Familial long QT syndrome: a case report

Josip Ereiz[1]*,
Katica Cvitkušić Lukenda[1,2],
Domagoj Vučić[1],
Ivan Bitunjac[1],
Josip Silović[1],
Blaženka Miškić[1,2]

[1]General Hospital "Dr. Josip Benčević", Slavonski Brod, Croatia
[2]Josip Juraj Strossmayer University of Osijek, Faculty of Dental Medicine and Health Osijek, Osijek, Croatia

KEYWORDS: genetic testing, long QT syndrome, sudden cardiac death.
CITATION: Cardiol Croat. 2024;19(3-4):126. | https://doi.org/10.15836/ccar2024.126

***ADDRESS FOR CORRESPONDENCE:** Josip Ereiz, Opća bolnica "Dr. Josip Benčević", Andrije Štampara 42, HR-35000 Slavonski Brod, Croatia. / Phone +385-99-534-4456 / E-mail: ereiz12345@gmail.com
ORCID: Josip Ereiz, https://orcid.org/0009-0002-2139-016X • Katica Cvitkušić Lukenda, https://orcid.org/0000-0001-6188-0708 Domagoj Vučić, https://orcid.org/0000-0003-3169-3658 • Ivan Bitunjac, https://orcid.org/0000-0002-4396-6628 Josip Silović, https://orcid.org/0009-0002-9918-7575 • Blaženka Miškić, https://orcid.org/0000-0001-6568-3306

Introduction: Long QT syndrome (LQTS) has a prevalence of 1:2000, with mutations in the KCNQ1, KCNH2, and SCN5A genes predominating in 90% of cases. The diagnosis should be suspected when the Schwartz score is ≥ 3.5 and can be confirmed by genetic testing.[1-3]

Case report: 50-year-old Caucasian woman was referred for a first cardiology evaluation (December 2018) after her 18-year-old daughter was diagnosed with a prolonged QT interval during a general medical examination. The patient, a healthy adult with a negative family history of sudden cardiac death (SCD), is a non-smoker with good exercise capacity and no history of syncope. In 12-lead resting electrocardiography (ECG) the QTc interval was 498ms (**Figure 1**). Physical examination and laboratory results were unremarkable, and she was not on QTc-prolonging therapy. Echocardiography showed no structural heart disease. On 24-hour Holter ECG, the predominant rhythm was sinus rhythm, with no premature ventricular contractions and an insignificant number of premature atrial contractions recorded. The average QTc was 482ms and the longest was 534ms. She underwent exercise testing, and in the fourth minute of recovery, the QTc was 511ms. Malignant arrhythmias were not recorded during the test. When we summarize the basic criteria for QTc duration and exercise testing, her Schwartz score was 4, which meets the diagnostic criteria for LQTS. The next step was a genetic test. The results obtained showed a heterozygous missense variant c.251G > A, p. (Arg174His) in KCNQ1, resulting in the final diagnosis of LQT1. Since the chance of inheriting LQTS is 50% (autosomal dominant), the patient's daughters (18 and 15 years old) also underwent genetic testing. The results were positive in both patients. They were asymptomatic with no recorded arrhythmias, and beta-blockers were introduced at the hospital.

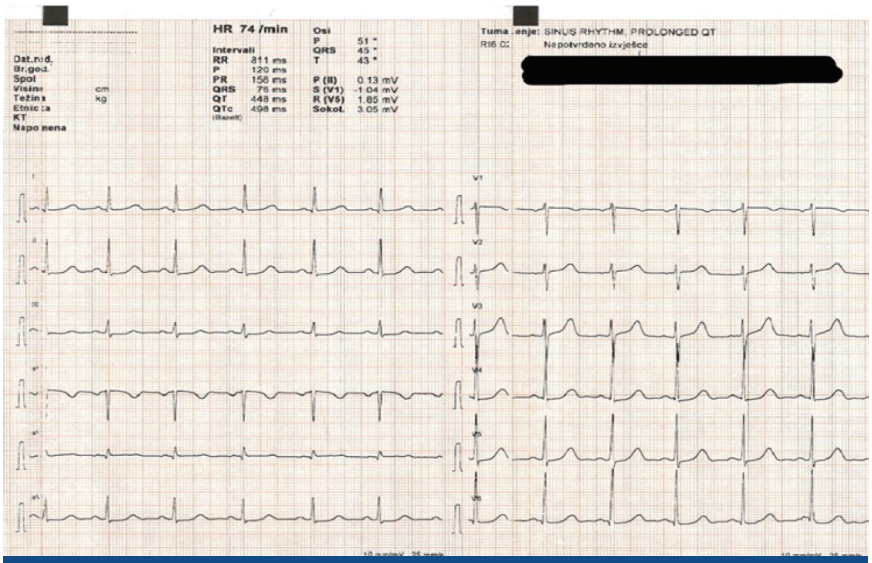

FIGURE 1. Maternal electrocardiographic record, QTc 498 ms (Bazett formula).

Conclusion: Because the patients were asymptomatic and had no family history of malignant arrhythmias or SCD, drug therapy with beta-blockers was recommended. Patients were educated about avoiding strenuous physical activity and about the list of medications that may prolong the QT interval. Acquisition of an automated external defibrillator for home use and regular cardiology follow-up were recommended.

RECEIVED:
October 19, 2023
ACCEPTED:
October 27, 2023

LITERATURE

1. Krahn AD, Laksman Z, Sy RW, Postema PG, Ackerman MJ, Wilde AAM, et al. Congenital Long QT Syndrome. JACC Clin Electrophysiol. 2022 May;8(5):687-706. https://doi.org/10.1016/j.jacep.2022.02.017
2. Schwartz PJ, Crotti L. QTc behavior during exercise and genetic testing for the long-QT syndrome. Circulation. 2011 Nov 15;124(20):2181-4. https://doi.org/10.1161/CIRCULATIONAHA.111.062182
3. Sy RW, van der Werf C, Chattha IS, Chockalingam P, Adler A, Healey JS, et al. Derivation and validation of a simple exercise-based algorithm for prediction of genetic testing in relatives of LQTS probands. Circulation. 2011 Nov 15;124(20):2187-94. https://doi.org/10.1161/CIRCULATIONAHA.111.028258