Genomics Education Programme

NHS
Health Education England

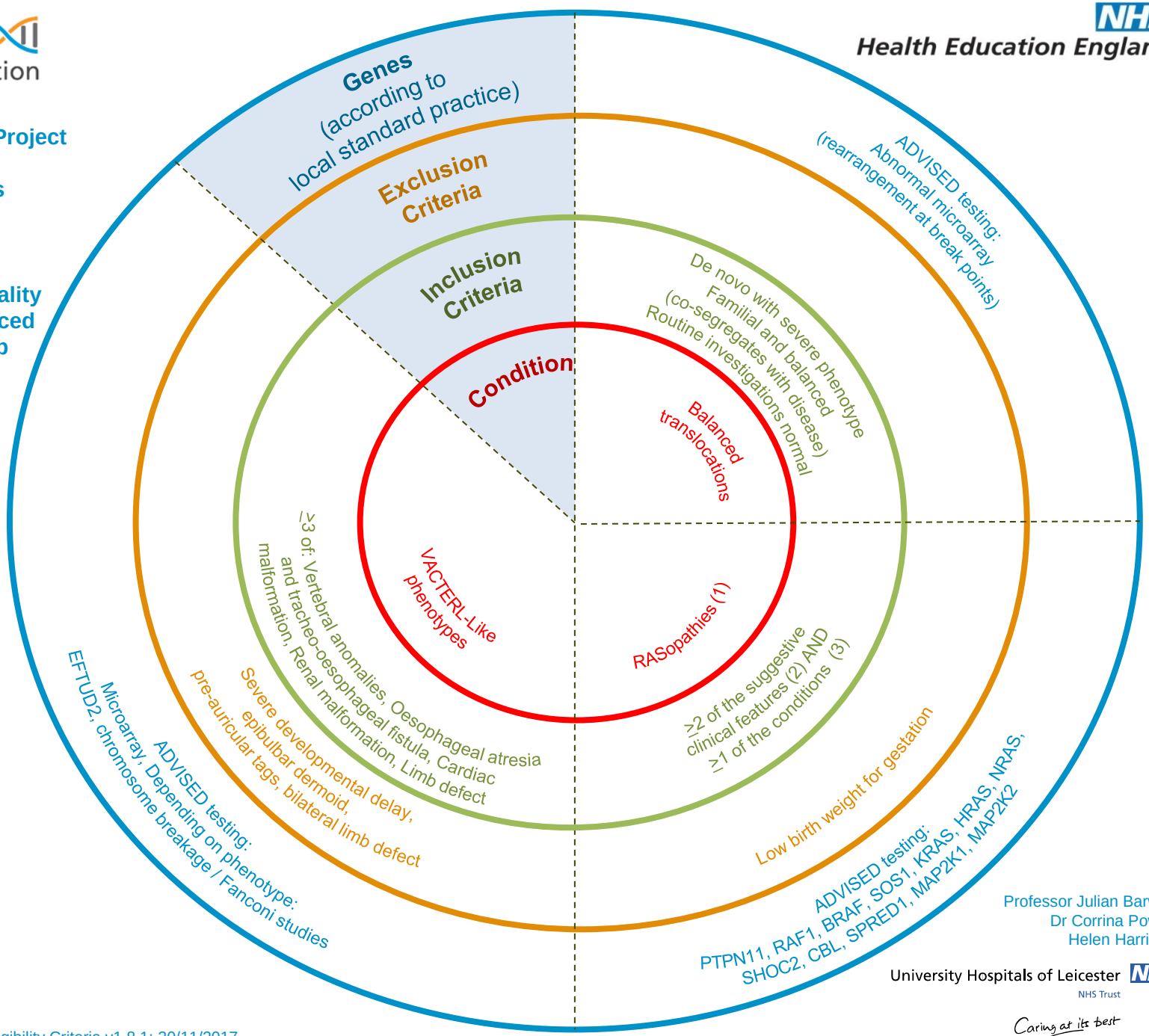
100,000 Genomes Project Eligibility wheels
A tool for clinicians

Rare Diseases: Dysmorphic and congenital abnormality syndromes - Balanced translocations, limb disorders and RASopathies

(1)
- Noonan syndrome
- Noonan syndrome plus other features
- Cardio-facio-cutaneous syndrome
- LEOPARD syndrome
- Costello syndrome
- Legius syndrome

(2)
- Early feeding difficulty/ failure to thrive
- Relative macrocephaly
- Short stature
- Developmental disability

(3)
- Cardiomyopathy
- Congenital heart disease
- Arrhythmia
- Malignancy:
  - Bladder carcinoma
  - Rhabdomyosarcoma
  - Leukaemia
  - Phaeochromocytoma
- Skin abnormalities:
  - Hyperkeratosis
  - Café au lait patches
  - Ulerythema oophorogenes
  - Keratosis pilaris
  - Excess palmar skin



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Caring at its best