What is Duchenne Muscular Dystrophy (DMD)?
DMD is a Neuromuscular disease caused by mutations in the dystrophin gene.

What is Gene therapy?
Use of viral vectors to deliver functional genes to muscle
- Adeno associated viral vector (AAV) is very safe and has a good tropism for human
- Dystrophin gene is too large for AAV
  → Dystrophin ‘microgenes’ were designed to exclude domains demonstrated to be dispensable for the functionality of the protein
  → Challenges to be faced:
    - Can ‘microgenes’ be delivered to all fibres in a muscle?
    - Can ‘microgenes’ be delivered bodywide?
    - Do ‘microgenes’ improve muscle strength?

Full length dystrophin gene

Microgene for dystrophin

Virus production

5'ITR Promoter Dystrophin pA 3'ITR

Transfection

AAV infection:

Virus purification

Tibialis Anterior (shin) muscles injected with AAV vector expressing microdystrophin