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| **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 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 | Channelopa  thies | 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/)

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| **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 | 550  3 genes 1500 |
| * Keratosis follicularis spinulosa decalvans (KFSD) |  | * MBTPS2 | 28 days | 550 |
| * TSH deficiency and macroorchidism, X-linked |  | * IGSF1 | 56 days | 550 |