

Symptoms and impacts of nonsense mutation Duchenne muscular dystrophy (DMD) at different stages of ambulation

DMD is a rare genetic disorder that causes progressive, irreversible muscular damage and degeneration.

DMD impacts patients and caregivers as the disease progresses.

Increasing health state severity was related to decreasing physical function and independent action, such as the ability to walk, run and jump.

Decreasing physical function and independent action impacted:

- Individuals' ability to take part in daily and social activities, and their emotional wellbeing
- Increased levels of care and emotional burden
- Health-related quality of life (HRQoL) of individuals with nmDMD and their caregivers

Interviews were conducted with caregivers of individuals with nonsense mutation DMD (nmDMD) across three of the health states:



- 1 Early Ambulatory**
- Can stand from supine
 - Can walk/run 10 metres

"At the end of a school day...he'd be really exhausted, we'd be carrying him, he wouldn't really be in a state to walk...he'd be really tired, he'd be complaining that his legs were hurting"



- 2 Late Ambulatory**
- Can walk/run 10 metres

"We don't go to sports day anymore...they got him involved with helping to manage it but he was still just, "This is just a big memory of what I can't do and I don't want to be here"



- 3 Transfers**
- Can remain standing and bear weight



"He doesn't get up independently now"

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