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The Mutations of Genes Associated with Sick Sinus Syndrome in Children

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Sick sinus syndrome (SSS) is life-threatening cardiac arrhythmia, which sometimes can manifest itself with syncope and needs a pacemaker implantation even in children. Sometimes, SSS is accompanied by structural heart diseases such as septal defects, cardiomyopathies, but often the heart is structurally normal. Some mutations of genes associated with high risk of SSS are known. At the same time, the etiology of the syndrome is unidentified and may be genetic caused in 50% of patients with SSS. There are no studies on the prevalence of SSS-associated mutations in children.

The aim of our work is to identify and study the types of mutations of genes associated with high risk of SSS in children.

Methods. We included in the study 19 children (31.5% boys) with severe SSS, from the database of the Russian Pediatric Arrhythmia Center. Personal and family history, physical examination, including ECG, stress test, Holter monitoring, ECHO and other tests, and whole exome sequencing were made. The average age was 8.1±4.5 (from 2 to 17).

Results. In 47% (9 pts) there was the combination of SSS and structural heart disease. 13 children (68%) had syncope, 9 pacemakers were implanted. 7 children (37%) had the mutations of genes associated with SSS: 3 – SCN5A, 2 - HCN4, 1 - TRPM4, 1 - PRDM16. Family history of cardiac diseases was positive in 5 pts; 2 pts had family members with implanted pacemakers.

Conclusion. We found the mutations of genes associated with SSS in 37% of children. Further research and larger patient samples are required to study the prevalence of genetic types of SSS and show the correlation of the genotype with the clinical prognosis.
In addition, our work will enable practitioners to identify children from families with family forms of SSS and sudden cardiac death. Further research can help us determine the criteria for selecting children for genetic testing.