Care Pathway Topic:Paediatric and Adult Duchenne Muscular Dystrophy Care Pathway

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This document is part of the
Care Pathways Toolkit for **Healthcare Professionals** & **Patient Representatives**

WP6 TASK 6.2

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Outline Pathway Scope

Rare Condition: Duchenne Muscular Dystrophy

Orphanet definition:

ORPHACODE: 98896 A rare, genetic, muscular dystrophy characterized by rapidly progressive muscle weakness and wasting due to degeneration of skeletal, smooth and cardiac muscle.

ORPHACODE: 206546 In females: a rare, genetic muscular dystrophy affecting female carriers and characterized by variable degrees of muscle weakness due to progressive skeletal myopathy, sometimes associated with dilated cardiomyopathy or left ventricle dilation.

Target population: Affected males with a confirmed diagnosis of DMD by pathogenic (P) or likely pathogenic (LP) dystrophin gene (*DMD)* variant or a muscle biopsy showing dystrophin absent, at-risk male relatives, at-risk female relatives, females who carry a P or LP dystrophin gene variant

Duration/Scope of the pathway: lifelong – from diagnosis, paediatric, transition, adult, pregnancy/perinatal, palliative care

Care Setting: Specialist hospital care services, Health & Social Care Professionals (HSCPs), Primary Care/General Practitioner**,** Community Disability services, Psychosocial Services & Supports including education / employment support services, Patient organisations

General outcome measures:

- Improved access to specialist neuromuscular MDT care for individuals affected with DMD; decrease diagnostic delays; identification and follow-up of at-risk female relatives
- Improved access to genotype-specific precision medicine therapies (e.g. Ataluren if eligible)
- Improved shared care model and integrated multi-disciplinary care
- Uniformity of care and equity of access to quality care. Tool for optimisation of healthcare commissioning
- Empowerment of PLWRDs to navigate their national health service
- increase knowledge, education and awareness of the condition for intersecting sectors and the community in general
- Enhance clarity of team members roles and responsibilities

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Please refer to this document to access all resources:

https://jardin-ern.eu/?resource=care-pathways-toolkit

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