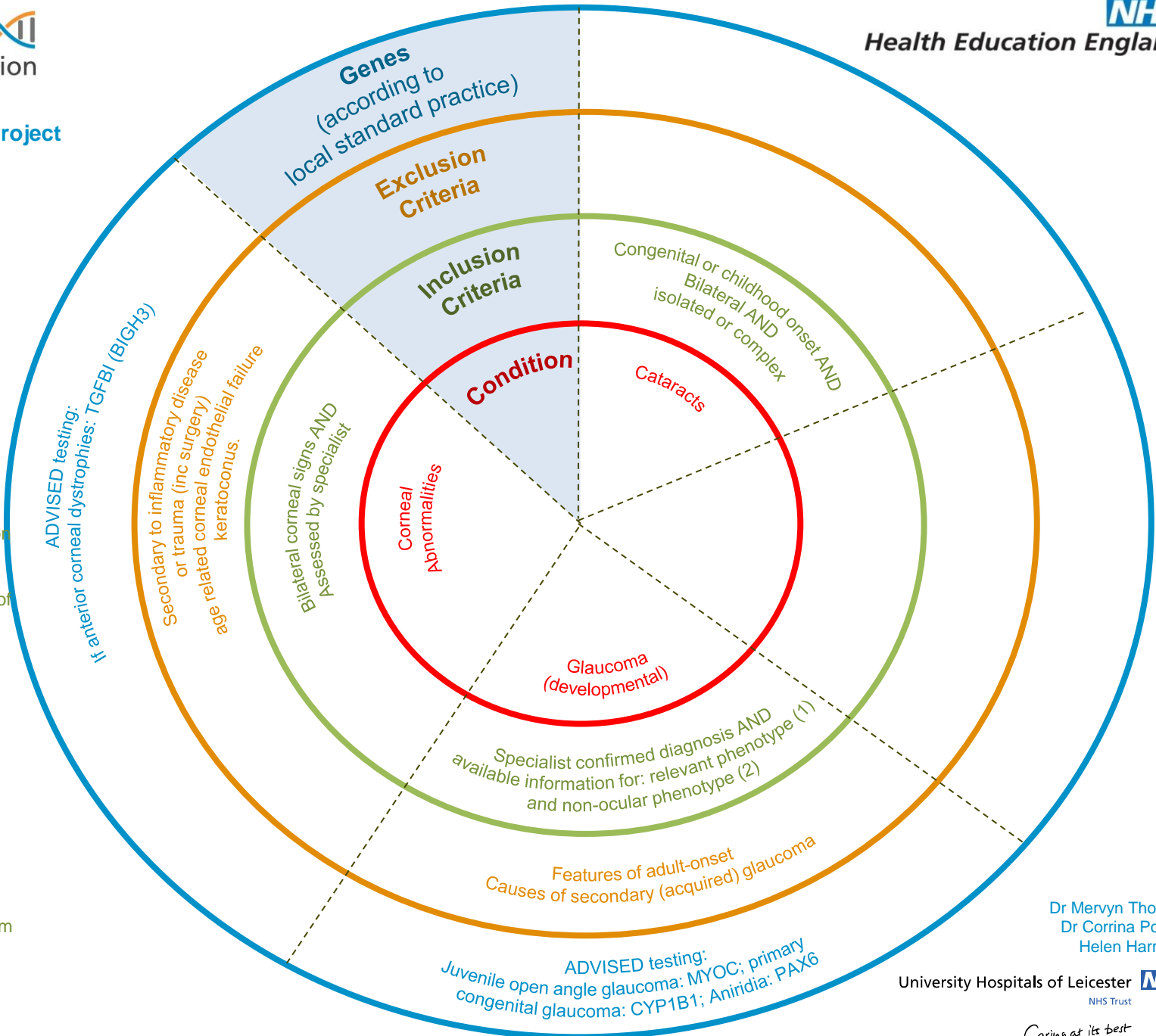




Rare Diseases: Ophthalmological disorders - Anterior segment abnormalities

- (1) Relevant phenotypic information:
- Structural assessment of anterior segment
 - Optic nerve head characteristics
 - IOP at first presentation
 - Highest recorded IOP (where available)
 - Slit lamp assessment of anterior segment
 - Pachymetry
 - ERG and VEP (if appropriate)

- (2) Non-ocular phenotypic information
- Presence/absence of maxillary hypoplasia
 - Umbilical anomaly
 - Cardiac anomalies (from history)
 - Growth delay
 - Dental anomalies
 - Metabolic disease (from history)



Dr Mervyn Thomas
Dr Corrina Powell
Helen Harrison