



Case report

Case report of heterotaxy syndrome with sinus node dysfunction and left ventricular hypertrabeculation: clinical and genetic insights

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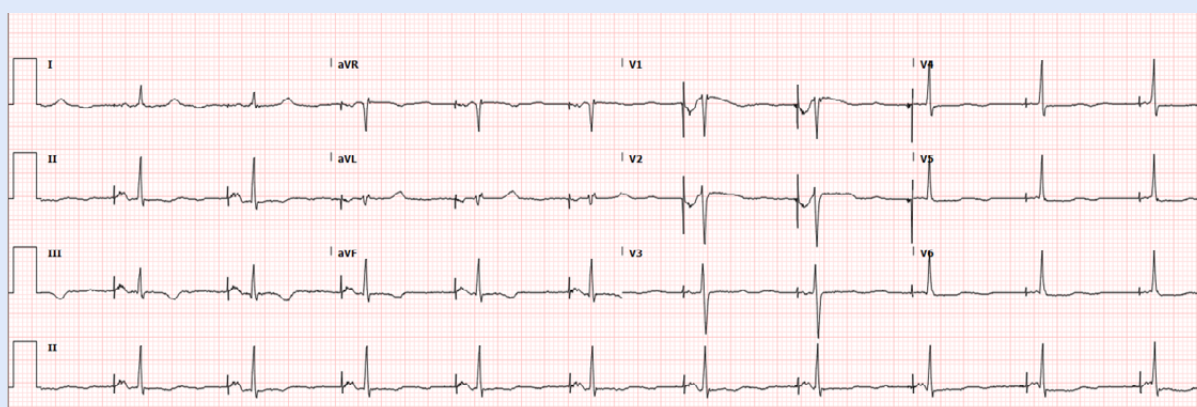


Figure 1S. ECG post-pacemaker implantation showing atrial-paced rhythm, ventricular-sensed beats, with a narrow QRS complex.

Table 1S. Genes associated with left ventricular hypertrabeculation. This table includes genes that have been classified as definitively associated with left ventricular hypertrabeculation (previously referred to as non-compaction), based on current genetic and experimental evidence.

Gene	Gene Name	Associated Syndrome(s)	Gene Function
ACTC1	Actin alpha cardiac muscle 1		Sarcomere function*
DES	Desmin	Myopathy, myofibrillar; Scapuloperoneal syndrome, neurogenic, Kaeser type	Sarcomere function*
DSP	Desmoplakin	Epidermolysis bullosa, lethal acantholytic; Keratosis palmoplantaris striata II; Skin fragility-woolly hair syndrome	Cellular junction protein
MIB1	Mindbomb E3 ubiquitin protein ligase 1		Protein degradation
MYBPC3	Myosin binding protein C3		Sarcomere function*
MYH7	Myosin heavy chain 7	Laing distal myopathy; Myopathy, myosin storage; Scapuloperoneal syndrome, myopathic type	Sarcomere function*
NONO	Non-POU domain containing octamer binding	Mental retardation, X-linked, syndromic	Transcriptional/translational regulator
RYR2	Ryanodine receptor 2	Ventricular tachycardia, catecholaminergic polymorphic	Sarcomere function*
TAZ	Tafazzin	Barth syndrome	Mitochondrial function
TPM1	Tropomyosin 1		Sarcomere function*
TTN	Titin	Muscular dystrophy, limb-girdle, autosomal recessive; Myopathy, myofibrillar; Salih myopathy; Tibial muscular dystrophy, tardive	Sarcomere function*

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Table 2S. Associated genes with sinus sick syndrome.

Gene	Clinical Features	Reference
SCN5A	Congenital/adult Sick sinus syndrome, Brugada syndrome	DOI:10.1172/JCI18062, DOI:10.4070/kcj.2016.46.1.63
HCN4	Sinus bradycardia, chronotropic incompetence, association with AF and left ventricular hypertrabeculation	DOI:10.1016/j.hrthm.2017.02.006, DOI:10.1016/j.hrthm.2017.01.020
EMD	Sick sinus syndrome and atrial arrhythmias in Emery-Dreifuss muscular dystrophy	DOI:10.3233/jnd-230172
CACNA1C	LQTS, family members with AF and SSS	DOI:10.1002/mgg3.1673
MT-TL1	Hypertrophic/dilated cardiomyopathy, preexcitation syndromes, conduction delay, sick sinus syndrome	DOI:10.1111/chd.12634
GNB5	Intellectual developmental disorder with cardiac arrhythmia syndrome; sick sinus syndrome	DOI:10.3760/cma.j.cn112140-20200421-00411
TTN/CCDC141	Sinus bradycardia, early repolarization, high susceptibility to atrial fibrillation, SSS	DOI:10.3892/mmr.2018.8773, DOI:10.1093/eurheartj/ehaa1108
KCNQ2	Familial sick sinus syndrome	DOI:10.1152/physiolgenomics.00132.2021
KRT8	Familial sick sinus syndrome	DOI:10.1093/eurheartj/ehaa1108
SCN10A	Atrial fibrillation and tachy-brady	DOI:10.1093/eurheartj/ehaa1108
LMNA	Sinus bradycardia, conduction defects, cardiomyopathy LGMD1B	DOI:10.1371/journal.pone.0155421
ZFHX3	Sick sinus syndrome and atrial fibrillation	DOI:10.1093/eurheartj/ehaa1108
RYR1	RYR1-related myopathy	DOI:10.1186/s13052-019-0756-1
KCNQ1	LQTS1, SQT2, AF	DOI: 10.4022/jafib151
MYH6	Aortic coarctation, BAV	DOI: 10.1093/eurheartj/ehaa1108