

Additional file 1. The SureSelect Custom Targeted Gene Panel.

CPLANE1, ACTA2, ACTC1, **ACVR2B**, AHI1, ALMS1, ANKS6, ARL3, ARL6, ARMC, ARMC9, B3GAT3, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BICC1, BMP4, C11ORF70, **C21ORF59**, C5ORF42, CC2D2A, **CCDC103**, **CFAP53**, **CCDC114**, **CCDC151**, **CCDC39**, **CCDC40**, **CCDC65**, **CCNO**, CCP110, CDC5L, CDK13, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP52, **CFC1**, CHD1L, CHD7, CRB2, **CRELD1**, CRKL, CSPP1, DHCR7, **DNAAF1**, **DNAAF2**, **DNAAF3**, **DNAAF5**, **DNAH1**, **DNAH11**, **DNAH5**, **DNAH9**, **DNAI1**, **DNAI2**, DNAJB11, DNAL1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, **DYX1C1**, DZIP1L, EHMT1, EYA1, FGF20, **FOXH1**, FRAS1, FREM1, FREM2, GANAB, **GAS8**, GATA3, GATA4, GATA5, GATA6, **GDF1**, GJA1, GJA5, GLI3, GLIS2, GREB1L, HAND1, HNF1B, **HYDIN**, IFT140, IFT172, IFT27, IFT52, IFT74, IFT80, IFT81, INPP5E, INVS, IQCB1, ITGA8, JAG1, ANOS1, KIAA0556, KIAA0586, KIF7, LAMA2, **LEFTY2**, LRP2, **LRRC56**, **LRRC6**, LZTFL1, **MCIDAS**, MCTP2, MED13L, MEIS2, MKKS, MKS1, **MMP21**, **MNS1**, NEK1, NEK8, NKX2-5, NKX2-6, **NME8**, **NODAL**, NOTCH1, NOTCH2, NPHP1, NPHP3, NPHP4, NR2F2, NSD1, **OFD1**, PAX2, PBX1, PDE6D, **PIH1D3**, PKD1, **PKD1L1**, PKD2, PKHD1, PRKD1, PTK7, RBFOX2, REN, RET, ROBO2, RPGR, RPGRIP1L, **RSPH1**, **RSPH3**, **RSPH4A**, **RSPH9**, SALL1, SALL4, SDCCAG8, SIX1, SIX2, SIX5, SLIT3, SOX17, **SPAG1**, **STK36**, TAB2, TBC1D32, TBX1, TBX20, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TFAP2B, TLL1, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNXB, TRAF3IP1, TRAP1, TRIM32, TTC21B, **TTC25**, TTC8, UMOD, UPK3A, WDPCP, WDR19, WDR34, WDR35, WDR60, WNT4, ZFPM2, **ZIC3**, **ZMYND10**, ZNF423

The panel comprises 206 genes implicated in isolated or syndromic congenital heart disease, heterotaxy, renal defects, motile and non-motile ciliopathies. The 40 genes described in association with Primary ciliary dyskinesia (PCD) cases are highlighted in yellow. Additionally, 11 heterotaxy-related genes are highlighted in green.