NHS
Health Education England

Genomics Education Programme

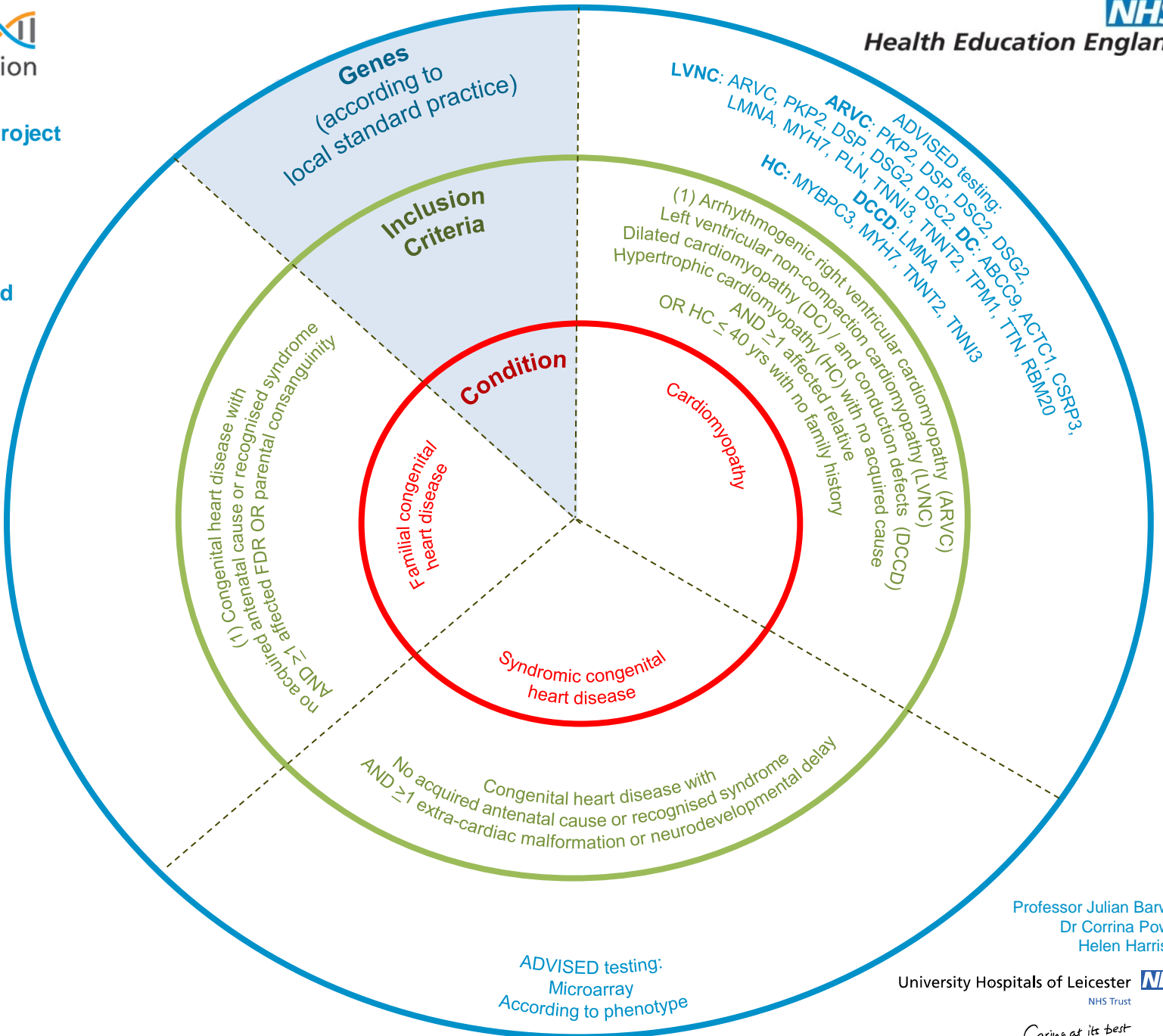**100,000 Genomes Project Eligibility wheels A tool for clinicians**

Rare Diseases: Cardiology - Cardiomyopathy and Congenital heart disease

(1)
Recruitment should NOT include unaffected relatives except in severe / syndromic cases. Multiple affected relatives are preferable to singleton cases.

Genes (according to local standard practice)

Inclusion Criteria

Condition

LVNC: ARVC, PKP2, DSP, DSG2, DSC2, DSG2,
ARVC: PKP2, DSP, DSG2, DSC2. **DC**: ABCC9, ACTC1, CSRP3,
LMNA, MYH7, PLN, TNNI3, TNNT2, TPM1, TTN, RBM20
HC: MYBPC3, MYH7, TNNT2, TNNI3
DCCD: LMNA
ADVISED testing:

(1) Arrhythmogenic right ventricular cardiomyopathy (ARVC) /
Left ventricular non-compaction cardiomyopathy (LVNC) /
Dilated cardiomyopathy (DC) / and conduction defects (DCCD)
Hypertrophic cardiomyopathy (HC) with no acquired relative
AND ≥1 affected relative
OR HC ≤ 40 yrs with no family history

Cardiomyopathy

(1) Congenital heart disease with
no acquired antenatal cause or recognised syndrome
AND ≥1 affected FDR OR parental consanguinity

Familial congenital heart disease

Syndromic congenital heart disease

Congenital heart disease with
No acquired antenatal cause or recognised syndrome
AND ≥1 extra-cardiac malformation or neurodevelopmental delay

ADVISED testing:
Microarray
According to phenotype

Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester **NHS**
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