Beyond Diagnosis: Standard of Care for Adult Duchenne Muscular Dystrophy (DMD) Patients in Wales, the United Kingdom (UK)

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BACKGROUND

Duchenne Muscular Dystrophy (DMD) is a progressive, debilitating and fatal neuromuscular disease caused by variants in DMD gene. With increase in life expectancy, there are growing numbers of adults with DMD whose complex medical needs have not been addressed in previous international standard of care. In this project, we aim to evaluate various aspects of adult DMD care in Wales by benchmarking against the 2021 Adult North Star Network Guidelines.

OBJECTIVES

- To evaluate whether the local health providers, institutions and facilities are adhering to the national guidelines for the care of adult DMD patients.
- To identify discrepancies and gaps including access to specialist services and holistic are.
- To develop achievable and specific recommendations for improvement based on the findings.

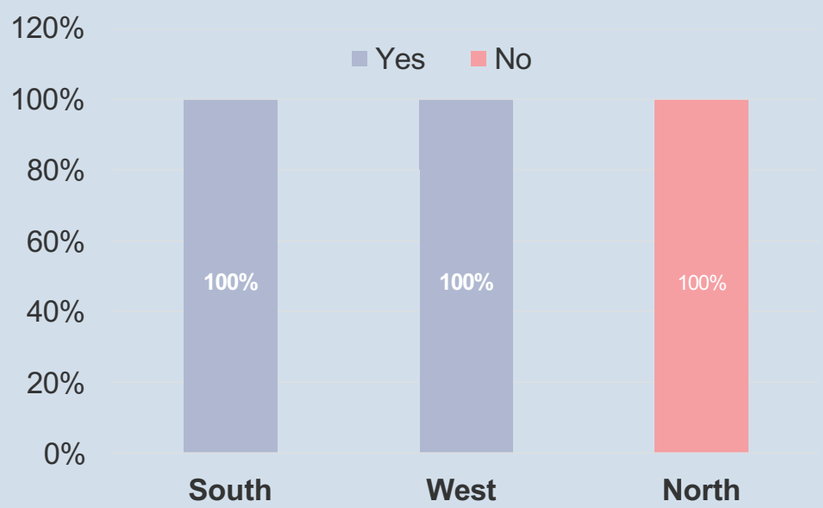
METHODOLOGY

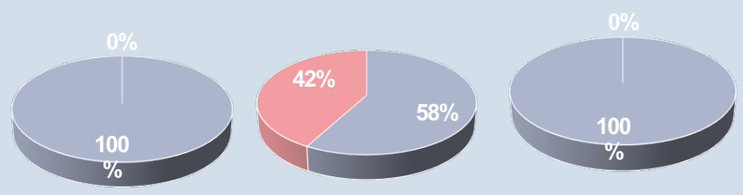
Data was collected retrospectively between June 2023–July 2024 from neuromuscular centres across Wales. Source data is from patients' notes and electronic patient records. All adult DMD patients aged ≥14 years were included (N=56). Results were scrutinised by stakeholders, neuromuscular team members, neurologists, public health and data experts and service user representatives.

RESULTS

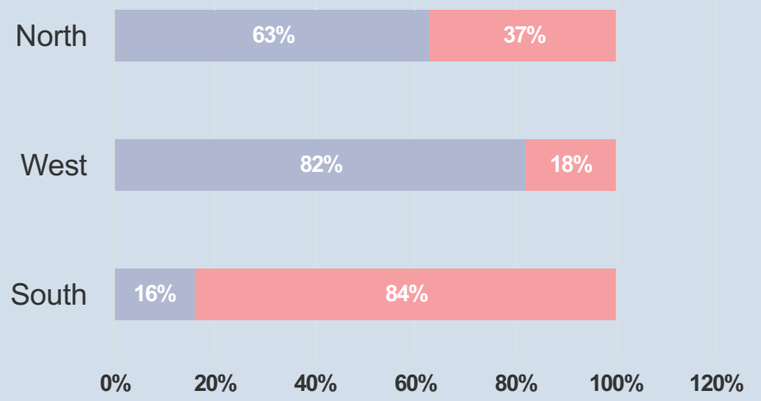
Topic	Yes	No	Not Applicable	Unknown
Regular Respiratory Input?	88%	12%		
Regular Physiotherapy Input?	86%	14%		
Regular Cardiology Input?	86%	14%		
Appropriate Anaesthetic Care?	76%	4%	12%	8%
Gastroenterology Input if needed?	68%	14%	16%	2%
Care co-ordinator involve in medical care?	67%	33%		
On Steroid treatment?	56%	44%		
Renal Function Monitored?	55%	45%		
Respite Care Available?	54%	46%		
Bone Specialist Input if needed?	53%	24%	23%	
Transitional Service Available?	33%	46%	21%	
Psychological Support if needed?	38%	62%		

Care Advisor involved in medical care?





Regular Cardiology Follow up?



Respite Care Available?

CONCLUSION

Findings show strengths in access to respiratory (88%) and physiotherapy care (86%), except in North Wales. Psychological support was identified as a critical unmet need across all regions, with 62% lacking access. Top challenges across specific regions of Wales include absence of clinical care coordination in North Wales (100%), insufficient cardiology follow-up in West Wales (42% require follow up), and limited respite care in South/Southeast Wales (84% without access).

DISCUSSION

These findings highlight critical regional disparities in adult Welsh DMD care and emphasize the need for targeted improvements in psychological, cardiac, and coordinated clinical services. Strengthening the care pathways in these areas is vital for achieving equity in care for DMD patients in Wales, the UK.

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