

Compound heterozygous or homozygous SCN5A mutation and atrial standstill in paediatric population: two cases report.

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Atrial standstill is a rare arrhythmogenic condition characterized by the absence of electrical and mechanical atrial activity. It occurs as idiopathic, familial or sporadic. Recently, studies have linked the familial forms of atrial standstill to the SCN5A gene, which encodes the alpha subunit of the Nav1.5 cardiac sodium channel protein.

We describe two pediatric cases of sinus node dysfunction and compound heterozygous SCN5A mutations.

Case1- A 3 years old boy with history of syncope was implanted with a dual chamber-epicardial pacemaker because of a sinus node dysfunction with severe bradycardia associated with pauses > 3 s. No family history of cardiological disease or sudden cardiac death. The parents are cousins. After implantation, pacemaker parameters showed atrial lead dysfunction and elevated ventricular capture. PM was programmed in VVI 40/min. During follow-up visits, PM interrogation showed progressive increasing ventricular stimulation up to 90%, no ventricular arrhythmias and a stable ventricular capture at 1.3 V. PM was then reprogrammed in VVIR 60-130/min. Genetic tests were positive for homozygote SCN5A mutation (c.4380C>A: p.Phe1460Leu). The family genetic test showed the presence of the same heterozygote mutation in both parents.

Case2- A 3 years-old boy hospitalized for a left sylvian ischemic stroke with right hemiparesis and left facial paralysis. The ECG showed a sinus node dysfunction characterized by a bradycardic junctional rhythm 40/min with pauses > 2.5 s and a Brugada pattern in the precordial lead V1. TE echocardiography showed a dilated left atrium with a large thrombus in the left appendage. Anticoagulation therapy was contraindicated a cause of the elevated risk of bleeding. The implantation of an endocardial PM DDD showed the presence of an atrial standstill. The PM was then programmed in VVIR 60-130/min. In the suspicion of a Brugada syndrome, a therapy with hydroquinidine was started. The genetic test found a double mutation of the gene SCN5A: c.559A>C (pThr187Pro), never described before, and c.4747C>T (p.Arg1583Cys). The child died of neurological complication.

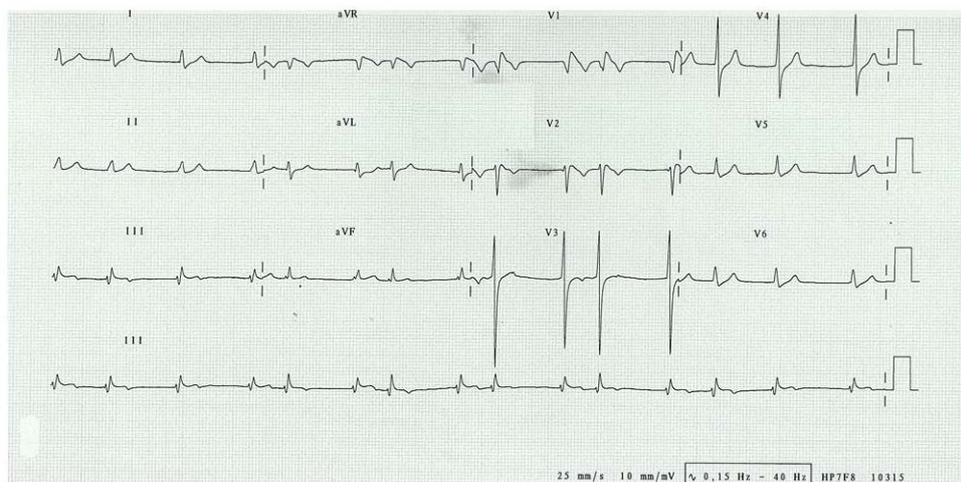


Figure 1: Admission ECG of patient 2.