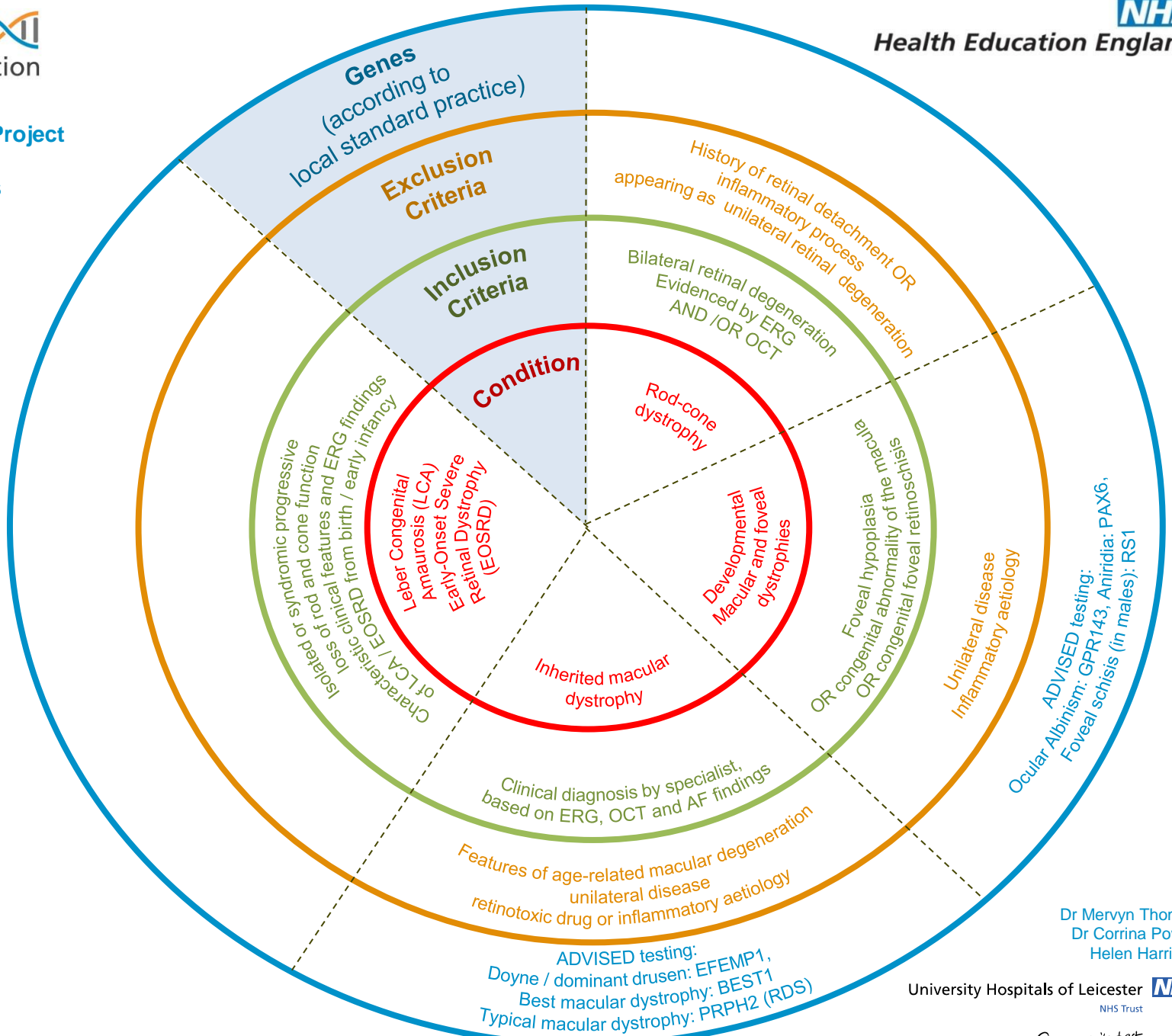




Genomics Education Programme

100,000 Genomes Project Eligibility wheels
A tool for clinicians

Rare Diseases: Ophthalmological disorders - Posterior segment abnormalities #2



ADVISED testing:
Ocular Albinism: GPR143, Aniridia: PAX6,
Foveal schisis (in males): RS1

ADVISED testing:
Doyme / dominant drusen: EFEMP1,
Best macular dystrophy: BEST1
Typical macular dystrophy: PRPH2 (RDS)

Dr Mervyn Thomas
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester **NHS**
NHS Trust

Caring at its best