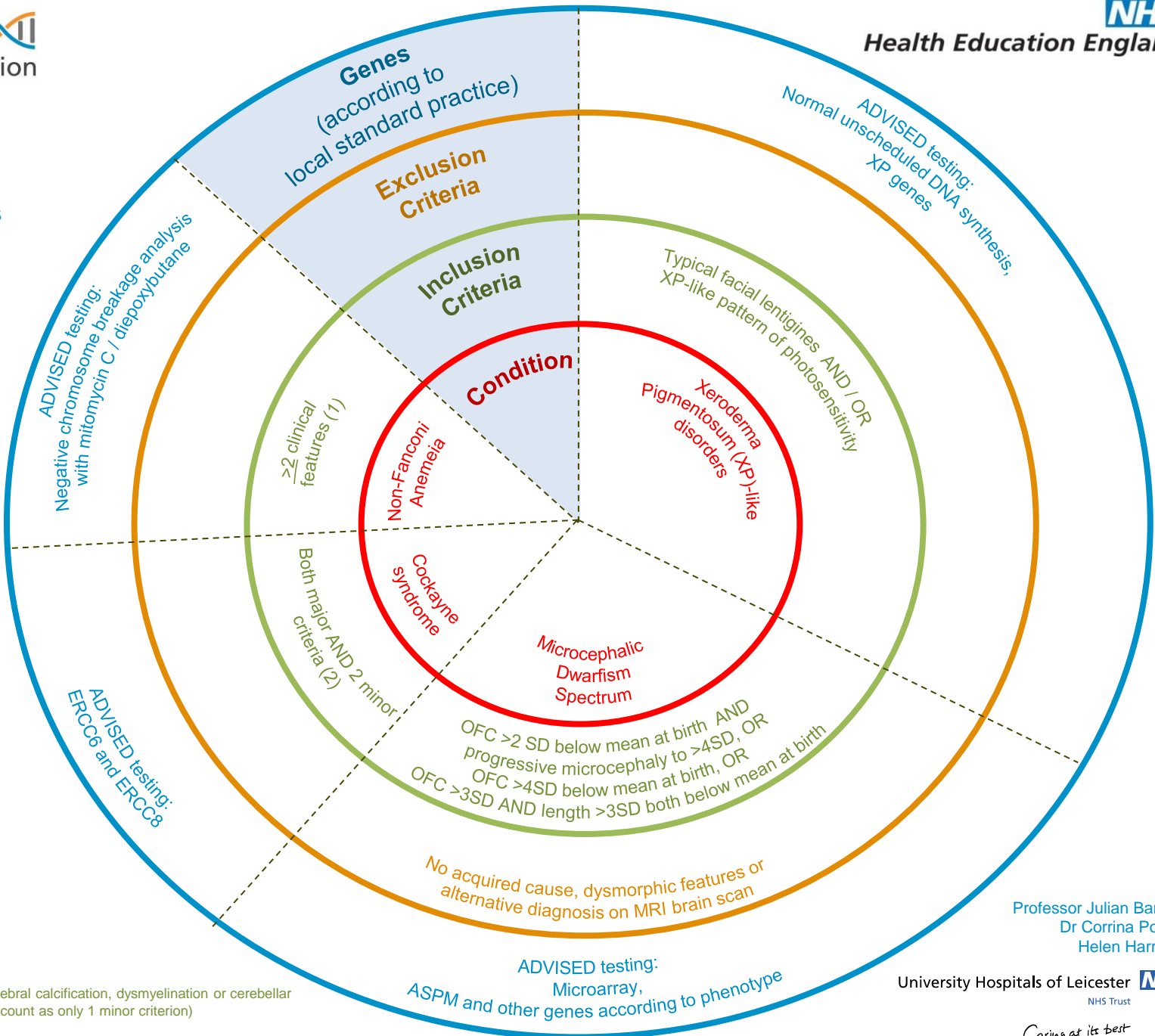




- (1)
Clinical features:
- Skin:
 - hyperpigmentation
 - Cafe au lait spots
 - Hypopigmentation
 - Microsomia:
 - short stature
 - Upper limbs:
 - Absent / hypoplastic thumbs or radii
 - Head and face:
 - Microcephaly
 - micrognathia
 - Hypogonitalia in males or females
 - Renal:
 - Ectopic or pelvic
 - abnormal, horseshoe
 - Haematology:
 - Unexplained cytopenia

- (2)
Major criteria (pre- or post-natal onset):
- Microcephaly
 - Small stature
- Minor criteria:
- Persistently cold peripheries
 - Tremor
 - Joint contractures
 - Progressive loss of body fat
 - Cataracts
 - Enophthalmia
 - Brain imaging abnormality: cerebral calcification, dysmyelination or cerebellar hypoplasia (if >1 present, still count as only 1 minor criterion)



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Caring at its best