|  |  |  |  |
| --- | --- | --- | --- |
| **Gene panels** | **Alias** | **Turnaround time** | **Prices €** |
| *See next pages for request of individual genes* |  |  |  |
| * 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 panelMODYScan | 90 days | 1500 |
| * Muscular dystrophies / myopathies
 | Muscle panelMuscleScan | 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/**](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 |
|  | 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 | 5503 genes 1500 |
| * Keratosis follicularis spinulosa decalvans (KFSD)
 |  | * MBTPS2
 | 28 days | 550 |
| * TSH deficiency and macroorchidism, X-linked
 |  | * IGSF1
 | 56 days | 550 |