

## GENERAL INFORMATION

**Below are list of Assays for Medical Genomic Services using NGS and Microarray.**

For Medical Pathogen Identification Services please refer to **MBG-C0019**.

For other assays not listed below, individual projects and any other research collaborations, please contact MBG Centre for further information.

**The recommended sample types are as per published recommendations. Please contact MBG Centre for alternative sample type suitability.**

**All samples should be freshly collected and transported at 4°C following below guidelines.**

### Sample Details:

EDTA blood:	>2.5ml of blood in EDTA tube
Fresh Tissue:	>50mg tissue in saline dampened gauze to be received within 2hours of operation; or snap frozen in liquid nitrogen within 2hours of operation and stored below -70°C. Transport on dry ice.
FFPE:	5x10um sections selected by a pathologist to contain >50% viable tumour (avoid necrotic areas).
Buccal Swab:	Do not eat / drink for >1hour before collection. Rub swab inside the mouth with firm pressure for 1min. Place in the collection tube, seal and store between -20°C to 25°C. Transport within 48 hours.

### Notes:

- The Turn-around times (TAT) indicated are only inclusive of working days.
- Samples delivered before 11:00am will begin processing immediately resulting in shorter turnaround times.
- For Price details please contact MBG.
- For whole genome sequencing assay; MBG Centre recommends analysing relatives (proband plus parents; or siblings) to provide the most accurate interpretation.

Assay Code	ASSAY NAME	No. Genes (Coverage)	Genes included	Min Sample No per run	Recommended sample types	TAT	Accreditation
<b>WHOLE GENOME SEQUENCING</b>							
HGG-125	Human Whole Genome Sequencing (incl. full bioinformatics analysis & report)	WGS (30x)	Whole Genome	1	EDTA Blood	25-30 days	Validated kit-based assay
HGG-157	Human Whole Genome Sequencing (FASTQ data only)	WGS (30x)	Whole Genome	1	EDTA Blood	25-30 days	Validated kit-based assay
<b>INHERITED DISORDERS – TARGETED PANELS</b>							
HGT-186	Targeted Clinical Exome Sequencing (incl. full bioinformatics analysis & report)	(>30X)	Includes >4800 clinically relevant genes	3	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-128	Comprehensive Autism Panel (incl. full bioinformatics analysis & report)	61 genes (>30X)	ADSL, AFF2, ALDH5A1, AP152, ARX, ATRX, BRAF, CACNA1C, CASK, CDKL5, CHD7, CHD8, CNTNAP2, CREBBP, CYP27A1, DHCR7, DMD, EHMT1, FGD1, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, HPRT1, KDM5C, L1CAM, MAGEL2, MBD5, MECP2, MED12, MEF2C, MID1, NHS, NIPBL, NLGN3, NLGN4X, NR1I3, NRXN1, NSD1, OPHN1, PAFAH1B1, PCDH19, PHF6, PNKP, PQBP1, PTCHD1, PTEN, PTPN11, RAB39B, RAI1, RELN, SCN1A, SLC2A1, SLC9A6, SMARCB1, SMC1A, TCF4, UBE2A, UBE3A, VPS13B	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-129	Comprehensive Cardio-vascular Panel (incl. full bioinformatics analysis & report)	29 genes (>30X)	ABCC9, ACTN2, ANK2, ANKRD1, BAG3, CACNA1C, DMD, DSG2, DSP, KCNE2, KCNH2, LDB3, MYBPC3, MYH6, MYL3, MYPN, PKP2, RYR2, SCN5A, SOS1, TTN, TTR, VCL, FBN1, CHD7, NOTCH1, COL4A3, EHMT1, SMAD4	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-130	Comprehensive Cardio-myopathy Panel (incl. full bioinformatics analysis & report)	67 genes (>30X)	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BRAF, CALR3, CASQ2, CAV3, CRYAB, CSRP3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FHL1, FKTN, GAA, GATAD1, GLA, HRAS, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYO22, MYPN, NEBL, NEXN, NRAS, PDLIM3, PKP2, PLN, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, SOS1, TAZ, TCAP, TGFB3, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-131	Sudden Cardiac Arrest Panel (incl. full bioinformatics analysis & report)	13 genes (>30X)	ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A, CALM1, CALM3, KCND3	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-133	Diabetes MODY Panel (incl. full bioinformatics analysis & report)	12 genes (>30X)	ABCC8, BLK, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	2	EDTA Blood, Buccal Swab	15-20 days	Validated kit-based assay
HGT-134	Comprehensive Epilepsy Panel (incl. full bioinformatics analysis & report)	64 genes (>30X)	ADSL, ALDH7A1, ARHGEF9, ARX, ATP1A2, ATP6AP2, CACNA1A, CASK, CDKL5, CHRNA2, CHRNA4, CHRN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DNAJC5, EPM2A, FOLR1, GABRA1, GABRG2, GAMT, GOSR2, GRIN2A, KCNJ10, KCNQ2, KCNQ3, KCTD7, LGI1, MBD5, MECP2, MEF2C, MFSD8, NHLRC1, NRXN1, OPHN1, PCDH19, PHF6, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SLC25A22, SLC2A1, SLC9A6, SPTAN1, STXBP1, TBC1D24, TCF4, TPP1, TSC1, UBE3A, ZEB2	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-135	Gastro-Intestinal Panel (incl. full bioinformatics analysis & report)	21 genes (>30X)	ABCB11, ABCB4, ABCC2, ATP7B, BAAT, EPCAM, FAH, G6PC, JAG1, SLC10A2, TJP2, UGT1A1, UGT1A10, UGT1A3, UGT1A4, UGT1A5, UGT1A6, UGT1A7, UGT1A8, UGT1A9, VIPAS39	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-136	Mitochondrial Disorder Panel (Nuclear genes only) (incl. full bioinformatics analysis & report)	35 genes (>30X)	ATPAF2, BCS1L, COX10, COX15, COX4I2, COX6B1, FASTKD2, FOXRED1, LRPPRC, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFAF1, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NUBPL, SCO1, SCO2, SDHAF1, SURF1, TACO1, TMEM70, TTC19, UQCRB, UQCRCQ	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay

Assay Code	ASSAY NAME	No. Genes (Coverage)	Genes included	Min Sample No per run	Recommended sample types	TAT	Accreditation
HGT-137	<b>Mitochondrial Whole Genome Analysis</b> (incl. full bioinformatics analysis & report)	14 genes (>30X)	MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR2	20	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-138	<b>Congenital Muscular Dystrophy Panel</b> (incl. full bioinformatics analysis & report)	30 genes (>30X)	CHKB, COL6A1, COL6A2, COL6A3, FKRP, FKTN, ITGA7, LAMA2, LMNA, RYR1, SEPN1, ACTA1, CFL2, DAG1, DPM1, DPM3, ISPD, KBTBD13, LARGE, MEGF10, MTM1, NEB, POMGNT1, POMT1, POMT2, SYNE1, TCAP, TNNT1, TPM2, TPM3	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-139	<b>LGMD Muscular Dystrophy Panel</b> (incl. full bioinformatics analysis & report)	34 genes (>30X)	ANO5, CAPN3, CAV3, DES, DMD, DYSF, FKRP, FKTN, GAA, LMNA, MYOT, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TRIM32, TTN, COL6A1, COL6A2, COL6A3, DAG1, DNAJB6, EMD, FHL1, FLNC, GNE, ISPD, PLEC, SYNE1, VCP	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-140	<b>Neuro-Muscular Panel</b> (incl. full bioinformatics analysis & report)	54 genes (>30X)	ACTA1, ANO5, BAG3, BIN1, CAPN3, CAV3, CFL2, COL6A1, COL6A2, COL6A3, CRYAB, DAG1, DES, DMD, DNM2, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GNE, IGHMBP2, ISPD, ITGA7, LAMA2, LARGE, LDB3, LMNA, MTM1, MYH7, MYOT, NEB, PLEC, POMGNT1, POMT1, POMT2, PYGM, RYR1, SCN4A, SEPN1, SGCA, SGCB, SGCD, SGCG, TCAP, TNNI2, TNNT1, TPM2, TPM3, TRIM32, TTN, VPK1	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-141	<b>Cystic Fibrosis &amp; Pulmonary Disease Panel</b> (incl. full bioinformatics analysis & report)	51 genes (>30X)	CFTR, SFTPB, ABCA3, ACVRL1, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMPR2, CCDC39, CCDC40, CSF2RA, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, EDN3, EFEMP2, ELN, ENG, FBLN5, FLCN, GDNF, HPS1, HPS3, HPS4, HPS5, HPS6, LTBPA, NME8, PHOX2B, RET, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPC, SFTPD, SMAD9, STAT3, TERT, TSC1, TSC2	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-143	<b>Deafness Panel</b> (incl. full bioinformatics analysis & report)	23 genes (>30X)	CDH23, CLDN14, COL4A4, EDN3, GJB2, HGF, LARS2, LOXHD1, LRTOMT, MARVELD2, MYO15A, MYO6, MYO7A, OTOF, PCDH15, POU4F3, PTPRQ, SCARF2, SLC26A4, TMC1, UBIAD1, USH2A, WFS1	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-144	<b>Dysmorphic - Dysplasia Panel</b> (incl. full bioinformatics analysis & report)	62 genes (>30X)	EVC, EXT1, EZH2, FBN1, FGFR2, FGFR3, FKBP10, FREM1, GALNS, GNPAT, HGSNAT, HRAS, HSPG2, IFT122, IRF6, KMT2D, LEPRE1, LIFR, LMNA, MASP1, MGAT2, MMP2, NEK1, NF1, NFIX, NIPBL, NSD1, OBSL1, PCNT, PEX13, PYCR1, RIPK4, ROR2, SERPINF1, SGSH, TCOF1, TMEM67, TNFRSF11B, UBR1, WISP3	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-145	<b>Endocrine Disorder Panel</b> (incl. full bioinformatics analysis & report)	5 genes (>30X)	BBS5, GHR, GHRHR, SHOX, TBCE	2	EDTA Blood, Buccal Swab	15-20 days	Validated kit-based assay
HGT-146	<b>Metabolic Disorder Panel</b> (incl. full bioinformatics analysis & report)	40 genes (>30X)	ACADM, AMT, ARG1, ARSB, ASAH1, ASL, ASS1, BCKDHA, BCKDHB, BTD, CBS, CLN8, CPS1, DBT, DPAGT1, FAH, G6PC, GAA, GBA, GCDH, GLB1, GNS, H6PD, HSD3B7, IVD, L2HGDH, MCCC2, MMACHC, MMADHC, MUT, OXCT1, PAH, PEX1, PGM1, PHKG2, PTS, PYGL, SLC22A5, SLC37A4, SLC3A1	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-147	<b>Neurological Disorder Panel</b> (incl. full bioinformatics analysis & report)	110 genes (>30X)	ABAT, ABCD1, ABHD12, ADCK3, ADK, ADRA2B, AGRN, AHI1, ALS2, ARFGF2, ARHGFB6, ARID1B, ARL13B, ARL14EP, ASPM, ATL1, ATN1, ATRX, C12orf57, C5orf42, CACNA1G, CAPN3, CASC5, CCDC88C, CDK5RAP2, CDKL5, CENPJ, CEP57, CLN6, COG6, COL6A1, COL6A2, COL6A3, CPA6, CYP27A1, DPYD, DYSF, ERLIN2, FKRP, FKTN, FLVCR2, FOXG1, GDAP1, GNE, HEPACAM, HEXA, HSD17B4, HUWE1, ISPD, KIAA0196, KIF1A, KIF7, L1CAM, LAMA2, LARGE, LRPPRC, MCPH1, MECP2, MED23, MFSD8, MPDZ, NDE1, NPC2, NTRK1, OCLN, OPTN, PANK2, PEX1, PEX12, PEX16, PEX26, PEX5, PEX6, PHGDH, PLA2G6, PLP1, PNPO, POMT1, POMT2, PSAP, RELN, RNASEH2B, RNASEH2C, RYR1, SCN2A, SEPN1, SGCA, SGCB, SLC25A19, SOD1, SPG11, SPG20, SPTBN2, STXBP1, SYNE2, SYNGAP1, TCTN1, TMEM237, TMEM67, TRAPPC9, TRMT1, TUBA1A, TYMP, VAPB, WDR45B, WDR62, WDR81, WNK1, YARS, ZNF526	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay

Assay Code	ASSAY NAME	No. Genes (Coverage)	Genes included	Min Sample No per run	Recommended sample types	TAT	Accreditation
HGT-148	<b>Pelvic Inflammatory Disease Panel</b> (incl. full bioinformatics analysis & report)	29 genes (>30X)	ADA, AICDA, AIRE, AK2, ATM, BTK, CD247, CD40, CD40LG, COLEC11, CYBB, DCLRE1C, DNMT3B, DOCK8, FLG, IL7R, JAK3, LYST, MLPH, NCF2, NHEJ1, PTPRC, RAB27A, RAG1, RAG2, RFXANK, SLC17A5, STAT3, WAS	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-149	<b>Renal Disease Panel</b> (incl. full bioinformatics analysis & report)	21 genes (>30X)	C5orf42, CC2D2A, CEP290, COL4A4, COL4A6, FRAS1, IQCB1, LAMB2, MKS1, NEK8, NPHP3, NPHP4, NPHS1, PKHD1, SLC4A4, SMARCAL1, TCTN2, TMEM237, TMEM67, UPK3A, XDH	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGT-151	<b>Warfarin sensitivity Panel</b> (incl. full bioinformatics analysis & report)	2 genes (>30X)	CYP2C9, VKORC1	2	EDTA Blood, Buccal Swab	15-20 days	Validated kit-based assay
HGT-150	<b>Visual Disorder Panel</b> (incl. full bioinformatics analysis & report)	80 genes (>30X)	ABCA4, ADAM9, AIPL1, ALMS1, ARL6, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C8orf37, CACNA1F, CDHR1, CEP290, CERKL, CHD7, CHM, CHST6, CNGA3, CNGB1, CNNM4, CRB1, CRX, CRYAA, CRYBA1, CRYGC, CRYGD, CYP1B1, EFEMP1, EPHA2, EYS, FAM161A, FZD4, GALT, GCNT2, GDF3, GPR179, GUCY2D, IGFBP7, IMPG2, KCNJ13, KCNV2, LCA5, LRAT, LRP5, LTBP2, MERTK, MFRP, MIP, MYO7A, NHS, NMNAT1, NPHP4, NR2E3, PAX6, PDE6C, PROM1, PRPH2, PRSS56, RAB3GAP1, RAB3GAP2, RDH12, RGS9, RLBP1, RP1, RP1L1, RP2, RPE65, RPGRI1, SLC16A12, SLC4A11, SNRNP200, SPATA7, TRPM1, TULP1, USH2A, WDR19	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
<b>INHERITED CANCER PANELS</b>							
HGC-152	<b>Inherited Breast Cancer Panel</b> (incl. full bioinformatics analysis & report)	6 genes (>30X)	BRCA1, BRCA2, CDH1, PALB2, PTEN, TP53	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
HGC-153	<b>Comprehensive Inherited Cancer Panel</b> (incl. full bioinformatics analysis & report)	56 genes (>30X)	ALK, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN2A, CHEK2, FH, FLCN, GPC3, MAX, MEN1, MET, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF2, PALB2, PHOX2B, PMS2, POLD1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RET, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WT1, XRCC2	2	EDTA Blood, Buccal Swab	30-35 days	Validated kit-based assay
<b>SOLID TUMOUR PANELS</b>							
HGC-154	<b>Somatic Cancer Panel (15 genes)</b> (incl. full bioinformatics analysis & report)	15 genes (>30X)	AKT1, BRAF, EGFR, ERBB2, FOXL2, GNA11, GNAQ, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, RET, TP53	8	Fresh tissue, FFPE	5-10 days	Validated kit-based assay
<b>SINGLE NUCLEOTIDE POLYMORPHISM ARRAYS</b>							
HGA-158	<b>Human Whole Genome SNP Array</b>	N/A	Various human SNP arrays including Loss-of-function variants, Pharmacogenomic markers, Inflammation and HLA markers, Copy number variants and other markers associated with human disease. Please contact MBG Centre for full details.	96	EDTA Blood	10-15 days	Validated kit-based assay
<b>METAGENOMICS</b>							
HGT-161	<b>Microbial Whole Genome Analysis</b>	N/A	Shotgun Sequencing	10	Isolates	15-20 days	Validated kit-based assay
HGT-162	<b>Metagenomics - Microbial 16S Analysis</b>	1 (>100X)	16s rRNA sequencing Bacterial Identification	90	Numerous	15-20 days	Validated in-house developed assay
HGT-203	<b>Metagenomics - Microbial 18S Analysis</b>	1 (>100X)	18s rRNA sequencing Bacterial Identification	90	Numerous	15-20 days	Validated in-house developed assay
<b>MITOCHONDRIAL ANALYSIS</b>							

Assay Code	ASSAY NAME	No. Genes (Coverage)	Genes included	Min Sample No per run	Recommended sample types	TAT	Accreditation
HGT-159	Ancestral Studies	WGS (mtDNA)	Whole Mitochondrial DNA (mtDNA) Sequencing	90	Blood, Tissue, Buccal Swab, Urine	30-35 days	Validated in-house developed assay