INCIDENCE OF SIGNIFICANT ARRHYTHMIC EVENTS IN PATIENTS WITH LONG QT SYNDROME

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INTRODUCTION

- **Congenital long QT syndrome (LQTS)** is a hereditary disease characterized by prolonged QTc interval and risk of ventricular tachycardia (VT), which may lead to **syncope, cardiac arrest, or sudden cardiac death (SCD) in young people**.

PURPOSE

- To report the experience and the incidence of significant arrythmias in patients (pts) with congenital LQTS in an inherited primary arrhythmic syndrome (IPAS) center from a Portuguese tertiary hospital.

METHODS

- **Prospective single-center study** of consecutive patients with LQTS, recruited between 1997 and 2021
- Clinical data and 12-lead ECG were registered
- Genetic screening was performed using DNA targeted sequencing for a panel which included KCNQ1, KCNH2, SCN5A and KCNE1
- **Outcome measures:** significant arrhythmic events such as SCD, symptomatic and asymptomatic arrhythmic events

RESULTS

15 patients with LQTS

Median follow-up of 4 [3,5-12,7] years

Baseline characteristics:
- 9 (67%) index-cases
- 9 (60%) females
- Mean age at diagnosis was 44±16 years-old (yo)
- 1 associated congenital abnormality - Andersen-Tawil syndrome (ATS)

Schwartz score distribution – probability of clinical diagnosis	**N**
≥3.5 - HIGH	12
1,5-3 – INTERMEDIATE	1
≤1– LOW	2

All on beta-blocker (BB)

6 (40%) received an ICD

- 2 due to aborted SCD at diagnosis
- 4 due to syncope recurrence, documented VT and/or QTc>500ms despite BB

6 (40%) had significant arrhythmic events (1 SCD) - 0.06%/year

Clinical features at diagnosis:
- 11 (73%) QT≥480 ms
- 3 (20%) notched T wave in 3 leads
- 9 (60%) were symptomatic: 7 (47%) previous syncope (only 2 with stress), 5 (33%) torsade de pointes
- 9 (60%) family history of unexplained SCD <30 yo and/or definite LQTS

Genetic screening (in 14 patients):
- 11 (79%) KCNH2
- 1 (7%) KCNJ2 (LQTS 7) (patient with ATS)
- 2 (14%) no mutation identified

Figure – Summary of diagnostic features, management and follow-up of 15 LQTS patients

Type of LQTS (mutated gene)	Schwarz score for clinical diagnosis	Torsade de pointes/aborted SCD at diagnosis	Follow-up (in years)	Events during follow-up	ICD implantation
LQT2 (KCNH2)	3.5	No	3.9	No	No
LQT2 (KCNH2)	7	Yes	7.2	No	Yes
LQT2 (KCNH2)	4	No	21.1	Unexplained syncope	Yes
LQT2 (KCNH2)	4	No	18.0	No	No
LQT2 (KCNH2)	4.5	No	3.7	No	No
LQT2 (KCNH2)	1	No	3.5	No	Yes
LQT2 (KCNH2)	5.5	No	4.0	No	No
LQT2 (KCNH2)	4.5	No	12.7	Sudden cardiac death	No
LQT2 (KCNH2)	1.5	No	1.7	No	No
LQT2 (KCNH2)	5.5	No	3.4	Unexplained syncope + documented NSVT	Yes
LQT2 (KCNH2)	8.5	Yes	11.2	Appropriate ICD therapies	Yes
LQTS7 (KCNJ2)	0	No	3.0	Documented NSVT	Programmed
No mutations found	5	Yes (with a metabolic and iatrogenic component)	4.0	No	No
No mutations found	6	Yes	6.0	Appropriate ICD therapies	Yes
No genetic screening performed	7	Yes (initial diagnosis in 1997)	23.2	No	No

Legend: ICD = Implantable cardia defibrillator, NSVT = non-sustained ventricular tachycardia, SCD = Sudden Cardiac Death

CONCLUSIONS

- The **KCNH2** was **the most prevalent mutation in this Portuguese cohort**.
- Care of congenital LQTS pts in an IPAS center was associated with a **low incidence of significant clinical events (0.06%/year).**