MEDGENOME

MedGenome
Test list

www.medgenome.com

About MedGenome

MedGenome is the largest throughput genomics diagnostics laboratory in India spearheading the decoding of genetic information to enhance personalized medicine.

A market leader in genomics-based diagnostics and research, MedGenome has worked with thousands of doctors, hospitals, and patients across India by applying Next-generation Sequencing (NGS) techniques in cancer, ophthalmic and neurological disorders, diabetes, cardiovascular diseases and rare genetic diseases.

MedGenome offers a broad range of pre-designed gene mutation panels which have been developed with in-depth disease understanding and are invaluable for deciphering the following:

- Disease biology/pathogenesis
- Familial association/inheritance pattern
- Prognosis
- Therapeutic response

MedGenome currently functions from two different laboratory locations. The NGS lab is located in Bangalore while the molecular diagnostic lab is located in Cochin. MedGenome offers seamless logistic services across the country with best-in-class pre-analytic specimen preservation, temperature control and turn around for transit of specimen from client's doorstep. All laboratory reports are available online/via email.

MedGenome has also established its state-of-the-art pre-test and post-test counselling services wherein the clinician can discuss intricacies of genetic tests and their results with the trained and informed personnel at the lab or with its dedicated Scientific Affairs team.

Besides molecular and genomic tests MedGenome also offers a broad range of Immunohistochemistry (IHC) tests which add trust to its Oncology diagnostic services.

For more information, please visit us at www.medgenome.com or call our customer support services (available between 9 am and 6 pm Monday through Saturday) at +91-80-67154932

258/A, 3rd Floor, Narayana Nethralaya Building, Narayana Health City,
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Plot no: SDF 17, First Floor, C Block, CSEZ, Kakkanad, Cochin, Kerala – 682037

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GENETIC TEST SELECTION LIST:

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CARDIOLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM001	Cardiac channelopathy gene panel	☐ NGS	4 WEEKS
MGM002	Cardiomyopathy gene panel	☐ NGS	4 WEEKS
MGM003	Cardiomyopathy predisposition - *MYBPC3* (25bp deletion)	☐ Sanger	7 DAYS
MGM004	Clopidogrel dosage: *CYP2C19**2 & *CYP2C19**3	☐ Sanger	10 DAYS
MGM005	Clopidogrel dosage: *CYP2C19**2 & *CYP2C19**3	☐ RT-PCR	7 DAYS
MGM007	Fabry disease (*GLA*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM006	Fabry disease (*GLA*) gene analysis	☐ NGS	4 WEEKS
	Pompe disease - (refer to metabolic disorders)		
MGM008	Statin induced myopathy predisposition - *SLCO1B1* p.(Val174Ala)	☐ Sanger	7 DAYS
MGM009	Warfarin dosage- *VKORC1* (c.-1639 G>A), *CYP2C9**2, *CYP2C9**3 & *CYP2C9**13	☐ Sanger	7 DAYS

CONNECTIVE TISSUE DISORDERS and SKELETAL DYSPLASIAS [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM301	Achondroplasia *FGFR3* G380R common mutation screening	☐ Sanger	2 WEEKS
MGM310	Hypochondroplasia *FGFR3* N540K common mutation screening	☐ Sanger	2 WEEKS
MGM010	Achondroplasia (*FGFR3*) gene analysis	☐ NGS	4 WEEKS
MGM011	Connective tissue disorder gene panel	☐ NGS	4 WEEKS
MGM012	Cutis-laxa gene panel	☐ NGS	4 WEEKS
MGM013	Ehler Danlos syndrome gene panel	☐ NGS	4 WEEKS
MGM014	Marfan's syndrome (*FBN1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM015	Marfan's syndrome (*FBN1*) gene analysis	☐ NGS	4 WEEKS
MGM016	Osteogenesis imperfecta gene panel	☐ NGS	4 WEEKS
MGM017	Osteopetrosis gene panel	☐ NGS	4 WEEKS
MGM018	Pachydermoperiostosis & primary hypertrophic osteoarthropathy gene panel	☐ NGS	4 WEEKS
MGM019	Skeletal dysplasia gene panel	☐ NGS	4 WEEKS

DERMATOLOGY (SKIN) [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM020	Ectodermal dysplasia gene panel	☐ NGS	4 WEEKS
MGM021	Epidermolysis bullosa gene panel	☐ NGS	4 WEEKS
MGM022	Icthyosis gene panel	☐ NGS	4 WEEKS
MGM023	Oculocutaneous albinism gene panel	☐ NGS	4 WEEKS
MGM024	Sjogren-Larsson syndrome (*ALDH3A2*) gene analysis	☐ NGS	4 WEEKS
MGM025	Xeroderma pigmentosum gene panel	☐ NGS	4 WEEKS

ENDOCRINOLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM026	Androgen receptor (*AR*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM027	Androgen receptor (*AR*) gene analysis	☐ NGS	4 WEEKS
MGM030	Congenital adrenal hyperplasia *CYP21A2* (21-0H) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM028	Congenital adrenal hyperplasia *CYP21A2* (21-0H) gene analysis	☐ Sanger	3 WEEKS
MGM029	Congenital adrenal hyperplasia *CYP21A2* (21-0H) gene analysis	☐ NGS	3 WEEKS
MGM292	Congenital hyperparathyroidism gene panel	☐ NGS	4 WEEKS
MGM031	Congenital hypopituitarism gene panel	☐ NGS	4 WEEKS
MGM309	Hereditary pancreatitis gene panel	☐ NGS	4 WEEKS
MGM032	Kallmann syndrome gene panel	☐ NGS	4 WEEKS
MGM033	Maturity-Onset Diabetes of the Young (MODY) & neonatal diabetes gene panel	☐ NGS	4 WEEKS

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ENT [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM034	Connexin-30 (*GJB6*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM035	Connexin-26 (*GJB2*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM036	Connexin-26 (*GJB2*) gene analysis (exon 2)	☐ Sanger	7 DAYS
MGM037	Deafness gene panel	☐ NGS	4 WEEKS
MGM038	Mondini defect (*SLC26A4*) gene analysis	☐ NGS	4 WEEKS
	Usher syndrome - refer to opthalmology		
MGM039	Waardenburg syndrome gene panel	☐ NGS	3 WEEKS

HAEMATOLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM040	Alpha thalassemia (*HBA1* & *HBA2*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM041	Alpha thalassemia (*HBA1* & *HBA2*) gene analysis	☐ Sanger	2 WEEKS
MGM042	Aplastic anemia gene panel	☐ NGS	4 WEEKS
MGM262	*ATRX* gene analysis	☐ NGS	4 WEEKS
MGM043	Beta thalassemia (*HBB*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM044	Beta thalassemia (*HBB*) gene analysis	☐ Sanger	2 WEEKS
MGM045	Congenital afibrinogenemia gene panel	☐ NGS	4 WEEKS
MGM046	Congenital dyserythropoietic anemia gene panel	☐ NGS	4 WEEKS
MGM047	Diamond blackfan anemia gene panel	☐ NGS	4 WEEKS
MGM048	Dyskeratosis congenita gene panel	☐ NGS	4 WEEKS
MGM049	Factor V Leiden mutation analysis (exon 10)	☐ Sanger	7 DAYS
MGM050	Factor VII deficiency gene analysis	☐ NGS	4 WEEKS
MGM051	Fanconi anemia gene panel	☐ NGS	4 WEEKS
MGM052	Haemophilia (*F8* & *F9*) gene panel	☐ NGS	4 WEEKS
MGM053	Haemophilia (*F8*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM306	Haemophilia A (*F8*) gene analysis (analysis of the *F8* inversion is not included)	☐ NGS	4 WEEKS
MGM307	Haemophilia B (*F9*) gene analysis	☐ NGS	4 WEEKS
MGM068	Hemophagocytic lymphohistiocytosis (*PRF1, STK11, UNC13D*)deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM067	Hemophagocytic lymphohistiocytosis gene panel	☐ NGS	4 WEEKS
MGM054	Hereditary elliptocytosis gene panel	☐ NGS	4 WEEKS
MGM293	Hereditary hemolytic anemia panel (For RBC membrane disorders and enzymopathies)	☐ NGS	4 WEEKS
MGM059	Hereditary spherocytosis gene panel	☐ NGS	4 WEEKS
MGM288	HLA typing high resolution (HLA A, B, C, DRB1, DQB1, DRB3,4,5, DPA1, DPB1)	☐ NGS	10 DAYS
MGM056	*MTHFR* gene analysis - 2 exons (5 & 8)	☐ Sanger	10 DAYS
MGM071	Primary immunodeficiency gene panel	☐ NGS	4 WEEKS
MGM319	Protein S deficiency (*PROS1* gene deletion/duplication analysis)	☐ MLPA	3 WEEKS
MGM072	Severe combined immunodeficiency (SCID) gene panel and analysis (*DOCK8, STAT3*)	☐ NGS	4 WEEKS
MGM057	Sickle cell anemia (*HBB*) gene analysis (exon 1)	☐ Sanger	10 DAYS
MGM058	Sideroblastic anaemia gene panel	☐ NGS	4 WEEKS
MGM060	Von Willebrand disease (*VWF*) gene analysis	☐ NGS	4 WEEKS

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IMMUNOLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM061	Agammaglobulinemia (*BTK*) gene analysis	☐ NGS	4 WEEKS
MGM062	Chediak-Higashi syndrome (*LYST*) gene analysis	☐ NGS	4 WEEKS
MGM063	Congenital neutropenia gene panel	☐ NGS	4 WEEKS
MGM065	Cystic fibrosis (*CFTR*) del508 mutation analysis	☐ Sanger	7 DAYS
MGM064	Cystic fibrosis (*CFTR*) gene analysis	☐ NGS	4 WEEKS
MGM066	Cystic fibrosis (*CFTR*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM069	Hyper-immunoglobulin E syndrome deletion/duplication	☐ MLPA	3 WEEKS

METABOLIC DISORDERS [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM073	Citrullinemia gene panel	☐ NGS	4 WEEKS
	Fabry disease - (refer to cardiology)		
MGM074	Fanconi bickel syndrome (*SLC2A2*) gene analysis	☐ NGS	4 WEEKS
MGM075	Fatty acid oxidation disorders gene panel	☐ NGS	4 WEEKS
MGM076	Gilbert syndrome (*UGT1A1*) gene analysis (both point mutation and repeat analysis)	☐ NGS/ Fragment analysis	4 WEEKS
MGM077	GLUT1 deficiency (*SLC2A1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM078	GLUT1 deficiency (*SLC2A1*) gene analysis	☐ NGS	4 WEEKS
MGM079	Glycine encephalopathy (*GLDC*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM080	Glycine encephalopathy gene panel	☐ NGS	4 WEEKS
MGM081	Glycogen storage disorder gene panel	☐ NGS	4 WEEKS
MGM082	Glycosylation (CDG) disorders gene panel	☐ NGS	4 WEEKS
MGM083	Hemochromatosis gene panel	☐ NGS	4 WEEKS
MGM084	Homocystinuria gene panel	☐ NGS	4 WEEKS
MGM086	Hyperargininemia (*ARG1*) gene analysis	☐ NGS	4 WEEKS
MGM085	Hypercholesterolemia gene panel	☐ NGS	4 WEEKS
MGM087	Leigh syndrome & mitochondrial encephalopathy gene panel	☐ NGS	4 WEEKS
MGM088	Maple syrup urine disease gene panel	☐ NGS	4 WEEKS
MGM089	McArdle disease (*PYGM*) gene analysis	☐ NGS	4 WEEKS
MGM090	Menkes disease (*ATP7A*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM091	Menkes disease (*ATP7A*) gene analysis	☐ NGS	4 WEEKS
MGM092	Methylmalonic aciduria gene panel	☐ NGS	4 WEEKS
MGM093	Mucopolysaccharidosis gene panel	☐ NGS	4 WEEKS
MGM094	Neurometabolic disorder (*MLC1*, *L2HGDH*, *D2HGDH*, *MLYCD*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM095	Niemann-Pick disease gene panel	☐ NGS	4 WEEKS
MGM096	Organic acidemia gene panel	☐ NGS	4 WEEKS
MGM097	Peroxisomal disorder gene panel (includes adrenoleukodystrophy)	☐ NGS	4 WEEKS
MGM098	Pompe disease (*GAA*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM099	Pompe disease (*GAA*) gene analysis	☐ NGS	4 WEEKS
MGM100	Progressive familial intrahepatic cholestasis gene panel	☐ NGS	4 WEEKS
MGM101	Urea cycle defects gene panel	☐ NGS	4 WEEKS
MGM102	Wilson disease (*ATP7B*) gene analysis	☐ NGS	4 WEEKS
MGM314	Niemann Pick disease (*NPC1, NPC2, SMPD1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM318	Propionic acidemia (*PCCA*) deficiency deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM325	Wilson disease (*ATP7B*) deletion/ duplication analysis	☐ MLPA	3 WEEKS

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NEPHROLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM103	Alport syndrome gene panel	☐ NGS	4 WEEKS
MGM104	Bartter syndrome gene panel	☐ NGS	4 WEEKS
MGM308	Hemolytic uremic syndrome (*CFH, CFHR1* & *CFHR3*) deletion duplication analysis	☐ MLPA	3 WEEKS
MGM105	Meckel Gruber syndrome gene panel	☐ NGS	4 WEEKS
MGM317	Polycystic kidney disease (*PKHD1*) deficiency deletion/duplication analysis	☐ NGS	3 WEEKS
MGM106	Polycystic kidney disease gene panel (ARPKD: *PKHD1*/ADPKD: *PKD1* & *PKD2*)	☐ NGS	4 WEEKS
MGM107	Primary hyperoxaluria gene panel	☐ NGS	4 WEEKS
MGM108	Xanthinuria (*XDH*) gene analysis	☐ NGS	4 WEEKS

NEUROLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM294	Chromosomal microarray - Affymetrix Cytoscan 750K genechip	☐ Microarray	4 WEEKS
MGM295	Chromosomal microarray - Affymetrix Cytoscan HD genechip	☐ Microarray	4 WEEKS
MGM213	Neurofibromatosis type 1 (*NF1*) gene analysis	☐ NGS	4 WEEKS
MGM214	Neurofibromatosis type 2 (*NF2*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM215	Neurofibromatosis type 2 (*NF2*) gene analysis	☐ NGS	4 WEEKS
MGM212	Neurofibromatosis type 1 (*NF1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM229	*TSC1 & TSC2* gene analysis	☐ NGS	4 WEEKS
MGM230	*TSC1 & TSC2* gene analysis	☐ Sanger	3 WEEKS
MGM231	*TSC1* deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM232	*TSC1* gene analysis	☐ Sanger	4 WEEKS
MGM233	*TSC2* deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM234	*TSC2* gene analysis	☐ Sanger	4 WEEKS

MOVEMENT DISORDERS

CODE	TEST	METHOD	TAT
MGM316	Pantothenate kinase-associated neurodegeneration (*PLA2G6*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM123	Ataxia-telangiectasia (*ATM*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM124	Ataxia-telangiectasia (*ATM*) gene analysis	☐ NGS	4 WEEKS
MGM125	Dystonia gene panel	☐ NGS	4 WEEKS
MGM126	Early-onset juvenile parkinsonism gene panel	☐ NGS	4 WEEKS

NEUROMUSCULAR

CODE	TEST	METHOD	TAT
MGM302	Charcot-Marie-Tooth type 4 (*EGR2, GDAP1, NEFL, PRX*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM131	Arthrogryposis & congenital myasthenic syndrome gene panel	☐ NGS	4 WEEKS
MGM133	Charcot-Marie-Tooth and *1A/HNPP* (*PMP22, COXI0, TEKT3*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM132	Charcot-Marie-Tooth and sensory neuropathies gene panel	☐ NGS	4 WEEKS
MGM134	Charcot-Marie-Tooth *PMP22* deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM135	Congenital Muscular Dystrophy *(LAMA2)* deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM136	Duchenne Muscular Dystrophy *(DMD)* deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM137	Duchenne Muscular Dystrophy *(DMD)* gene analysis	☐ NGS	4 WEEKS

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NEUROLOGY (continued)

CODE	TEST	METHOD	TAT
	OTHERS **(NEUROLOGY)**		
MGM324	WAGR syndrome (*PAX6*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM145	4H syndrome gene panel	☐ NGS	4 WEEKS
MGM146	Aicardi-Goutieres syndrome gene panel	☐ NGS	4 WEEKS
MGM147	Cockayne syndrome gene panel	☐ NGS	4 WEEKS
MGM148	Comprehensive neurology panel	☐ NGS	6 WEEKS
MGM304	Cystic megalencephaly (*MLC1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM150	Cystic megalencephaly (*MLC1*) gene analysis	☐ NGS	4 WEEKS
MGM138	Dysferlinopathy/LGMD2A (*CAPN3*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM139	Dysferlinopathy/LGMD2B (*DYSF*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM127	Episodic ataxia gene panel	☐ NGS	4 WEEKS
MGM151	Familial hemiplegic migraine gene panel	☐ NGS	4 WEEKS
MGM152	Hereditary spastic paraplegia gene panel	☐ NGS	4 WEEKS
MGM128	Hyperekplexia gene panel	☐ NGS	4 WEEKS
MGM153	Joubert syndrome gene panel	☐ NGS	4 WEEKS
MGM120	Krabbe disease (*GALC*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM119	Krabbe disease (*GALC*) gene analysis	☐ NGS	4 WEEKS
MGM121	Leukodystrophy gene panel	☐ NGS	4 WEEKS
MGM140	Limb-girdle muscular dystrophy deletion/duplication analysis (*SGCA, SGCB,SGCD, SGCG* & *FKRP*)	☐ MLPA	3 WEEKS
MGM154	Lissencephaly gene panel	☐ NGS	4 WEEKS
MGM122	Metachromatic leukodystrophy gene panel	☐ NGS	4 WEEKS
MGM155	Microcephaly gene panel	☐ NGS	4 WEEKS
MGM141	Muscular dystrophy & congenital myopathy gene panel	☐ NGS	4 WEEKS
MGM142	Myotonia congenita gene panel	☐ NGS	4 WEEKS
MGM313	Neuronal ceroid lipofuscinosis (*CLN3, PPT1, TPP1*) deficiency deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM156	Neuronal ceroid lipofuscinosis gene panel	☐ NGS	4 WEEKS
MGM157	Neuronal migration disorder gene panel	☐ NGS	4 WEEKS
MGM158	*NOTCH3* (CADASIL) gene analysis	☐ NGS	4 WEEKS
MGM159	*NOTCH3* (CADASIL) gene analysis -2 exons (2 & 3)	☐ Sanger	10 DAYS
MGM129	Pantothenate kinase-associated degeneration (*PANK2*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM130	Pantothenate kinase-associated neurodegeneration gene panel	☐ NGS	4 WEEKS
MGM160	Pontocerebellar hypoplasia gene panel	☐ NGS	4 WEEKS
MGM161	Rett syndrome (*MECP2*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM162	Rett syndrome gene panel	☐ NGS	4 WEEKS
MGM144	Spinal muscular atrophy (*SMN1*) gene analysis	☐ Sanger	4 WEEKS
MGM143	Spinal muscular atrophy (*SMN1/SMN2*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM321	Spinal muscular atrophy gene panel	☐ NGS	4 WEEKS
MGM279	Spinocerebellar ataxia 1 (*ATXN1*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM283	Spinocerebellar ataxia 10 (*ATXN10*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM284	Spinocerebellar ataxia 12 (*PPP2R2B*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM280	Spinocerebellar ataxia 2 (*ATXN2*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM281	Spinocerebellar ataxia 3 (*ATXN3*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM282	Spinocerebellar ataxia 6 (*CACNA1A*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM322	Spinocerebellar ataxia 7 (*ATXN7*) repeat expansion analysis	☐ Fragment analysis - PCR	2 WEEKS
MGM285	Spinocerebellar ataxia repeat expansion analysis: Any two of (SCA1, SCA2, SCA3, SCA6, SCA7, SCA10, SCA12)	☐ Fragment analysis - PCR	2 WEEKS
MGM286	Spinocerebellar ataxia repeat expansion analysis: SCA1, SCA2, SCA3, SCA12	☐ Fragment analysis - PCR	2 WEEKS
MGM287	Spinocerebellar ataxia repeat expansion analysis: SCA1, SCA2, SCA3, SCA6, SCA7, SCA10, SCA12	☐ Fragment analysis - PCR	4 WEEKS
MGM163	Tay-Sachs disease (*HEXA*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM164	Tay-Sachs disease (*HEXA*) gene analysis	☐ NGS	4 WEEKS
MGM166	X-linked mental retardation gene panel	☐ NGS	4 WEEKS
MGM165	X-linked spastic paraplegia-2 (PLP1) deletion/duplication analysis	☐ MLPA	3 WEEKS

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NEUROLOGY (continued)

CODE	TEST	METHOD	TAT
	Neurology Immunology (**Drug Reactions**)		
MGM337	HLA-B* 1502 for carbamazepine toxicity	☐ NGS	10 DAYS
	Neurology-Epilepsy		
MGM109	Benign infantile epilepsy gene panel	☐ NGS	4 WEEKS
MGM111	Dravet syndrome (*SCN1A*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM110	Dravet syndrome (*SCN1A*) gene analysis	☐ NGS	4 WEEKS
MGM305	Early infantile epileptic encephalopathy-4 (*STXBP1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM112	Epileptic encephalopathy gene panel	☐ NGS	4 WEEKS
MGM113	Familial female mental retardation/epilepsy gene panel	☐ NGS	4 WEEKS
MGM114	Progressive myoclonic epilepsy gene panel	☐ NGS	4 WEEKS
	Neurology-leukodystrophies		
MGM117	Canavan disease (*ASPA*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM116	Canavan disease (*ASPA*) gene analysis	☐ NGS	4 WEEKS
MGM118	Hypomyelination syndrome gene panel	☐ NGS	4 WEEKS

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ONCOLOGY

CODE	TEST	METHOD	TAT	PREFERRED SAMPLE TYPE
MGM168	ALL risk stratification gene panel - B-ALL	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow / Purified Genomic DNA
MGM169	ALL risk stratification gene panel - T-ALL	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow / Purified Genomic DNA
MGM170	AML risk stratification gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow / Purified Genomic DNA
MGM171	*APC* (Adenomatous polyposis coli) gene analysis	☐ NGS	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM172	*APC* (Adenomatous polyposis coli) gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM173	*BCR-ABL* qualitative gene fusion analysis	☐ Qualitative RT-PCR	5 DAYS	Peripheral Blood / Bone Marrow
MGM174	*BCR-ABL* quantitative (International Scale) gene fusion analysis	☐ RT-PCR	7 DAYS	Peripheral Blood / Bone Marrow
MGM175	*BCR-ABL* quantitative gene fusion analysis	☐ RT-PCR	7 DAYS	Peripheral Blood / Bone Marrow
MGM334	Bladder cancer - sensitivity to mTOR inhibitors	☐ NGS	7 WEEKS	Fresh frozen tissue/FFPE blocks & Peripheral Blood
MGM176	*BRAF* gene analysis - 2 exons (11 & 15)	☐ Sanger	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM177	*BRAF* V600E mutation analysis	☐ RT-PCR	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM178	*BRCA1 & BRCA2* deletion/duplication analysis	☐ MLPA	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM179	*BRCA1 & BRCA2* gene analysis	☐ NGS	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM180	*BRCA1 & BRCA2* gene analysis	☐ Sanger	6 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM181	*BRCA1* gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM182	*BRCA2* gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM335	Breast cancer - NGS theranostic panel	☐ NGS	8 WEEKS	Fresh frozen tissue/FFPE blocks & Peripheral Blood
MGM183	Carney complex (*PRKAR1A*) gene analysis	☐ NGS	4 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM330	CEL/HES (Chronic eosinophilic leukemia/hyper eosinophilic syndrome) diagnostic and prognostic gene panel	☐ NGS	4 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM184	CMPD (Chronic myeloproliferative disorder gene panel)	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM185	CLL prognostication/risk stratification gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM186	CML/CNL (chronic myeloid leukemia/chronic neutrophilic leukemia - atypical) - prognostication gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM329	CMML (Chronic myelomonocytic leukemia) risk stratification gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM332	Colorectal cancer - theranostic panel	☐ NGS	5 WEEKS	FFPE blocks & Peripheral Blood
MGM340	*DPYD* (IVS14+1G>A) mutation analysis	☐ Sanger	7 DAYS	Peripheral Blood / Purified Genomic DNA
MGM339	*DYPD* gene analysis (for 5 FU toxicity evaluation)	☐ NGS	4 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM189	*EGFR* gene analysis - 4 exons (18, 19, 20, 21):	☐ Sanger	10 DAYS	Fresh frozen tissue/FFPE blocks
MGM190	*EGFR* gene analysis (Hot Spot)	☐ RT-PCR	7 DAYS	Fresh frozen tissue/FFPE blocks
MGM193	*FLT3* gene internal tandem duplication analysis and D835 point mutation analysis	☐ Fragment analysis & Sanger	2 WEEKS	Bone Marrow / Purified Genomic DNA

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MEDGENOME

GENETIC TEST SELECTION LIST:

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Oncology (continued)

CODE	TEST	METHOD	TAT	PREFERRED SAMPLE TYPE
MGM333	GIST (Gastrointestinal stromal tumor) - theranostic panel (Hot Spot)	☐ NGS	6 WEEKS	FFPE blocks & Peripheral Blood
MGM187	Hereditary cancer gene panel - extended	☐ NGS	6 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM194	Hereditary cancer gene panel - focussed	☐ NGS	6 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM197	*IDH1* gene analysis (exon 4)	☐ Sanger	7 DAYS	Peripheral Blood; Fresh frozen tissue/FFPE blocks
MGM198	Imatinib Resistance (*ABL* kinase) gene analysis:	☐ Sanger	10 DAYS	Peripheral Blood / Bone Marrow
MGM199	*JAK2* gene analysis - 2 exons (12, 14)	☐ Sanger	7 DAYS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM200	JMML (Juvenile myelomonocytic leukemia) risk stratification gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM201	*KIT* gene analysis - 2 exons (9,11)	☐ Sanger	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM202	*KIT* gene analysis - 4 exons (9, 11, 13, 17)	☐ Sanger	10 DAYS	Fresh frozen tissue / FFPE blocks
MGM204	*KRAS* gene analysis - 2 exons (2 & 3)	☐ Sanger	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM203	*KRAS* gene analysis (Hot Spot)	☐ RT-PCR	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM205	LCH/hairy cell leukemia - *BRAF* V600E mutation analysis	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow
MGM070	Lymphoproliferative disorder gene panel	☐ NGS	4 WEEKS	Fresh frozen tissue / FFPE blocks
MGM311	Lynch syndrome *(MLH1, MSH2, EPCAM)* deletion/duplication analysis	☐ MLPA	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM167	Lynch syndrome gene panel	☐ NGS	4 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM206	MDS (Myelodysplastic syndrome) prognostication and risk stratification gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow
MGM191	MDS-AML (Myelodysplastic syndrome-acute myeloid leukemia) familial gene panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM192	MDS-AML (Myelodysplastic Syndrome-Acute myeloid leukemia) Familial gene panel-reflex	☐ MLPA	3 WEEKS	Peripheral Blood / Bone Marrow/ Purified Genomic DNA
MGM336	Melanoma - theranostic panel	☐ NGS	9 WEEKS	FFPE blocks & Peripheral Blood
MGM207	*MGMT* gene methylation analysis (Temozolomide resistance)	☐ RT-PCR	2 WEEKS	FFPE blocks
MGM208	Myeloproliferative neoplasms - *CALR* (exon 9) gene analysis	☐ Fragment analysis	7 DAYS	Peripheral Blood / Bone Marrow
MGM209	Myeloproliferative neoplasms - *JAK2* V617F mutation analysis	☐ Sanger	7 DAYS	Peripheral Blood / Bone Marrow
MGM210	Myeloproliferative neoplasms - *MPL* (exon 10) gene analysis	☐ Sanger	7 DAYS	Peripheral Blood / Bone Marrow
MGM211	Myeloproliferative neoplasms prognostication and risk stratification panel	☐ NGS	3 WEEKS	Peripheral Blood / Bone Marrow
MGM290	Multiple myeloma gene panel	☐ NGS	4 WEEKS	Peripheral Blood / Bone marrow/ Purified Genomic DNA
MGM289	MPN (Myeloproliferative neoplasm) reflex Panel	☐ Sanger	10 DAYS	Peripheral Blood / Bone Marrow
MGM331	NSCLC (Non small cell lung cancer) NGS Panel (Hot Spot)	☐ NGS	4 WEEKS	FFPE blocks & Peripheral Blood

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Oncology (continued)

CODE	TEST	METHOD	TAT	PREFERRED SAMPLE TYPE
MGM216	*NPM1* gene analysis (Hot Spot - exon 12)	☐ Fragment analysis	2 WEEKS	Peripheral Blood / Bone Marrow
MGM217	*NRAS* gene analysis (Hot Spot)	☐ RT-PCR	7 DAYS	Fresh frozen tissue / FFPE blocks
MGM218	*PDGFRA* gene analysis - 3 exons (12, 14, 18)	☐ Sanger	10 DAYS	Peripheral blood/Bone marrow/Fresh frozen tissue / FFPE blocks
MGM219	*PML-RARα* gene fusion analysis (Qualitative)	☐ RT-PCR	10 DAYS	Peripheral Blood / Bone Marrow
MGM220	*PTEN* gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM291	*RAS* extended profiling analysis by NGS	☐ NGS	12 DAYS	Fresh frozen tissue/FFPE blocks
MGM223	*RET* oncogene - 2 exons (10, 11)	☐ Sanger	10 DAYS	Peripheral Blood / Purified Genomic DNA
MGM224	*RET* oncogene - 8 exons (5, 8, 10, 11, 13, 14, 15, 16)	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM225	*ROS1* gene fusion analysis	☐ RT-PCR	2 WEEKS	FFPE blocks
MGM196	Somatic cancer mutation - Hot Spot tumor panel	☐ NGS	4 WEEKS	Fresh frozen tissue/FFPE blocks & Peripheral Blood in 2 Streck Tubes
MGM226	*STK11* gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
Tuberous sclerosis - refer to neurology				
MGM227	*TP53* gene analysis	☐ NGS	4 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM228	*TP53* gene analysis	☐ Sanger	3 WEEKS	Peripheral Blood / Purified Genomic DNA
MGM235	*VHL* (Von Hippel-Lindau syndrome) gene analysis	☐ Sanger	10 DAYS	Peripheral Blood / Purified Genomic DNA
MGM323	*VHL* (Von Hippel-Lindau syndrome) deletion/duplication analysis	☐ MLPA	3 WEEKS	Peripheral Blood / Purified Genomic DNA
IMMUNOHISTOCHEMISTRY*				
MGM236	*ALK* D5F3	☐ IHC	4 DAYS	FFPE blocks
MGM237	*BRAF* V600E IHC analysis	☐ IHC	4 DAYS	FFPE blocks
MGM239	Breast prognostic/predictive IHC1 panel (ANY ONE OF - ER, PR,Her2/neu, MIB-1/Ki67)	☐ IHC	4 DAYS	FFPE blocks
MGM240	Breast prognostic/predictive IHC2 panel (ER, PR)	☐ IHC	4 DAYS	FFPE blocks
MGM241	Breast prognostic/predictive IHC3 panel (ER, PR, Her2/neu)	☐ IHC	4 DAYS	FFPE blocks
MGM242	Breast prognostic/predictive IHC4 panel (ER, PR, Her2/neu, MIB-1/Ki67)	☐ IHC	4 DAYS	FFPE blocks
MGM238	c-MET IHC analysis	☐ IHC	4 DAYS	FFPE blocks
MGM312	Lynch syndrome mismatch repair (MMR) 4 gene (MLH1, MLH2, MSH6 & PMS2) panel - IHC [Microsatellite instability]	☐ IHC	4 DAYS	FFPE blocks
MGM243	ROS1 IHC analysis	☐ IHC	4 DAYS	FFPE blocks

***Note:** If we receive test requests for two or more non ER, PR IHC markers for a single patient (example: ALK, ROS1 and cMET) - the TAT will be 6 days.

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OPHTHALMOLOGY [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM303	Corneal dystrophy gene panel	☐ NGS	4 WEEKS
MGM244	Bardet-Biedl syndrome gene panel	☐ NGS	4 WEEKS
MGM245	Cataract gene panel (congenital/ developmental)		4 WEEKS
MGM249	Congenital stationary night blindness gene panel	☐ NGS	4 WEEKS
MGM252	Leber congenital amaurosis gene panel	☐ NGS	4 WEEKS
MGM254	Microphthalmia and anophthamia gene panel	☐ NGS	4 WEEKS
MGM256	Optic atrophy gene panel:	☐ NGS	4 WEEKS
MGM221	*RB1* gene analysis	☐ NGS	4 WEEKS
MGM222	*RB1* gene deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM257	Retinal degeneration gene panel	☐ NGS	4 WEEKS
MGM260	Usher syndrome (*USH2A*) deletion/ duplication analysis	☐ MLPA	3 WEEKS
MGM259	Usher syndrome gene panel	☐ NGS	4 WEEKS

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MISCELLANEOUS (includes syndromes) [preferred sample type - EDTA blood/DNA]

CODE	TEST	METHOD	TAT
MGM277	Additional family member (investigational) testing	☐ Sanger	4 WEEKS
MGM261	Alagille syndrome gene panel	☐ NGS	4 WEEKS
MGM272	Clinical exome - 20MB (80-100x)	☐ NGS	6 WEEKS
MGM328	Clinical exome with reflex sanger for investigational - 20MB (80-100x)	☐ NGS & Sanger	6 WEEKS
MGM263	Ciliopathy gene panel	☐ NGS	4 WEEKS
MGM264	Cohen's syndrome (*VPS13B*) gene analysis	☐ NGS	4 WEEKS
MGM265	Cornelia de Lange syndrome gene panel	☐ NGS	4 WEEKS
MGM268	Craniosynostosis gene panel	☐ NGS	4 WEEKS
MGM315	Non-immune hydrops gene panel	☐ NGS	4 WEEKS
MGM266	Noonan syndrome gene panel	☐ NGS	4 WEEKS
MGM267	Pallister Hall syndrome (*GLI3*) gene analysis	☐ NGS	4 WEEKS
MGM269	Smith-Lemli-Opitz syndrome (*DHCR7*) gene analysis	☐ NGS	4 WEEKS
MGM271	Sotos syndrome (*NSD1*) deletion/duplication analysis	☐ MLPA	3 WEEKS
MGM270	Sotos syndrome gene panel	☐ NGS	4 WEEKS
MGM278	Raw data charges	☐ NA	7 DAYS
MGM326	TRIO - Whole exome sequencing - 45-50MB (80-100x)	☐ NGS	6 WEEKS
MGM327	TRIO Reflex - Clinical Exome sequencing - 20MB (80-100x)	☐ NGS & Sanger	6 WEEKS
MGM274	Whole exome sequencing - 50MB(80-100x)	☐ NGS	6 WEEKS
MGM275	Whole genome sequencing (mean 30x)	☐ NGS	8 WEEKS
MGM276	Whole mitochondrial genome sequencing	☐ NGS	2 WEEKS

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Panorama™ Non invasive prenatal screening test

CODE	TEST	METHOD	TAT
MGM273	Panorama™ Non invasive prenatal screening test or (NIPS) (Trisomy 21, Trisomy 18, Trisomy 13, Monosomy X and Triploidy) For NIPS, 2 x 10 ml blood in Streck tubes should be collected from the pregnant woman and a cheek swab from the father of the pregnancy (optional) [Note: Please request for an exclusive kit, that includes the Streck tubes, cheek swab, TRF, syringe and needle as well as sample collection instructions from a MedGenome representative].	☐ NGS	12 DAYS

Pre-Implantation Genetic Screening (PGS)

CODE	TEST	METHOD	TAT
MGM297	Pre-Implantation Genetic Screening (PGS) For PGS, A few trophectoderm cells from a day 5 embryo [Note: Please request for an exclusive PGS kit, that includes the PCR tubes, Cell wash solution(CWS), 1 x PBS for collecting biopsy, TRF, and instruction material from a MedGenome Representative].	☐ NGS	10 DAYS

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RT-PCR: Real-Time PCR**, IHC:** Immunohistochemistry

SAMPLE REQUIREMENT:

- ✓ Blood: 3-4 ml EDTA anticoagulated blood, packed securely to avoid spillage, must be shipped to reach within 48 hours after collection on ice (4-8^{0}C).
- ✓ In case of *BCR-ABL*, *PML-RARα* and *IRMA* testing, 2-3 ml blood collected in PaxGene/EDTA tubes (provided on request) must be packed securely to avoid spillage, must be shipped to reach within 48 hours after collection on ice at (4-8^{0}C).
- ✓ FFPE tissue: 3-4 mm sections of FFPE tumor tissue shipped in ambient temperature.
- ✓ We accept 1 stained slide (H & E) and 10 unstained non-baked slides with 10-um thick sections of FFPE tumor tissue.
- ✓ Tumor section should contain at least 70% tumor content as verified by the pathologist. Pathology report should accompany specimen.
- ✓ Tissue in FFPE blocks/slides may not be available after testing. If available, the sample shall be sent back provided the courier charges are borne by the patient.
- ✓ Fresh frozen tissue: The tumor should be sectioned and a pathology report to be generated that includes an assessment of tumor content Frozen tissue should be shipped in dry ice. Please call us prior to shipping fresh frozen samples.
- ✓ For tumor panel test, FFPE/fresh frozen tissue should be accompanied by matched normal (blood) sample of the patient.
- ✓ DNA of the patient can also be sent directly. If DNA is tumor derived, please confirm if the tumor content was > = 70% and 10-ug high quality DNA A260/A280 value in the range 1.7 - 1.9.
- ✓ Please provide relevant clinical indications for accurate testing. In case of Genome sequencing, Exome sequencing, NGS Panels and testing for germline mutations (ex: BRCA) on blood, provision of the clinical details and family history are mandatory.

MedGenome offers broadest range of genomic tests for diverse clinical specialties

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