Genomics Education Programme

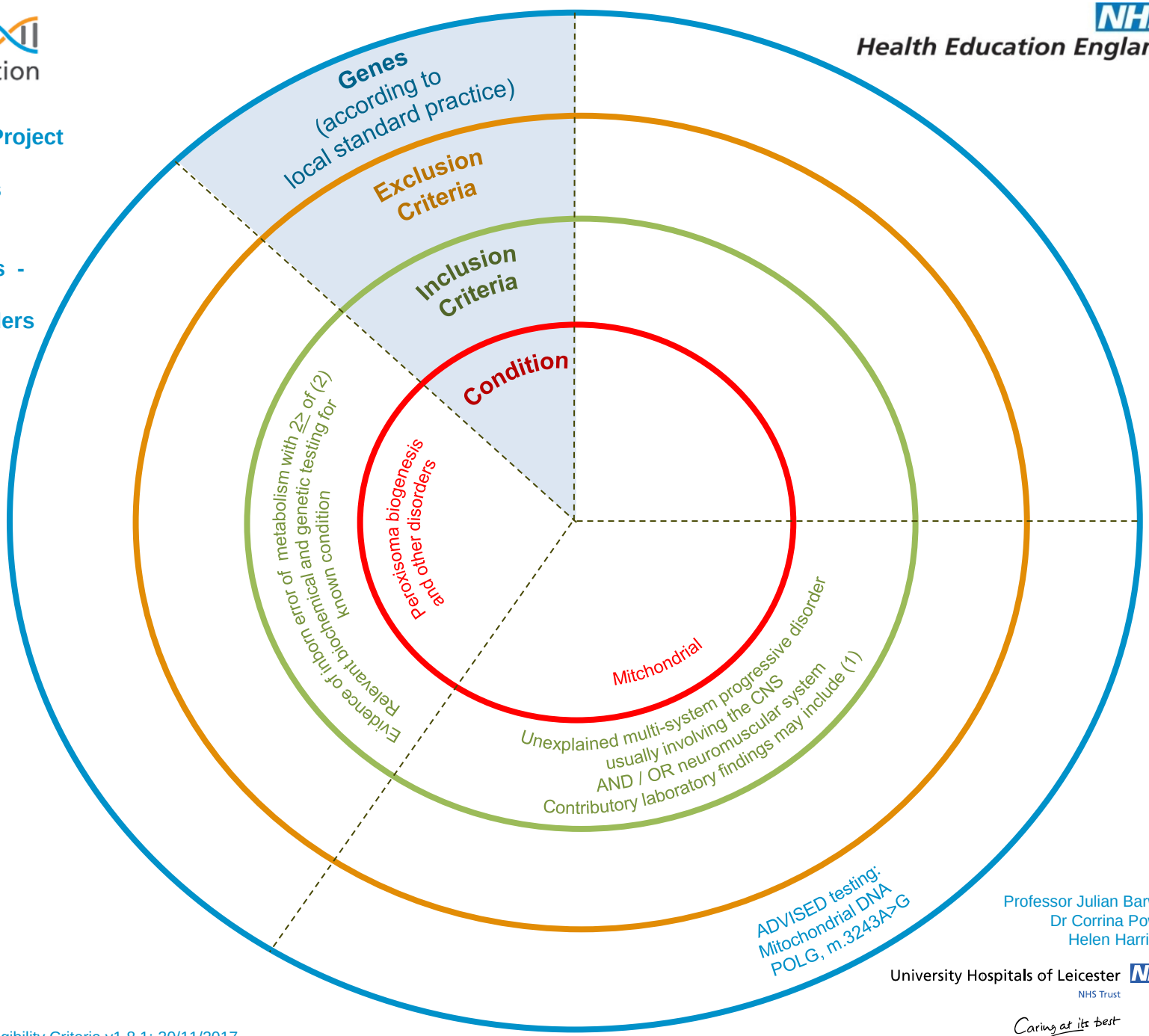
NHS
Health Education England

100,000 Genomes Project Eligibility wheels A tool for clinicians

Rare Diseases: Metabolic disorders - Mitochondrial and Peroxisomal disorders

(1)
- Characteristic brain MR imaging.
- Raised serum, CSF or urinary organic acid biomarkers
- Muscle biopsy diagnostic of mitochondrial dysfunction inc histochemical (COX-deficient fibres, ragged-red fibres) and biochemical (respiratory chain enzyme deficiencies) markers of disease pathology

(2)
- Clinical presentation
- Biochemical
- Haematological
- Radiological



Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS
NHS Trust
Caring at its best