Genomics Education Programme

NHS
Health Education England

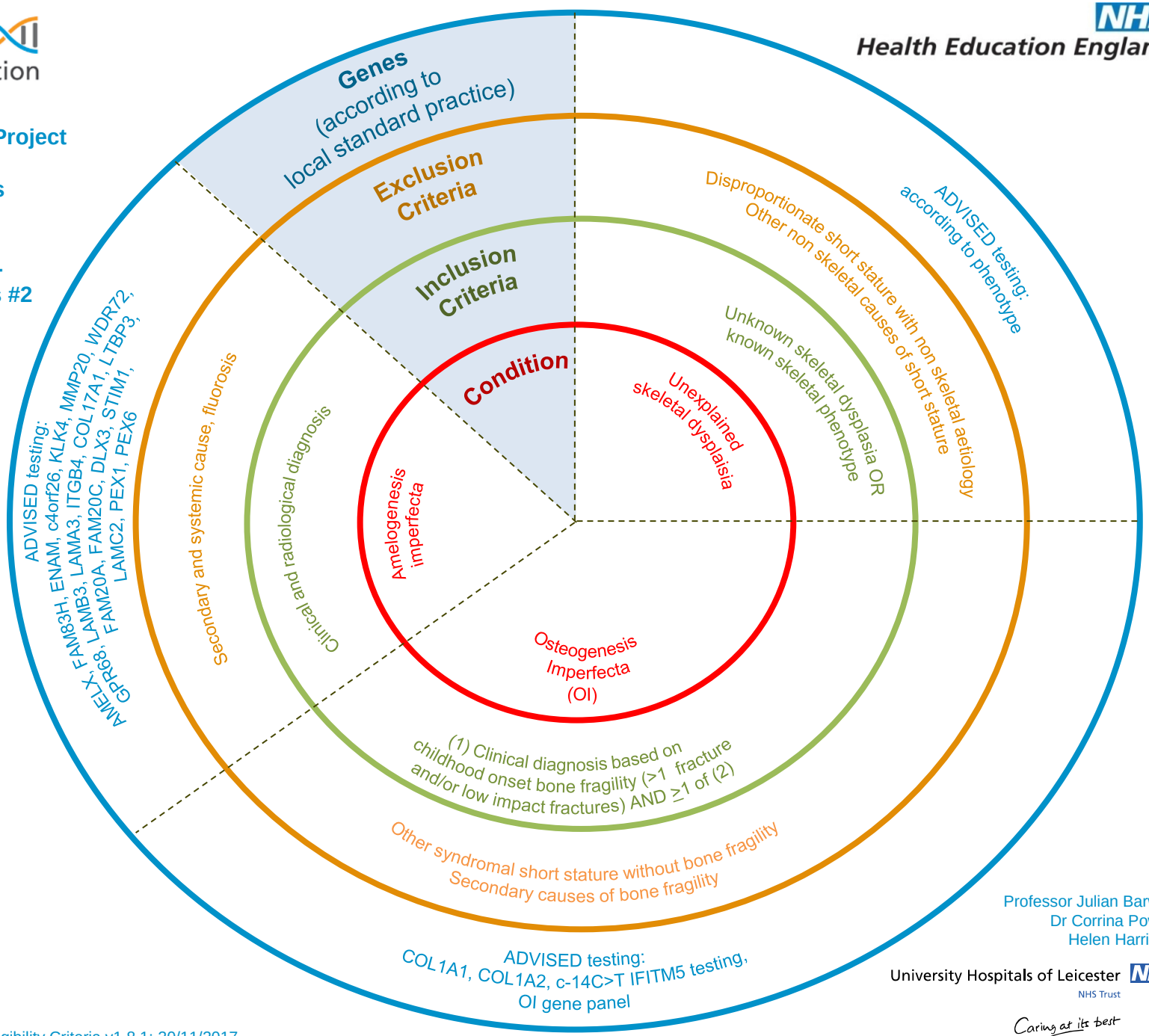
# 100,000 Genomes Project
# Eligibility wheels
# A tool for clinicians

## Rare Diseases:
## Skeletal disorders -
## Skeletal dysplasias #2

(1)
If no previous genetic testing, advise recruitment as singleton. However, if previous genetic testing was normal then recruit with affected relatives

(2)

- Short stature (no other syndromic cause)
- Dentinogenesis Imperfecta
- Blue sclerae
- Joint hypermobility
- Deafness
- Facial Dysmorphism characteristic of OI / bone fragility
- Unexplained joint contractures





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Caring at its best