Health Education England Genes local standard practice) (according to **Genomics Education Programme** ADVISED TO SITTO 100,000 Genomes Project Exclusion **Eligibility wheels** Criteria A tool for clinicians Radiological son and dysfunction)

Ratiological son and dysfunction)

Ratiological son and dysfunction)

Sensorine user a with our cannot dysfunction with its gentinal sensory dysfunction. Inclusion **Rare Diseases: Neurology and** Criteria neurodevelopmental disorders -Condition **Neurodevelopmental** disorders #1 Major criteria Hypomelanotic macules (≥3 at least 5mm) • Angiofibromas (≥3) or fibrous cephalic plaque 1 major AMD 72 minor cities Unqual fibromas (≥2) Classico · Shagreen patch sclerosis Multiple retinal hamartomas · Cortical dysplasias Intellectual Subependymal nodules .vioderate to Severe case of delay

.vioderate to Severe case of delay

(unless x-linked family heatings

(unless x-linked developmentalities

Childhood onset, developmentalities Moderate to Severe rate of A Subependymal giant cell astrocytoma ADVISED TOCK · Cardiac rhabdomyoma Lymphangioleiomyomatosis Known antenatal cause teathors

Known antenatal or neonatal incause

Known more hour cause

Known more hour cause (LAM) Renal angiomyolipomas (≥2) Minor criteria · "Confetti" skin lesions Dental enamel pits (>3) Intraoral fibromas (≥2) ADVISED resting test as according to phenotype Retinal achromic patch · Multiple renal cysts Nonrenal hamartomas Professor Julian Barwell (2)Dr Corrina Powell Recruitment should NOT include unaffected relatives Helen Harrison except in severe / syndromic cases. Multiple affected relatives are preferable to singleton cases. University Hospitals of Leicester NHS

Caring at its best