Neurology reference document: Test Directory indications

NHS North West
NHS Genomic Laboratory Hub

Please locate your R code and complete the relevant documents to activate testing.
For any cascade/familial/prenatal testing, please use the standard referral form.

R54: Hereditary ataxia with onset in adulthood	WGS referral forms
R55: Hereditary ataxia with onset in childhood	WGS referral forms
R56: Adult onset dystonia/movement disorder	WGS referral forms
R57: Childhood onset dystonia/movement disorder	WGS referral forms
R58: Adult onset neurodegenerative disorder	WGS referral forms
R59: Early onset or syndromic epilepsy	WGS referral forms
R60: Adult onset hereditary spastic paraplegia	WGS referral forms
R61: Childhood onset hereditary spastic paraplegia	WGS referral forms
R62: Adult onset leukodystrophy	WGS referral forms
R66: Paroxysmal central nervous system disorders*	Standard referral form
R68: Huntington disease	Standard referral form
R70: Spinal muscular atrophy type 1 diagnostic test	Standard referral form
R71: Spinal muscular atrophy type 1 rare mutation testing	Standard referral form
R72: Myotonic dystrophy type 1	Standard referral form
R73: Duchenne/Becker muscular dystrophy (DMD/BMD)	Standard referral form
R74: Facioscapulohumeral muscular dystrophy	Standard referral form
R75: Oculopharyngeal muscular dystrophy	Standard referral form
R76: Skeletal muscle channelopathy*	Standard referral form
R77: Hereditary neuropathy – PMP22 copy number	Standard referral form
R78: Hereditary neuropathy – NOT PMP22 copy number	WGS referral forms
R79: Congenital muscular dystrophy**	Standard referral form + Viapath proforma
R80: Congenital myasthenic syndrome**	Standard referral form
R81: Congenital myopathy**	Standard referral form + Viapath proforma
R82: Limb girdle muscular dystrophy**	Standard referral form + Newcastle proforma
R83: Arthrogryposis	WGS referral forms
R84: Cerebellar anomalies	WGS referral forms
R85: Holoprosencephaly – NOT chromosomal	WGS referral forms
R86: Hydrocephalus	WGS referral forms
R87: Cerebral malformation	WGS referral forms
R88: Severe microcephaly	WGS referral forms
R109: Childhood onset leukodystrophy (super-panel)	WGS referral forms
R221: Familial tumours of the nervous system	Standard referral form
R222: Neurofibromatosis type 1	Standard referral form
R228: Tuberous sclerosis	Standard referral form
R252: SMA carrier testing at population risk for partners of known carriers	Standard referral form
R294: Ataxia telangiectasia – DNA repair testing	Standard referral form
R295: Ataxia telangiectasia – mutation testing	Standard referral form
R336: Cerebral vascular malformations	Standard referral form
R337: CADASIL	Standard referral form
R345: Facioscapulohumeral muscular dystrophy - extended testing	Standard referral form
R371: Malignant hyperthermia	Standard referral form
R376: Segmental/atypical neurofibromatosis type 1	Standard referral form
R378: Linkage testing for Duchenne or Becker muscular dystrophy	Standard referral form
R381: Other rare neuromuscular disorders (super-panel)	WGS referral forms
R383: Linkage testing for Huntington disease	Standard referral form
R410: Myotonic dystrophy type 2 (DM2)	Standard referral form
R419: Acute rhabdomyolysis*/**	Standard referral form

**Can be included as a secondary WGS indication alongside an appropriate primary WGS indication*
***Form part of the R381 super-panel and can therefore be requested as part of WGS testing via this clinical indication*