Health Education England Genes (according to local standard practice) Genomics Education ADVISED testing. TOP 74 **Programme** 100,000 Genomes Project Exclusion Underlying cause identified **Eligibility wheels** Criteria A tool for clinicians Affecting any body onset <31 yrs Op Segment Usland Presponsite Glady Spring Resident Spring Re Inclusion **Rare Diseases: Neurology** Criteria Motor disorders of the **CNS #1** Common trinucleotide repeat disorders.
ATXN2, ATXN3, CACNA1A, ATXN2, P.T.YN1, F.Y.Y. (recessive history), F.M.R.1
P.Y.N. F.XN (recessive history), F.M.R.1
P.Y.N. F.XN (recessive history), F.M.R.1 Condition (1) inflammatory lesions on brain MRy youral of ""s, malignancy, alcohology, promise a property of the property of t Recruitment should NOT include (1) Unexplained cerebellar unaffected relatives except in ADVISED testing: ataxia for >2 yrswith / without (2) severe/syndromic Hereditary cases. Multiple ataxia affected relatives are preferable to singleton cases. Spasticity Structural or it Peripheral neuropathy Cerebellar Bulbar dysfunction hypoplasia Cerebellar or ponto-cerebellar or ponto-cerebe rebellar or ponto-cereberah hypoplasia on MRI brain scan Findings characteristic of Joubert syndrome, evidence of courses a constant infection ungs characteristic of Joubert syndrum evidence of causative prenatal infection ADVISED testing:
ADVISED testing:
Gene panel: ITPR1, SPTBN2, KCNC3, CASK, ORINY Professor Julian Barwell Dr Corrina Powell Helen Harrison University Hospitals of Leicester NHS

Caring at its best