Genomics Education Programme

NHS
Health Education England

100,000 Genomes Project Eligibility wheels A tool for clinicians

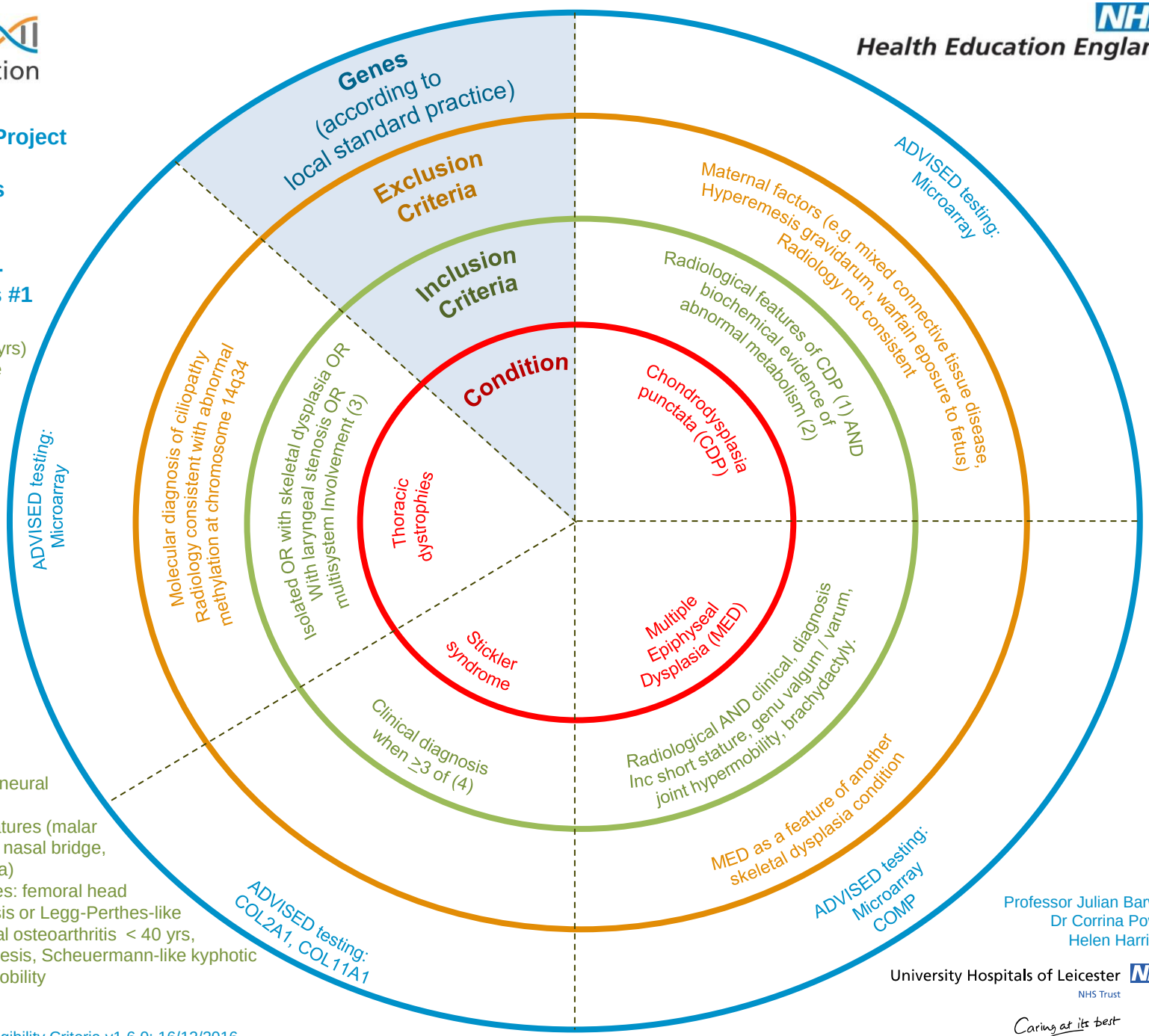
Rare Diseases: Skeletal disorders - Skeletal dysplasias #1

(1)
- Stippling (resolves ≤ 2 yrs)
- Rhizomelia, short tibiae and metacarpals
- Vertebral body clefting (remnants)

(2)
- VLCFA profile
- Abnormal cholesterol biosynthesis/ arylsulphatase E

(3)
- Renal cysts
- Hepatic cysts
- Retinal dystrophy
- Laterality defect
- Polydactyly (>1 limb)

4)
- Cleft palate
- Characteristic retinal or vitreous changes
- High frequency sensorineural hearing loss
- Characteristic facial features (malar hypoplasia, broad / flat nasal bridge, and micro / retrognathia)
- Musculoskeletal features: femoral head failure (slipped epiphysis or Legg-Perthes-like disease), radiographical osteoarthritis < 40 yrs, scoliosis, spondylolisthesis, Scheuermann-like kyphotic deformity, joint hypermobility



Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS
NHS Trust
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