

# GENETIC TESTING TO IDENTIFY DYSTROPHIN MUTATIONS IN DUCHENNE MUSCULAR DYSTROPHY (DMD)

Genetic testing and identification of mutations in DMD is important because it allows for genetic counselling for families and access to standards of care

First stage testing only detects large mutations (deletion or duplication) in the dystrophin gene. If you suspect DMD, a negative first stage test should always be followed up by full genetic sequencing to detect small / single nucleotide mutations (such as nonsense mutations, splice site mutations, small insertions or deletions)

The Medical Genetics Unit at the University Hospital of Ferrara (Italy) offers a service supported by PTC Therapeutics to provide full gene sequence testing for patients with a suspected diagnosis of DMD

*If you are interested in making use of the service, for full details and how it works please contact:*

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