



Accreditation Scope

**Biogenix Labs Omics Centre of Excellence - a unit of G42 Laboratory
LLC, NAM 020
Medical Laboratory, (ISO 15189:2012)**

Masdar City – Abu Dhabi, UAE

Issue Date: 21-04-2022

Expiry Date: 20-04-2025

Issue No: 01

Medical Field	Biological Sample	Components / Analytes	Examination technique	Examination Procedure Reference
Molecular Pathology	Whole Blood (EDTA)	Cardiomyopathy familial dilated gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Dilated cardiomyopathy gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Long QT syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Congenital ichthyosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Cutis Laxa gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Epidermolysis bullosa gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Hermansky-Pudlak syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Ichthyosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Cleft lip/Cleft Palate gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Congenital deafness gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Usher Syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Disorders of 46,XY gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Hyperlipidemia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Kallman syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	MODY Panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Obesity gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Pseudohypoaldosteronism gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Diamond-Blackfan anemia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Fetal and neonatal alloimmune thrombocytopenia	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Hemophilia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Shwachman-Diamond Syndrome	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Spherocytosis/Elliptocytosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Thrombocytopenia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Thrombophilia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Hyper IgE Syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Primary immunodeficiency gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Severe combined immunodeficiency gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Infertility gene panel (Blood) Common: Male & Female	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Infertility gene panel (Blood): Female	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Infertility gene panel (Blood): Male	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-



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				SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Fatty acid oxidation disorders gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Lysosomal storage disorder gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Maple syrup urine disease gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Peroxisomal disorder gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Alport syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Autosomal Dominant and Recessive Polycystic Kidney Disease gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Bardet-Biedl Syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Bartter Syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Nephrolithiasis and Nephrocalcinosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Nephrolithiasis and Nephrocalcinosis	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	3-Methylcrotonyl-CoA Carboxylase Deficiency gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	All Neuro gene panel: 1500 genes x 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Craniosynostosis and craniofacial gene panel: 628 genes x1.5	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Dravet Syndrome	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Dystroglycan-Related Congenital Muscular Dystrophy gene panel; 350 X 2 (Includes Mitochondrial DNA)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Epilepsy (generalized) with febrile seizures gene panel: 600 X2 (Includes Mitochondrial DNA)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Epileptic encephalopathy gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Facioscapulohumeral muscular dystrophy	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Familial Hemiplegic Migraine gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Holoprosencephaly, Nonsyndromic gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Hyperammonemia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Joubert syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Leber Congenital Amaurosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Leukodystrophy gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Metabolic myopathies gene panel: 90 GENES (Including Mitochondrial) X 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Microcephaly gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Mitochondrial Complex IV Deficiency gene panel (Nuclear and Mitochondrial Genes)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Muscular dystrophy plus panel: 350 X 2 (Includes Mitochondrial DNA)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Myoclonic dystonia gene panel; 85 genes X 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Pontocerebellar hypoplasia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Pyruvate Dehydrogenase Complex Deficiency gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Spastic paraplegia complete gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Treacher-Collins syndrome 3	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Urea Cycle Disorders gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	BRCA1/BRCA2 NGS plus gene panel: 2 genes X2 + MLPA	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Cancer Panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Colon Cancer Panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Comprehensive Cancer Panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Gastric cancer, hereditary diffuse	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	HBOC (Hereditary Breast and Ovarian Cancer) gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Hereditary nonpolyposis colorectal cancer (HNPCC)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Leukemia minimal residual disease NGS panel: 31	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Lung cancer targeted gene panel with rearrangement tumor: 70	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Myeloid tumor panel: 64 GENES X 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Neurofibromatosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Pancreatitis panel: 30 genes x2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Achromatopsia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Glaucoma gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Microcornea, myopic chorioretinal atrophy and telecanthus (sequence analysis of ADAMTS18 gene)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Oculomotor apraxia gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Optic atrophy gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Retinal cone dystrophy type 3B	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Retinal degeneration gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Retinitis Pigmentosa gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Stargardt disease gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Stickler Syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Expiry Date: 20-04-2025

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Medical Field	Biological Sample	Components / Analytes	Examination technique	Examination Procedure Reference
Molecular Pathology	Whole Blood (EDTA)	Alagille syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Arthrogryposis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Comprehensive pulmonary disease gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Osteogenesis imperfecta and low bone density disorders gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Periodic Fever Syndromes gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Primary Ciliary Dyskinesia (PCD)/Immotile Cilia Syndrome and Cystic Fibrosis gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Progressive Familial Intrahepatic Cholestasis (PFIC) and Alagille syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Bardet-Biedl syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Cornelia de Lange syndrome (sequence analysis of NIPBL gene)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Disorders of Sex Development gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Marfan syndrome type 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Meckel Syndrome	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Mitochondrial genes - WGS	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Noonan Syndrome	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Waardenburg syndrome	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Medical Field	Biological Sample	Components / Analytes	Examination technique	Examination Procedure Reference
Molecular Pathology	Whole Blood (EDTA)	Pre-PGT-Monogenic diseases (blood)	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Surfactant metabolism dysfunction gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	3-Methylcrotonyl-CoA Carboxylase Deficiency gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Aicardi-Goutieres syndrome gene panel	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Solid and Pediatric Tumor Panel: 150 genes x 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Whole Genome Analysis, Solo	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Whole Genome Sequencing, Duo	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING



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Molecular Pathology	Whole Blood (EDTA)	Whole Genome Sequencing, Trio Plus	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Whole Genome Sequencing, Trio	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
Molecular Pathology	Whole Blood (EDTA)	Autoimmune lymphoproliferative syndrome NGS panel: 35 genes X 2	NGS (Next Generation Sequencing)	BGX-SOP-ILM-001 ILLUMINA DNA PCR-FREE LIBRARY PREPARATION - HYBEX; BGX-SOP-ILM-002 ILLUMINA NOVASEQ 6000 SEQUENCING
END				