

## Laboratory turnaround times and prices

(not valid within the Netherlands)

Version January 2026

The turnaround times are calculated from the day of receipt to the day that the result leaves the laboratory. In urgent cases, shorter time periods can be arranged. Please contact the laboratory.

When several genes, for the same disorder, are simultaneously requested, the price will not exceed €1500,-.

Diagnostics	Type	Turnaround time*	Price €
Hemoglobinopathy diagnostics	Hematology and DNA testing	60 days	750
Postnatal genome diagnostics	Confirmation/exclusion of a known mutation	28 days (FSHD 90 days)	350 (FSHD 1000)
	Prenatal testing (known mutation)	14 days (exception FSHD1)	700
	scanning	For more details, see page 3 and onwards	
	QF-PCR	2 working days	On request
	Karyotyping urgent	5 working days	On request
	Whole Genome Sequencing (single patient)	On request	1600
	Whole Genoom Sequencing (trio analysis)	On request	3200
	Other techniques	28 days	On request

Gene panels	Alias	Turnaround time	Prices €
<i>See next pages for request of individual genes</i>			
o Basal cell carcinoma	BCC panel	56 days	1200
o Breast and ovarium cancer panel	HBOC panel	42 days	1200
o Cerebral angiopathies / adult-onset leukoencephalopathies (including CADASIL)	CHA panel	90 days	1500
o Coffin-Siris / Nicolaidis-Baraitser syndrome	CSS panel	90 days	1500

o Colorectal carcinoma	CRC panel	70 days	1500
o Episodic Ataxia	EA panel	56 days	1500
o FAMMM (Familial Atypical Multiple Mole-Melanoma)	Melanoma panel	56 days	1200
o Familial pancreatic carcinoma	PACA panel	42 days	1500
o Short stature, basic gene panel	Growth panel	56 days	1500
o Hereditary Multiple Osteochondromas	HMO panel	56 days	1500
o LYNCH syndrome	LYNCH panel	56 days	1200
o Lipodystrophy	LIPO panel	90 days	1500
o Migraine, familial hemiplegic	FHM panel	56 days	1500
o MODY (Maturity Onset Diabetes of the Young)	Diabetes panel MODYScan	90 days	1500
o Muscular dystrophies / myopathies	Muscle panel MuscleScan	56 days	1500
o Paragangliomas and/or pheochromocytomas	PGL panel	56 days	1200
o Polyglutamin repeat disorders	PolyQ	56 days	650
o Polyposis coli, adenomatous*	Polyp panel	56 days	1200
o Polycystic kidney disease	PKD panel	90 days	1500
o Skeletal Muscle Channelopathies	Channelopathies	56 days	1500

For an overview of all genes in the gene panels see <https://www.lumc.nl/en/afdelingen/clinical-genetics/gene-panels/>

## Turnaround times mutational scanning and prices

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Disorder/Referral	Type	Gene/Test	Turnaround time	Prices €
<b>Blood diseases</b>				
○ Hemochromatosis	Type 1	○ HFE	28 days	350
○ Hemoglobinopathies / Thalassemia Please use "Requisition form Hemoglobinopathy analysis"				2100
○ Hemophilia (Please send in 2 tubes of EDTA blood)	Type A	○ F8	56 days	650
	Type B	○ F9	56 days	550
<b>Cancer genetics</b>				
<i>*Requests only by a consultant clinical geneticist</i>				
○ 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
		○ PTEN	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:	28 days	350

		JAK2 exon 12 & exon 14 p.(Val617Phe), MPL exon 10 and CALR exon 9		
○ Parangliomas 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	56 days	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
<b>Channelopathies</b>				
○ 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
<b>Diabetes</b>				
○ 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
<b>Growth and skeletal defects</b>				
○ 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
<b>Immune system</b>				
○ 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
<b>Metabolic diseases</b>				
○ Adrenal hypoplasia, congenital		○ NR0B1 (DAX1)	56 days	750
○ Cystinuria		○ SLC3A1	56 days	550
		○ SLC7A9	56 days	550
<b>Muscular dystrophies/ Myopathies</b>				

○ 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	○ RAPSIN	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 1/2	○ 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
<b>Neurogenetics</b>				
○ 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

<b>Polycystic kidney disease</b>				
○ 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
<b>Syndromes</b>		○		
○ 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 (B3GALT1)	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
<b>Other</b>				
○ Hypocalciuric Hypercalcemia, Familial (FHH)		○ CASR ○ GNA11 ○ AP2S1	56 days	550 3 genes 1500
○ Keratosis follicularis spinulosa decalvans (KFSD)		○ MBTPS2	28 days	550