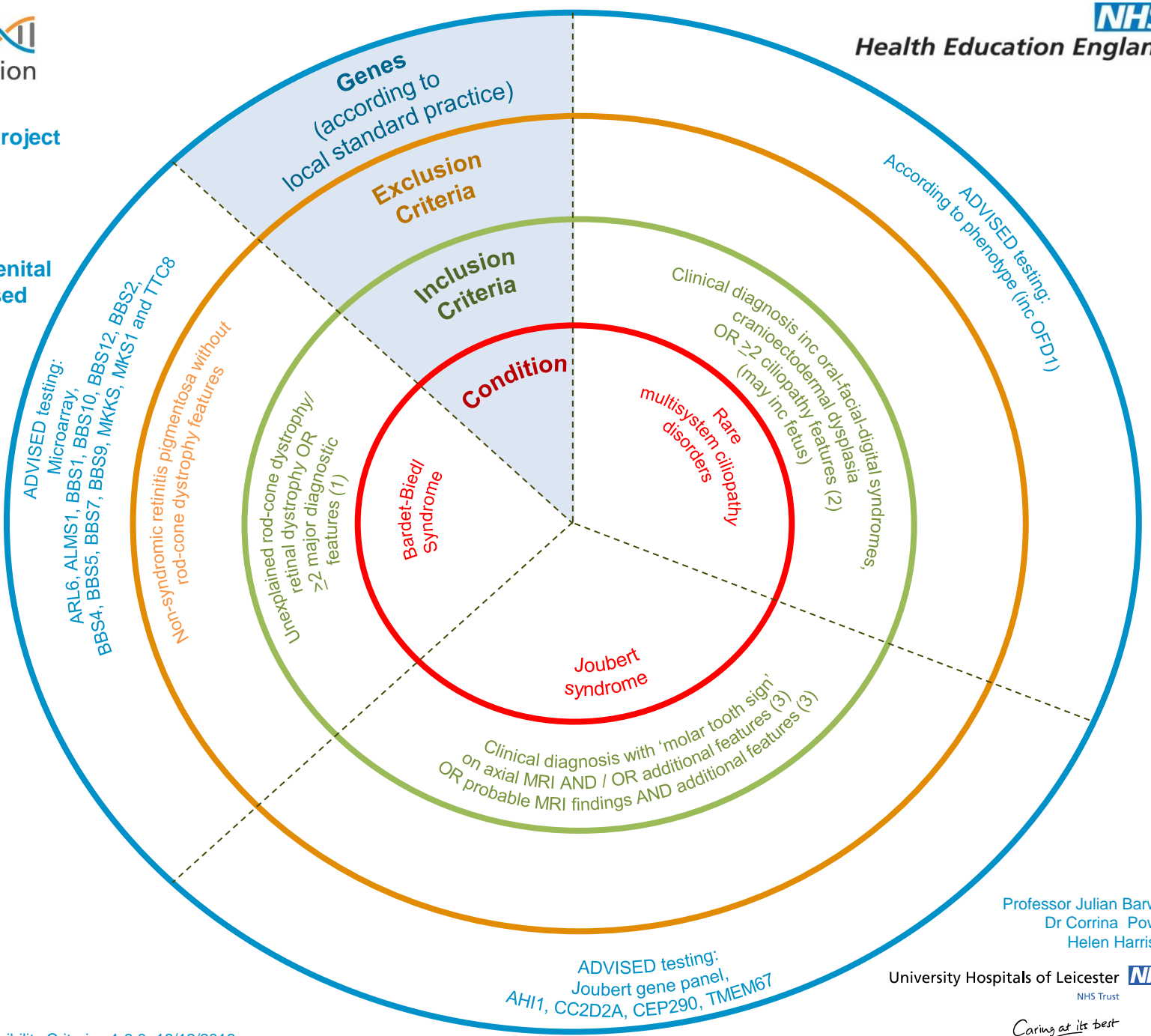




Rare Diseases: Ciliopathies - Congenital malformations caused by ciliopathies

- (1)
 - Obesity
 - Polydactyly
 - Rod-cone dystrophy/ retinal dystrophy / retinitis pigmentosa
 - Hypogenitalism
 - Renal dysplasia
- (2)
 - Polycystic or 'bright' kidneys
 - Cerebellar hypoplasia (especially vermis where visible) / Dandy-Walker malformation or variant
 - Polydactyly
 - Short limbs and/or short ribs
 - Occipital encephalocele
 - Retinal dystrophy
- (3)
 - Polydactyly
 - Renal cysts
 - Retinal dystrophy
 - Oculomotor apraxia
 - Characteristic breathing abnormality



Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS Trust

Caring at its best