

NEXT GENERATION SANGER SEQUENCING PLATFORMS (NGS)

Category	Disease Category	Disease	Number of Genes	
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Liddle Syndrome Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Hyperlipidemia Core Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Short QT Syndrome (SQTS) Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Hereditary Hemorrhagic Telangiectasia (HHT) Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Brugada Syndrome Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Pan	10	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Long QT Syndrome (LQTS) Panel	18	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Atrial Fibrillation Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Hyperlipidemia Panel	20	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Pulmonary Artery Hypertension (PAH) Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel	33	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Heterotaxy and Situs Inversus Panel	34	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Noonan Syndrome Panel	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Marfan Syndrome Panel	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Ehlers-Danlos Syndrome Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Aorta Panel	53	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Arrhythmia Panel	62	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Hypertrophic Cardiomyopathy (HCM) Panel	92	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Congenital Structural Heart Disease Panel	125	850
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Dilated Cardiomyopathy (DCM) Panel	130	990
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Cardiomyopathy Panel	217	990
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	Comprehensive Cardiology Panel	260	990
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Hereditary Acrodermatitis Enteropathica Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Tuberous Sclerosis Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Pachyonychia Congenita Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Waardenburg Syndrome Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Adams-Oliver Syndrome Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Neurofibromatosis Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Xeroderma Pigmentosum Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Cutis Laxa Panel	10	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Dyskeratosis Congenita Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Progeria and Progeroid Syndromes Panel	17	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Hereditary Melanoma and Skin Cancer Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Hermansky-Pudlak Syndrome Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Ectodermal Dysplasia Panel	25	690
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Albinism Panel	26	850
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Epidermolysis Bullosa Panel	26	850

Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Palmoplantar Keratoderma Panel	26	850
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Ehlers-Danlos Syndrome Panel	32	850
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	Ichthyosis Panel	39	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Pendred Syndrome Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Branchio-Oto-Renal (BOR) Syndrome Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Alport Syndrome Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Hereditary Hemorrhagic Telangiectasia (HHT) Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Waardenburg Syndrome Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Stickler Syndrome Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Usher Syndrome Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Non-Syndromic Hearing Loss Panel	138	990
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Syndromic Hearing Loss Panel	138	990
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-nose-throat	Comprehensive Hearing Loss and Deafness Panel	288	990
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Glucocorticoid Deficiency Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Congenital Adrenal Hyperplasia Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Hyperparathyroidism Panel	14	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Premature Ovarian Failure Panel	16	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Hyperlipidemia Panel	18	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Hypomagnesemia Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Hypothyroidism and Resistance to Thyroid Hormone Panel	22	690
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Kallmann Syndrome Panel	31	850
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Monogenic Obesity Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	50	850
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MODY Panel	54	850
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Comprehensive Monogenic Diabetes Panel	67	850
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	Abnormal Genitalia/ Disorders of Sex Development Panel	73	850
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Polycystic Liver Disease Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Pancreatitis Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Gastrointestinal Atresia Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Hirschsprung Disease Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Congenital Diarrhea Panel	29	850
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Cholestasis Panel	52	850
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	Congenital Hepatic Fibrosis Panel	52	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Red Blood Cell Membrane Disorder Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Dyskeratosis Congenita Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Diamond-Blackfan Anemia Panel	14	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Hemophagocytic Lymphohistiocytosis Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Coagulation Factor Deficiency Panel	16	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Hermansky-Pudlak Syndrome Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Platelet Function Disorder Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Fanconi Anemia Panel	24	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Congenital Neutropenia Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Thrombocytopenia Panel	37	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Hereditary Leukemia Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Bleeding Disorder/Coagulopathy Panel	71	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Anemia Panel	88	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Bone Marrow Failure Syndrome Panel	156	990
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Comprehensive Hematology Panel	270	990

Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Comprehensive Hematology and Hereditary Cancer Panel	369	990
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	Comprehensive Immune and Cytopenia Panel	642	990
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Tuberous Sclerosis Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Lung Cancer Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Pancreatic Cancer Core Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Breast Cancer High Risk Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Neurofibromatosis Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Xeroderma Pigmentosum Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Paraganglioma-Pheochromocytoma Panel	11	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Melanoma and Skin Cancer Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Endocrine Cancer Panel	22	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Pancreatic Cancer Panel	22	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Colorectal Cancer Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Renal Cancer Panel	26	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Breast and Gynecological Cancer Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Cancer High Risk Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Leukemia Panel	42	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Gastrointestinal Cancer Panel	43	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Hereditary Pediatric Cancer Panel	71	850
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Comprehensive Hereditary Cancer Panel	160	990
Next Generation and Sanger Sequencing Platforms (NGS)	Hereditary Cancer	Comprehensive Hematology and Hereditary Cancer Panel	348	990
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Chronic Granulomatous Disease Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Dyskeratosis Congenita Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Hemophagocytic Lymphohistiocytosis Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Congenital Neutropenia Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Autoinflammatory Syndrome Panel	47	850
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Complement System Disorder Panel	80	850
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Severe Combined Immunodeficiency Panel	80	850
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Bone Marrow Failure Syndrome Panel	135	990
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Primary Immunodeficiency Panel	336	990
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCI	383	990
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	Comprehensive Immune and Cytopenia Panel (contracted only)	642	990
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Exostosis and Related Disorders Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Cerebral Cavernous Malformation Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Septo-Optic Dysplasia Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Cornelia de Lange Syndrome Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Meier-Gorlin Syndrome Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Seckel Syndrome Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Adams-Oliver Syndrome Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Kabuki Syndrome Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Neurofibromatosis Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Chondrodysplasia Punctata Panel	10	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Holoprosencephaly Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Lymphatic Malformations and Related Disorders Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Metaphyseal Dysplasia Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Gastrointestinal Atresia Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Hirschsprung Disease Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	16	690

Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Vascular Malformations Panel	16	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Short Rib Dysplasia / Asphyxiating Thoracic Dysplasia Panel	18	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Polymicrogyria Panel	20	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Brachydactyly / Syndactyly Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Cleft Lip/Palate and Associated Syndromes Panel	22	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	3-M Syndrome / Primordial Dwarfism Panel	24	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Lissencephaly Panel	24	690
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Facial Dysostosis and Related Disorders Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Micromelic Dysplasia Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Osteopetrosis and Dense Bone Dysplasia Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Osteogenesis Imperfecta Panel	33	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Heterotaxy and Situs Inversus Panel	34	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Skeletal Dysplasia with Abnormal Mineralization Panel	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Spondylometaphyseal / Spondyloepi-(meta)-physeal Dysplasia Pane	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Craniosynostosis Panel	38	850
Next Generation and Sanger Sequencing Platforms (NGS)	Mationations	Macrocephaly / Overgrowth Syndrome Panel	48	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Limb Malformations Panel	50	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Neuronal Migration Disorder Panel	59	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Arthrogryposes Panel	78	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Microcephaly and Pontocerebellar Hypoplasia Panel	78	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Comprehensive Short Stature Syndrome Panel	100	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Skeletal Dysplasias Core Panel	113	850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Comprehensive Skeletal Dysplasias and Disorders Panel	411	990
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Discover Dysplasia (live 1/1/21)	419	990
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	Comprehensive Growth Disorders / Skeletal Dysplasias and Disorder	510	990
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Cystinuria Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Creatine Metabolism Deficiency Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Tyrosinemia Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Homocystinuria Core Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Periodic Paralysis Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Hereditary Hemochromatosis Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Hyperphenylalaninemia Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Aicardi-Goutières Syndrome Panel	7	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Congenital Mono- and Disaccharide Disorders Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Porphyria Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Congenital and Familial Lipodystrophy Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Coenzyme q10 Deficiency Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Metabolic Liver Failure Panel	16	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Hypomagnesemia Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Purine and Pyrimidine Metabolism Disorders Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Fatty Acid Oxidation Syndrome Panel	26	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Peroxisomal Disorders Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Glycogen Storage Disorder Panel	29	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Nephrolithiasis Panel	35	850
	Metabolic Disorders	Monogenic Obesity Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)				
Next Generation and Sanger Sequencing Platforms (NGS) Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Congenital Disorders of Glycosylation Panel	48	850

Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	50	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Organic Acidemia/Aciduria & Cobalamin Deficiency Panel	54	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Mitochondrial DNA Depletion Syndrome Panel	62	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Lysosomal Disorders and Mucopolysaccharidosis Panel	102	850
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Metabolic Myopathy and Rhabdomyolysis Panel	127	990
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	Comprehensive Metabolism Panel	505	990
Next Generation and Sanger Sequencing Platforms (NGS)	Mitochondrial Disorders	Mitochondrial Genome Test	37	850
Next Generation and Sanger Sequencing Platforms (NGS)	Mitochondrial Disorders	Mitochondrial DNA Depletion Syndrome Panel	62	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Liddle Syndrome Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Diabetes Insipidus Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Primary Hyperoxaluria Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Branchio-Oto-Renal (BOR) Syndrome Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Renal Tubular Acidosis Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Alport Syndrome Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Hemolytic Uremic Syndrome Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Senior-Loken Syndrome Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Bartter Syndrome Panel	10	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Pseudohypoaldosteronism Panel	10	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Hypophosphatemic Rickets Panel	13	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Meckel Syndrome Panel	13	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Polycystic Kidney Disease Panel	13	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Hypomagnesemia Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Nephronophthisis Panel	20	690
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Bardet-Biedl Syndrome Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Renal Malformation Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Joubert Syndrome Panel	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Monogenic Obesity Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Cystic Kidney Disease Panel	43	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Nephrolithiasis Panel	45	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Primary Ciliary Dyskinesia Panel	47	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Nephrotic Syndrome Panel	96	850
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	Ciliopathy Panel	119	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Tuberous Sclerosis Panel	2	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Creatine Metabolism Deficiency Panel	3	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Cerebral Cavernous Malformation Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Periodic Paralysis Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Septo-Optic Dysplasia Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Collagen Type VI-Related Disorders Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Emery-Dreifuss Muscular Dystrophy Panel	6	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Porphyria Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Holoprosencephaly Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Nemaline Myopathy Panel	13	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Coenzyme q10 Deficiency Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Polymicrogyria Panel	20	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Congenital Myasthenic Syndromes Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Lissencephaly Panel	24	690
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Spinal Muscular Atrophy Panel	30	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	NCL and Progressive Myoclonic Epilepsy Panel	31	850

Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Amyotrophic Lateral Sclerosis Panel	35	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Idiopathic Generalized and Focal Epilepsy Panel	35	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Migraine Panel	47	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Macrocephaly / Overgrowth Syndrome Panel	48	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	LGMD and Congenital Muscular Dystrophy Panel	56	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Dementia Panel	58	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Neuronal Migration Disorder Panel	59	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Dystonia Panel	68	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Autism Spectrum Disorders Panel	75	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Spastic Paraplegia Panel	75	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Microcephaly and Pontocerebellar Hypoplasia Panel	78	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Parkinson Disease Panel	82	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Metabolic Epilepsy Panel	84	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Neuro-Ophthalmology Panel	87	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Metabolic Myopathy and Rhabdomyolysis Panel	90	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	X-linked Intellectual Disability Panel	106	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Leukodystrophy and Leukoencephalopathy Panel	118	850
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Charcot-Marie-Tooth Neuropathy Panel	153	990
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Comprehensive Muscular Dystrophy / Myopathy Panel	161	990
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Epileptic Encephalopathy Panel	203	990
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Ataxia Panel	257	990
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Beyond Paediatric Epilepsy Panel – for Europe and Middle East	511	990
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	Comprehensive Epilepsy Panel	511	990
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Septo-Optic Dysplasia Panel	4	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Achromatopsia Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Stickler Syndrome Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Senior-Loken Syndrome Panel	9	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Flecked Retina Disorders Panel	12	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Ectopia Lentis Panel	14	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Glaucoma Panel	19	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Congenital Stationary Night Blindness Panel	20	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Usher Syndrome Panel	21	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Albinism Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Vitreoretinopathy Panel	24	690
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Bardet-Biedl Syndrome Panel	27	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Leber Congenital Amaurosis Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Macular Dystrophy Panel	28	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Corneal Dystrophy Panel	29	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Joubert Syndrome Panel	36	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Cone Rod Dystrophy Panel	44	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis P	61	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Optic Atrophy Panel	76	850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Neuro-Ophthalmology Panel	97	850
Next Generation and Sanger Sequencing Platforms (NGS) Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology Ophthalmology	Cataract Panel	113	850
Next Generation and Sanger Sequencing Platforms (NGS) Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Retinitis Pigmentosa Panel	115	850
		Retinal Dystrophy Panel	351	990
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	My Retina Tracker Program Panel (US only)	351	990
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	Neonatal Respiratory Distress - Surfactant Dysfunction Panel	5	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	neonatat Respiratory Distress - Sunactant Dysignetion Panel	Э	090

Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Cystic Lung Disease Panel	8	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Central Hypoventilation and Apnea Panel	15	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Bronchiectasis Panel	22	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Hermansky-Pudlak Syndrome Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Pulmonary Artery Hypertension (PAH) Panel	23	690
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Interstitial Lung Disease Panel	30	850
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Primary Ciliary Dyskinesia Panel	41	850
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	Comprehensive Pulmonology Panel	114	850