NE&Y GLH Rare Disease WGS Eligibility & Sample Requirements - 2021

NHSE will fund WGS analysis for rare disease patients for the specific clinical indications listed below.

Indications:

This table shows the eligible disease groups including their test code. The document published by NHSE entitled 'Rare and Inherited Disease Eligibility Criteria' sets out which patients should be considered for testing under that indication, the optimal family structure (singleton or trio) and the specialist clinicians who would be expected to request the test.

https://www.england.nhs.uk/wp-content/uploads/2018/08/Rare-and-inherited-disease-eligibility-criteria-2021-22-v2.pdf

Speciality Group	Test code	Clinical Indication
Endocrinology	R143.4	Neonatal diabetes
Metabolic	R98.2	Likely inborn error of metabolism - targeted testing not possible
Musculoskeletal	R104.3	Skeletal dysplasia
Musculoskeletal	R100.3	Rare syndromic craniosynostosis or isolated multisuture synostosis
Neurology	R54.3	Hereditary ataxia with onset in adulthood
Neurology	R55.4	Hereditary ataxia with onset in childhood
Neurology	R59.3	Early onset or syndromic epilepsy
Neurology	R61.4	Childhood onset hereditary spastic paraplegia
Neurology	R83.2	Arthrogryposis
Neurology	R381.2	Other rare neuromuscular disorders
Neurology	R84.4	Cerebellar anomalies
Neurology	R85.2	Holoprosencephaly (not chromosomal)
Neurology	R86.3	Hydrocephalus
Neurology	R87.3	Cerebral malformation
Neurology	R88.3	Severe microcephaly
Neurology	R109.3	Childhood onset leukodystrophy

Speciality Group	Test code	Clinical Indication
Neurology	R56.3	Adult onset movement disorder WGS
Neurology	R57.5	Movement disorders - childhood onset WGS
Neurology	R58.4	Neurodegenerative disorders - adult onset WGS
Neurology	R60.3	Hereditary spastic paraplegia - adult onset WGS
Neurology	R62.2	White matter disorders - adult onset WGS
Neurology	R78.4	Hereditary neuropathy WGS
Renal	R193.4	Cystic renal disease
Renal	R257.2	End-stage renal disease - childhood onset WGS
Core	R29	Intellectual disability
Core	R27	Congenital malformation and dysmorphism syndromes
Core	R69.5	Hypotonic infant with a likely central cause
Core	R89	Ultra-rare and atypical monogenic disorders
Cardiology	R135.2	Paediatric or syndromic cardiomyopathy WGS
Opthalmology	R31.3	Cataracts WGS
Opthalmology	R32.2	Retinal disorders WGS
Opthalmology	R33.3	Retinal disorders WGS
Opthalmology	R34.3	Retinal disorders WGS
Opthalmology	R35.3	Retinal disorders WGS
Opthalmology	R36.2	Structural eye disease WGS
Immunology	R15.4	Primary immunodeficiency WGS

Sample Requirements:

A completed request card and record of discussion is required to accompany each sample. The standard sample requirements to ensure sufficient sample quality and quantity are stated below.

Referral	Volume of Blood	Blood tube Preservative Type
Adult	3-5ml	EDTA
Paediatric	>3ml	EDTA
Infant	1-3ml	EDTA

In exceptional circumstances where individuals are unable to provide blood samples the following samples may be permitted.

Fibroblast samples

DNA extracted from fibroblast cultures may be submitted for individuals who have undergone bone marrow transplantation or circumstances where other options such as stored DNA are unavailable. Fibroblasts should be collected, processed and stored using a locally validated process within a UKAS ISO 15189:2012 (or previously CPA (UK) Ltd accredited laboratory) accredited laboratory. The sample type indicating DNA fibroblast must be stated on rare disease test request form.

Stored DNA samples

In cases where an individual is difficult to bleed, has had a bone marrow transplant, is deceased or the DNA extracted fails to meet the WGS DNA sample requirements then it is acceptable to use stored DNA samples which have been extracted and stored in a UKAS ISO 15189:2012 accredited laboratory provided they are in line with the requirements for WGS DNA sample requirements. In such cases, this must be stated on the rare disease test request form.