To study the prevalence of cardiac channelopathies in children aged 5-18 years undergoing treatment for epilepsy

Jaskaran Singh Gujral
Nitish Naik
Gautam Sharma
Shefali Gulati

Introduction: Cardiac channelopathies, most commonly Long QT syndrome may be misdiagnosed as refractory epilepsy, when in reality, these events represent convulsive syncope. In addition, both cardiac channelopathies and epilepsy may coexist. The objective of the study was to observe the prevalence of cardiac channelopathies in children aged 5-18 years undergoing treatment for epilepsy.

Methods: All patients aged 5-18 years undergoing treatment for epilepsy under the division of Pediatric Neurology at a tertiary care centre in North India and with non-contributory neuroimaging/electroencephalogram were included in the study. All patients underwent a focused history, physical examination and a 12-lead electrocardiogram. Patients with ECG findings suggestive of Long QT syndrome (QTc ≥ 480 ms – prepubertal, ≥ 470 ms – post pubertal males, ≥ 480 ms – post pubertal females, as per Bazett formula and with modified Schwartz score > 3.5), Brugada syndrome (type 1 pattern), short QT syndrome (QTc < 360 ms at heart rate < 100 bpm)/ Arrythmogenic Right Ventricular cardiomyopathy (2010 task force criteria), underwent blood sampling for genetic analysis by whole exome sequencing.

Result: A total of 1000 patients were enrolled in the study. 5 cases suggestive of Long QT syndrome were identified. All the identified patients were boys with a mean age of 9.2+2.8 years at the time of diagnosis. The mean lag period between symptom onset and final diagnosis was 4 ± 3.1 years. The mean QTc was 500 + 23 ms and the mean Schwartz score was 4.8 + 1.3. Of them, 3/5 had a history of syncope, which was exertional in 2 patients. The seizure semiology was generalized tonic clonic seizures in most of the cases (3/5). One of the patients also had congenital bilateral sensorineural hearing loss. T wave alternans and exercise induced polymorphic ventricular tachycardia was observed in one patient each. All patients were started on oral propranolol therapy (weight based) and one of the patients was advised permanent pacemaker implantation. All the patients were asymptomatic during a mean follow up of 8 months. Antiepileptics were tapered off in all the patients except the one with left focal seizures. The results of whole exome sequencing are available for 3 of the patients out of which a de-novo heterozygous variant with damaging effect (c.950A>G) was found in exon 7 of KCNQ1 gene in one of the patients, suggestive of Long QT syndrome 1.

Conclusion: The 12-lead electrocardiogram is a cost effective investigation which should be sought in all patients presenting with seizures of unknown causation especially in the pediatric population.