Genomics Education Programme

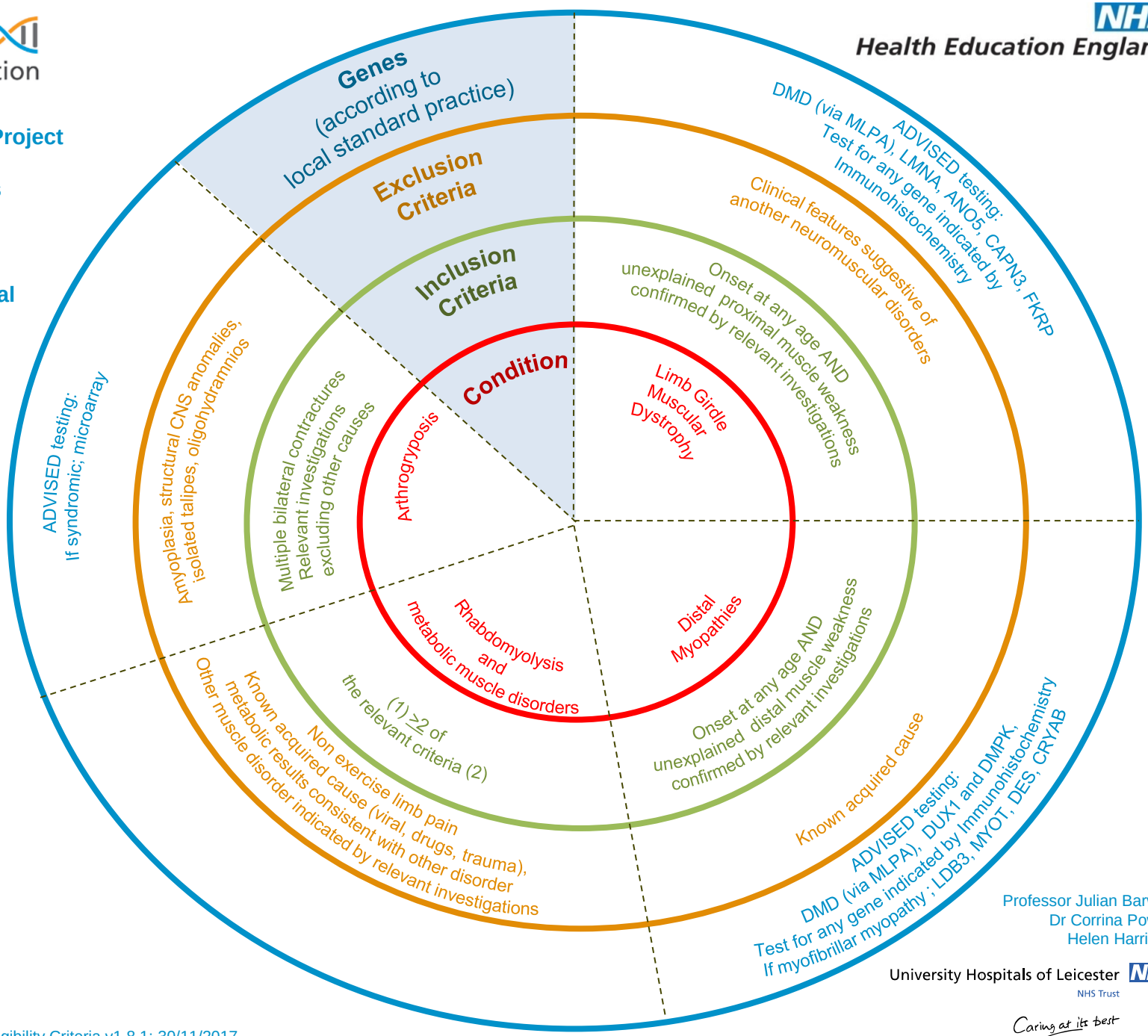
NHS Health Education England

100,000 Genomes Project Eligibility wheels A tool for clinicians

Rare Diseases: Neurology and Neurodevelopmental disorders – Neuromuscular disorders #2

(1)
Recruitment should NOT include unaffected relatives unless part of a TRIO/DUO. Multiple affected relatives are preferable to singleton cases.
(2)

- Symptom triggers: exercise, fasting, fever and heat
- Variable CK with exacerbations (>10x)
- Myoglobinuria
- Treatment for acute rhabdomyolysis +/ - acute renal failure
- Progressive muscle weakness
- Muscle pain with atrophy and weakness
- Malignant hyperpyrexia syndrome
- Neuroleptic Malignant syndrome
- Acute compartment Syndrome



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Caring at its best