# Disease translation in DMD: Neuromuscular rare disease translational research in patients with DMD.

# Disease translation in DMD

## Hub Summary

This study is designed to study a number of genes considered to be modifiers for DMD. This translational research will identify and obtain DNA samples and clinical information from 400 cases with DMD. This data will then be grouped into clinically and genetically defined groups. The DNA of the participants will be analysed and correlated to motor performance, age at loss of ambulation, severity of respiratory failure and severity of cardiac impairment.

## Study Number: Not on clinicaltrials.gov

## Description by Great Ormond Street Hospital NHS Foundation Trust

Genetic disease modifiers have been recently described in DMD, whilst other have been identified but not validated yet. The expression of these genes in subjects with DMD and the individual genetic profile appears to determine the severity of clinical phenotype and response to treatment with steroids.

In this study we aim to study a number of genes considered to be modifiers for DMD. To pursue our objective we will identify and obtain DNA samples and clinical information from 400 cases with DMD (300 children and 100 adults) and will stratify them into clinically and genetically defined groups. The participants' DNA will be analysed by Single Nucleotide Polymorphism (SNP) profiling and correlated to motor performance, age at loss of ambulation, severity of respiratory failure and severity of cardiac impairment.

## Can I take part?

For contact details and to find out more, please refer to dmdhub.org.

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dmdhub.org

**Trial Status**
Recruiting

**UK Locations**
London - GOSH, Recruiting, Alder Hey, Fully recruited, Birmingham, Fully recruited, Bristol, Recruiting, Cambridge, Recruiting, Newcastle, Recruiting, Oswestry, Recruiting

**Trial Sponsor**
Great Ormond Street Hospital NHS Foundation Trust

**Age**
>5 years old

**Mutation Specific**
Non-mutation specific therapies

**Muscle Biopsy**
No Muscle Biopsy Required

**MRI**
No

**Phase**
Non-therapeutic

**Recruitment Target**
300 children, 100 adults