Comprehensive suit of Precision Disease Diagnostic testing

Igenomix®
WITH SCIENCE ON YOUR SIDE

FAMILY: PRECISION DISEASE DIAGNOSTIC
PRODUCT CATEGORY: PRECISION PANELS

Product Name	Panels	Technology
Cardiology	Cardiomyopathy gene panel	NGS
	Cardiac Channelopathy gene panel	NGS
Connective Tissue Disorders	Ehlers-Danlos syndrome gene panel	NGS
	Marfan syndrome	NGS
	Cutis Laxa gene panel	NGS
	Connective tissue disorder gene panel	NGS
Dermatology	Ectodermal dysplasia gene panel	NGS
	Epidermolysis bullosa gene panel	NGS
	Ichthyosis gene panel	NGS
	Oculocutaneous albinism gene panel	NGS
	Xeroderma pigmentosum gene panel	NGS
	Tuberous Sclerosis (TSC1 & TSC2) gene panel	NGS
	TSC1 deletion/duplication	MLPA
	TSC2 deletion/duplication	MLPA
Endocrinology	Monogenic and syndromic obesity gene panel	NGS
	Hyperlipidemia gene panel	NGS
	Maturity-onset diabetes of the young (MODY) & neonatal diabetes gene panel	NGS
	Disorders of Sex Development (Abnormal Genitalia) Panel	NGS
	Hereditary pancreatitis gene panel	NGS
	"Congenital adrenal hyperplasia CYP21A2 (21-0H) NGS and deletion/duplication analysis"	NGS & MLPA
ENT	Deafness (syndromic & non-sydromic) gene panel	NGS
	Waardenburg syndrome gene panel	NGS
	Usher syndrome gene panel	NGS
	Hereditary Hemorrhagic Telangiectasia	NGS
	Broncio-Oto-Renal syndrome panel	NGS
Gastroenterology	Alagille syndrome gene panel	NGS
	Congenital hepatic fibrosis gene panel	NGS
	Gilbert or Crigler-Najjar syndrome (UGT1A1) gene analysis (only point mutations)	NGS
	UGT1A1 - repeat analysis	Fragment analysis
	Hemochromatosis gene panel	NGS
	Progressive familial intrahepatic cholestasis gene panel	NGS
	Wilson disease (ATP7B) gene analysis	NGS
	ATP7B deletion/duplication	MLPA
Haematology	Congenital afibrinogenemia gene panel	NGS
	Bone marrow failure syndrome gene panel	NGS
	Anemia gene panel	NGS
	Haemophilia (F8 & F9) gene panel	NGS
	F8 intron 22 inversion	PCR
	Hemophagocytic lymphohistiocytosis (HLH) gene panel	NGS
	Beta-thalassemia (HBB) gene analysis	NGS
	HBB deletion/duplication	MLPA
	Alpha-thalassemia (HBA1/2) gene analaysis	Sanger

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	HBA1 & HBA2 deletion/duplication	MLPA
	Von Willebrand disease (VWF) gene analysis	NGS
	Thrombocytopenia gene panel	NGS
	Thrombophilia gene panel	NGS
Immunology	IKBKG deletion/duplication analysis	MLPA
	Primary immunodeficiency gene panel	NGS
	Severe combined immunodeficiency gene panel	NGS
Metabolic Disorders	Fatty acid oxidation disorders gene panel	NGS
	Glycine encephalopathy gene panel	NGS
	Glycogen storage disorder gene panel	NGS
	Glycosylation (CDG) disorders gene panel	NGS
	Methylmalonic aciduria gene panel	NGS
	Organic acidemia gene panel	NGS
	Inborn Errors of Metabolic (IEM) disorder gene panel	NGS
	Leigh syndrome & mitochondrial encephalopathy gene panel	NGS
	Ornithine transcarbamylase deficiency (OTC) deletion/duplication analysis	MLPA
	Lysosomal storage disorder gene panel	NGS
	Urea cycle defects gene panel	NGS
Nephrology	Alport syndrome gene panel	NGS
	Bartter syndrome gene panel	NGS
	Meckel Gruber syndrome gene panel	NGS
	Nephrotic syndrome gene panel	NGS
	Polycystic kidney disease gene panel	NGS
	Primary hyperoxaluria gene panel	NGS
	Primary ciliary diskinesia gene panel	NGS
Neurology	Comprehensive neurology panel	NGS
	Neuronal migration disorder gene panel	NGS
Neurology Epilepsy	Aicardi-Goutieres syndrome gene panel	NGS
	Comprehensive epilepsy gene panel	NGS
	Rett syndrome gene panel	NGS
Neurology Movement Disorders	Ataxia-telangiectasia (ATM) gene analysis	NGS
	Ataxia-telangiectasia (ATM) deletion/duplication	MLPA
	Dystonia gene panel	NGS
	Early-onset juvenile parkinsonism gene panel	NGS
	Hereditary spastic paraplegia gene panel	NGS
	Hyperekplexia gene panel	NGS
	Neurofibromatosis (NF1 and NF2) gene analysis	NGS
	Neurofibromatosis type 1 (NF1) deletion/duplication	MLPA
	Neurofibromatosis type 2 (NF2) deletion/duplication	MLPA
Neurology Neurodegenerative	Adrenoleukodystrophy (ABCD1) gene analysis	NGS
	Joubert syndrome gene panel	NGS
	Leukodystrophy gene panel	NGS
	Metachromatic leukodystrophy gene panel	NGS

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Product Name	Panels	Technology
Neurology Neuromuscular	Neurodegeneration with brain iron accumulation (NBIA) gene panel	NGS
	Neurodegenration with brain iron accumulation 2B (PLA2G6) deletion/duplication analysis	MLPA
	Pantothenate kinase-associated degeneration (PANK2) deletion/duplication analysis	MLPA
	Arthrogryposis & congenital myasthenic syndrome gene panel	NGS
	Charcot-Marie-Tooth and sensory neuropathies gene panel	NGS
	Muscular dystrophy & congenital myopathy gene panel	NGS
	Duchenne muscular dystrophy (DMD) gene analysis	NGS
	Duchenne Muscular Dystrophy (DMD) deletion/duplication	MLPA
	Myotonia congenita gene panel	NGS
	PMP22 deletion/duplication analysis	MLPA
	Spinal Muscular Atrophy (SMN1) gene analysis	Sanger
	Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication	MLPA
Ophthalmology	Leber congenital amaurosis gene panel	NGS
	Optic atrophy gene panel	NGS
	Retinal degeneration gene panel	NGS
	Congenital cataract gene panel	NGS
	Cone-rod dystrophy gene panel	NGS
	Retinitis Pigmentosa gene panel	NGS
Rare disorders	Bardet-Biedl syndrome gene panel	NGS
	Cornelia de Lange syndrome gene panel	NGS
	DiGeorge syndrome deletion/duplication analysis	NGS
	Cystic Fibrosis (CFTR) gene analysis	NGS
	Cystic fibrosis (CFTR) gene deletion/duplication	MLPA
	Noonan syndrome gene panel	NGS
	Prader-Willi/Angelman syndrome deletion/duplication	MS-MLPA
	Stickler syndrome panel	NGS
Skeletal disorders	Skeletal dysplasia gene panel	NGS
	Osteogenesis imperfecta gene panel	NGS
	Osteopetrosis gene panel	NGS
Hereditary Cancer panel		NGS
New born genetic screening panel		NGS