UKGTN Testing Criteria

Test name: (for UKGTN administration to complete) **Mitochondrial Genome 37 Gene Panel**	
Approved name and symbol of disorder/condition(s): Mitochondrial Genome 37 Gene Panel – Appendix 1	**OMIM number(s):** Appendix 1
Approved name and symbol of gene(s): Appendix 1	**OMIM number(s):** Appendix 1

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer:	
Title/Position:	**Lab ID:**

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant Neurologist	
Consultant Ophthalmologist (adult and paediatric)	
Consultant Paediatric Neurologist	
Consultant Metabolic Paediatrician	
Consultant Intensive Care Paediatrician	
Consultant Cardiologist	
Consultant Hepatologist	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Characteristic clinical features of mitochondrial disease (a combination of: fatigue, muscle weakness, CPEO, ptosis, neuropathy, ataxia, epilepsy, encephalopathy, learning difficulties, dementia, optic atrophy, retinopathy, cardiac disturbance, hearing loss, diabetes, thyroid problems)	
AND/OR characteristic muscle biopsy	
AND/OR characteristic biochemical findings	

The referral proforma should be obtained from:

Newcastle lab:
www.mitochondrialncg.nhs.uk/newcastle_index.html

London Institute of Neurology:
http://www.mitochondrialncg.nhs.uk/documents/London_Referral_Form.pdf

Oxford laboratory:
http://www.ouh.nhs.uk/services/referrals/genetics/genetics laboratories/documents/Mitochondrialprform.pdf

Additional Information:
For panel tests: At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation.

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.

Testing Criteria Appendix 1

Genes and diseases in panel test: Information and references with respect to pathogenic and polymorphic mutations in the mt genome is available on the MITOMAP website (http://www.mitomap.org/MITOMAP) and its' respective links.

HGNC standard name and symbol of the gene	HGNC number	OMIM number	OMIM standard name of condition and symbol	OMIM number
mitochondrially encoded tRNA phenylalanine; **MT-TF**	**7481**	590070	MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES; **MELAS**	540000
mitochondrially encoded tRNA valine; **MT-TV**	**7500**	590105	MITOCHONDRIAL MYOPATHY; **Mitochondrial Inherited Leigh/Leigh like Syndrome**	251900
mitochondrially encoded 16S RNA; **MT-RNR2**	**7471**	561010	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded 12S RNA; **MT-RNR1**	**7470**	561000	DEAFNESS, NONSYNDROMIC SENSORINEURAL, MITOCHONDRIAL	500008
mitochondrially encoded tRNA leucine 1 (UUA/G); **MT-TL1**	**7490**	590050	MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES; **MELAS;** KEARNS-SAYRE SYNDROME; **KSS;** DIABETES AND DEAFNESS, MATERNALLY INHERITED; **MIDD; Chronic Progressive Opthalmoplegia; CPEO**	540000; 530000; 520000
mitochondrially encoded NADH dehydrogenase 1; **MT-ND1**	**7455**	516000	MITOCHONDRIAL MYOPATHY; **LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded tRNA isoleucine; **MT-TI**	**7488**	590045	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA glutamine; **MT-TQ**	**7495**	590030	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA methionine; **MT-TM**	**7492**	590065	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded NADH dehydrogenase 2;	**7456**	516001	MITOCHONDRIAL MYOPATHY;	251900

MT-ND2			**LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	535000
mitochondrially encoded tRNA tryptophan; **MT-TW**	**7501**	590095	MITOCHONDRIAL MYOPATHY; **Mitochondrial Inherited Leigh/Leigh like Syndrome**	251900
mitochondrially encoded tRNA alanine; **MT-TA**	**7475**	590000	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA asparagine; **MT-TN**	**7493**	590010	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA cysteine; **MT-TC**	**7477**	590020	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA tyrosine; **MT-TY**	**7502**	590100	MITOCHONDRIAL MYOPATHY; **Chronic Progressive Opthalmoplegia; CPEO**	251900
mitochondrially encoded cytochrome c oxidase I; **MT-CO1**	**7419**	516030	MITOCHONDRIAL MYOPATHY; **LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded tRNA serine 1 (UCN); **MT-TS1**	**7497**	590080	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA aspartic acid; **MT-TD**	**7478**	590015	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded cytochrome c oxidase II; **MT-CO2**	**7421**	516040	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA lysine; **MT-TK**	**7489**	590060	MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS; **MERRF; Mitochondrial Inherited Leigh/Leigh like Syndrome**	545000 535000

mitochondrially encoded ATP synthase 8; **MT-ATP8**	**7415**	516070	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded ATP synthase 6; **MT-ATP6**	**7414**	516060	NEUROPATHY, ATAXIA, AND RETINITIS PIGMENTOSA; **NARP; Mitochondrial Inherited Leigh Syndrome MILS; LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	551500 535000
mitochondrially encoded cytochrome c oxidase III; **MT-CO3**	**7422**	516050	MITOCHONDRIAL MYOPATHY; **Mitochondrial Inherited Leigh/Leigh like Syndrome; LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded tRNA glycine; **MT-TG**	**7486**	590035	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded NADH dehydrogenase 3; **MT-ND3**	**7458**	516002	MITOCHONDRIAL MYOPATHY; **Mitochondrial Inherited Leigh/Leigh like Syndrome**	251900
mitochondrially encoded tRNA arginine; **MT-TR**	**7496**	590005	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded NADH dehydrogenase 4L; **MT-ND4L**	**7460**	516004	MITOCHONDRIAL MYOPATHY; **LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded NADH dehydrogenase 4; **MT-ND4**	**7459**	516003	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA histidine; **MT-TH**	**7487**	590040	MITOCHONDRIAL MYOPATHY	251900

mitochondrially encoded tRNA serine 2 (AGU/C); **MT-TS2**	**7498**	590085	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded tRNA leucine 2 (CUN); **MT-TL2**	**7491**	590055	MITOCHONDRIAL MYOPATHY	251900
mitochondrially encoded NADH dehydrogenase 5; **MT-ND5**	**7461**	516005	MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES; **MELAS;** WOLFRAM SYNDROME, MITOCHONDRIAL FORM; **Mitochondrial Inherited Leigh/Leigh like Syndrome; LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	540000; 598500 535000
mitochondrially encoded NADH dehydrogenase 6; **MT-ND6**	**7462**	516006	MITOCHONDRIAL MYOPATHY; **Mitochondrial Inherited Leigh/Leigh like Syndrome; LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded tRNA glutamic acid; **MT-TE**	**7479**	590025	MITOCHONDRIAL MYOPATHY, INFANTILE, TRANSIENT; **MMIT**	500009
mitochondrially encoded cytochrome b; **MT-CYB**	**7427**	516020	MITOCHONDRIAL MYOPATHY; **LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000
mitochondrially encoded tRNA threonine; **MT-TT**	**7499**	590090	MITOCHONDRIAL MYOPATHY	251900

mitochondrially encoded tRNA proline; **MT-TP**	**7494**	590075	MITOCHONDRIAL MYOPATHY **LHON where the 3 common point mutations have been excluded at m.3460G>A; m.11778G>A and m.14484T>C**	251900 535000