



List of Services

Test Code	Test Name / Description	Sample Type, Volume	TAT (days)
Enzymology			
ENZ01	Aromatic L-amino acid Decarboxylase Enzyme Analysis (Plasma)	Frozen Plasma, 1 mL	21
ENZ06	Thymidine phosphorylase Enzyme Analysis (Blood)	Whole Blood, 5 mL	
Metabolic analyses			
MET01	Amino acids (CSF)	Frozen CSF, 10 mL	14-18
MET02	Amino acids (Plasma)	Frozen Plasma, 1 mL	
MET03	Amino acids (Urine)	Urine, 5-10 mL	
MET04	Coenzyme Q10 (Leukocytes)	Whole Blood, 3 mL	
MET05	Coenzyme Q10 (Muscle)	Frozen Muscle, 75 mg	
MET07	Lactate (CSF)	Frozen CSF, 1 mL	
MET08	Lactate (plasma)	Frozen Plasma, 1 mL	
MET09	Phenylalanine Loading Assay (Plasma)		
MET10	Pyruvate (Blood)	Whole Blood, Treated, 1 mL	
MET11	Pyruvate (CSF)	Frozen CSF, 1 mL	
MET12	Thymidine/Deoxyuridine analytes (Plasma)	Frozen Plasma, 1 mL	
MET19	Creatine and Guanidinoacetate (Urine)	Urine, 1 mL	
MET20	Alpha amino adipic semialdehyde (Urine)	Frozen CSF, 1 mL	
MET21	Sialic Acid (Urine)	Urine, 1 mL	
MET23	Creatine and Guanidinoacetate (Plasma)	Frozen Plasma, 1 mL	
MET24	Glucose (Plasma)		
MET29	3-O-Methyldopa (Plasma)		
Neurochemistry analyses			
NC01	5-Methyltetrahydrofolate (CSF)	Frozen CSF, 1 mL	10-14
NC02	Neopterin (CSF)		
NC03	Neopterin/Tetrahydrobiopterin (CSF)		
NC04	Neurotransmitter Metabolites (5HIAA. HVA. 3OMD) (CSF)		
NC05	Pyridoxal 5'-phosphate (CSF)		
NC06	Succinyladenosine (CSF)		
NC07	Sialic Acid (CSF)		
NC08	Alpha-amino adipic semialdehyde (CSF)		
NC10	Glucose (CSF)		
Gene sequencing (SANGER), Deletion/Duplication (MLPA), Point mutations – Individual genes & panels			
MOL001	Mitochondrial DNA Depletion Testing (Muscle)	Frozen Muscle, 75 mg	14-28
MOL002	Mitochondrial DNA Deletion Analysis	Whole Blood (3 mL) or Extracted DNA	
MOL021	Mitochondrial Genome Sequencing		
MOL025	DDC Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL026	ABCB7 Full Gene Sequencing Analysis		
MOL027	ACADM Full Gene Sequencing Analysis		
MOL028	ACADS Full Gene Sequencing Analysis		
MOL029	ACADVL Full Gene Sequencing Analysis		
MOL030	ALDH7A1 Full Gene Sequencing Analysis		
MOL031	SLC25A4 Full Gene Sequencing Analysis		
MOL032	ATP1A2 Full Gene Sequencing Analysis		
MOL033	CACNA1A Full Gene Sequencing Analysis		
MOL034	CAPN3 Full Gene Sequencing Analysis		
MOL035	CAV3 Full Gene Sequencing Analysis		
MOL036	NDUFS1 Full Gene Sequencing Analysis		



List of Services

dynacare.ca

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MOL037	NDUFS4 Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
MOL038	NDUFS7 Full Gene Sequencing Analysis		
MOL039	NDUFS8 Full Gene Sequencing Analysis		
MOL040	NDUFV1 Full Gene Sequencing Analysis		
MOL041	SDHA Full Gene Sequencing Analysis		
MOL042	SDHB Full Gene Sequencing Analysis		
MOL043	SDHC Full Gene Sequencing Analysis		
MOL044	SDHD Full Gene Sequencing Analysis		
MOL045	BCS1L Full Gene Sequencing Analysis		
MOL046	COQ2 Full Gene Sequencing Analysis		
MOL047	COX10 Full Gene Sequencing Analysis		
MOL048	COX15 Full Gene Sequencing Analysis		
MOL049	CPT1A Full Gene Sequencing Analysis		
MOL050	CPT2 Full Gene Sequencing Analysis		
MOL051	DGUOK Full Gene Sequencing Analysis		
MOL052	DYSF Full Gene Sequencing Analysis		
MOL053	TYMP Full Gene Sequencing Analysis		
MOL054	EIF2B1 Full Gene Sequencing Analysis		
MOL055	EIF2B2 Full Gene Sequencing Analysis		
MOL056	EIF2B3 Full Gene Sequencing Analysis		
MOL057	EIF2B4 Full Gene Sequencing Analysis		
MOL058	EIF2B5 Full Gene Sequencing Analysis		
MOL059	FXN Full Gene Sequencing Analysis		
MOL060	GCH1 Full Gene Sequencing Analysis		
MOL061	GYS2 Full Gene Sequencing Analysis		
MOL062	HADHA Full Gene Sequencing Analysis		
MOL063	HADHB Full Gene Sequencing Analysis		
MOL064	KCNA1 Full Gene Sequencing Analysis		
MOL065	LAMA2 Full Gene Sequencing and Copy Number Analysis		
MOL066	LMNA Full Gene Sequencing Analysis		
MOL067	MECP2 Full Gene Sequencing Analysis		
MOL068	MPV17 Full Gene Sequencing Analysis		
MOL069	OPA1 Full Gene Sequencing Analysis		
MOL070	PDHA1 Full Gene Sequencing Analysis		
MOL071	PDSS1 Full Gene Sequencing Analysis		
MOL072	PDSS2 Full Gene Sequencing Analysis		
MOL073	C10ORF2 Full Gene Sequencing Analysis		
MOL074	PNPO Full Gene Sequencing Analysis		
MOL075	POLG Full Gene Sequencing and Copy Number Analysis		
MOL076	PYGM Full Gene Sequencing Analysis		
MOL077	RRM2B Full Gene Sequencing Analysis		
MOL078	SGCA Full Gene Sequencing Analysis		
MOL079	SGCB Full Gene Sequencing Analysis		
MOL080	SGCD Full Gene Sequencing Analysis		
MOL081	SGCG Full Gene Sequencing Analysis		
MOL082	SCO1 Full Gene Sequencing Analysis		
MOL083	SCO2 Full Gene Sequencing Analysis		
MOL085	SLC25A20 Full Gene Sequencing Analysis		



List of Services

dynacare.ca

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MOL086	HSPD1 Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
MOL087	SPG7 Full Gene Sequencing Analysis		
MOL088	SUCLA2 Full Gene Sequencing Analysis		
MOL089	SUCLG1 Full Gene Sequencing Analysis		
MOL090	SURF1 Full Gene Sequencing Analysis		
MOL091	TH Full Gene Sequencing Analysis		
MOL092	TPH2 Full Gene Sequencing Analysis		
MOL093	UBE3A Full Gene Sequencing Analysis		
MOL094	DARS2 Full Gene Sequencing Analysis		
MOL095	SLC18A2 Full Gene Sequencing Analysis		
MOL096	PTS Full Gene Sequencing Analysis		
MOL097	SLC6A3 Full Gene Sequencing Analysis		
MOL098	TIMM8A Full Gene Sequencing Analysis		
MOL099	MRPS16 Full Gene Sequencing Analysis		
MOL100	UQCRB Full Gene Sequencing Analysis		
MOL101	NDUFAF2 Full Gene Sequencing Analysis		
MOL102	NDUFA1 Full Gene Sequencing Analysis		
MOL103	NDUFS2 Full Gene Sequencing Analysis		
MOL104	NDUFS3 Full Gene Sequencing Analysis		
MOL105	NDUFV2 Full Gene Sequencing Analysis		
MOL106	NDUFAF1 Full Gene Sequencing Analysis		
MOL107	PUS1 Full Gene Sequencing Analysis		
MOL108	TUFM Full Gene Sequencing Analysis		
MOL109	POLG2 Full Gene Sequencing Analysis		
MOL110	APTX Full Gene Sequencing Analysis		
MOL111	TAZ Full Gene Sequencing Analysis		
MOL112	ATPAF2 Full Gene Sequencing Analysis		
MOL113	GFM1 Full Gene Sequencing Analysis		
MOL114	MFN2 Full Gene Sequencing Analysis		
MOL115	LRPPRC Full Gene Sequencing Analysis		
MOL116	GLRA1 Full Gene Sequencing Analysis		
MOL117	QDPR Full Gene Sequencing Analysis		
MOL118	RNASEH2B Full Gene Sequencing Analysis		
MOL119	TREX1 Full Gene Sequencing Analysis		
MOL120	GLRB Full Gene Sequencing Analysis		
MOL121	SLC2A1 Full Gene Sequencing Analysis		
MOL122	GFAP Full Gene Sequencing Analysis		
MOL124	RNASEH2A Full Gene Sequencing Analysis		
MOL125	ALDH5A1 Full Gene Sequencing Analysis		
MOL126	SPR Full Gene Sequencing Analysis		
MOL127	SLC6A5 Full Gene Sequencing Analysis		
MOL128	COQ3 Full Gene Sequencing Analysis		
MOL129	COQ4 Full Gene Sequencing Analysis		
MOL131	COQ6 Full Gene Sequencing Analysis		
MOL132	COQ7 Full Gene Sequencing Analysis		
MOL133	COQ9 Full Gene Sequencing Analysis		
MOL137	TPM3 Full Gene Sequencing Analysis		
MOL138	ZMPSTE24 Full Gene Sequencing Analysis		



List of Services

dynacare.ca

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MOL139	MTM1 Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
MOL140	ADCK3 Full Gene Sequencing Analysis		
MOL141	DBH Full Gene Sequencing Analysis		
MOL142	ETFDH Full Gene Sequencing Analysis		
MOL143	POMT2 Full Gene Sequencing Analysis		
MOL144	MYOT Full Gene Sequencing Analysis		
MOL145	EMD Full Gene Sequencing Analysis		
MOL146	HCCS Full Gene Sequencing Analysis		
MOL147	BRAF Full Gene Sequencing Analysis		
MOL148	TCAP Full Gene Sequencing Analysis		
MOL149	FKRP Full Gene Sequencing Analysis		
MOL150	FKTN Full Gene Sequencing Analysis		
MOL151	NDUFAF5 Full Gene Sequencing Analysis		
MOL152	RARS2 Full Gene Sequencing Analysis		
MOL153	POMT1 Full Gene Sequencing Analysis		
MOL154	RYR1 Full Gene Sequencing and Copy Number Analysis		
MOL155	GFER Full Gene Sequencing Analysis		
MOL156	TMEM126A Full Gene Sequencing Analysis		
MOL157	NDUFS6 Full Gene Sequencing Analysis		
MOL158	NDUFAF3 Full Gene Sequencing Analysis		
MOL159	TNNI2 Full Gene Sequencing Analysis		
MOL160	NDUFA2 Full Gene Sequencing Analysis		
MOL161	NDUFA11 Full Gene Sequencing Analysis		
MOL162	NDUFAF4 Full Gene Sequencing Analysis		
MOL163	ETFA Full Gene Sequencing Analysis		
MOL164	ETFB Full Gene Sequencing Analysis		
MOL165	FASTKD2 Full Gene Sequencing Analysis		
MOL166	FOLR1 Full Gene Sequencing Analysis		
MOL167	GARS Full Gene Sequencing Analysis		
MOL168	RNASEH2C Full Gene Sequencing Analysis		
MOL169	SLC25A19 Full Gene Sequencing Analysis		
MOL170	SLC17A5 Full Gene Sequencing Analysis		
MOL171	MTHFR Full Gene Sequencing Analysis		
MOL172	SAMHD1 Full Gene Sequencing Analysis		
MOL173	TMEM70 Full Gene Sequencing Analysis		
MOL174	UQCRCQ Full Gene Sequencing Analysis		
MOL182	COX6B1 Full Gene Sequencing Analysis		
MOL183	AIFM1 Full Gene Sequencing Analysis		
MOL184	PHOX2B Full Gene Sequencing Analysis		
MOL185	AMPD1 Full Gene Sequencing Analysis		
MOL186	SLC2A1 MLPA Duplication/Deletion Analysis		
MOL187	MECP2 MLPA Duplication/Deletion Analysis		
MOL189	Mitochondrial Genome Sequencing + Deletion Analysis		
MOL208	CACNA1A Copy Number Analysis		
MOL209	ADSL Full Gene Sequencing Analysis		
MOL210	FOXRED1 Full Gene Sequencing Analysis		
MOL211	NUBPL Full Gene Sequencing Analysis		
MOL212	GLDC Full Gene Sequencing Analysis		



List of Services

dynacare.ca

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MOL213	GCDH Full Gene Sequencing	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL214	ATP1A2 MLPA Duplication/Deletion Analysis		
MOL215	GCH1 MLPA Duplication/Deletion Analysis		
MOL216	KCNQ2 MLPA Duplication/Deletion Analysis		
MOL217	PDHA1 MLPA Duplication/Deletion Analysis		
MOL218	SCN1A MLPA Duplication/Deletion Analysis		
MOL219	GLDC MLPA Duplication/Deletion Analysis		
MOL222	PDHB Full Gene Sequencing Analysis		
MOL223	PDP1 Full Gene Sequencing Analysis		
MOL224	DLAT Full Gene Sequencing Analysis		
MOL225	DLD Full Gene Sequencing Analysis		
MOL226	PC Full Gene Sequencing Analysis		
MOL227	CACNB4 Full Gene Sequencing Analysis		
MOL228	SLC1A3 Full Gene Sequencing Analysis		
MOL229	KCNQ2 Full Gene Sequencing Analysis		
MOL231	SLC2A1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis	Whole Blood or Extracted DNA	
MOL232	Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Leukocyte)		
MOL233	SLC12A3 Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
MOL234	GCH1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL235	PDHA1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL236	GLDC Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL237	DHFR Full Gene Sequencing Analysis		
MOL238	FOXP2 Full Gene Sequencing Analysis		
MOL239	CAPN3 MLPA Duplication/Deletion Analysis		
MOL240	CAPN3 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL241	TACO1 Full Gene Sequencing Analysis		
MOL242	ACAD8 Full Gene Sequencing Analysis		
MOL243	ACAD9 Full Gene Sequencing Analysis		
MOL244	SLC12A3 MLPA Duplication/Deletion Analysis		
MOL245	SLC12A3 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL246	SLC6A8 Full Gene Sequencing Analysis		
MOL247	SGCE Full Gene Sequencing Analysis		
MOL248	SCN1A Full Gene Sequencing and Copy Number Analysis		
MOL249	Known Familial Variant (KFV) Analysis (each mutation)		
MOL250	MPZ Full Gene Sequencing Analysis		
MOL251	GPHN Full Gene Sequencing Analysis		
MOL252	SLC6A4 Full Gene Sequencing Analysis		
MOL253	PGAM2 Full Gene Sequencing Analysis		
MOL254	CHKB Full Gene Sequencing Analysis		
MOL255	SCN1B Full Gene Sequencing Analysis		
MOL256	SPAST Full Gene Sequencing Analysis		
MOL257	MECP2 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL258	SCN9A Full Gene Sequencing and Copy Number Analysis		
MOL259	Friedreich Ataxia Genetic Testing (Repeat Expansion)		
MOL260	ACADVL MLPA Duplication/Deletion Analysis		



List of Services

dynacare.ca

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MOL261	ACADVL Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
MOL262	SLC22A5 MLPA Duplication/Deletion Analysis		
MOL264	SPAST MLPA Duplication/Deletion Analysis		
MOL265	SPAST full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL266	ATL1/SPG3 MLPA Duplication/Deletion Analysis		
MOL267	ATL1/SPG3 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL268	CLCNKA Full Gene Sequencing Analysis		
MOL270	KCNJ1 Full Gene Sequencing Analysis		
MOL271	BSND Full Gene Sequencing Analysis		
MOL272	PFKM Full Gene Sequencing Analysis		
MOL274	PDXK Full Gene Sequencing Analysis		
MOL276	PMP22 MLPA Duplication/Deletion Analysis		
MOL277	GJB1 (CX32) MLPA Duplication/Deletion Analysis		
MOL278	MPZ MLPA Duplication/Deletion Analysis		
MOL281	DOK7 Full Gene Sequencing Analysis		
MOL282	SGCE MLPA Duplication/Deletion Analysis		
MOL283	SGCE Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL286	CLCNKB MLPA Duplication/Deletion Analysis		
MOL287	CLCNKB full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL289	MTM1 MLPA Duplication/Deletion Analysis		
MOL290	MTM1 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL291	OPA1 MLPA Duplication/Deletion Analysis		
MOL292	OPA1 Full Gene Sequencing Analysis + MLPA Duplication/Deletion Analysis		
MOL293	MFN2 MLPA Duplication/Deletion Analysis		
MOL294	POLG MLPA Duplication/Deletion Analysis		
MOL295	ATP5E Full Gene Sequencing Analysis		
MOL299	Myotonic Dystrophy 1 (DMPK) Genetic Testing (Repeat expansion)		
MOL301	Spinal Muscular Atrophy (SMN1/SMN2) MLPA Duplication/Deletion Analysis		
MOL302	Dystrophin MLPA Duplication/Deletion Analysis		
MOL303	Myotonic Dystrophy 2 (ZNF9/CNBP) Genetic Testing (Repeat Expansion)		
MOL305	TK2 Full Gene Sequencing Analysis		
MOL306	ARHGEF9 Full Gene Sequencing Analysis		
MOL307	CACNA1S Full Gene Sequencing Analysis		
MOL308	GLUL Full Gene Sequencing Analysis		
MOL309	ADAR Full Gene Sequencing Analysis		
MOL313	C10ORF2 MLPA Duplication/Deletion Analysis		
MOL314	FKRP MLPA Duplication/Deletion Analysis		
MOL315	GARS MLPA Duplication/Deletion Analysis		
MOL316	HSPB1 MLPA Duplication/Deletion Analysis		
MOL317	LAMA2 MLPA Duplication/Deletion Analysis		
MOL318	MPV17 MLPA Duplication/Deletion Analysis		



List of Services

dynacare.ca

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MOL319	NEFL MLPA Duplication/Deletion Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL320	PRX MLPA Duplication/Deletion Analysis		
MOL321	RAB7A MLPA Duplication/Deletion Analysis		
MOL322	RRM2B MLPA Duplication/Deletion Analysis		
MOL324	SGCA MLPA Duplication/Deletion Analysis		
MOL325	SGCB MLPA Duplication/Deletion Analysis		
MOL326	SGCD MLPA Duplication/Deletion Analysis		
MOL327	SGCG MLPA Duplication/Deletion Analysis		
MOL328	SLC25A4 MLPA Duplication/Deletion Analysis		
MOL329	TK2 MLPA Duplication/Deletion Analysis		
MOL330	KCNQ2 Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL331	SPTLC1 Full Gene Sequencing Analysis		
MOL332	GJB1 (CX32) Full Gene Sequencing Analysis		
MOL333	GJB1 (CX32) Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL334	Mitochondrial DNA Depletion Testing (Leukocyte)	Whole Blood (3 mL)	14-28
MOL335	LITAF Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL336	SEPT9 Full Gene Sequencing and Copy Number Analysis		
MOL339	DARS Full Gene Sequencing Analysis	Frozen Muscle, 75 mg	
MOL340	Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Muscle)		
MOL341	GNE Full Gene Sequencing Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL342	DPYS Full Gene Sequencing Analysis		
MOL343	SPG11 MLPA Duplication/Deletion Analysis		
MOL344	PMP22 Full Gene Sequencing Analysis		
MOL345	PMP22 Full Gene Sequencing + MLPA Duplication/Deletion Analysis		
MOL346	SCN10A Full Gene Sequencing Analysis		
MOL347	SCN11A Full Gene Sequencing Analysis		
MOL348	SCN5A Full Gene Sequencing Analysis		
MOL349	PAH Full Gene Sequencing Analysis		
MOL350	PKD1/PKD2 MLPA Duplication/Deletion Analysis		
MOL351	GAMT Full Gene Sequencing Analysis		
MOL352	GATM Full Gene Sequencing Analysis		
MOL354	DEPDC5 Full Gene Sequencing Analysis		
MOL355	CLCN1 Full Gene Sequencing Analysis		
MOL356	SCN4A Full Gene Sequencing Analysis		
MOL360	PEX6 Full Gene Sequencing Analysis		
MOL362	TUBB4A Full Gene Sequencing Analysis		
MOL364	C9orf72 Repeat Expansion Analysis		
MOL366	Huntington Disease: HTT Repeat Expansion Analysis		
MOL367	PROSC Full Gene Sequencing Analysis		
MOL368	SCA1 (ATXN1) Genetic Testing (Repeat expansion)		
MOL369	SCA2 (ATXN2) Genetic Testing (Repeat expansion)		
MOL370	SCA3 (ATXN3) Genetic Testing (Repeat expansion)		
MOL371	SCA6 (CACNA1A) Genetic Testing (Repeat expansion)		
MOL372	SCA7 (ATXN7) Genetic Testing (Repeat expansion)		
MOL373	SCA8 (ATXN8) Genetic Testing (Repeat expansion)		
MOL374	SCA10 (ATXN10) Genetic Testing (Repeat expansion)		



List of Services

dynacare.ca

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MOL375	SCA12 (PPP2R2B) Genetic Testing (Repeat expansion)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL376	SCA17 (TBP) Genetic Testing (Repeat expansion)		
MOL377	SCA36 (NOP56) Genetic Testing (Repeat expansion)		
MOL378	DRPLA (ATN1) Genetic Testing (Repeat expansion)		
MOL379	Spinocerebellar Ataxia Repeat Panel (SCA 8, 10, 12, 17, 36, & DRPLA)	Whole Blood (3 mL) or Extracted DNA	14-28
MOL380	Comprehensive Spinocerebellar Ataxia Repeat Expansion Panel (SCA 1, 2, 3, 6, 7, 8, 10, 12, 17, 36, & DRPLA)		
MOL381	Fragile X Repeat Expansion + Methylation Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
MOL388	CYP21A2 (Congenital Adrenal Hyperplasia) Deletion/Duplication and Selected Variant Analysis		
MOL389	GBA (Gaucher Disease) Deletion/Duplication Analysis		
MOL390	HBA1 and HBA2 (Alpha-thalassemia) Deletion/Duplication and Selected Variant Analysis		
MOL391	Comprehensive Ataxia Repeat Expansion Panel (SCA 1, 2, 3, 6, 7, 8, 10, 12, 17, 36, DRPLA & FRDA)	Whole Blood (3 mL) or Extracted DNA	21-28
MOL392	Huntington-like Disease Type 2: JPH3 Repeat Expansion Analysis	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
Tissue culture			
TC01	Cryopreservation of cells	N/A	14-28
TC03	Thaw & Expand cells		
TC04	Tissue Culture of Lymphocytes with EBV Transformation		
TC05	Mycoplasma testing of cells		
TC06	Tissue Culture of skin fibroblasts (includes skin biopsy processing)		
Next generation sequencing (NGS) panels			
NGS197	Coenzyme Q10 Deficiency (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
NGS198	Comprehensive mtDNA Depletion Syndromes (NGS Panel and Copy Number Analysis)		
NGS301	Comprehensive Cellular Energetics Defects (NGS Panel and Copy Number Analysis + mtDNA)		
NGS302	Carbohydrate Metabolism Deficiency (NGS Panel and Copy Number Analysis + mtDNA)		
NGS303	Lipid Metabolism Deficiency (NGS Panel and Copy Number Analysis + mtDNA)		
NGS304	Pyruvate Metabolism Disorders (NGS Panel and Copy Number Analysis + mtDNA)		
NGS305	PDH/Tricarboxylic Acid Cycle (TCA) Defects (NGS Panel and Copy Number Analysis + mtDNA)		
NGS306	Oxidative Phosphorylation (OXPHOS) Defects (NGS Panel and Copy Number Analysis + mtDNA)		
NGS307	Ceroid Lipofuscinosis Disorders (NGS Panel and Copy Number Analysis)		
NGS308	Creatine Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS309	Cobalamin/Homocysteine/Methionine Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS310	GABA Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS311	Glutaric Acidemia Disorders (NGS Panel and Copy Number Analysis)		
NGS312	Ketone Body Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS313	Lysosomal Disease (NGS Panel and Copy Number Analysis)		



List of Services

dynacare.ca

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NGS314	Methylmalonic Acid Metabolism Deficiency (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
NGS315	Neurotransmitter Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS316	Dopamine Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS317	Serotonin Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS318	Tetrahydrofolate Metabolism Deficiency (NGS Panel and Copy Number Analysis)		
NGS319	Fever Syndromes (NGS Panel and Copy Number Analysis)		
NGS320	Tyrosinemia (NGS Panel and Copy Number Analysis)		
NGS321	Urea Cycle Disorders (NGS Panel and Copy Number Analysis)		
NGS323	Amyotrophic Lateral Sclerosis (NGS Panel and Copy Number Analysis)		
NGS324	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA)		
NGS325	Comprehensive Intellectual Disability/Autism (NGS Panel and Copy Number Analysis + mtDNA)		
NGS327	Congenital Glycosylation Disorders (NGS Panel and Copy Number Analysis)		
NGS330	Comprehensive Muscular Dystrophy/Myopathy (NGS Panel and Copy Number Analysis + mtDNA)		
NGS331	Congenital Myasthenic Syndromes (NGS Panel and Copy Number Analysis)		
NGS332	Hypokalemic and Hyperkalemic Periodic Paralysis Disorders (NGS Panel and Copy Number Analysis)		
NGS333	Malignant Hyperthermia (NGS Panel and Copy Number Analysis)		
NGS335	Neurofibromatosis (NGS Panel and Copy Number Analysis)		
NGS337	Spastic Paraplegia (NGS Panel and Copy Number Analysis + mtDNA)		
NGS343	Peroxisomal Disease (NGS Panel and Copy Number Analysis)		
NGS344	Aicardi-Goutieres Syndrome (NGS Panel and Copy Number Analysis)		
NGS345	Charcot-Marie-Tooth Disease (NGS Panel and Copy Number Analysis + mtDNA)		
NGS345A	Charcot-Marie-Tooth Disease, Axonal (NGS Panel and Copy Number Analysis + mtDNA)		
NGS345D	Charcot-Marie-Tooth Disease, Demyelinating (NGS Panel and Copy Number Analysis + mtDNA)		
NGS346	Hereditary Sensory & Autonomic Neuropathy (NGS Panel and Copy Number Analysis)		
NGS347	Spinal Muscular Atrophy (NGS Panel and Copy Number Analysis)		
NGS348	Fetal Akinesia, Arthrogryposis, or Contractures (NGS Panel and Copy Number Analysis)		
NGS349	Nonsyndromic Intellectual Disability (NGS Panel and Copy Number Analysis)		
NGS350	Syndromic Intellectual Disability (NGS Panel and Copy Number Analysis)		
NGS351	Leigh Disease (NGS Panel and Copy Number Analysis + mtDNA)		
NGS352	Comprehensive Ophthalmoplegia Syndromes (NGS Panel and Copy Number Analysis + mtDNA)		
NGS353	Cellular Energetics Ophthalmoplegia Syndromes (NGS Panel and Copy Number Analysis + mtDNA)		



List of Services

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NGS354	Non-mitochondrial Comprehensive Ophthalmoplegia Syndromes (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
NGS355	Cytochrome C Oxidase Deficiency (NGS Panel and Copy Number Analysis + mtDNA)		
NGS356	Alzheimer Disease/Frontotemporal Dementia (NGS Panel and Copy Number Analysis)		
NGS357	Parkinson's Disease/Parkinsonism (NGS Panel and Copy Number Analysis)		
NGS358	Comprehensive Dystonia (NGS Panel and Copy Number Analysis + mtDNA)		
NGS359	Primary Dystonia (NGS Panel and Copy Number Analysis)		
NGS360	Basal Ganglia Calcification Dystonia (NGS Panel and Copy Number Analysis)		
NGS361	OXPPOS Defect Dystonia (NGS Panel and Copy Number Analysis + mtDNA)		
NGS362	Neurodegeneration with Brain Iron Accumulation (NGS Panel and Copy Number Analysis)		
NGS363	Comprehensive Cardiomyopathy (NGS Panel and Copy Number Analysis + mtDNA)		
NGS364	Left Ventricular Noncompaction Cardiomyopathy Syndromes (NGS Panel and Copy Number Analysis)		
NGS365	Hereditary Cardiac Arrhythmia (NGS Panel and Copy Number Analysis)		
NGS366	Hereditary Ventricular Tachycardia Syndromes (NGS Panel and Copy Number Analysis)		
NGS367	Arrhythmogenic Right Ventricular Dysplasia (NGS Panel and Copy Number Analysis)		
NGS368	Long and Short QT Syndrome (NGS Panel and Copy Number Analysis)		
NGS369	Brugada Syndrome (NGS Panel and Copy Number Analysis)		
NGS370	Congenital Heart Defects (NGS Panel and Copy Number Analysis)		
NGS371	Congenital Central Hypoventilation Syndromes (NGS Panel and Copy Number Analysis)		
NGS372	Comprehensive Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis + mtDNA)		
NGS373	Non-Mitochondrial Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis)		
NGS374	Mitochondrial Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis + mtDNA)		
NGS375	Vanishing White Matter, Dysmyelinating, and Hypomyelinating Leukodystrophy (NGS Panel and Copy Number Analysis)		
NGS376	Comprehensive Dementia (NGS Panel and Copy Number Analysis)		
NGS377	Ehlers Danlos, Ehlers Danlos-like Syndromes, and Aneurysm Syndromes (NGS Panel and Copy Number Analysis)		
NGS378	Marfan and Marfan-like Syndromes (NGS Panel and Copy Number Analysis)		
NGS379	Polycystic Kidney Disease (NGS Panel and Copy Number Analysis)		
NGS380	Amyloid Related Disorders (NGS Panel and Copy Number Analysis)		
NGS381	Mucopolysaccharidosis and Mucolipid Disorders (NGS Panel and Copy Number Analysis)		
NGS383	Comprehensive Metabolic Disease Hepatomegaly (NGS Panel and Copy Number Analysis + mtDNA)		
NGS384	Carbohydrate Metabolism Hepatomegaly (NGS Panel and Copy Number Analysis)		



List of Services

dynacare.ca

Test Code	Test Name / Description	Sample Type, Volume	TAT (days)
NGS385	Comprehensive Epilepsy (NGS Panel and Copy Number Analysis + mtDNA)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
NGS386	Epileptic Encephalopathy (NGS Panel and Copy Number Analysis)		
NGS387	Comprehensive Neuronal Migration Disorders (NGS Panel and Copy Number Analysis + mtDNA)		
NGS388	Non-Mitochondrial Neuronal Migration Disorders (NGS Panel and Copy Number Analysis)		
NGS389	Mitochondrial Neuronal Migration Disorders (NGS Panel and Copy Number Analysis + mtDNA)		
NGS392	Bartter/Gitelman Syndromes (NGS Panel and Copy Number Analysis)		
NGS393	Maple Syrup Urine Disease (NGS Panel and Copy Number Analysis)		
NGS394	Joubert Syndrome (NGS Panel and Copy Number Analysis)		
NGS395	Meckel Syndrome (NGS Panel and Copy Number Analysis)		
NGS396	Porphyria Disorders (NGS Panel and Copy Number Analysis)		
NGS397	Osteogenesis Imperfecta (NGS Panel and Copy Number Analysis)		
NGS398	Macrocephaly and Overgrowth Syndromes (NGS Panel and Copy Number Analysis)		
NGS399	Heterotaxy Syndromes (NGS Panel and Copy Number Analysis)		
NGS400	Pain Syndromes (NGS Panel and Copy Number Analysis)		
NGS401	Stickler Syndrome (NGS Panel and Copy Number Analysis)		
NGS402	Usher Syndrome (NGS Panel and Copy Number Analysis)		
NGS404	Hypothyroidism (NGS Panel and Copy Number Analysis)		
NGS405	Amyotrophic Lateral Sclerosis (NGS Panel and Copy Number Analysis + C9orf72 Repeat Expansion Analysis)		
NGS406	Alzheimer Disease/Frontotemporal Dementia (NGS Panel and Copy Number Analysis + C9orf72 Repeat Expansion Analysis)		
NGS407	Comprehensive Dementia (NGS Panel and Copy Number Analysis + C9orf72 Repeat Expansion Analysis)		
NGS408	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA + HTT Repeat Expansion Analysis)		
NGS409	Comprehensive Dystonia (NGS Panel and Copy Number Analysis + mtDNA + HTT Repeat Expansion Analysis)		
NGS410	Comprehensive Dementia (NGS Panel and Copy Number Analysis + HTT Repeat Expansion Analysis)		
NGS411	Comprehensive Dementia (NGS Panel and Copy Number Analysis + C9orf72 & HTT Repeat Expansion Analysis)		
NGS412	Myoclonic Epilepsy (NGS Panel and Copy Number Analysis)		
NGS413	Congenital Myopathies (NGS Panel and Copy Number Analysis)		
NGS414	Noonan Syndrome (NGS Panel and Copy Number Analysis)		
NGS417	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA + SCA & HTT Repeat Expansion Analysis)	Whole Blood (3 mL) or Extracted DNA	
NGS418	MNG Xpress™ Actionable Epilepsy (NextGen Sequencing Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
NGS419	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA + FRDA Repeat Expansion Analysis)	Whole Blood (3 mL) or Extracted DNA	
NGS420	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA + SCA & FRDA Repeat Expansion Analysis)		
NGS421	Congenital Muscular Dystrophies (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	
NGS422	Limb-Girdle Muscular Dystrophy (NGS Panel and Copy Number Analysis)		



List of Services

Test Code	Test Name / Description	Sample Type, Volume	TAT (days)		
NGS423	Emery-Dreifuss Muscular Dystrophy (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab			
NGS424	Duchenne/Becker Muscular Dystrophy (NGS Panel and Copy Number Analysis)				
NGS425	Microcephaly (NGS Panel and Copy Number Analysis)				
NGS426	Hydrocephalus (NGS Panel and Copy Number Analysis)				
NGS427	X-linked Intellectual Disability (NGS Panel and Copy Number Analysis + Fragile X Repeat Expansion & Methylation)				
NGS428	Tuberous Sclerosis (NGS Panel and Copy Number Analysis)				
NGS429	Familial Hemiplegic Migraine (NGS Panel and Copy Number Analysis + mtDNA)				
NGS430	Stroke (NGS Panel and Copy Number Analysis + mtDNA)	Whole Blood (3 mL) or Extracted DNA			
NGS431	Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA + SCA Repeat Expansion Analysis)				
NGS432	Comprehensive Intellectual Disability/Autism (NGS Panel and Copy Number Analysis + mtDNA + Fragile X Repeat Expansion and Methylation)	Whole Blood (3 mL), Extracted DNA or Buccal Swab			
NGS445	Comprehensive Neuropathies (NGS Panel and Copy Number Analysis + mtDNA)				
NGS446	Dopa-Responsive Dystonia (NGS Panel and Copy Number Analysis)				
NGS447	Sarcoglycanopathies (NGS Panel and Copy Number Analysis)				
NGS448	Hyperreflexia (NGS Panel and Copy Number Analysis)				
NGS449	Hyperphenylalaninemia (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28		
NGS452	Familial Hypercholesterolemia (NGS Panel and Copy Number Analysis)				
NGS453	Cornelia de Lange Syndrome (NGS Panel and Copy Number Analysis)				
NGS454	Polymicrogyria (NGS Panel and Copy Number Analysis)				
NGS455	Lissencephaly (NGS Panel and Copy Number Analysis)				
NGS456	Cerebral Cavernous Malformations (NGS Panel and Copy Number Analysis)				
NGS457	Holoprosencephaly (NGS Panel and Copy Number Analysis)				
NGS458	Comprehensive Hearing Loss (NGS Panel and Copy Number Analysis + mtDNA)				
NGS459	Waardenburg Syndrome (NGS Panel and Copy Number Analysis)				
NGS460	Alport Syndrome (NGS Panel and Copy Number Analysis)				
NGS461	Pendred Syndrome (NGS Panel and Copy Number Analysis)	Whole Blood (3 mL), Extracted DNA or Buccal Swab			
NGS462	Perrault Syndrome (NGS Panel and Copy Number Analysis)				
NGS463	Treacher-Collins Syndrome (NGS Panel and Copy Number Analysis)				
NGS464	Comprehensive Vision Loss & Eye Disorders (NGS Panel and Copy Number Analysis + mtDNA)				
NGS465	Dysautonomia (NGS Panel and Copy Number Analysis)				
NGS466	Comprehensive Anemia (NGS Panel and Copy Number Analysis)				
NGS9001	Known Familial Variant (KFV) Analysis (each mutation) - Copy Number				
NGS9003	Known Familial Variant (KFV) Analysis (each mutation) - Mitochondrial				
Whole exome sequencing					
WES001	MNG Exome TRIO Sequencing			Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
WES001X	MNG Xpress Exome™ TRIO Sequencing				
WES011	MNG Exome™ TRIO Sequencing (no mtDNA)				



List of Services

dynacare.ca

Test Code	Test Name / Description	Sample Type, Volume	TAT (days)
WES002	MNG Exome™ Additional Comparator (only available when trio is ordered)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
WES003	MNG Exome™ Proband Only Sequencing		
WES003X	MNG Xpress Exome™ Proband Only Sequencing		
WES013	MNG Exome™ Proband Only Sequencing (no mtDNA)		
WES008	MNG Exome™ DUO Sequencing		
WES008X	MNG Xpress Exome™ DUO Sequencing		
WES018	MNG Exome™ DUO Sequencing (no mtDNA)		
Carrier screening			
WES006	MNG Carrier Exome™ Sequencing and Copy Number Analysis + mtDNA (for couples only)	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
Healthy whole exome sequencing			
WES007	MNG Healthy Exome™ Sequencing and Copy Number Analysis + mtDNA	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-28
RNA sequencing			
RNA001	MNG Transcriptome™	RNA stabilized whole blood (6 mL), cultured fibroblasts, skin or muscle	14-28
RNA001A	MNG Answers™ - MNG Transcriptome™		
RNA002	Panel-Specific RNA Sequencing		
RNA003	Gene-Specific RNA Sequencing Targeted Analysis		
RNA003A	MNG Answers™ - Gene-Specific RNA Sequencing		
Whole genome sequencing			
WGS001	MNGenome® TRIO Sequencing	Whole Blood (3 mL), Extracted DNA or Buccal Swab	14-42
WGS001A	MNG Answers™ - MNGenome® TRIO Sequencing		
WGS001X	MNGenome Xpress™ TRIO Sequencing		
WGS002	MNGenome® Additional Comparator (only available when trio is ordered)		
WGS003	MNGenome® Proband Only Sequencing		
WGS003A	MNG Answers™ - MNGenome® Proband Only Sequencing		
WGS003X	MNGenome Xpress™ Proband Only Sequencing		
WGS008	MNGenome® DUO Sequencing		
WGS008X	MNGenome Xpress™ DUO Sequencing		
STAT services			
STAT100	NC MET STAT (Except MET22 and MET25)	N/A	10-14
STAT200	NC STAT		
STAT300	NC STAT		
STAT500	NGS STAT		
STAT200M	MOL STAT		
STAT250	NGS Panel STAT		