The presence of multiple variants affects the clinical phenotype and prognosis in left ventricular noncompaction after surgery

Keiichi Hirono, MD, PhD,^a Takehiko Sakai, MD,^b Yukiko Hata, PhD,^c and Naonori Nishida, MD, PhD,^a Toyama and Fukui, Japan

From the Departments of ^aPediatrics, and ^cLegal Medicine, Graduate School of Medicine, University of Toyama, Toyama; and ^bDepartment of Pediatrics, Fukui Prefectural Hospital, Fukui, Japan.

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Address for reprints: Keiichi Hirono, MD, PhD, Department of Pediatrics, Graduate School of Medicine, University of Toyama, 2630 Sugitani, Toyama City, Toyama, 930-0194 Japan (E-mail: khirono1973@gmail.com).

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Massive pericardial effusion and prominent trabeculation of the left ventricle can be seen.

Left ventricular noncompaction (LVNC) is a hereditary cardiomyopathy morphologically characterized by numerous prominent trabeculations and deep intertrabecular recesses communicating with the left ventricular cavity.¹ Here, we report the first case of prenatal echocardiography diagnosis of LVNC, which deteriorated after surgical intervention and was associated with variants in the *MYH7* and *CD36* genes.

CASE REPORT

A 30-year-old multigravida with singleton clomiphene pregnancy was referred at 26 weeks of gestation due to the existence of ascites, and fetal echocardiography revealed that the fetus had LVNC and double outlet of the right ventricle, in addition to the interruption of the aorta and aortic stenosis (Figure 1). The patient was born as a full-term female infant with a birth weight of 2.53 kg and Apgar scores of 4 and 9 at 1 and 5 minutes, respectively. No other extracardiac anomalies were observed. No postnatal resuscitation was required, and intravenous prostaglandin E1 was started to retain ductus arteriosus.

As the baby became gradually symptomatic with tachypnea and tachycardia at 6 days of age, she underwent the bilateral pulmonary artery banding. After surgery, heart failure worsened with pericardial effusion, pleural effusion, and ascites. In addition, there was prominent trabeculation of the left ventricle and blood signals within the intertrabecular region (Figure 1). Diuretics, inotropic support, and ventilation support were initiated. The pericardial effusion, pleural effusion, and ascites continued for 77 days. The family history was examined, and it revealed that her father and her grandmother had LVNC (Figure 1). The patient is

now 2 years old, has severe heart failure, and receives intravenous prostaglandin E1.

Once informed consent from the parents was obtained, blood samples of the patient and parents were taken to perform genetic testing using next-generation sequencing, including 4813 genes (Table 1). A novel heterozygous splicing variant in the *MYH7* gene (c.896-1G>A) was identified in the patient and her father (Figure 2). A heterozygous missense variant in the *CD36* gene (c.760T>C, p.Phe254Leu) that was previously reported was identified in the patient and her mother (Figure 2).² Variants were considered likely pathogenic by multiple in silico algorithms (Table 2). The patient and her mother showed reduced *CD36* expression compared with the father in both monocytes and platelets (Figure 2).

DISCUSSION

This is the first fetal case of LVNC associated with variants in *MYH7* and *CD36* genes. *MYH7* defects are a major cause of genetic cardiomyopathies, including LVNC.³ The variant in *MYH7* (c.896-1G>A) identified in this study affects an amino acid located in the myosin head region,

Central Message

We report a fetal case of left ventricular noncompaction associated with variants in the *MYH7* and *CD36* genes. Identification of the molecular defect is important before and after cardiac surgery.

FIGURE 1. Echocardiography image shows an abnormal, highly trabeculated left ventricular myocardium at 30 weeks of gestation (A), at 0 days of age (B), and at 38 days of age in 4-chamber view (C) and short-axis view (D). E, Pedigrees of the patient. *Filled symbols* indicate individuals with LVNC; *open symbols*, unaffected status. RV, Right ventricle; LV, left ventricle; RA, right atrium; LA, left atrium; PE, pericardial effusion.

which contains all the necessary elements to generate movement of actin relative to myosin during ATP hydrolysis. Defective *CD36* is a likely candidate gene for

cardiomyopathy.² Therefore, we hypothesize that the reason why our case with multiple variants was young at diagnosis and presented with heart failure is that our case

A2M, A4GALT, A4GNT, AAAS, AADAC, AADAACL2, AANAT, AARS, AARS2, AASS, ABAT, ABCA1, ABCA10, ABCA12, ABCA13, ABCA14, ABCA15, ABCA16, ABCA17, ABCB1, ABCB11, ABCB4, ABCB6, ABCB7, ABCC1, ABCC11, ABCC2, ABCC3, ABCC4, ABCC6, ABCC8, ABCC9, ABCD1, ABCD3, ABCD4, ABCG1, ABCG2, ABCG5, ABCG8, ABHD12, ABHD5, ABI3BP, ABO, ACACA, ACACB, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADV, ACAN, ACAT1, ACAT2, ACBD5, ACBD6, ACCS, ACE, ACHE, ACLY, ACO2, ACOX1, ACP5, ACSF3, ACSL4, ACSL5, ACSM2B, ACSM3, ACTA1, ACTA2, ACTB, ACTC1, ACTGI, ACTN2, ACTN3, ACTN4, ACVR1, ACVR1B, ACVR1C, ACVR2B, ACVRL1, ACY1, ADA, ADAM10, ADAM12, ADAM17, ADAM19, ADAM33, ADAM7, ADAM9, ADAMTS10, ADAMTS13, ADAMTS16, ADAMTS17, ADAMTS18, ADAMTS2, ADAMTSL2, ADAMTSL3, ADAMTSL4, ADAR, ADARBI, ADC, ADCK3, ADCY10, ADCY3, ADCY5, ADCY6, ADCY9, ADCYAP1, ADD1, ADH1B, ADH1C, ADH4, ADH5, ADH7, ADIPOQ, ADIPOR1, ADK, ADM, ADNP, ADORA1, ADORA2A, ADORA3, ADRA1A, ADRA2A, ADRA2B, ADRA2C, ADRB1, ADRB2, ADRB3, ADRBK2, ADSL, ADTRP, AFF2, AFF3, AFG3L2, AFP, AGA, AGBL4, AGER, AGGF1, AGK, AGL, AGMO, AGO1, AGO2, AGPAT2, AGPS, AGRN, AGRP, AGT, AGTRI, AGTR2, AGXT, AHCY, AHII, AHR, AHRR, AHSG, AHSP, AICDA, AIFI, AIFM1, AIMP1, AIP, AIPL1, AIRE, AKI, AK2, AKAP10, AKAP13, AKAP2, AKAP9, AKR1B1, AKRIC2, AKRIC3, AKRIC4, AKRID1, AKR7A2, AKR7A3, AKT1, AKT2, AKT3, ALAD, ALAS2, ALB, ALCAM, ALDH16A1, ALDH18A1, ALDH1A1, ALDH1A2, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG10B, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALK, ALMS1, ALOX12, ALOX12B, ALOX15, ALOX5, ALOX5AP, ALOXE3, ALPL, ALS2, ALS2CL, ALX1, ALX3, ALX4, AMACR, AMELX, AMELY, AMER1, AMH, AMHR2, AMN, AMPD1, AMPD3, AMT, ANAPC1, ANG, ANGPT1, ANGPTL3, ANGPTL4, ANGPTL5, ANK1, ANK2, ANK3, ANKH, ANKK1, ANKRD1, ANKRD11, ANKRD26, ANKS1A, ANKS1B, ANO10, ANO5, ANO6, ANO7, ANTXR1, ANTXR2, ANXA11, ANXA5, AOA, AOC1, APIS1, APIS2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APAF1, APBA2, APBB1, APBB3, APC, APCDD1, APEX1, APHA, APHB, APLNR, APOA1, APOA2, APOA4, APOA5, APOB, APOBEC3B, APOBEC3G, APOBEC3H, APOC1, APOC2, APOC3, APOC4, APOD, APOE, APOH, APOL1, APOL3, APOM, APP, APR, APTX, AQP1, AQP2, AQP3, AQP4, AQP5, AQP7, AR, AREL1, ARF4, ARFGF2, ARG1, ARHGAP24, ARHGAP31, ARHGAP6, ARHGAP9, ARHGEF10, ARHGEF11, ARHGEF12, ARHGEF6, ARHGEF7, ARHGEF9, ARID1A, ARID1B, ARL11, ARL13B, ARL14EP, ARL6, ARL6IP5, ARMS2, ARPC3, ARSA, ARSB, ARSE, ARSF, ART4, ARVCF, ARX, AS3MT, ASAHI, ASAH2, ASB10, ASCC1, ASCC3, ASCL1, ASIC3, ASIP, ASL, ASMT, ASMTL, ASNS, ASPA, ASPM, ASPN, ASPRV1, ASS1, ASTN2, ASXL1, ATCA, ATF1, ATF3, ATF5, ATF6, ATG16L1, ATG7, ATIC, ATL1, ATM, ATN1, ATOH7, ATP10A, ATP10D, ATP13A2, ATP13A4, ATP1A2, ATP1A3, ATP1B1, ATP2A1, ATP2A2, ATP2A3, ATP2B2, ATP2B4, ATP2C1, ATP5E, ATP5SL, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0A4, ATP6V1B1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRNL1, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN3L, ATXN7, ATXN8OS, AUH, AURKA, AURKC, AUTS2, AVP, AVPRIA, AVPR1B, AVPR2, AXIN1, AXIN2, AXL, B2M, B3GALNT1, B3GALT6, B3GALT, B3GAT3, B3GNT1, B3GNT3, B4GALT1, B4GALT7, B9D1, B9D2, BAALC, BAAT, BACE1, BAG3, BAG6, BANF1, BANK1, BAP1, BARD1, BAX, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAM, BCAP31, BCAT1, BCAT2, BCHE, BCKDHA, BCKDHB, BCL10, BCL11A, BCL2, BCL2A1, BCL2L1, BCL2L2, BCL9, BCMO1, BCOR, BCORL1, BCR, BCS1L, BDKR2, BDNF, BEAN1, BEST1, BEX4, BFSP1, BFSP2, BHLHA9, BHLHE41, BHMT, BICC1, BICD1, BIN1, BIRC5, BLK, BLM, BLMH, BLNK, BLOC1S3, BLOC1S6, BLVRA, BM1, BMP1, BMP10, BMP15, BMP2, BMP2K, BMP4, BMP5, BMP7, BMPER, BMPRIA, BMPRI1B, BMPRI2, BNC2, BOLA3, BPGM, BPI, BP1FA1, BPY2, BRAF, BRAP, BRAT1, BRCA1, BRCA2, BRCC3, BRD1, BRIPI, BRSK2, BRWD1, BRWD3, BSCL2, BSG, BSND, BST1, BTAF1, BTBD9, BTC, BTD, BTK, BTLA, BTNIA1, BTN2A1, BTNL2, BTRC, BUB1B, C10orf11, C10orf137, C10orf2, C12orf10, C12orf57, C12orf65, C19orf12, CIGALT1, CIQA, CIQB, C1QC, C1QTNF5, CIR, CIS, C2, C2orf71, C3, C3ARI, C4, C4B, C4BPA, C5, CSAR2, C5orf42, C6, C6orf15, C7, C7orf10, C8A, C8B, C8orf37, C9, C9orf72, CA1, CA12, CA2, CA4, CA6, CA8, CABIN1, CABP4, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1H, CACNA1S, CACNA2D1, CACNA2D3, CACNA2D4, CACNB2, CACNB4, CACNG2, CADM1, CALCA, CALCR, CALHM1, CALM1, CALM3, CALR, CALR3, CAMK4, CAMKK1, CAMKK2, CAMP, CAMTA1, CANT1, CAPN10, CAPN13, CAPN3, CARD14, CARD8, CARD9, CARTPT, CASC16, CASC5, CASK, CASP1, CASP10, CASP12, CASP2, CASP3, CASP5, CASP8, CASP9, CASQ2, CASR, CAST, CAT, CATSPER1, CATSPER2, CATSPER3, CATSPER4, CAV1, CAV3, CBFB, CBL, CBLB, CBRI, CBR3, CBS, CBX2, CBX4, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC14, CCDC170, CCDC22, CCDC28B, CCDC39, CCDC40, CCDC50, CCDC8, CCDC88C, CCHCR1, CCK, CKAR, CCKBR, CCL11, CCL13, CCL17, CCL2, CCL22, CCL26, CCL3L1, CCL4L1, CCL5, CCL7, CCM2, CCNA2, CCND1, CCR1, CCR2, CCR3, CCR5, CCR6, CCR7, CCRL2, CCT5, CD109, CD14, CD151, CD177, CD19, CD1A, CD1E, CD207, CD209, CD22, CD226, CD24, CD244, CD247, CD27, CD2AP, CD320, CD36, CD38, CD3D, CD3E, CD3EAP, CD3G, CD4, CD40, CD40LG, CD44, CD46, CD5, CD55, CD58, CD59, CD72, CD79A, CD79B, CD81, CD86, CD8A, CD96, CDA, CDAN1, CDC42BPB, CDC6, CDC73, CDH1, CDH12, CDH13, CDH15, CDH23, CDH3, CDH5, CDH8, CDHR1, CDK11A, CDK4, CDK5R1, CDK5RAP2, CDK5RAP3, CDK7, CDKL3, CDKL5, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2B-AS1, CDKN2C, CDON, CDN, CDT1, CDY1, CDY2A, CEACAM16, CEBA, CEBPE, CEL, CELSR1, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP57, CEP63, CEP68, CEP85L, CER1, CERKL, CERS6, CES1, CES2, CETP, CFB, CFC1, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFL2, CFLAR, CFP, CFTR, CGA, CGB, CHAT, CHD1L, CHD2, CHD3, CHD6, CHD7, CHD8, CHDH, CHEK2, CHFR, CHGA, CHGB, CHI3L1, CHI3L2, CHIA, CHIT1, CHK, CHL1, CHM, CHMP2B, CHMP4B, CHN1, CHRDL1, CHRFAM7A, CHRM1, CHRM2, CHRM3, CHRNA1, CHRNA2, CHRNA3, CHRNA4, CHRNA5, CHRNA7, CHRNA9, CHRN1, CHRN2, CHRN4, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHST8, CHSY1, CHUK, CIB2, CIC, CIDEA, CIDEA, CIITA, CILP, CIRH1A, CISD2, CISH, CITED2, CKM, CLCA1, CLCA2, CLCF1, CLCN1, CLCN2, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN14, CLDN16, CLDN19, CLEC11A, CLEC2D, CLEC4M, CLEC7A, CLIC2, CLK2, CLMP, CLN3, CLN5, CLN6, CLN8, CLNK, CLOCK, CLPS, CLPTM1, CLRNI, CLSTN2, CLTCL1, CLU, CLUL1, CLYBL, CMA1, CMPK1, CNBP, CNDP1, CNGA1, CNGA3, CNGB1, CNGB3, CNKSR1, CNKSR2, CNNM2, CNNM4, CNOT3, CNOT4, CNPY3, CNR1, CNR2, CNTF, CNTN1, CNTN4, CNTNAP2, CNTNAP4, CNTNAP5, COA5, COCH, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL17A1, COL18A1, COL1A1, COL1A2, COL25A1, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL4A6, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL6A4P2, COL6A5, COL7A1, COL8A2, COL9A1, COL9A2, COL9A3, COLECI1, COLQ, COMMD1, COMP, COMT, COQ2, COQ4, COQ6, COQ9, CORIN, CORO1A, COX10, COX14, COX15, COX4I1, COX4I2, COX6B1, COX7A1, COX7A2, CP, CPA4, CPA6, CPB2, CPE, CPLX2, CPNI,

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TABLE 1. Continued

CPOX, CPS1, CPT1A, CPT1B, CPT2, CPZ, CRI, CR2, CRADD, CRB1, CRBN, CREB1, CREB3L3, CREBBP, CRELD1, CRH, CRHR1, CRISP2, CRK, CRKL, CRLF1, CRP, CRTAP, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGEP, CRYGS, CRYM, CSAG1, CSDE1, CSF1, CSF1R, CSF2, CSF2RA, CSF2RB, CSF3R, CSGALNACT1, CSH1, CSMD1, CSMD3, CSNK1A1L, CSNK1D, CSNK1E, CSNK2A2, CSNK2A3, CSRP3, CST3, CSTA, CSTB, CSTF2T, CTC1, CTDPI, CTF1, CTGF, CTH, CTHRC1, CTLA4, CTNNA3, CTNNB1, CTNND1, CTNS, CTRC, CTSA, CTSB, CTSC, CTSD, CTSF, CTSK, CTSZ, CTTNBP2, CUBN, CUL3, CUL4B, CUL5, CUL7, CX3CR1, CXCL10, CXCL11, CXCL12, CXCL16, CXCL5, CXCR1, CXCR3, CXCR4, CYB5A, CYB5R3, CYBA, CYBB, CYBRD1, CYCS, CYLD, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1A1, CYP1A2, CYP1B1, CYP21A2, CYP24A1, CYP26A1, CYP26B1, CYP27A1, CYP27B1, CYP2A13, CYP2A6, CYP2B6, CYP2C18, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP2D7P1, CYP2E1, CYP2F1, CYP2G1P, CYP2J2, CYP2R1, CYP2W1, CYP3A4, CYP3A43, CYP3A5, CYP3A5P1, CYP3A7, CYP46A1, CYP4A11, CYP4A22, CYP4B1, CYP4F12, CYP4F2, CYP4F22, CYP4F3, CYP4V2, CYP7A1, CYP7B1, CYSLTR1, CYSLTR2, D2HGDH, DAG1, DAO, DAOA, DAPK1, DARC, DARS2, DAZ1, DAZ2, DAZ3, DAZL, DBH, DBI, DBT, DCAF13, DCAF17, DCC, DCDC2, DCK, DCLK1, DCLRE1C, DCN, DCP1B, DCTD, DCTN1, DCX, DCXR, DDAH1, DDAH2, DDB2, DDC, DDOST, DDR1, DDR2, DDX11, DDX20, DDX25, DDX39B, DDX3Y, DDX5, DDX53, DDX58, DEAF1, DEC1, DECRI, DEFB1, DEFB126, DEFB4A, DES, DFNA5, DFN3B1, DFN59, DGAT1, DGCR14, DGCR2, DGCR5, DGCR6, DGCR8, DGKD, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHH, DHODH, DHRS4L1, DHX16, DHX36, DHX37, DIABLO, DIAPH1, DIAPH2, DIAPH3, DICER1, DIO1, DIO2, DIP2A, DIP2B, DIP2C, DIRC2, DIS3L2, DISC1, DISP1, DKC1, DKK2, DKK3, DLAT, DLD, DLG3, DLG5, DLGAP2, DLGAP3, DLL1, DLL3, DLX3, DLX5, DLX6, DMBT1, DMC1, DMD, DMGDH, DMP1, DMPK, DMRT1, DMXL1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJ4, DNAJB2, DNAJB6, DNAJC19, DNAJC5, DNAJC6, DNAL1, DNASE1, DNASE1L3, DNASE2, DND1, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DNMT3B, DNMT3L, DOC2A, DOCK3, DOCK4, DOCK6, DOCK8, DOCK9, DOK7, DOLK, DPAGT1, DPM1, DPM3, DPP10, DPP6, DPY19L2, DPYD, DPYS, DPYSL2, DRD1, DRD2, DRD3, DRD4, DRD5, DROSHA, DRP2, DSC2, DSC3, DSCAM, DSCR8, DSG1, DSG2, DSG4, DSP, DSPP, DST, DTNA, DTNBP1, DUOX2, DUOX2A, DUSP23, DUX4, DXO, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, DYX1C1, DYX1C1-CCPG1, EARS2, EBAG9, EBP, ECE1, EC1I, ECM1, ECM2, ECSIT, EDA, EDA2R, EDAR, EDARADD, EDN1, EDN2, EDN3, EDNRB, EDNRB, EEF1B2, EEF2K, EFCAB5, EFEMP1, EFEMP2, EFHC1, EFHC2, EFNB1, EFR3A, EFTUD2, EGF, EGFR, EGLN1, EGR2, EGR3, EHD2, EHMT1, EHMT2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3H, EIF4E, EIF4G1, ELAC2, ELANE, ELAVL2, ELF4, ELK1, ELN, ELOVL4, ELP2, ELP4, EMC4, EMD, EME1, EMG1, EMX2, EN2, ENAM, ENG, ENO1, ENO3, ENPP1, ENSA, ENTPD1, EOMES, EP300, EPAS1, EPB41, EPB41L1, EPB42, EPC2, EPCAM, EPHA2, EPHA3, EPHA5, EPHA7, EPHB2, EPHB6, EPHX1, EPHX2, EPM2A, EPO, EPOR, EPX, ERAP1, ERAP2, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN2, ERMAPP, ESAM, ESCO2, ESPN, ESR1, ESR2, ESRRB, ESRRG, ETFA, ETFB, ETFDH, ETHE1, ETNPPL, ETS1, EVC, EVC2, EVI5, EXO1, EXO5, EXOC4, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EYA4, EYS, EZH2, F10, F11, F12, F13A1, F13B, F2, F2R, F2RL1, F3, F5, F7, F8, F9, FA2H, FAAH, FAH2, FABP1, FABP2, FABP3, FABP4, FABP6, FABP7, FADD, FADS2, FAH, FAM120A, FAM126A, FAM134B, FAM161A, FAM205A, FAM20A, FAM20C, FAM58A, FAM83H, FAM8A1, FAM91A1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FARS2, FAS, FASLG, FASN, FASTKD2, FBLIM1, FBLN1, FBLN5, FBN1, FBN2, FBN3, FBP1, FBXL6, FBXO10, FBXO18, FBXO7, FBXW11, FBXW4, FBXW7, FCAR, FCER1A, FCER2, FCGR1A, FCGR2A, FCGR2B, FCGR3A, FCGR3B, FCGRT, FCN2, FCN3, FCRL3, FDFT1, FECH, FEM1A, FEN1, FERMT1, FERMT3, FEV, FEZF2, FFAR1, FFAR4, FGA, FGB, FGD1, FGD3, FGD4, FGF1, FGF10, FGF14, FGF2, FGF20, FGF23, FGF3, FGF8, FGF9, FGFBP1, FGFR1, FGFR2, FGFR3, FGFR4, FGFR1L, FGG, FH, FHL1, FHL2, FIG4, FIGLA, FIP1L1, FKBP10, FKBP14, FKBP5, FKBP6, FKBP6L, FKR1, FKTN, FLCN, FLG, FLNA, FLNB, FLNC, FLT1, FLT3, FLT4, FLVCR1, FLVCR2, FMN1, FMN2, FMO1, FMO2, FMO3, FMO4, FMO5, FMO6P, FMRI, FN1, FN3K, FOLH1, FOLR1, FOXA1, FOXA2, FOXA3, FOXC1, FOXC2, FOXD3, FOXE1, FOXE3, FOXF1, FOXF2, FOXG1, FOXH1, FOXI1, FOXI2, FOXL2, FOXN1, FOXPI, FOXF2, FOXF3, FOXRED1, FPGS, FPR1, FPR2, FRA10AC1, FRAS1, FREM1, FREM2, FREM3, FRG1, FRK, FRMD6, FRMD7, FRMPD4, FRY, FRZB, FSCB, FSCN2, FSHB, FSHR, FTCD, FTH1, FTHL17, FTL, FTO, FTSJ1, FUCA1, FURIN, FUS, FUT1, FUT2, FUT3, FUT6, FUT7, FUT8, FUZ, FXN, FXYD2, FXYD6, FYCO1, FZD1, FZD3, FZD4, FZD6, FZD9, G6PC, G6PC2, G6PC3, G6PD, GAA, GAB2, GABBR1, GABRA1, GABRA5, GABRA6, GABRB3, GABRD, GABRG1, GABRG2, GABRG3, GABRR2, GAD1, GAD2, GADD45B, GAK, GAL3ST3, GALT, GALE, GALK1, GALNS, GALNT12, GALNT18, GALNT2, GALNT3, GALNT9, GALNTL5, GALP, GALT, GAMT, GAN, GAP43, GARS, GAS1, GAS2L2, GAS6, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD1, GATM, GBA, GBA3, GBE1, GBGT1, GC, GCDH, GCGR, GCH1, GCK, GCKR, GCLC, GCLM, GCM2, GCNT2, GCSH, GDAP1, GDF1, GDF15, GDF3, GDF5, GDF6, GDF9, GDI1, GDNF, GEMIN2, GEMIN4, GFAP, GFER, GFII, GFIIIB, GFM1, GFPT1, GFPT2, GFRA1, GGCX, GGH, GGT5, GH1, GH2, GHR, GHRH, GHRHR, GHRL, GHSR, GIF, GIGYF2, GIMAP8, GIP, GIPC3, GIPR, GIT1, GJA1, GJA3, GJA4, GJA5, GJA8, GJB1, GJB2, GJB3, GJB4, GJB6, GJC2, GJC3, GJD2, GK, GLA, GLB1, GLCCII, GLDC, GLE1, GLI1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLO1, GLP1R, GLRA1, GLRB, GLRX5, GLS, GLTSCR1, GLUD1, GLUD2, GLUL, GLYCTK, GM2A, GMIP, GNA14, GNAI2, GNAQ, GNAS, GNAS-AS1, GNAT1, GNAT2, GNB1L, GNB3, GNB5, GNE, GNMT, GNPTAB, GNPTG, GNRH1, GNRHR, GNS, GOLGA3, GOLGA5, GON4L, GORAB, GOSR2, GOT1, GP1BA, GP1BB, GP2, GP6, GP9, GPAM, GPANK1, GPATCH8, GPBARI, GPC3, GPC4, GPC6, GPD1, GPD1L, GPD2, GPHN, GPI, GPIHBP1, GPR1, GPR12, GPR139, GPR143, GPR179, GPR33, GPR55, GPR56, GPR68, GPR98, GPS1, GPSM2, GPT, GPX1, GPX4, GRB10, GREM1, GRHL2, GRHPR, GRIA3, GRID1, GRIK1, GRIK2, GRIK3, GRIK4, GRIN1, GRIN2A, GRIN2B, GRIN3A, GRIP1, GRK1, GRK4, GRK5, GRM1, GRM3, GRM5, GRM6, GRM7, GRM8, GRN, GRPR, GRXCRI, GSC, GSDMA, GSDMB, GSE1, GSK3B, GSN, GSPT1, GSPT2, GSR, GSS, GSTA1, GSTA2, GSTA3, GSTK1, GSTM1, GSTM3, GSTM4, GSTO1, GSTO2, GSTP1, GSTT1, GSTT2, GSTT2B, GSTZ1, GTDC2, GTF2E1, GTF2H1, GTF2H5, GTF2IRD1, GTF2IRD2, GUCA1A, GUCA1B, GUCY2C, GUCY2D, GUSB, GYG1, GYLTL1B, GYPA, GYPB, GYPC, GYPE, GYS1, GYS2, GZMB, H19, H2BFWT, H6PD, HABP2, HACE1, HADH, HADHA, HADHB, HAL, HAMP, HAND1, HAND2, HARS, HARS2, HAS1, HAVCR1, HAX1, HBA1, HBA2, HBB, HBD, HBE1, HBEGF, HBG1, HBG2, HBM, HBS1L, HBZ, HCCS, HCK, HCLS1, HCN1, HCN2, HCN4, HCP5, HCRT, HCRT1, HCRT2, HDAC4, HDAC8, HDAC9, HDC, HDLBP, HDX, HEATR2, HELQ, HEPACAM, HERC2, HES6, HES7, HESX1, HEXA, HEXB, HEY1, HFE, HFE2, HGD, HGF, HGSNAT, HHEX, HHIP, HIBCH, HIF1A, HIF1AN, HIGD2A, HIP1, HIST1H2AE, HIST3H3, HK1, HK2, HLA-A, HLA-B, HLA-C, HLA-DMB, HLA-DOA, HLA-DPB1, HLA-DPB2, HLA-DQA1,

(Continued)

TABLE 1. Continued

HLA-DQB1, HLA-DRA, HLA-DRB1, HLA-DRB5, HLA-E, HLA-G, HLCS, HLX, HMBS, HMCN1, HMGAI, HMGAI2, HMGCL, HMGCR, HMGCS2, HMHA1, HMOX1, HMOX2, HMSD, HMX1, HMX2, HNF1A, HNF1B, HNF4A, HNMT, HNRNP3, HNRNPU, HOGAI, HOMER2, HOXA1, HOXA10, HOXA11, HOXA13, HOXA2, HOXA4, HOXB13, HOXB6, HOXD10, HOXD13, HOXD4, HP, HPD, HPGD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HR, HRAS, HRC, HRG, HRH2, HRH3, HS1BP3, HS6ST1, HSD11B1, HSD11B2, HSD17B1, HSD17B10, HSD17B2, HSD17B3, HSD17B4, HSD3B1, HSD3B2, HSD3B7, HSF4, HSFY1, HSN2, HSP90AA1, HSP90B1, HSPA1A, HSPA1B, HSPA1L, HSPA5, HSPA8, HSPA9, HSPB1, HSPB3, HSPB6, HSPB7, HSPB8, HSPD1, HSPG2, HTN3, HTRA1, HTRA2, HTR1B, HTR2A, HTR2B, HTR2C, HTR3A, HTR3B, HTR3C, HTR3E, HTR5A, HTR6, HTR7, HTRA1, HTRA2, HTT, HUS1B, HUWE1, HVCN1, HYAL1, HYDIN, HYL1, HYMAI, IAPP, IBSP, ICAM1, ICAM4, ICAM5, ICK, ICOS, ID3, IDE, IDH1, IDH2, IDH3B, IDO1, IDS, IDUA, IER3IP1, IFI30, IFI44L, IFIH1, IFITM3, IFITM5, IFNA10, IFNA17, IFNA2, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IFNL3, IFRD1, IFT122, IFT140, IFT43, IFT80, IGBP1, IGF1, IGF1R, IGF2, IGF2R, IGFBP1, IGFBP3, IGFBP5, IGFBP7, IGHA1, IGHG2, IGHM, IGHMBP2, IGKV, IGLL1, IHH, IKBP, IKBKAP, IKBKKG, IL10, IL10RA, IL10RB, IL11RA, IL12A, IL12B, IL12RB1, IL12RB2, IL13, IL16, IL17A, IL17F, IL17RA, IL17RB, IL17REL, IL18, IL18R1, IL18RAP, IL19, IL1A, IL1B, IL1R1, IL1RAPL1, IL1RL1, IL1RN, IL2, IL20RA, IL20RB, IL21, IL21R, IL23R, IL2RA, IL2RG, IL3, IL31RA, IL36RN, IL4, IL4R, IL5, IL6, IL6R, IL7, IL7R, IL8, IL9, IL9R, ILDR1, ILK, IMM2P2L, IMMT, IMPA2, IMPAD1, IMPDH1, IMPDH2, IMPG2, INF2, INHA, INMT, INPP4A, INS, INSIG1, INSIG2, INSL3, INSL6, INSR, INVS, IQCB1, IQGAP1, IQGAP2, IQGAP3, IQSEC2, IRAK1, IRAK3, IRAK4, IRF1, IRF2, IRF4, IRF5, IRF6, IRF7, IRF8, IRGM, IRS1, IRS2, IRS4, IRX4, IRX5, ISCU, ISL1, ISPD, ISYNA1, ITCH, ITGA11, ITGA2, ITGA2B, ITGA3, ITGA4, ITGA6, ITGA7, ITGA9, ITGAE, ITGAM, ITGB2, ITGB3, ITGB4, ITIH1, ITIH3, ITIH4, ITIH6, ITK, ITM2B, ITPA, ITPKC, ITPRI, ITPR3, ITS2, IVD, IYD, JAG1, JAG2, JAK2, JAK3, JAM3, JMD1C, JPH2, JPH3, JRK, JUN, JUP, KAL1, KALRN, KANK1, KANSL1, KARS, KAT6B, KATNAL2, KBTBD13, KCNA1, KCNA3, KCNA5, KCNAB2, KCNC3, KCND2, KCND3, KCNE1, KCNE1L, KCNE2, KCNE3, KCNE4, KCNH2, KCNIP4, KCNJ1, KCNJ10, KCNJ11, KCNJ13, KCNJ15, KCNJ2, KCNJ3, KCNJ5, KCNJ6, KCNJ8, KCNJ9, KCNK18, KCNK6, KCNK9, KCNMA1, KCNMB1, KCNMB3, KCNN3, KCNQ1, KCNQ2, KCNQ3, KCNQ4, KCNS1, KCNS3, KCNV2, KCTD13, KCTD7, KDM4C, KDM5A, KDM5C, KDM5D, KDM6A, KDM6B, KDR, KEL, KERA, KHDC3L, KHK, KIAA0100, KIAA0196, KIAA0226, KIAA0232, KIAA0319, KIAA0513, KIAA1033, KIAA1199, KIAA1279, KIAA1377, KIAA1432, KIAA1462, KIAA2022, KIF11, KIF17, KIF18A, KIF1A, KIF1B, KIF21A, KIF22, KIF5A, KIF5B, KIF6, KIF7, KIFAP3, KIR2DL1, KIR2DL3, KIR2DL4, KIR3DL1, KIR3DL2, KIRREL3, KISS1, KISS1R, KIT, KITLG, KL, KLB, KLF1, KLF10, KLF11, KLF5, KLF6, KLF7, KLHDC8B, KLHL10, KLHL3, KLHL7, KLHL9, KLK1, KLK12, KLK15, KLK3, KLK4, KLK7, KLKB1, KLRK1, KMT2C, KMT2D, KMT2E, KNG1, KPNA1, KRAS, KRIT1, KRT1, KRT10, KRT12, KRT13, KRT14, KRT16, KRT17, KRT18, KRT2, KRT3, KRT31, KRT37, KRT38, KRT4, KRT5, KRT6A, KRT6B, KRT6C, KRT74, KRT75, KRT8, KRT81, KRT83, KRT85, KRT86, KRT9, KRTAP1-1, KYNU, L1CAM, L2HGDH, L3MBTL1, LAMA1, LAMA2, LAMA3, LAMA4, LAMA5, LAMB1, LAMB2, LAMB3, LAMC1, LAMC2, LAMC3, LAMP2, LAMTOR2, LARGE, LARS2, LBP, LBR, LCA5, LCAT, LCE3B, LCE3C, LCE5A, LCN10, LCT, LDB3, LDHA, LDHB, LDLR, LDLRAD4, LDLRAP1, LEFTY2, LEMD3, LEP, LEPR, LEPRE1, LEPREL1, LETM1, LFNG, LGALS13, LGALS2, LGALS3, LGI1, LHB, LHCGR, LHFPL5, LHX1, LHX3, LHX4, LHX8, LIAS, LIF, LIFR, LIG1, LIG3, LIG4, LILRA3, LIM2, LIMK1, LIN28A, LIN28B, LINS, LIPA, LIPC, LIPE, LIPG, LIPH, LIPI, LIPN, LITAF, LLGL1, LMAN1, LMBR1, LMBRD1, LMF1, LMNA, LMNB1, LMNB2, LMTK3, LMX1B, LNX2, loc344967, LOR, LOX, LOXHD1, LOXLI, LOXL2, LPA, LPAR1, LPAR6, LPIN1, LPIN2, LPIN3, LPL, LPP, LRAT, LRCH1, LRFN5, LRP1, LRP2, LRP4, LRP5, LRP6, LRP8, LRPAP1, LRPPRC, LRRC4, LRRC6, LRRC8A, LRRK2, LRSAM1, LRTOMT, LTA, LTBP1, LTBP2, LTBP3, LTBP4, LTC4S, LTF, LTK, LTN1, LUM, LY96, LYN, LYST, LYZ, LZTFL1, LZTS1, MACROD2, MAD1L1, MAD2L1, MADD, MAF, MAFB, MAGEE2, MAGEL2, MAGI2, MAGT1, MAK, MAMLD1, MAN1A2, MAN1B1, MAN2B1, MANBA, MAOA, MAOB, MAP2, MAP2K1, MAP2K2, MAP2K3, MAP2K4, MAP3K1, MAP3K15, MAP4K5, MAP6, MAP7D3, MAPK10, MAPK8IP1, MAPT, MARS2, MARVELD2, MASP1, MASP2, MAST4, MASTL, MAT1A, MATN3, MATR3, MAVS, MAX, MBD1, MBD3, MBD4, MBD5, MBL2, MBTPS2, MC1R, MC2R, MC3R, MC4R, MCCC1, MCCC2, MCEE, MCF2L2, MCFD2, MCHR1, MCL1, MCM3AP, MCM4, MCM5, MCM6, MCOLN1, MCPH1, MDH1, MDM2, MDM4, MDN1, MECP2, MED12, MED13, MED13L, MED17, MED23, MED25, MEF2A, MEF2C, MEFV, MEGF10, MEGF11, MEIS1, MEIS2, MEN1, MEP1B, MERTK, MESDC2, MESP2, MEST, MET, MFF, MFGE8, MF12, MFN2, MFRP, MFSD2A, MFSD8, MGAT1, MGAT2, MGAT4C, MGEA5, MGLL, MGMT, MGP, MGST2, MGST3, MIA3, MIAT, MICA, MICB, MID1, MIF, MIIP, MINPP1, MIP, MIPOL1, MIR106B, MIR124-1, MIR125A, MIR126, MIR140, MIR146A, MIR16-1, MIR17, MIR17HG, MIR182, MIR184, MIR191, MIR196A2, MIR206, MIR24-1, MIR27A, MIR2861, MIR30C1, MIR34B, MIR499A, MIR502, MIR510, MIR890, MIR892B, MIR934, MIR96, MIRLET7E, MITF, MKKS, MKL1, MKRN3, MKS1, MLC1, MLH1, MLH3, MLLT3, MLPH, MLXIPL, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MME, MMEL1, MMP1, MMP10, MMP12, MMP13, MMP14, MMP2, MMP20, MMP3, MMP7, MMP8, MMP9, MNX1, MOCOS, MOCS1, MOCS2, MOG, MOGS, MOK, MPDUI, MPDZ, MPG, MPHOSPH8, MPI, MPL, MPLKIP, MPO, MPP3, MPP4, MPP6, MPP7, MPST, MPV17, MPZ, MR1, MRAP, MRC1, MRE11A, MRPL3, MRPL48, MRPS16, MRPS22, MRRF, MS4A1, MS4A12, MS4A2, MS4A3, MS4A6A, MS4A6E, MSH2, MSH3, MSH4, MSH5, MSH6, MSMB, MSMO1, MSR1, MSRB3, MST1, MST1R, MSTN, MSX1, MSX2, MT1A, MT2A, MTAP, MTCH2, MTFMT, MTHFD1, MTHFD1L, MTHFR, MTHFS, MTM1, MTMR14, MTMR2, MTMR9, MTNR1A, MTNR1B, MTO1, MTPAP, MTR, MTRR, MTPP, MTUS1, MUC1, MUC13, MUC2, MUC3A, MUC4, MUC5B, MUC6, MUC7, MURC, MUSK, MUT, MUTHY, MVK, MX1, MYB, MYBL2, MYBPC1, MYBPC3, MYC, MYCL, MYCN, MYD88, MYF6, MYH11, MYH13, MYH14, MYH15, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL2, MYL3, MYLIP, MYLK, MYLK2, MYO15A, MYO18B, MYO1A, MYO1C, MYO1E, MYO1F, MYO3A, MYO5A, MYO5B, MYO6, MYO7A, MYO7B, MYO9B, MYOC, MYOCD, MYOM1, MYOT, MYOZ2, MYPN, MYT1, MYT1L, NAA10, NAGA, NAGLU, NAGPA, NAGS, NAIP, NAMPT, NAPRT1, NARS2, NAT1, NAT2, NAT8L, NAV2, NBAS, NBEA, NBEAL2, NBN, NBPFI, NCALD, NCAM1, NCAN, NCAPD2, NCF1, NCF2, NCF4, NCKAP1, NCOA1, NCOA3, NCR3, NCS1, NCSTN, NDE1, NDN, NDOR1, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB1, NDUFB3, NDUFB6, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NEB, NEBL, NEDD4L, NEDD9, NEFH, NEFL, NEFM, NEGR1, NEIL1, NEIL2, NEK1, NEK8, NELFA, NELL1, NEU1, NEU2, NEUROD1, NEUROG3, NEXN, NF1, NF2, NFATC4, NFE2L2, NFIA, NFIX,

(Continued)

TABLE 1. Continued

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OFD1, OGG1, OLFM2, OLR1, OMG, OPA1, OPA3, OPCML, OPHN1, OPN1LW, OPN1MW, OPN1SW, OPN4, OPRD1, OPRK1, OPRL1, OPRM1, OPTC, OPTN, OR13G1, OR7D4, ORAI1, ORC1, ORC4, ORC6, ORMDL3, OSMR, OSR1, OSTM1, OTC, OTOA, OTOF, OTOR, OTX2, OVGP1, OXCT1, OXTR, P2RX1, P2RX4, P2RX5, P2RX7, P2RY11, P2RY12, P2RY4, PABPC4L, PABPN1, PACRG, PAFAH1B1, PAFAH1B3, PAH, PAK3, PAK7, PALB2, PALLD, PANK2, PAPD7, PAPSS2, PARD3B, PARD6A, PARK2, PARK7, PARL, PARP1, PARP2, PASK, PAWR, PAX1, PAX2, PAX3, PAX4, PAX5, PAX6, PAX7, PAX8, PAX9, PC, PCBD1, PCCA, PCCB, PCDH11X, PCDH15, PCDH18, PCDH19, PCDH9, PCDHA10, PCDHA13, PCDHB4, PCK1, PCK2, PCM1, PCMT1, PCNT, PCOLCE, PCSK1, PCSK2, PCSK9, PDCD1, PDCD10, PDCD5, PDE10A, PDE11A, PDE4B, PDE4D, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDE7B, PDE8B, PDGFB, PDGFC, PDGFRA, PDHA1, PDHB, PDHX, PDLIM3, PDLIM5, PDP1, PDPK1, PDSS1, PDSS2, PDX1, PDXK, PDYN, PDZD7, PEAR1, PECAM1, PECR, PEMT, PENK, PEPD, PER1, PER2, PER3, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, 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FIGURE 2. A, Sanger sequence analysis of the patient. Flow cytometry analysis for surface expression of CD36 in monocytes (B) and platelets (C) from the patient and parents.

could be caused by a novel variant in the *MYH7* gene, and the heart failure, which worsened after surgical intervention, could be caused by the *CD36* abnormality.

In conclusion, our data support the hypothesis that the presence of multiple variants can affect the clinical

phenotype and prognosis in LVNC. Our case indicates that the evaluation of cardiac function, the presence of additional congenital malformations and family history, along with identification of the molecular defect, are likely to be important before and after cardiac surgery.

TABLE 2. Mutations identified in this patient

Gene	Protein	Coding	dbSNP	fathmm	SIFT	Polyphen-2	Align GVGD	Mutation Taster	PROVEAN	CADD	Minor allele frequency			Disease data base	
											ExAC, %	ExAC (East Asian), %	HGVD, %	Clin Var	HGMD
MYH7	splicesite_5	c.896-1C>T*		-	-	-	-	-	-	24.9	-	0	-	-	-
CD36	p.Phe254Leu	c.760T>C	rs142186404	Tolerated	Damaging	Probably damaging	C15 (GV: 0.00 – GD: 21.82)	Disease- causing	Deleterious	33.0	0.0033	0.046	0.18	Pathogenic (AR) Platelet glycoprotein deficiency IV deficiency	CM020668 (DM) CD36

dbSNP, Single Nucleotide Polymorphism database; fathmm, functional analysis through hidden Markov models; SIFT, Scale-Invariant Feature Transform; Polyphen-2, Polymorphism Phenotyping v2; PROVEAN, Protein Variation Effect Analyzer; CADD, Combined Annotation Dependent Depletion; ExAC, Exome Aggregation Consortium; HGVD, Human Gene Mutation Database; HGMID, Human Gene Mutation Database; GV, Grantham variation; GD, Grantham deviation.

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