



### DNA REQUIREMENTS

Please send us the following DNA amounts for the following tests. The 260/280 ratio should be above 1.80. Please contact us if you would like more information about sending us DNA.

Achromatopsia Panel, 5 genes (CNGA3, CNGB3, GNAT2, PDE6C, PDE6H)	10µg
Ashkenazi Jewish Panel, 7 genes (CLRN1-N48K, DHDDS-K42E, MAK-K429insAlu, FAM161A-c.1355-6delCA, FAM161A-c.1567C>T, LCA5-Q279X, PCDH15-R245X, CACNA2D4-delExon17-26, TRPM1-delExon2-7)	10µg
Anterior Segment Dysgenesis Panel, 14 genes (DCDC1, ELP4, FOXE3, PAX6, WT1, LAMB2, PITX2, PITX3, FOXC2, FOXC1, CYP1B1, COL4A1, BMP4, B3GALTL)	10µg
Bardet-Biedl Syndrome (BBS) Panel, 16 genes (BBS1 - BBS16)	10µg
Cockayne Syndrome Panel, 2 genes (ERCC6, ERCC8)	10µg
Common Hearing Loss Panel, 3 genes (SLC26A4, GJB2, GJB6)	10µg
Cone-Rod Dystrophy Panel, 26 genes (ABCA4, ADAM9, AIPL1, BEST1, c8ORF37, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, CRX, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2/RDS, RAX2, RDH5, RIMS1, RPGRIP1, SEMA4A, UNC119)	20µg
Congenital Stationary Night Blindness (CSNB) Panel, 14 genes (CABP4, CACNA1F, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1)	10µg
Hereditary Hemorrhagic Telangiectasia (HHT) Panel, 3 genes (ENG, ACVRL1, SMAD4)	10µg
Hermansky-Pudlak Syndrome and Oculocutaneous Albinism Panel, 15 genes (HPS1, HPS2, HPS3, HPS4, HPS5, HPS6, HPS7, HPS8, HPS9, OCA1, OCA2, OCA3, OCA4, OA1, LYST)	10µg
Joubert Syndrome Panel, 18 genes, (CEP290, NPHP1, OFD1, AHI1, RPGIP1L, ARL13B, C5ORF42, CC2D2A, CEP41, INPP5E, KIF7, TCTN1, TCTN2, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B)	20µg
Leber Congenital Amaurosis (LCA) Panel, 19 genes (AIPL1, CABP4, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1)	10µg
Neurofibromatosis Panel, 3 genes (KIT, NF1, SPRED1)	10µg

Neuronal Ceroid-Lipofuscinosis (NCL) Panel, 9 genes (PPT1/CLN1, TPP1/CLN2, CLN3, DNAJC5/CLN4, CLN5, CLN6, MFSD8, CLN8, CTSD/CLN10)	10µg
Optic Atrophy Panel, 2 genes (OPA1, OPA3)	10µg
Osteogenesis Imperfecta Panel, 4 genes (COL1A1, COL1A2, CRTAP, LEPRE1)	10µg
Retinal Dystrophy Panel, 127 genes, please see website for gene list	60µg
Rubinstein-Taybi Syndrome Panel, 3 genes (CREBBP, EP300, SRCAP)	10µg
Senior-Loken Syndrome Panel, 4 genes (CEP290, IQCB1, NPHP1, NPHP4)	10µg
Septo-Optic Dysplasia (SOD) Panel, 4 genes (HESX1, OTX2, SOX2, PAX6)	10µg
Stargardt/Macular Dystrophy Panel, 8 genes (ABCA4, BEST1, EFEMP1, ELOVL4, IMPG1, IMPG2, PROM1, RDS)	10µg
Tuberous Sclerosis (TSC) Panel, 2 genes (TSC1, TSC2)	10µg
Usher Syndrome Panel, 13 genes (ABHD12, CDH23, CLRN1, DFNB31, GPR98, HARS/USH3B, MYO7A, PCDH15, PDZD7, USH1C, USH1G USH1J/CIB2, USH2A)	20µg
Waardenburg Syndrome Panel, 7 genes (EDN3, EDNRB, MITF, PAX3, RET, SNAI2, SOX10)	10µg
<i>ARRAY CGH, qPCR deletion/duplication</i>	10µg
<i>Single genes, familial mutations</i>	10µg

Last updated 9/4/2014