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Case report

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

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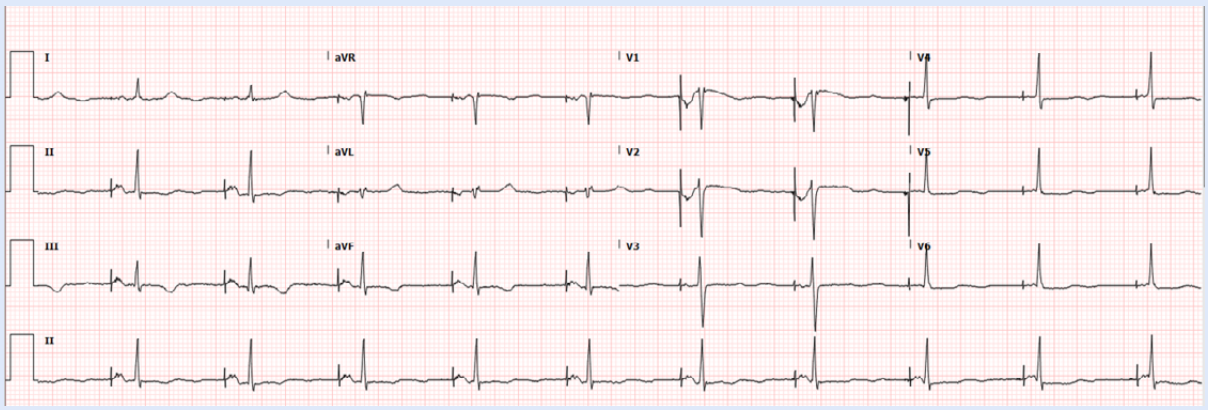


**Figure 1S.** ECG post-pacemaker implantation showing atrial-paced rhythm, ventricular-sensed beats, with a narrow QRS complex.

Adapted from: Rojanasopondist P, Nesheiwat L, Piombo S, Porter GA Jr, Ren M, Phoon CKL. Genetic Basis of Left Ventricular Noncompaction. Circ Genom Precis Med. 2022;15(3):e003517. doi:10.1161/CIRCGEN.121.003517.

**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 |
| KCNG2 | 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 |