Multigene sequencing for condition	Genes included	Variants Detected	PolyPhen-2	SIFT/PROVEAN	Mutation Taster
Cholestasis	*ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, AKR1D1, ALDOB, AMACR, ATP8B1, BAAT, CC2D2A, CFTR, CLDN1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, EHHADH, FAH, GPBAR1, HNF1B, HSD17B4, HSD3B7, INVS, JAG1, LIPA, MKS1, MPV17, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKHD1, POLG, SCP2, SERPINA1, SLC10A1, SLC10A2, SLC25A13, SLC27A5, SMPD1, TJP2, TMEM216, TRMU, UGT1A1, VIPAS39, VPS33B*	*EHHADH* gene A336T (c.1006 G>A) heterozygous variant	probably damaging	neutral/damaging	disease causing
		TMEM216 gene F97I (c.289T>A) heterozygous variant	benign	neutral/tolerated	polymorphism
Nephronopthisis	*AHI1, CEP290, GLIS2, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, TMEM67, TTC21B, XPNPEP3, BICC1, CRB2, EYA1, HNF1B, PAX2, PKD1, PKD2, PKHD1, SIX5, UMOD*	*TMEM216* gene F97I (c.289T>A) heterozygous variant	benign	neutral/tolerated	polymorphism
		IQCB1 gene E481K (c.1441G>A) heterozygous variant			
Cardiomyopathy	*ACTC1, ACTN2, BRAF, CSRP3, GLA, HRAS, KRAS, LAMP2, MAP2K1, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, NRAS, PLN, PRKAG2, PTPN11, RAF1, RIT1, SHOC2, SOS1, TNNC1, TNNI3, TNNT2, TPM1*	*MYL2 gene* A13T (c.37G>A) heterozygous variant	Probably damaging	Tolerated/neutral	Disease causing

Table 1: Showing the results of the multigene panels and whole exome sequencing conducted on the proband's blood.