

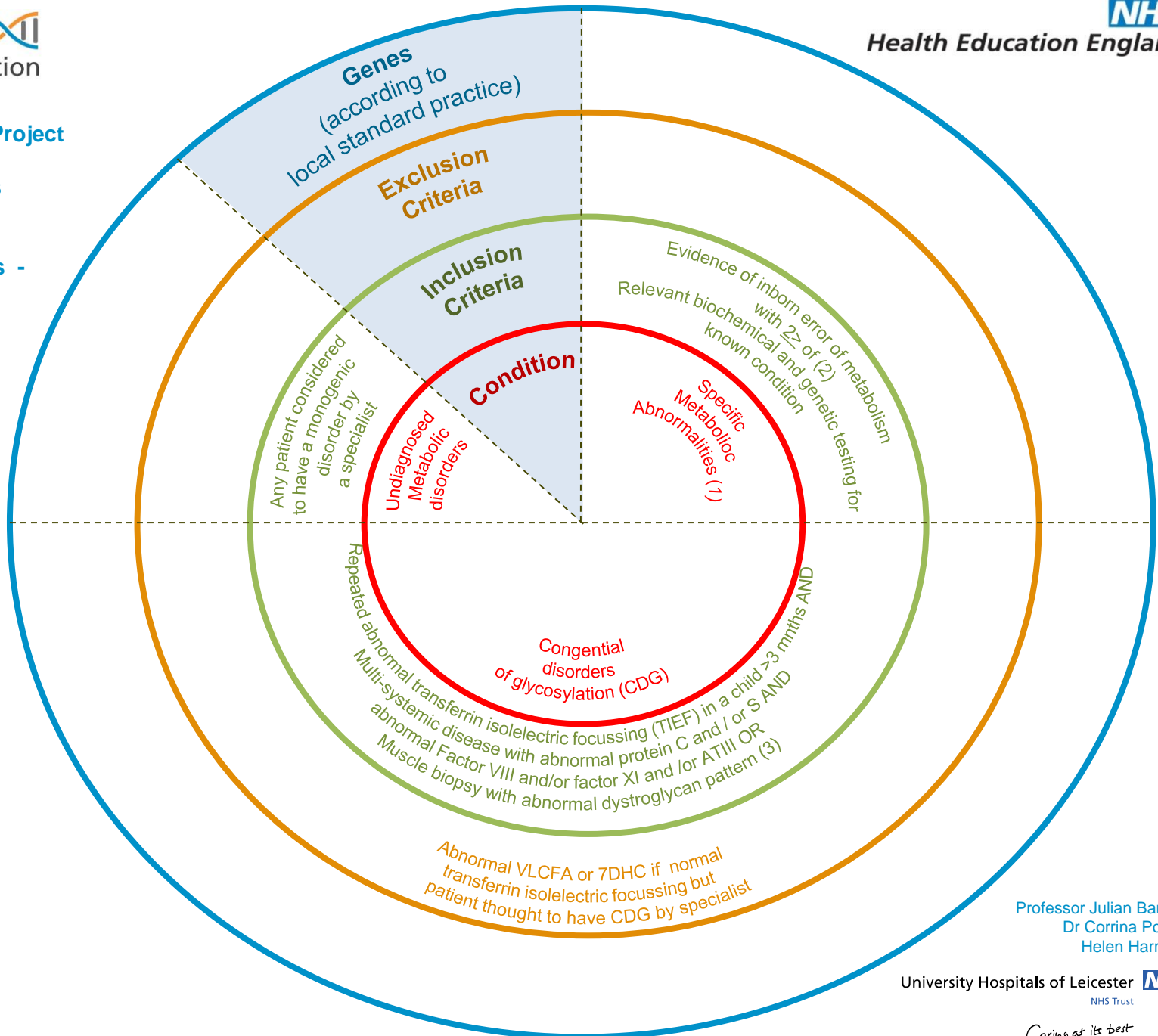


Rare Diseases: Metabolic disorders - Specific metabolic abnormalities

- (1)
 - Ketotic hypoglycaemia
 - Lactic acidosis
 - Cerebral folate deficiency

- (2)
 - Clinical presentation
 - Biochemical
 - Haematological
 - Radiological

(3)
 If TIEF abnormal: confirm by second sample and exclude PMM2 deficiency
 If TIEF normal: the following should be normal; 7DHC, VLCFA, protein C and S, factor VIII and XI and ATIII, T4 and TSH.
 If suspect isolated O glycosylation or combined N- and O-glycosylation defect: ApoC III



Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS Trust

Caring at its best