Table 1 Selected examples of monogenic cardiovascular diseases (Vrablík et al., 2021)

Gene(s)	CVD	Manifestation
LDLR, APOB, PCSK9	Familial hypercholesterolemia	High concentrations of LDL and total cholesterol; xanthomas; arcus lipoides cornae; xanthalesmas; coronary heart disease
ABCG5, ABCG8	Sitosterolemia	High plasma sitosterol, campesterol; hypercholesterolemia; premature coronary heart disease; xanthomas
MYH7, MYBPC3, TNNT2, TPM1, MYL2, MYL3, PLN	Hypertrophic cardiomyopathy	Hypertrophy of left ventricle, shortness of breath, diastolic dysfunction, left ventricular outflow ischemia
PKP2, DSP, DSG2, JUP, TMEM43	Arrhythmogenic right ventricular cardiomyopathy	Ventricular arrhythmias, right ventricular cardiomyopathy
MYH7, MYBPC3, TNNT2, MYH6, MYPN, ANKRD1, RAF1, DES, DMD	Familial dilated cardiomyopathy	Diastolic dysfunction, left ventricular hypertrophy, atrial fibrillation, congestive heart failure
FBN1, TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3, SKI	Marfan' s syndrome	Aortic aneurysm or dissection, valvular heart disease, enlargement of the proximal pulmonary artery, congestive heart failure, arrhythmias
ACTA2, FBN1, MYH11, TGFBR1/2, LOX, COL3A1, TGFB2/3	Thoracic aortic aneurysm and dissection	Chest pain, renal cysts, thumb-palm sign, temporal arteritis, bicuspid aortic valve, abdominal aneurysm, intracranial aneurysm
BMPR2, BMPR1B, CAV1, KCNK3, SMAD9, ACVRL1, ENG, EIF2AK4	Pulmonary arterial hypertension	Right ventricular failure, impaired brachial artery flow-mediated dilation, increased pulmonary vascular resistance
KCNQ1/H2/E1/J2, SCN5A, CAV3, CALM1/2	Long QT syndrome	Malignant arrhythmia, palpitations, syncope, anoxic seizures secondary to ventricular arrhythmia
KCNH2	Short QT syndrome	Abbreviated QTc interval on the ECG, propensity for atrial and ventricular arrhythmias
SCN5A	Brugada syndrome	Elevation of the ST, ventricular fibrillation, syncope, arrhythmia