Dhiti Omics Technologies

TEST MENU - 2022

V1-0422

RARE DISEASES

Test Code	Test Name
DN01	Whole Genome Sequencing [WGS]

- ✓ **Comprehensive Screening for Rare Genetic Disorders. Mainly for prognostic, diagnostic, preventive & therapy modulation of diseases.**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- WGS checks for SNVs, indels, CNVs, and SVs across the whole genome. It may be used for diagnosis, screening, treatment selection in prenatal, paediatric, critical care, rare disease, or adult medicine.

Test Code	Test Name
DN02	Whole Exome Sequencing [WES]

✓ **Mainly for prognostic, diagnostic, preventive & therapy modulation of diseases.**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- WES sequences the human protein coding regions.

Test Code	Test Name
DN42	Hirschsprung Disease Panel

- ✓ **This test is used for the diagnosis of Hirschsprung disease or aganglionic megacolon**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks
Gene List	*BDNF, CELSR3, EDN3, EDNRB, KIF1BP, L1CAM, MITF, NRG1, NRTN, PAX3, PHOX2B, RET, RMRP, SOX10, ZEB2*

MITOCHONDRIAL DISORDERS

Test Code	Test Name
DN05	Mitochondrial Disorder Panel

✓ Targets the full mitochondrial genome, without amplifying any nuclear encoded genes.

Sample	4mL whole blood in EDTA tube / gDNA
Technology	PCR+NGS
TAT	4 weeks

Screens for

- Chronic progressive external ophthalmoplegia [CPEO]
- Kearns-Sayre syndrome [KSS]
- Leber hereditary optic neuropathy [LHON]
- Mitochondrial encephalomyopathy with lactic
- acidosis and stroke-like episodes [MELAS]
- Myoclonic epilepsy myopathy sensory ataxia [MEMSA]
- Myoclonic epilepsy with ragged-red fibers [MERRF]
- Mitochondrial recessive ataxia syndrome [MIRAS]
- Neurogenic weakness with ataxia and retinitis pigmentosa [NARP]
- Sensory ataxia neuropathy, dysarthria, ophthalmoplegia [SAND])
- Spinocerebellar ataxia with epilepsy [SCAE]

Test Code	Test Name
DN05	Mitochondrial Disorder Panel

Diseases	Gene List
Chronic progressive external ophthalmoplegia (CPEO)	*ABCB7, ACAD9, ALAS2, APTX, ATP5E, ATPAF2, BCS1L, BOLA3, COQ2, COQ4, COQ9, COX10, COX15, COX6B1, DARS2, DGUOK, ETHE1, FASTKD2, FOXRED1, GLRX5, HSPD1, ISCU, KIF5A, LRPPRC, MFN2, MPV17, MRPS16, MRPS22, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFAF1(CIA30), NDUFAF2(B17.2L), NDUFAF3, NDUFAF4(HRPAP20), NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OPA1, PDHA1, PDSS1, PDSS2, POLG, POLG2 (PEOA4), PUS1, RARS2, RMRP, RRM2B (PEOA5), SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TK2, TMEM70, TSFM, TUFM, TYMP (ECGF1), UQCRB, UQCRQ, YARS2*
Kearns-Sayre syndrome (KSS)	
Leber hereditary optic neuropathy (LHON)	
Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS)	
Myoclonic epilepsy myopathy sensory ataxia (MEMSA)	
Myoclonic epilepsy with ragged-red fibers (MERRF)	
Mitochondrial recessive ataxia syndrome (MIRAS)	
Neurogenic weakness with ataxia and retinitis pigmentosa (NARP)	
Sensory ataxia neuropathy, dysarthria, ophthalmoplegia (SANDO)	
Spinocerebellar ataxia with epilepsy (SCAE)	

V1-0422

METABOLIC DISORDERS

Test Code	Test Name
DN07	Metabolic Disorder Panel

- ✓ Accurate diagnosis of Metabolism Errors and other Paediatric-Onset Genetic Disorders.
- ✓ Sequencing coding regions of 300+ genes linked to 120+ such disorders

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- Lipid Metabolism Disorders
- Acidurias
- Amino Acid Metabolic Disorders
- Carbohydrate Metabolic Disorders
- Fatty Acid Oxidation Disorders
- Metal Metabolism Disorders
- Enzyme Deficiency
- Mitochondrial Deficiency
- Organic Acid Disorders
- Urea Cycle Defects
- Peroxisomal Disorders
- Other Metabolic Disorders

ENDOCRINE DISORDERS

Test Code	Test Name
DN15	Endocrine Disorder Panel

✓ Several endocrine disorders have a strong genetic component. Genetic tests can guide treatment in patients, and pave way for carrier screening in families.

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- Abnormal Genitalia/ Disorders of Sex Development
- Congenital Adrenal Hyperplasia
- Glucocorticoid Deficiency
- Hyperlipidemia
- Hyperparathyroidism
- Hypoglycemia and Hyperinsulinism
- Hypomagnesemia
- Hypothyroidism and Resistance to Thyroid Hormone
- Kallmann Syndrome/ Hypogonadotropic hypogonadism
- Premature Ovarian Failure
- Combined pituitary hormone deficiency

Test Code	Test Name
DN15	Endocrine Disorder Panel

Diseases	Gene List
Hypogonadotropic hypogonadism	*FGFR1, GNRHR, KISS1R, GNRH1, FGF8, PROKR2, CHD7, PROK2, KISS1, WDR11, HS6ST1, NSMF,TAC3,TACR3, SEMA3A*
Kallmann syndrome	*KAL1*
Androgen insensitivity	*AR*
Growth hormone deficiency	*GH1, GHRHR, GHR*
Combined pituitary hormone deficiency	*POU1F1, PROP1, HESX1, LHX2, LHX4*
Multiple endocrine neoplasia, Type I and Type II	*RET, MEN1, CDKN1B*

Diseases	Gene List
Congenital Adrenal hyperplasia	*HSD3B2, CYP11B1, CYP21A, CYP17A1*
Hypoaldosteronism	*CYP11B2*
Congenital adrenal hypoplasia	*DAX1, NR0B1*
Congenital goitre	*TG, TPO, SLaC5A5*
Congenital hypothyroidism	*THRA, PAX8*
Congenital hyperthyroidism	*TSHR*
Thyroid hormone resistance	*THRB*
Male limited precautious puberty	*LHCGR*

CARDIOLOGY

Test Code	Test Name
DN03	Cardiac Disorder Panel

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Who should be tested?

1. Individuals presenting with the most common symptoms of cardiovascular disease
2. Individuals with a positive family history of cardiovascular disease or sudden (unexplained) death
3. Individuals without a positive family history but with symptoms resembling the specific disease indication

Screens for

- Cardiac Channelopathies
- Cardiomyopathies
- Congenital heart diseases

Test Code	Test Name
DN03	Cardiac Disorder Panel

Diseases	Gene List
Long QT syndrome (Types 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15)	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*
Short QT Syndrome (Type 1, 2, 3)	*KCNH2, KCNJ2, KCNQ1*
Brugada Syndrome (Type 1, 2, 3, 4, 5, 6, 7, 8, 9)	*CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE5, KCNE1L, KCNE3, KCNJ8, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, TRPM4*
Catecholaminergic polymorphic ventricular tachycardia (CPVT) (Type 1, 2, 4, 5)	*CALM1, CASQ2, RYR2, TRDN*
Congenital heart disease	*CFC1, CITED2, CRELD1, FOXH1, GATA4, GATA6, GDF1, GJA1, HAND1, MED13L, NKX2-5, NKX2-6, NOTCH1, SMAD6, TBX1, TBX20, ZFPM2*
Inherited Cardiac Myopathies	*ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CASQ2, CAV3, CRYAB, CSRP3, CTF1, DES, DSC2, DSG2, DSP, DTNA, EMD, FHL2, GATA4, GATAD1, GLA, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, PKP2, PLN, PRKAG2, RBM20, RYR2, SGCD, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL*

NEUROLOGY

Test Code	Test Name
DN04	Neurological Disorder Panel

✓ **Targeted analysis for 1200 genes associated with neurological disorders.**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- Neurocognitive/ Intellectual disorders (e.g. Autism, ASD, ADD/ADHD)
- Neuromuscular disorders (e.g. DMD, BMD, LGMD)
- Epileptic disorders
- Mitochondrial disorders*

Who should be tested?

1. Individuals presenting with the most common symptoms of a neurological disease
2. Individuals with a positive family history of neurological disease
3. Individuals without a positive family history but with symptoms resembling the specific disease indication
4. Individuals with a negative, but suspected, family history, in order to perform genetic counselling (prenatal analyses are recommended in families of affected individuals)

Test Code	Test Name
DN09	Neuromuscular Disorder Panel

- ✓ **Confirms diagnosis of patients with muscular dystrophies, carrier testing of family members or for prenatal testing in offspring**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks
Gene List	*SGCA, DYSF, CAPN3, FKRP, DMD, SGCB, SGCD, SGCG, DYSF, CAPN3, FKRP*

Screens for

- Duchenne Muscular Dystrophy – DMD
- Becker Muscular Dystrophy – BMD
- Limb Girdle Muscular Dystrophy – LGMD [Type 2A, 2B, 2C, 2D, 2E, 2F, & 5C]

Test Code	Test Name
DN17	CGH-Microarray

- ✓ Copy number changes including deletions and amplifications in developmental delays, mental retardation, and related conditions

Sample	3mL whole blood in EDTA / Saliva / buccal swab
Technology	Microarray
TAT	4 weeks

NEPHROLOGY

Test Code	Test Name
DN06	Inherited Common Nephrological Disorders

✓ **Screens the coding regions of 57 genes associated with inherited renal diseases**

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- Accurate diagnosis - helps you decide next course of treatment
- Prognosis - informed decision on transplant
- Assessment of recurrence chance
- Carrier status of family members
- Focussed test with an affordable price

Test Code	Test Name
DN06	Inherited Common Nephrological Disorders

Diseases	Gene List
Alport Syndrome	*COL4A4, COL4A5, COL4A3*
Polycystic Kidney Disease [PKD]	*PKD1, PKD2, PKHD1, GANAB*
Focal Segmental Glomerulosclerosis [FSGS]	*ACTN4, ADCK4, ANLN, APOL1, ARHGDIA, CD2AP, CFHR5, CRB2, DGKE, EMP2, INF2, LAMB2, MYO1E, NPHS1, NPHS2, NUP107, NUP205, NUP93, PAX2, PLCE1, PTPRO, TRPC6, WT1*
Atypical Hemolytic Uremic Syndrome [aHUS]	*CD46,CFB,CFHR1,CFHR3,CFH,C3,CFI,THBD*
Nephrotic Syndrome	*ACTN4,ADCK4,ANLN,APOL1,ARHGDIA,CD2AP,CFHR5, CRB2, DGKE, EMP2, INF2, LAMB2, MYO1E, NPHS1, NPHS2, NUP107, NUP205, NUP93, PAX2, PLCE1, PTPRO, TRPC6, WT1, APOA1*
Primary Hyperoxaluria	*AGXT,GRHPR,HOGA1*
Medullary Cystic Kidney Disease	*MUC1, UMOD*
Nephronophthisis/Joubert Syndrome/Senior Loken Syndrome	*INVS, NPHP1, NPHP3, NPHP4, TMEM67*
Other Diseases	*LMX1B – Nail-patella Syndrome* *NOTCH2 – Alagille syndrome* *BICC1 – Renal dysplasia* *HNF1B – Renal cysts* *MYH9 – Fechtner syndrome, Epstein syndrome PLG- Acute Nephritis* *FAN1- Interstitial Nephritis Karyomegalic*

GYNECOLOGY

Test Code	Test Name
DN12	Pre-natal Testing

Sample	4mL whole blood in EDTA tube / 10mL Amniotic Fluid / CVS
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

First trimester

- Increased NT
- Spina bifida Holoprosencephaly
- Encephalocele Microcephaly
- Facial clefting
- Agenesis of corpus callosum Renal agenesis
- Skeletal
- Limb reduction defects Absent nasal bone
- Anencephaly
- Acrania
- Micrognathia Omphalocele Gastroschisis
- Megacystis Hydrocephalus

Second trimester

- Tetralogy of Fallot
- Atrioventricular canal
- Right heart defects
- Great arteries defects
- Hydronephrosis
- Osteochondrodysplasias
- Arthrogryposis
- Hydrops
- Syndactyly
- Talipes
- Hydronephrosis

Test Code	Test Name
DN13	Mutation Testing for Pre – Natal validation (Includes MCC)

- ✓ VNTR/ STR analysis by PCR to rule out maternal cell contamination (MCC) in a prenatal sample under analysis.

Sample	4mL whole blood in EDTA tube (mother) and 10mL Amniotic Fluid/CVS
Technology	Sanger sequencing
TAT	2 weeks

Test Code	Test Name
DN14	Cystic Fibrosis Hotspot Testing

- ✓ Detects F508del mutation found in >80% of CF Patients. This is clinically actionable, with treatments such as lumacaftor/tezacaftor that help the defective CFTR fold correctly.

Sample	4mL whole blood in EDTA tube
Technology	Sanger sequencing
TAT	2 weeks

ONCOLOGY

Test Code	Test Name
DN11	Inherited Cancer Panel

✓ Screens for a whole gamut of inherited cancers, such as Lynch Syndrome, Breast and Ovarian Cancers etc.

Sample	4mL whole blood in EDTA tube / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Screens for

- Lynch Syndrome
- Breast Cancer
- Ovarian Cancer
- Colorectal Cancer
- Uterine Cancer
- Gastric Cancer
- Skin Cancer
- Familial Adenomatous Polyposis

How will genetic testing help?

- Helps the clinician to make a precise diagnosis
- Helps the clinician to decide the most suitable therapy and support for the patient.
- Early intervention and good surveillance can help the patient in leading a good life
- Results of genetic testing can be useful for future family planning

Test Code	Test Name
DN11	Inherited Cancer Panel

Cancer Type	Gene List
Breast cancer	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, FGFR2, NBN, PALB2, PTEN, RAD51C, STK11, TP53, TERT, TOX3, TP53, XRCC2*
Ovarian cancer	*AKT1, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2*
Colorectal cancer	*APC, ATM, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SCG5/GREM1, SMAD4, STK11, TP53*
Uterine cancer	*EPCAM, MLH1, MSH2, MSH6, PMS1, PMS2, PTEN*
Gastric cancer	*BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, PMS1, PMS2, SMAD4*
Skin cancer	*CDKN2A, EPCAM, MC1R, MITF, MLH1, MSH2, MSH6, PMS1, PMS2, POT1, PTCH1, XRCC3*
Familial Adenomatous Polyposis	*APC, ATM, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SCG5/GREM1, SMAD4, STK11, TP53*

Type	Hematological Cancer
Sample	4mL whole blood / 2 ml bone marrow in EDTA tube
Technology	RT-PCR
TAT	4 Days

Test Code	Test Name	Gene List
DN27	AML Comprehensive PCR Panel	*BCR-ABL1, PML-RaRa, AML-ETO, Inv16, FLT3 (ITD/TKD), NPM1, c-KIT*
DN28	ALL Comprehensive PCR Panel	*BCR-ABL1, TEL-AML1, MLL-AF4, MLL-AF9, MLL- ENL, E2A-PBX1*
DN29	BCR-ABL Qualitative [Major, Minor & Micro]	*BCR-ABL1*
DN30	BCR-ABL1 Quantitative [Major, Minor & Micro], WHO International Scale	*BCR-ABL1*
DN32	JAK 2 Detection [Exons 12-15]	*JAK 2*
DN33	PML-RARA Qualitative	*BCR1, BCR2 & BCR3*
DN34	PML-RARA Quantitative	*BCR1, BCR2 & BCR3*

Test Code	Test Name
DN31	*BCR/ABL1*, Kinase Domain Sequencing

Sample	4mL whole blood / 2 ml bone marrow in EDTA tube
Technology	Sanger sequencing
TAT	14 Days

Test Code	Test Name
DN35	KRAS Qualitative
DN36	NRAS Qualitative
DN37	BRAF Qualitative
DN38	EGFR Qualitative

Type	Solid Tumor
Sample	FFPE tissue block/unstained slides
Technology	RT-PCR
TAT	7 Days

Test Code	Test Name
DN22	Solid Tumours Panel_Somatic

Sample	FFPE tissue blocks/unstained slides
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Genes Covered

ALK, APC, ATM, BRAF, CDH1, CDKN2A, CTNNB1, DDR2, EGFR, ERBB2, ERBB4, EZH2, FBAKT1XW7, FGFR1, FGFR2, FGFR3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KDR, KIT, KRAS, MAP2K1, MET, MTOR, NOTCH1, NRAS, NTRK1, PDGFRA, PIK3CA, PTEN, RB1, RET, ROS1, SMAD4, SMO, STK11, TERT promoter, TP53, VHL

Resections:	Transport 8 unstained 5-micron slides. (Min: 5 slides)
Small Biopsies:	Transport 15 unstained 5-micron slides. (Min: 10 slides)
Transport Temperature:	Room temperature.
Unacceptable Conditions:	<10 percent tumor, Decalcified specimens, FNA smears with less than 50 tumor cells.
Additional Requirement:	Include surgical pathology report.

Test Code	Test Name
DN23	Non-small Cell Lung Carcinoma [NSCLC] Panel_Somatic

Sample	FFPE tissue blocks/unstained slides
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Genes Covered

FDA Approved and NCCN Recommended Therapies Guided by Genetic Markers

Erlotinib, Gefitinib	*EGFR, ERBB2, KRAS, MET*	Alectinib	*ALK*
Afatinib	*EGFR, ERBB2, KRAS, MET*	Bevacizumab	*KRAS, VHL*
Osimertinib	*EGFR*	Vemurafenib	*BRAF*
Crizotinib	*ALK*	Dabrafenib	*BRAF*
Ceritinib	*ALK*	Trastuzumab	*EGFR, ERBB2, MET, PIK3CA*

Resections:	Transport 8 unstained 5-micron slides. (Min: 5 slides)
Small Biopsies:	Transport 15 unstained 5-micron slides. (Min: 10 slides)
Transport Temperature:	Room temperature.
Unacceptable Conditions:	<10 percent tumor, Decalcified specimens, FNA smears with less than 50 tumor cells.
Additional Requirement:	Include surgical pathology report.

Test Code	Test Name
DN24	Colorectal Carcinoma [CRC] Panel_Somatic

Sample	FFPE tissue blocks/unstained slides
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Genes Covered

FDA Approved and NCCN Recommended Therapies Guided by Genetic Markers

5-Fluorouracil	*APC, SMAD4*
Cetuximab, Panitumumab	*BRAF, EGFR, KRAS, MET, NRAS, PIK3CA, PTEN*
Oxiplatin	*KRAS*
Bevacizumab	*KRAS*

Resections:	Transport 8 unstained 5-micron slides. (Min: 5 slides)
Small Biopsies:	Transport 15 unstained 5-micron slides. (Min: 10 slides)
Transport Temperature:	Room temperature.
Unacceptable Conditions:	<10 percent tumor, Decalcified specimens, FNA smears with less than 50 tumor cells.
Additional Requirement:	Include surgical pathology report.

Test Code	Test Name
DN25	Myeloid Tumours Panel_Somatic

Sample	3mL whole blood or bone marrow in EDTA tube
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Genes Covered

ANKRD26, ASXL1, ATM, CBL, CDKN2A, CEBPA, CREBBP, DNMT3A, ETV6, EZH2, FLT3, GATA2, HRAS, IDH1, IDH2, JAK2, KIT, KRAS, NF1, NOTCH1, NPM1, NRAS, PDGFRB, PHF6, PTPN11, RAD21, RUNX1, SF3B1, SMC1A, SMC3, SRSF2, STAG2, TET2, TP53, U2AF1, WT1

If smaller panel needed: *ANKRD26, ASXL1, CEBPA, DDX41, DNMT3A, ETV6, FLT3, GATA2, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, PDGFRA, RUNX1, TP53, WT1*

Transport Temperature: Room temperature. Ship over cool packs during summer/hot weather.
Unacceptable Conditions: Serum, plasma, grossly hemolyzed specimens, buccal brush or swab, FFPE tissue.

Test Code	Test Name
DN26	Lymphocytic Tumours Panel_Somatic

Sample	3mL whole blood or bone marrow in EDTA tube
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Genes Covered

ABL1, ASXL1, ATM, ATRX, BCOR, BCORL1, BRAF, CALR, CBL, CBLB, CBLC, CDKN2A, CEBPA, CREBBP, CSF3R, CUX1, DNMT3A, ETV6, EZH2, FBXW7, FLT3, GATA1, GATA2, GNAS, HRAS, IDH1, IDH2, IKZF1, JAK2, JAK3, KDM6A, KIT, KMT2A, MGM173, MGM175 KMT2D, KRAS, MPL, MYD88, MGM1060 NOTCH1, NPM1, NRAS, PDGFRA, PHF6, PTEN, PTPN11, RAD21, RUNX1, SETBP1, SF3B1, SMC1A, SMC3, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, ZRSR2

Transport Temperature: Room temperature. Ship over cool packs during summer/hot weather.

Unacceptable Conditions: Serum, plasma, grossly homolysed specimens, buccal brush or swab, FFPE tissue.

INFECTIOUS DISEASE

Test Code	Test Name
DN39	SARS-CoV-2 RT-PCR Test
DN41	SARS-CoV-2 Tapestry Algorithmic Pooling

Sample	Nasopharyngeal & Oropharyngeal Swab
Technology	RT-PCR
TAT	24 hours

Test Code	Test Name
DN40	SARS-CoV-2 Rapid Antigen Test

Sample	Nasopharyngeal/Nasal Swab
Technology	POC
TAT	24 hours

Test Code	Test Name
DN43	Rapid testing of Infectious diseases [Bacteria, Virus, Fungus]

Sample	Blood/Pus Swab/gDNA
Technology	Nanopore sequencing [Whole Genome Sequencing]
TAT	48 hours

- ✓ We conduct Whole Genome Metagenome Sequencing (WGMS) and Analysis in clinical samples such as blood / biological fluids (Cerebrospinal fluid, Urine etc) / gDNA.
- ✓ The test is run using the Oxford Nanopore Technology (ONT) platform, and provides rapid and accurate identification of the pathogens and AMR where applicable.

Test Code	Test Name
DN44	Human Immuno-deficiency Virus [HIV]
DN45	Hepatitis B Virus [HBV]
DN46	Hepatitis C Virus [HCV]
DN47	Human Papilloma Virus [HPV]

Sample	• 6mL whole blood in EDTA / 2mL Plasma for **HIV, HBV, HCV**. [Unacceptable specimens: Serum and CSF]. • FFPE tissue block or 5 unstained 4-5 micron slides for **HPV** in head and neck squamous cell carcinoma [Unacceptable specimens: FFPE specimens without tumour tissue / FFPE specimens fixed in formalin substitutes (ie., Bouen or B5 fixatives), alternative fixatives or heavy metal fixatives (B-4 or B-5) / Decalcified specimens]. • 3mL Cervical, anal, or vaginal specimen with brush or spatula from ThinPrep or SurePath collection kits for **HPV.**
Technology	RT-PCR
TAT	48 hours

OTHER TESTS

Test Code	Test Name
DN16	Carrier Testing

- ✓ Targeted mutation testing in family, when the mutation has already been established in proband

Sample	4mL whole blood in EDTA tube / Saliva / buccal swab / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Test Code	Test Name
DN18	Sickle Cell Anaemia Testing

- ✓ Detection of Sickle Cell Anaemia mutations

Sample	4mL whole blood in EDTA tube / Saliva / buccal swab / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Test Code	Test Name
DN19	Alpha Thalassemia Testing

✓ Detects point mutations in HBA1 & HBA2 genes

Sample	4mL whole blood in EDTA tube / Saliva / buccal swab / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Test Code	Test Name
DN20	Beta Thalassemia Testing

✓ Detects point mutations in HBB gene

Sample	4mL whole blood in EDTA tube / Saliva / buccal swab / gDNA
Technology	Next-Generation Sequencing [NGS]
TAT	4 weeks

Test Code	Test Name
DN21	Single Gene Sequencing

Sample	4mL whole blood in EDTA tube / Saliva / buccal swab / gDNA
Technology	Sanger / Nanopore
TAT	2 weeks

Test Code	Test Name	TAT
DN48	Data Reanalysis	1 week
DN49	Result Interpretation	1 week
DN50	Genetic Counselling	1 week

Dhiti Omics Technologies

Precision Molecular Diagnostics & Genetic Testing

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