Version March 2024

Gene panels	Alias	Turnaround time	Prices €
See next pages for request of individual genes			
○ Basal cell carcinoma	BCC panel	56 days	1200
○ Breast and ovarium cancer panel	HBOC panel	42 days	1200
○ Cerebral angiopathies / adult-onset leukoencephalopathies (including CADASIL)	CHA panel	90 days	1500
○ Coffin-Siris / Nicolaides-Baraitser syndrome	CSS panel	90 days	1500
○ Colorectal carcinoma	CRC panel	70 days	1500
○ Episodic Ataxia	EA panel	56 days	1500
○ FAMMM (Familial Atypical Multiple Mole-Melanoma)	Melanoma panel	56 days	1200
○ Familial pancreatic carcinoma	PACA panel	42 days	1500
○ Short stature, basic gene panel	Growth panel	56 days	1500
○ Hereditary Multiple Osteochondromas	HMO panel	56 days	1500
○ LYNCH syndrome	LYNCH panel	56 days	1200
○ Lipodystrophy	LIPO panel	90 days	1500
○ Migraine, familial hemiplegic	FHM panel	56 days	1500
○ MODY (Maturity Onset Diabetes of the Young)	Diabetes panel MODYScan	90 days	1500
○ Muscular dystrophies / myopathies	Muscle panel MuscleScan	56 days	1500
○ Paragangliomas and/or pheochromocytomas	PGL panel	56 days	1200
○ Polyglutamin repeat disorders	PolyQ	56 days	650
○ Polyposis coli, adenomatous*	Polyp panel	56 days	1200
○ Polycystic kidney disease	PKD panel	90 days	1500
○ Skeletal Muscle Channelopathies	Channelopathies	56 days	1500

For an overview of all genes in the gene panels see https://www.lumc.nl/over-het-lumc/afdelingen/klinische-genetica/genpanels/

Disorder/Referral	Type	Gene/Test	Turnaround time	Prices €
Blood diseases				
○ Hemochromatosis	Type 1	○ HFE	28 days	350

○ Hemoglobinopathies / Thalassemia Please use “Requisition form Hemoglobinopathy analysis”				750
○ Hemophilia (Please send in 2 tubes of EDTA blood)	Type A	○ F8	56 days	650
	Type B	○ F9	56 days	550
Cancer genetics				
**Requests only by a consultant clinical geneticist*				
○ Breast- and ovarian cancer, hereditary *		○ ATM	56 days	550
		○ BARD1	56 days	550
		○ BRCA1	56 days	750
		○ BRCA2	56 days	750
		○ BRIP1	56 days	550
		○ CHEK2	56 days	550
		○ PALB2	56 days	550
		○ RAD51C	56 days	550
		○ RAD51D	56 days	550
○ Clear cell meningioma/ Familial Multiple Meningioma*	CCM	○ SMARCE1	56 days	750
		○ SMARCB1	56 days	750
○ FAMMM (Familial Atypical Multiple Mole-Melanoma)*		○ CDKN2A	56 days	350
		○ CDK4	56 days	350
		○ POT1	56 days	550
		○ BAP1	56 days	550
		○ MITF	56 days	350
○ Gastrointestinal Stromal Tumors (GIST, Carney-Stratakis syndrome)		○ SDHA	56 days	550
○ Hyperparathyroidism-jaw tumor syndrome (HPT-JT/HRPT2)		○ CDC73	56 days	750
○ Lynch syndrome (HNPCC)*		○ MLH1	56 days	750
		○ MSH2 (incl. EPCAM)	56 days	750
		○ MSH6	56 days	750
		○ PMS2	56 days (RNA 120 days)	750 (750)
○ Myeloproliferative diseases (MPDs, somatic mutation)		○ JAK2 (p.Val617Phe)	28 days	350
		○ MPN-combi: JAK2 exon 12 & exon 14 p.(Val617Phe), MPL exon 10 and CALR exon 9	28 days	350
○ Paragangliomas and/or pheochromocytomas		○ MAX	56 days	550
		○ SDHA	56 days	550
		○ SDHAF2	56 days	750
		○ SDHB	56 days	750
		○ SDHC	56 days	750

		○ SDHD	56 days	750
		○ TMEM127	56 days	550
○ Polyposis coli, adenomatous*	FAP1	○ APC (incl. GREM1)	56 days	900
	MAP	○ MUTYH	56 days	750
	NAP	○ NTHL1		550
	PPAP	○ POLD1	56 days	550
	PPAP	○ POLE	56 days	550
	FAP4	○ MSH3	56 days	650
○ Renal Cell Carcinoma (RCC), hereditary		○ SDHB	56 days	750
○ Rhabdoid tumor predisposition syndrome (RTPS)*	RTPS1	○ SMARCB1	56 days	750
	RTPS2	○ SMARCA4	56 days	650
○ Small cell carcinoma of the ovary, hypercalcemic type*	SCCOHT	○ SMARCA4	56 days	650
	SCCOHT	○ SMARCB1	56 days	750
○ Schwannomatosis*		○ SMARCB1	56 days	750
Channelopathies				
○ Hyperkalemic periodic paralysis (HYPP)		○ SCN4A	56 days	750
○ Hypokalemic periodic paralysis (HOKPP)	Type 1	○ CACNA1S	28 days	750
	Type 2	○ SCN4A	56 days	750
○ Myotonia congenita (Thomsen, Becker disease)		○ CLCN1	56 days	750
○ Myotonia permanens/fluctuans		○ SCN4A	56 days	750
○ Paramyotonia congenita		○ SCN4A	56 days	750
Diabetes				
○ Hyperproinsulinemia		○ INS	56 days	750
○ Insulin dependent diabetes		○ INS	56 days	750
○ MIDD (Maternally Inherited Diabetes and Deafness)		○ m.3243A>G tRNALEU/UUR	28 days	750
○ MODY (Maturity Onset Diabetes of the Young)	Type 1	○ HNF4A	56 days	750
	Type 2	○ GCK	56 days	750
	Type 3	○ HNF1A	56 days	750
	Type 4	○ PDX1 (IPF1)	56 days	750
	Type 5	○ HNF1B	56 days	750
	Type 6	○ NEUROD1	56 days	750
	Type 10	○ INS	56 days	750
○ PNDM (Permanent Neonatal Diabetes Mellitus)		○ GCK	56 days	750
		○ INS	56 days	750
		○ KCNJ11	56 days	750
○ Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)		○ GCK	56 days	750
		○ KCNJ11	56 days	750
Growth and skeletal defects				
○ Achondroplasia		○ FGFR3	56 days	750

○ Acromesomelic dysplasia	Type Maroteaux	○ NPR2	56 days	750
○ Hereditary Multiple Osteochondromas		○ EXT1	56 days	750
		○ EXT2	56 days	750
○ NPR2- related tall stature		○ NPR2	56 days	750
○ Hypochondroplasia		○ FGFR3	56 days	650
○ Langer mesomelic dysplasia (Leri-Weill dyschondrosteosis)		○ SHOX	56 days	750
○ Multiple epiphyseal dysplasia		○ COMP	56 days	550
○ Pseudoachondroplastic dysplasia		○ COMP	56 days	550
○ Short stature (proportionate)		○ GH1	56 days	750
		○ GHR	56 days	750
		○ GHSR	56 days	550
		○ IGF1	56 days	750
		○ IGF1R	56 days	750
		○ IGFALS	56 days	750
		○ STAT5B	56 days	750
○ Short stature (osteochondritis dissecans)		○ ACAN	56 days	650
○ Tall stature		○ NPR2	56 days	750
○ Thanatophoric dysplasia		○ FGFR3	56 days	650
○ Van Buchem disease		○ VBCH	28 days	750
Immune system				
○ Chilblain lupus	Type 1	○ TREX1	28 days	550
○ Granulomatous disease, chronic, X-linked		○ CYBB	56 days	550
○ Lymphoproliferative syndrome, X-linked		○ XLP	28 days	550
○ Mediterranean fever, familial (FMF)		○ MEFV	56 days	550
○ Wiskott-Aldrich syndrome		○ WAS	28 days	550
Metabolic diseases				
○ Adrenal hypoplasia, congenital		○ NR0B1 (DAX1)	56 days	750
○ Cystinuria		○ SLC3A1	56 days	550
		○ SLC7A9	56 days	550
Muscular dystrophies/ Myopathies				
○ Slow-channel congenital myasthenic syndrome-4A (CMS4A)	Type 4A	○ CHRNE	56 days	350
○ Congenital myasthenic syndrome-5 (CMS5)	Type 5	○ COLQ	56 days	350
○ Congenital myasthenic syndrome-9 (CMS9) associated with AChR deficiency	Type 9	○ MUSK	56 days	350
○ Congenital myasthenic syndrome-10 (CMS10)	Type 10	○ DOK7	56 days	350
○ Congenital myasthenic syndrome-11 associated with acetylcholine receptor deficiency (CMS11)	Type 11	○ RAPSN	56 days	350
○ Congenital myasthenic syndrome-14 (CMS14)	Type 14	○ ALG2	56 days	350

○ Congenital myasthenic syndrome-15 (CMS15)	Type 15	○ ALG14	56 days	350
○ Duchenne and Becker		○ DMD MLPA only	28 days	350
		○ DMD Sequencing only	56 days	650
		○ DMD MLPA, if negative directly followed by sequencing	56 days	350 or 900
○ Emery-Dreifuss (X-linked)		○ EMD	28 days	550
○ Facioscapulohumeral (FSHD) (Please send in 2 tubes of EDTA blood)	Type 1	○ Rearrangement chromosome 4	90 days	1000
		○ Permissive haplotype analysis (4qA/B)	90 days	1000
	Type 2	○ SMCHD1	56 days	750
		○ LRIF1	56 days	
		○ DNMT3B	56 days	
○ Limb Girdle	Myofibrillar myopathy	○ MYOT	56 days	550
	Emery–Dreifuss muscular dystrophy (EDMD)	○ LMNA	56 days	550
	Rippling muscle disease	○ CAV3	28 days	550
	LGMD D4 / R1	○ CAPN3	56 days	750
	LGMD R2	○ DYSF	56 days	750
	LGMD R5	○ SGCG	56 days	550
	LGMD R3	○ SGCA	56 days	550
	LGMD R4	○ SGCB	56 days	550
	LGMD R6	○ SGCD	56 days	550
	LGMD R7	○ TCAP	28 days	550
	LGMD R8	○ TRIM32	56 days	550
	LGMD R9	○ FKRP	28 days	550
	LGMD R12	○ ANO5	56 days	550
○ Miyoshi (MMD3)		○ ANO5	56 days	550
○ Myopathy with extrapyramidal signs		○ MICU1	28 days	550
Neurogenetics				
○ Aicardi-Goutières syndrome	Type 1	○ TREX1	28 days	550
○ Alternating Hemiplegia of Childhood	Type 2	○ ATP1A3	56 days	550

○ CADASIL		○ NOTCH3	56 days	650
○ CARASIL/ CADASIL	Type 2	○ HTRA1	56 days	650
○ Cerebral hemorrhage with amyloidosis (HCHWA-D)		○ APP	28 days	350
○ Dentatorubral-pallidoluysian atrophy (DRPLA)		○ ATN1	28 days	350
○ Episodic ataxia	Type 2	○ CACNA1A	56 days	750
○ Huntington disease		○ HTT	28 days	350
○ Huntington, disease-like 2 (HDL2)		○ JPH3	28 days	350
○ Hyperekplexia (familial Startle disease)		○ GLRA1	56 days	750
		○ GLRB	56 days	750
		○ SLC6A5	56 days	750
○ Migraine, familial hemiplegic (FHM)		○ ATP1A2	56 days	750
		○ CACNA1A	56 days	750
		○ SCN1A	56 days	750
○ Myoclonus dystonia syndrome		○ SGCE	56 days	750
○ Neuronal ceroid lipofuscinosis (NCL)	Juvenile	○ CLN3	56 days	550
	Late infantile	○ TPP1 (CLN2)	56 days	550
	Late infantile	○ CLN6	56 days	550
	Late infantile	○ CLN8	56 days	550
	Late infantile / adult	○ PPT1 (CLN1)	56 days	550
○ Paroxysmal torticollis		○ CACNA1A	56 days	750
○ Polyglutamin repeat disorders		○ CACNA1A, TBP, ATXN1, ATXN7, ATXN2, ATXN3 en ATN1	56 days	650
○ Retinal vasculopathy with cerebral leukodystrophy (RVCL)		○ TREX1	28 days	550
Polycystic kidney disease				
○ Autosomal dominant Polycystic kidney disease (ADPKD)	Dominant	○ PKD1	90 days	900
	Dominant	○ PKD2	56 days	750
○ Autosomal dominant Polycystic kidney and liver disease (ADPKD)	Dominant	○ GANAB	56 days	750
○ Autosomal recessive Polycystic kidney (ARPKD)	Recessive	○ PKHD1	56 days	900
○ Renal cysts and diabetes syndrome (RCAD)	Dominant	○ HNF1B	56 days	750
Syndromes		○		
○ Coffin-Siris syndrome		○ ARID1A	56 days	750
		○ ARID1B	56 days	750
		○ SMARCA4	56 days	650
		○ SMARCB1	56 days	550
		○ SMARCE1	56 days	550
○ Ellis van Creveld syndrome		○ EVC	56 days	550
		○ EVC2	56 days	550
○ Filippi syndrome		○ CKAP2L	56 days	550

○ Marshall-Smith syndrome		○ NFIX	56 days	750
○ Nicolaides-Baraitser syndrome		○ SMARCA2	56 days	650
○ Peters Plus syndrome		○ B3GLCT (B3GALTL)	56 days	750
○ Pitt-Hopkins syndrome		○ TCF4	56 days	750
○ Rubinstein - Taybi syndrome		○ CREBBP	56 days	750
		○ EP300	56 days	750
○ Sotos syndrome		○ NSD1	56 days	750
○ Sotos-like syndrome		○ DNMT3A	56 days	550
		○ NFIX	56 days	750
		○ SETD2	56 days	650
		○ HIST1H1E	56 days	550
○ TAR (thrombocytopenia-absent radius) syndrome		○ 1q21.1 deletion and RBM8A SNP	28 days	550
○ Weaver syndrome		○ EZH2	56 days	550
Other				
○ Hypocalciuric Hypercalcemia, Familial (FHH)		○ CASR ○ GNA11 ○ AP2S1	56 days	550 3 genes 1500
○ Keratosis follicularis spinulosa decalvans (KFSD)		○ MBTPS2	28 days	550
○ TSH deficiency and macroorchidism, X-linked		○ IGSF1	56 days	550