Genomics Education Programme

NHS
Health Education England

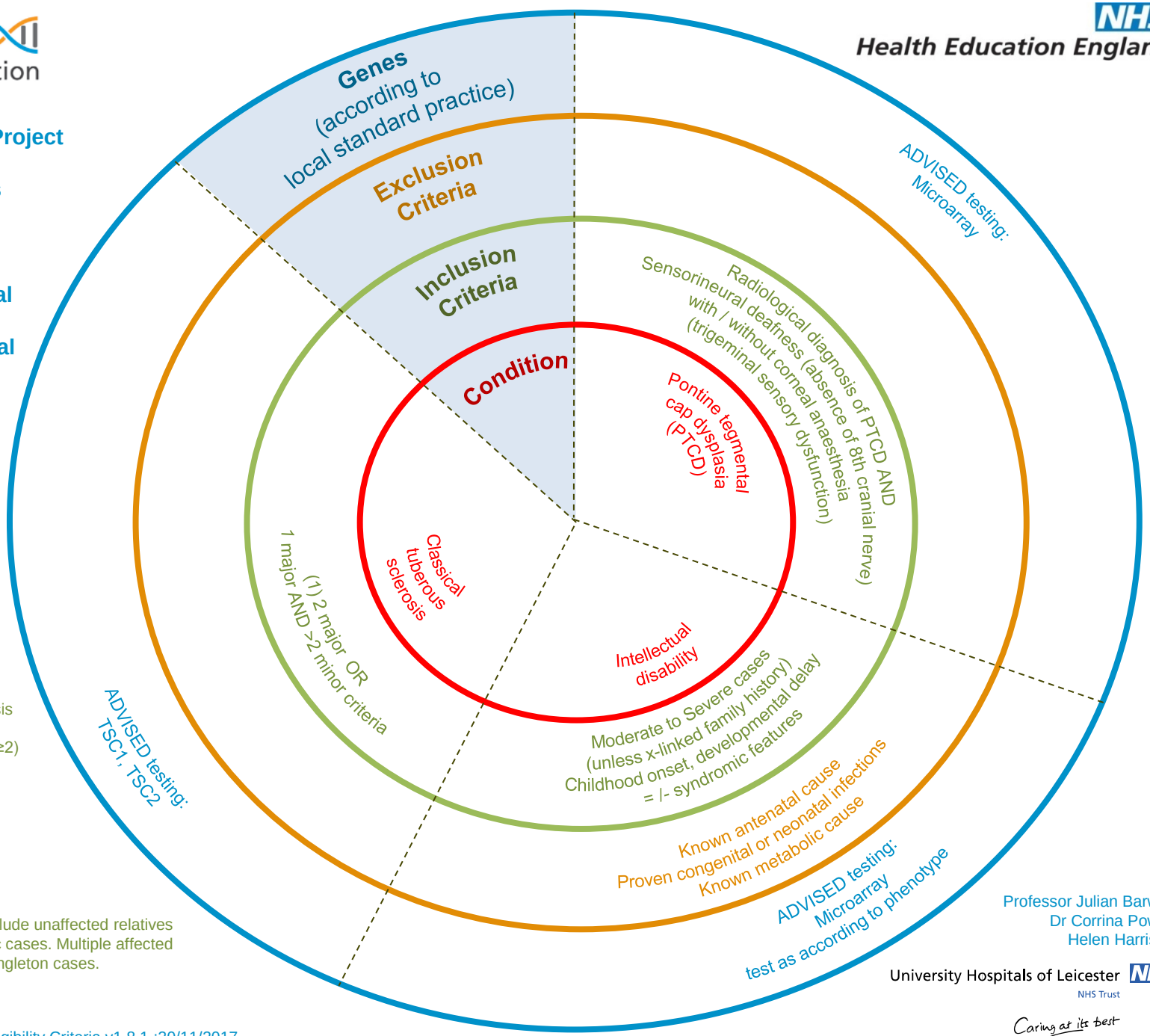
# 100,000 Genomes Project Eligibility wheels A tool for clinicians

## Rare Diseases: Neurology and neurodevelopmental disorders - Neurodevelopmental disorders #1

(1)
Major criteria
- Hypomelanotic macules (≥3 at least 5mm)
- Angiofibromas (≥3) or fibrous cephalic plaque
- Ungual fibromas (≥2)
- Shagreen patch
- Multiple retinal hamartomas
- Cortical dysplasias
- Subependymal nodules
- Subependymal giant cell astrocytoma
- Cardiac rhabdomyoma
- Lymphangioleiomyomatosis (LAM)
- Renal angiomyolipomas (≥2)

Minor criteria
- "Confetti" skin lesions
- Dental enamel pits (>3)
- Intraoral fibromas (≥2)
- Retinal achromic patch
- Multiple renal cysts
- Nonrenal hamartomas

(2)
Recruitment should NOT include unaffected relatives except in severe / syndromic cases. Multiple affected relatives are preferable to singleton cases.





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