Genomics Education Programme

NHS
Health Education England

100,000 Genomes Project
Eligibility wheels
A tool for clinicians

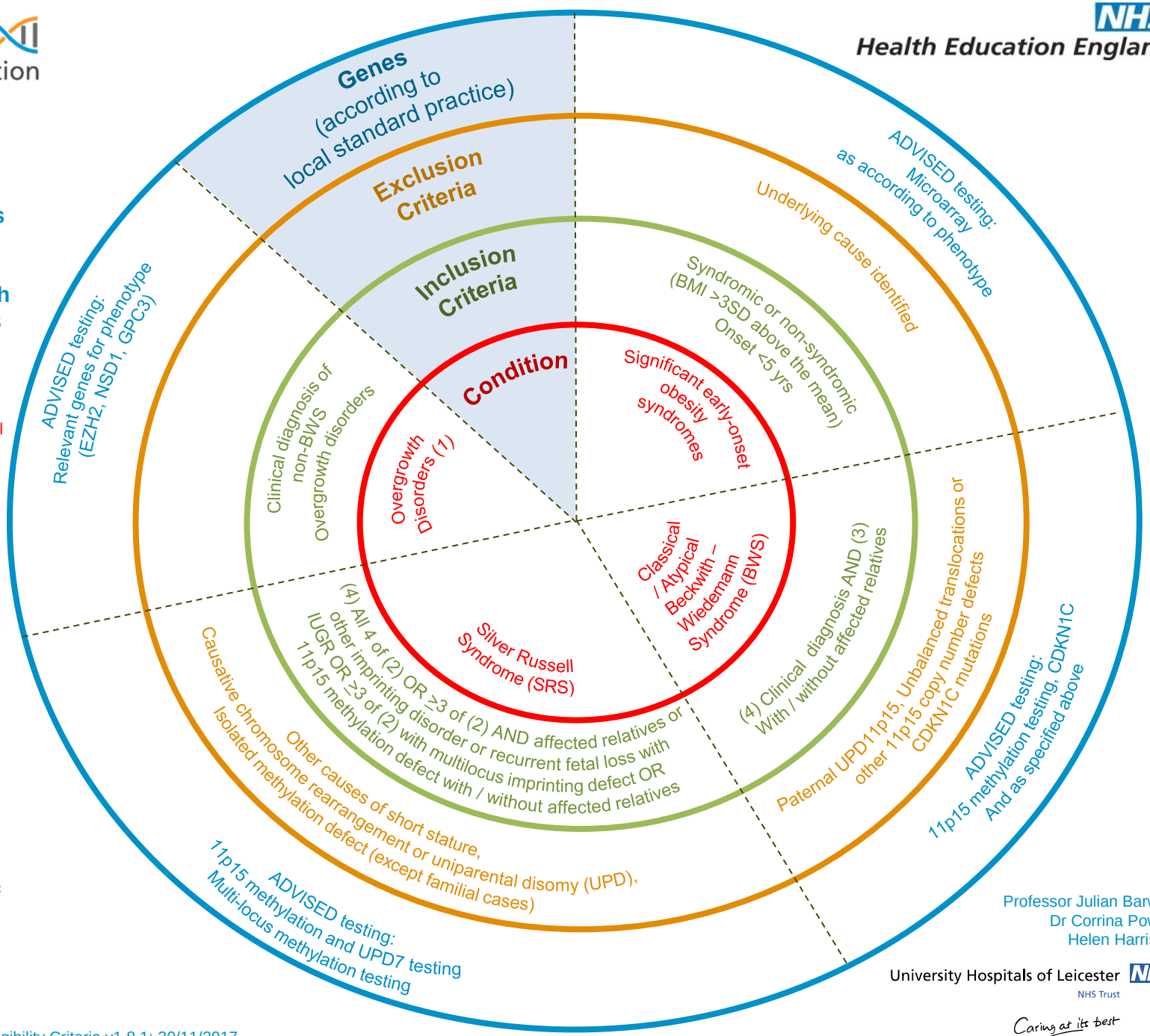
Rare Diseases: Endocrine – Growth hormone disorders and obesity syndromes

(1)
- Simpson - Golabi-Behmel Syndrome
- Sotos Syndrome
- Weaver Syndrome

(2)
Clinical features of SRS:
- Small for gestational age with or without IUGR
- Post-natal short stature
- Body asymmetry
- Marked feeding difficulties in infancy / childhood with or without poor growth

(3)
- Multilocus methylation defect (in trans imprinting defect) OR
- Balanced chromosome aberration OR
- 11p15 methylation defect
- No detectable cause

(4)
Recruitment should NOT include unaffected relatives except in severe / syndromic cases. Multiple affected relatives are preferable to singleton cases



Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS
NHS Trust
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