.g: Patient Journey |  |

**Please note:** this is an example of the types of parameters which might be considered for inclusion and detailing in the care pathway. This will help to inform the disciplines required for representations in the core writing group/pathway development group.