



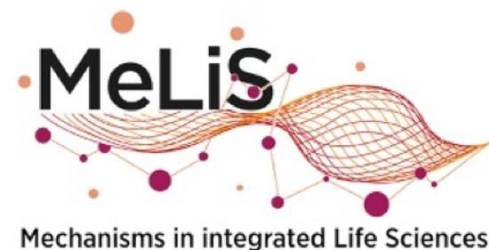
**SESSION SPORT  
& CARDIOPATHIES**  
*LE POINT SUR LES  
RECOMMANDATIONS*

# CANALOPATHIES CARDIAQUES

**Antoine DELINIERE, MD, PhD**  
*Lyon*

**HCL**  
HOSPICES CIVILS  
DE LYON

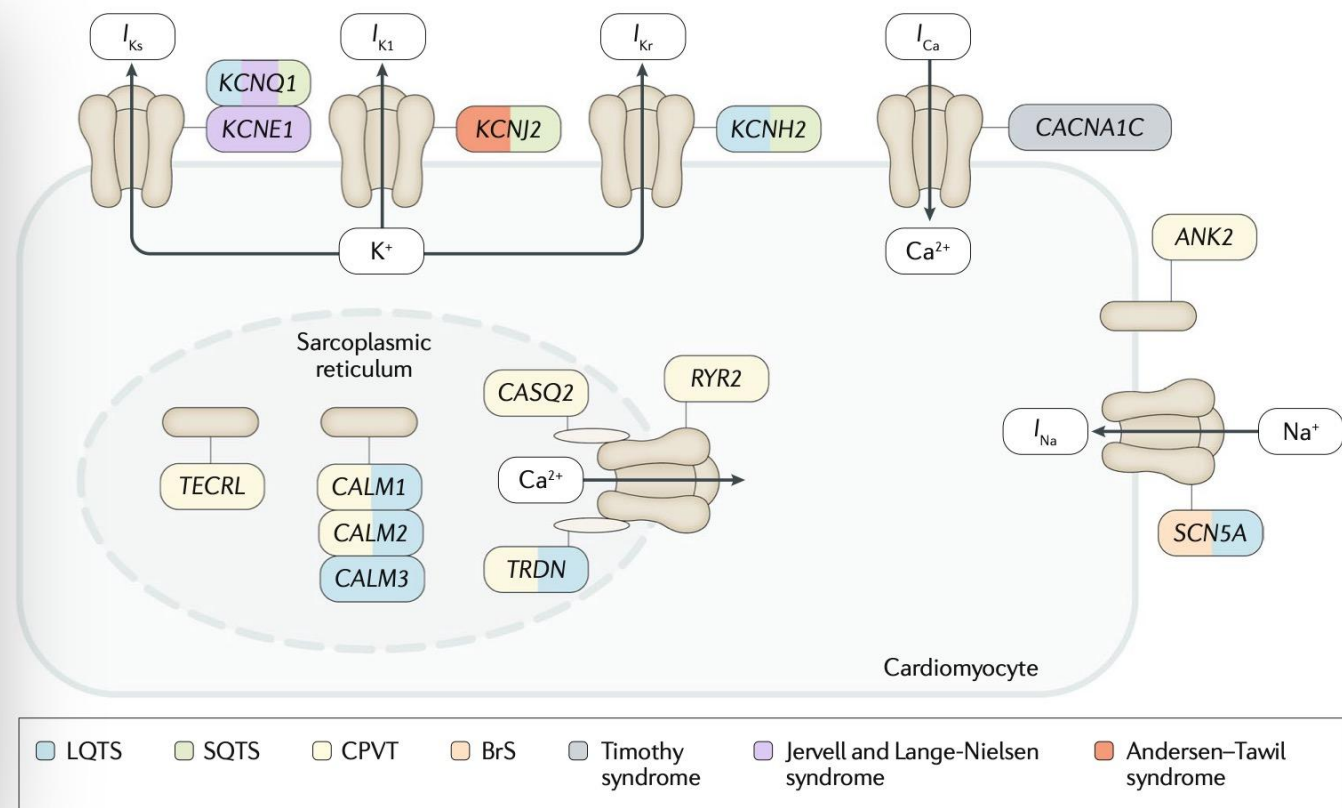
**L'INSTITUT DE  
CARDIOLOGIE**



## GENERALITES

- Syndrome du **QT long congénital (LQTS)**
- Syndrome de **Brugada (BrS)**
- **Tachycardie ventriculaire catécholergique (CPVT)**
- Syndrome du **QT court (SQTS)**
- Syndrome d'**Andersen-Tawil (ATS)**
- Syndrome de **Timothy (TS)**
- Syndrome de **Jervell et Lange-Nielsen (JLNS)**
- **Calmodulinopathies (CALM)**
- Trouble de la **conduction cardiaque familial progressif (PCCD)**
- Syndrome de **repolarisation précoce (ERS)**
- ... *et bien d'autres*

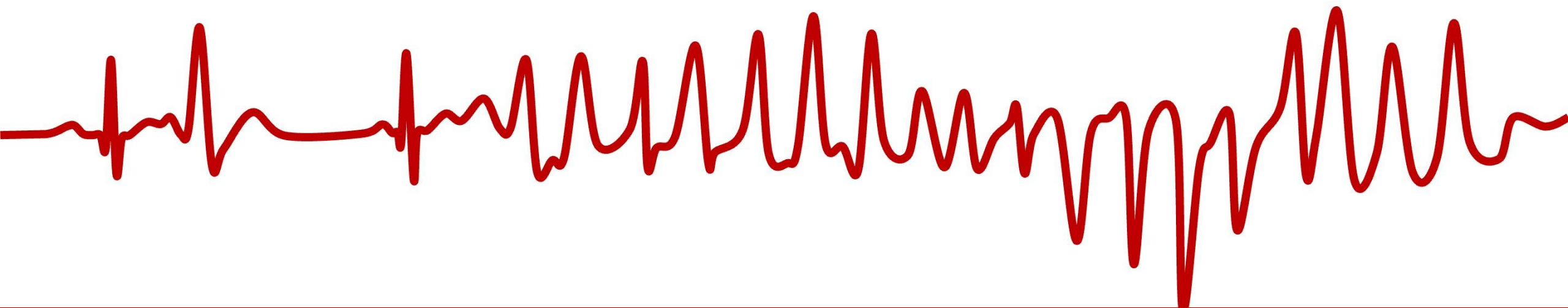
**Au total, 19 familles  
de maladies rythmiques héréditaires**



Schwartz PJ et al. Nature Reviews Disease Primers (2020)

## ▪ GENERALITES

- Prolongation de la durée du potentiel d'action ventriculaire : intervalle QTc long
- Incidence: **1 / 2 000** naissances
- Torsades de pointes → fibrillation ventriculaire
- 50% de risque de **syncope** ou **mort subite** <40 ans sans prise en charge
- Phénotype et prise en charge **génotype-spécifiques**

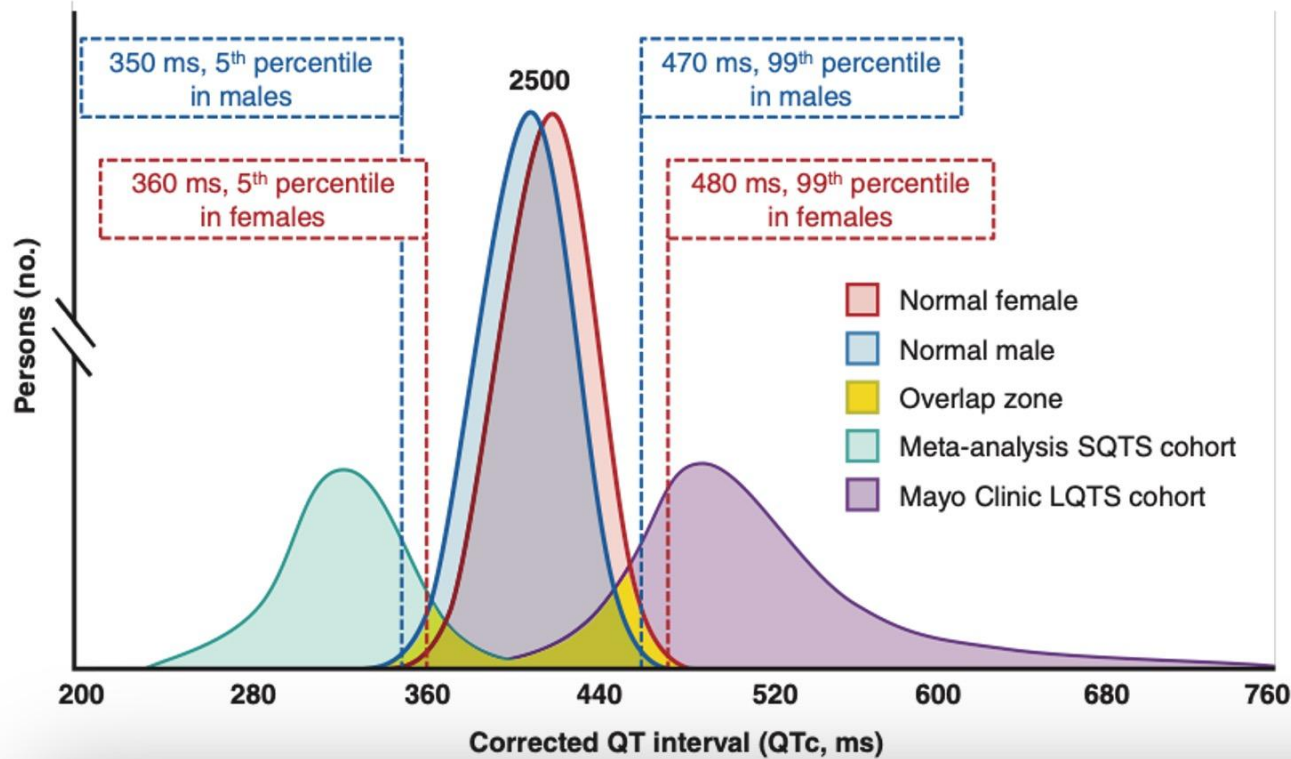


**GENERALITES**

IN DEPTH

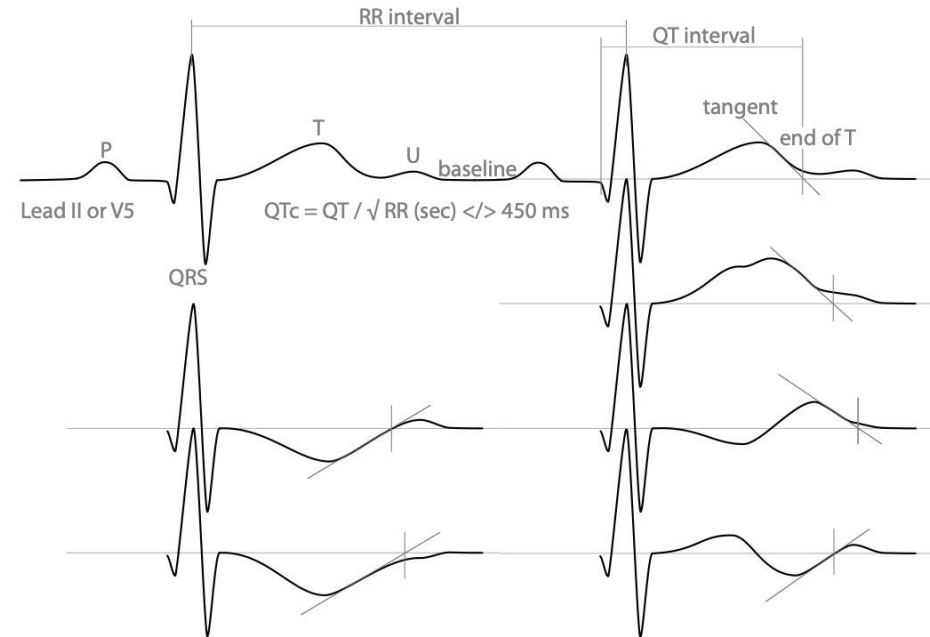
**Sex-Related Differences in Cardiac Channelopathies**

Implications for Clinical Practice



**Accurate electrocardiographic assessment of the QT interval: Teach the tangent**

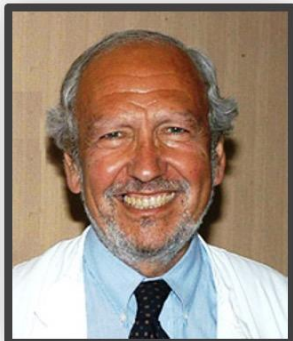
Pieter G. Postema, MD, Jonas S.S.G. De Jong, MD, Ivo A.C. Van der Bilt, MD, Arthur A.M. Wilde, MD, PhD



**Intervalle QTc allongé (99<sup>ème</sup> percentile)**

- Femmes  $\geq 480 \text{ ms}$  (borderline : 460 - 480 ms)
- Hommes  $\geq 470 \text{ ms}$  (borderline : 450 - 470 ms)

GENERALITES



MY APPROACH to the long QT syndrome (LQTS)\*

« When I suspect the possible presence of LQTS, I not only measure the QT interval [...],  
**I also look at it.** »

Peter J. Schwartz

**QTc allongé**



**Morphologie anormale de la repolarisation**

ORIGINAL RESEARCH ARTICLE

Determination and Interpretation of the QT Interval

Comprehensive Analysis of a Large Cohort of Long QT Syndrome Patients and Controls

	Controls n=592	LQT1 n=301	LQT2 n=370	LQT3 n=138	P Value
QTc <sub>Tangent</sub>					
Bazett, ms (±SD)	399 (±26)	447 (±40)	457 (±42)	435 (±44)	<0.001

**Sensibilité de seulement 19%**

Vink AS et al. Circulation. 2018

Spectrum of ST-T-Wave Patterns and Repolarization Parameters in Congenital Long-QT Syndrome  
 ECG Findings Identify Genotypes

Li Zhang, MD; Katherine W. Timothy, BS; G. Michael Vincent, MD; Michael H. Lehmann, MD; Jolene Fox, RN; Lisa C. Giuli, BS; Jiaying Shen, BS; Igor Splawski, PhD; Silvia G. Priori, MD; Steven J. Compton, MD; Frank Yanowitz, MD; Jesaia Benhorin, MD; Arthur J. Moss, MD; Peter J. Schwartz, MD; Jennifer L. Robinson, MS; Qing Wang, PhD; Wojciech Zareba, MD, PhD; Mark T. Keating, MD; Jeffrey A. Towbin, MD; Carlo Napolitano, MD; Aharon Medina, MD

Electrocardiographic Features in Andersen-Tawil Syndrome  
 Patients With *KCNJ2* Mutations  
 Characteristic T-U-Wave Patterns Predict the *KCNJ2* Genotype

Li Zhang, MD; D. Woodrow Benson, MD, PhD; Martin Tristani-Firouzi, MD; Louis J. Ptacek, MD; Rabi Tawil, MD; Peter J. Schwartz, MD; Alfred L. George, MD; Minoru Horie, MD, PhD; Gregor Andelfinger, MD; Gregory L. Snow, PhD; Ying-Hui Fu, PhD; Michael J. Ackerman, MD, PhD; G. Michael Vincent, MD

	LQT1 (n=179)	LQT2 (n=148)	LQT3 (n=60)
Overall: ECG patterns typical to genotype, %	87.7	87.8	65

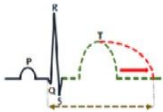
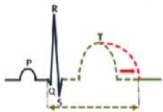
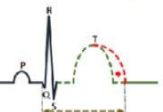
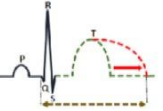
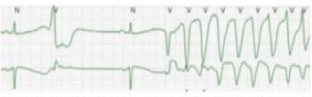

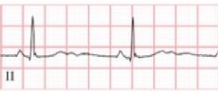
**Sensibilité**




**LQT1 61%**    **LQT2 62%**    **LQT3 33%**    **Andersen-Tawil 84%**

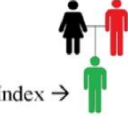
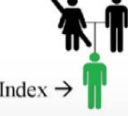
Zhang L et al. Circulation. 2000 & 2005

GENERALITES


Score de Schwartz

Electrocardiographic findings <i>(in absence of acquired causes)</i>		
<b>QTc duration</b> <i>(calculated by Bazett formula)</i>	≥ 480 msec 	3
	460-479 msec 	2
	450-459 msec <i>(men)</i> 	1
<b>QTc 4<sup>th</sup> minute of recovery from exercise test</b>	≥ 480 msec 	1
<b>Torsades de Pointes*</b> <i>(mutually exclusive)</i>		2
<b>T-wave alternans</b>		1
<b>Notched T wave in 3 leads</b>		1
<b>Low resting heart rate</b>	Below the 2 <sup>nd</sup> percentile for age	0.5

Clinical history		
<b>Syncope*</b> <i>(mutually exclusive)</i>	With stress 	2
	Without stress 	1
<b>Congenital deafness</b>		0.5

Family history <i>(the same family member cannot be counted twice for the rows below)</i>		
<b>Family members with definite LQTS</b>		1
<b>Unexplained SCD &lt;30 years among immediate family members</b>		0.5

- **LQTS certain : score ≥ 3,5**
- **Probabilité intermédiaire : score > 1 et < 3,5**
- **Faible probabilité : score ≤ 1**


 It is recommended that LQTS is diagnosed in the presence of a pathogenic mutation, irrespective of the QT duration.
 

I	C
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Schwartz PJ & Crotti L. *Circulation*. 2011  
 Wilde AAM et al. *Heart*. 2022  
 Zeppenfeld K et al. *European Heart Journal*. 2022

**CORRELATION GENOTYPE-PHENOTYPE**

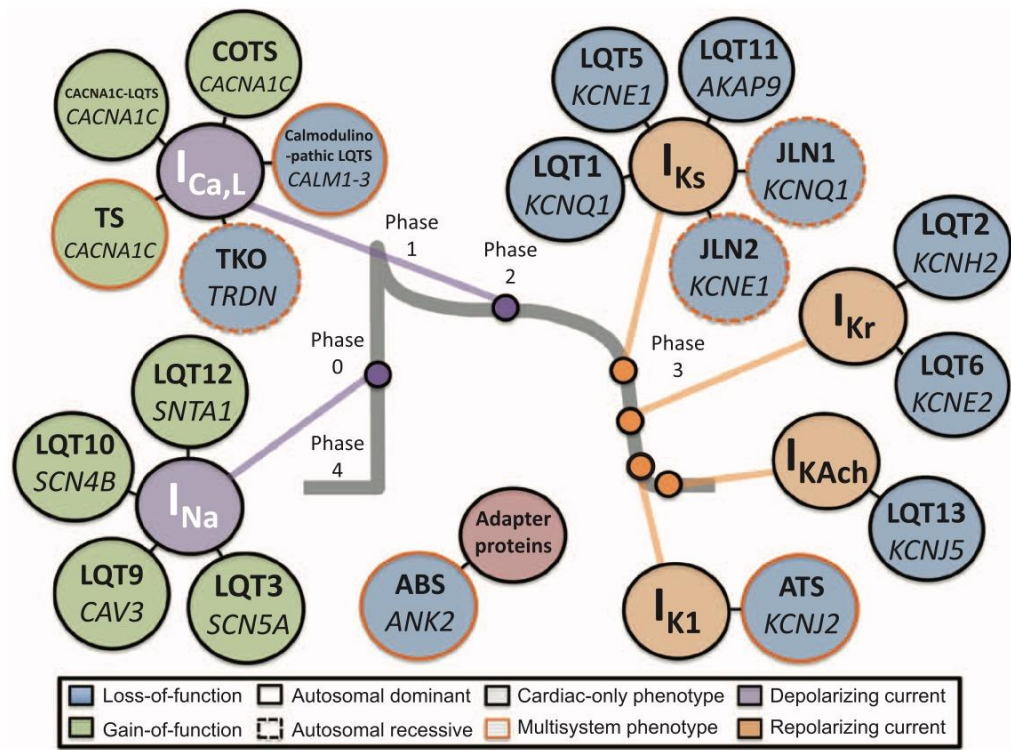
17 gènes impliqués



17 maladies différentes (LQT1-17)



Prise en charge gène-spécifique



Giudicessi JR & Ackerman MJ. Circ Arrhythm Electrophysiol (2016)

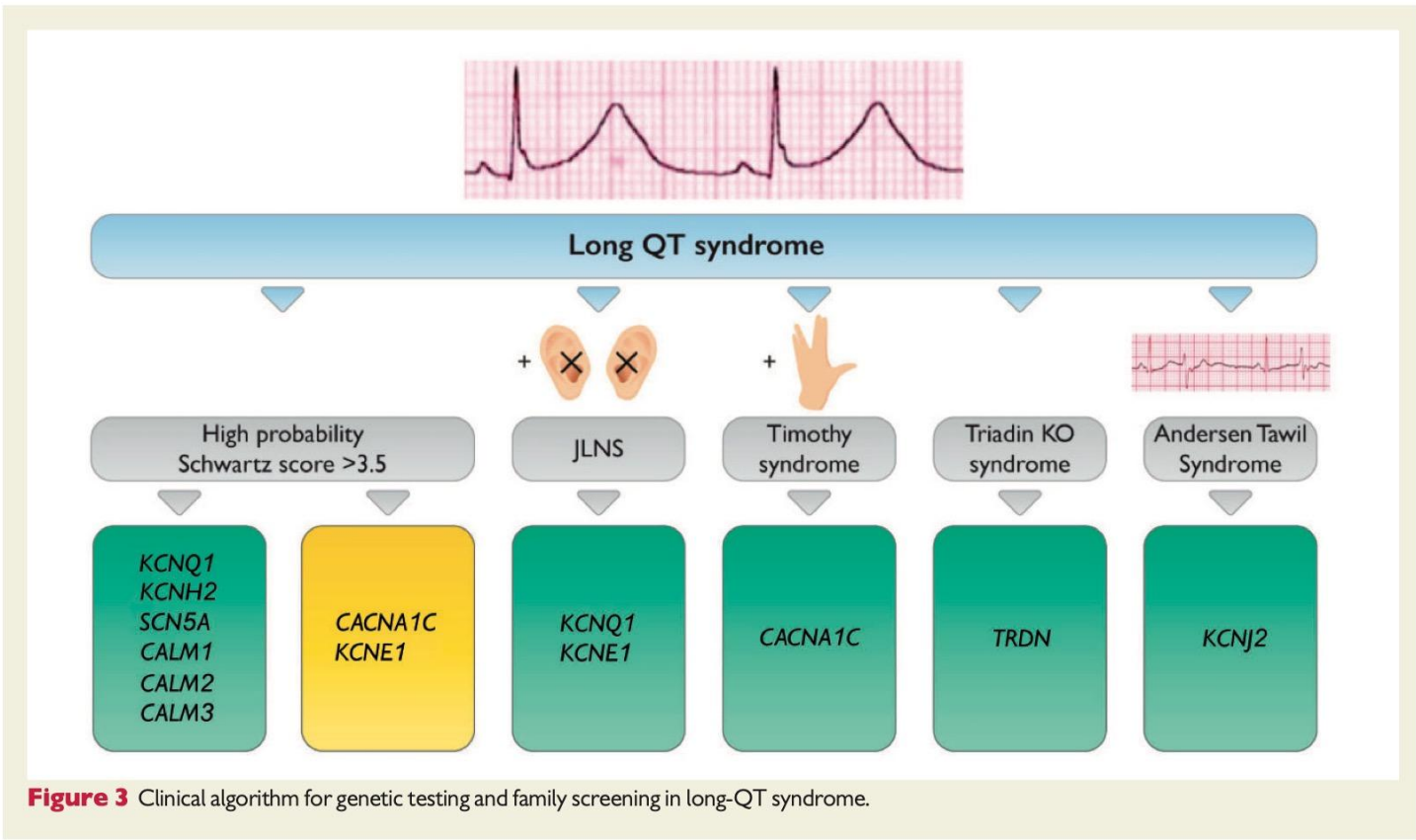


Figure 3 Clinical algorithm for genetic testing and family screening in long-QT syndrome.

Wilde AAM et al. Europace (2022)

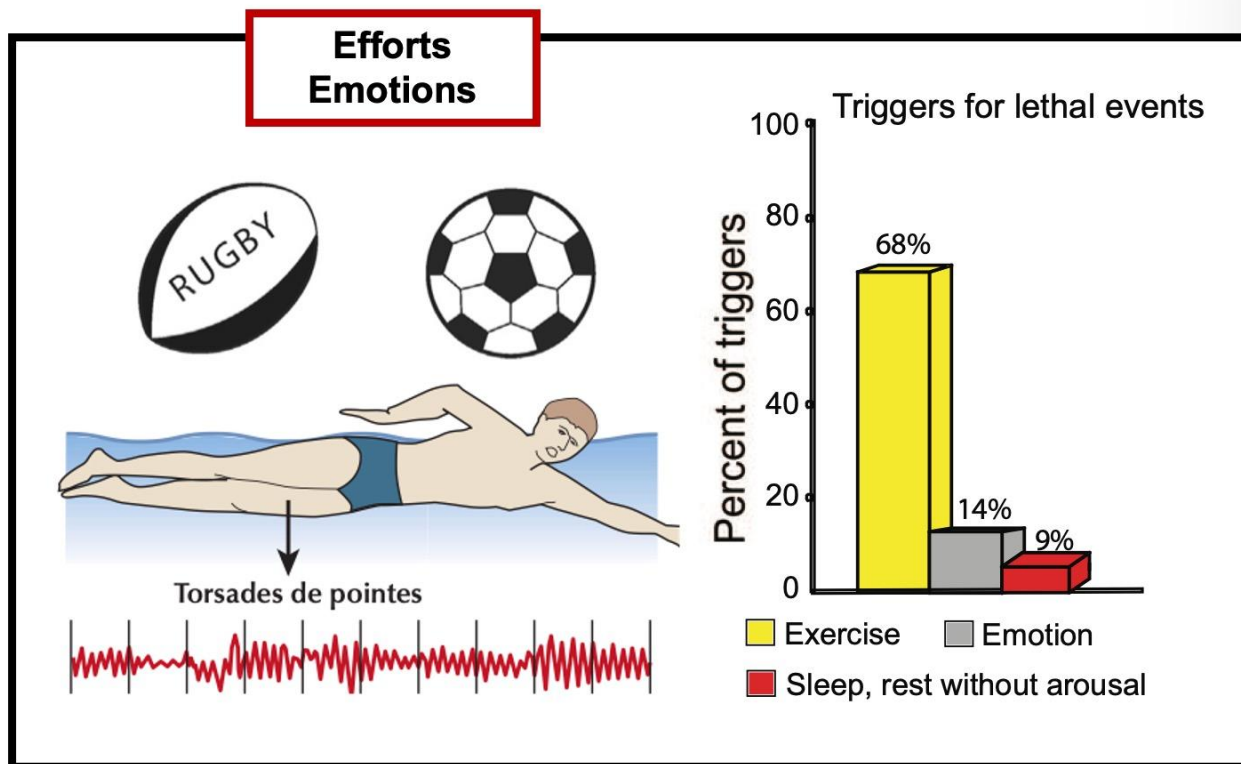
**LQTS type 1**

KCNQ1

**LQT1 = 35%**



Wilde AAM et al. Heart (2022)



Ackerman MJ. Nat Med (2004) – Ann Thompson  
 Ruan Y et al. Circ AE (2008)  
 Skinner JR et al. Heart Lung and Circulation (2019)  
 Schwartz PJ et al. Circulation (2001)

**RISQUE SUPERIEUR**

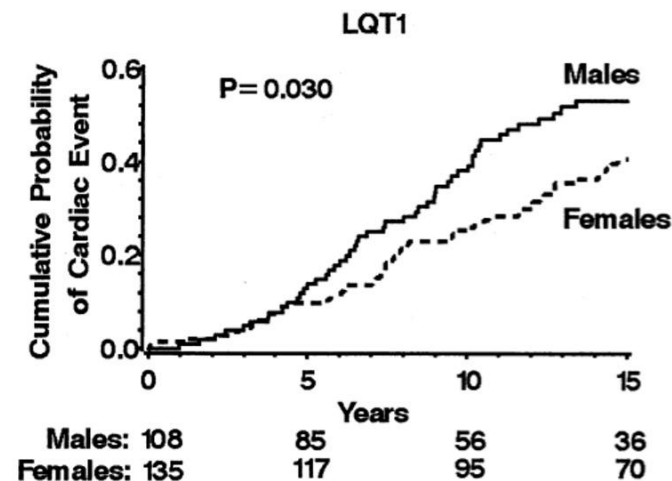
**Garçons ≤ 15 ans**

Age of onset arrhythmias

Childhood



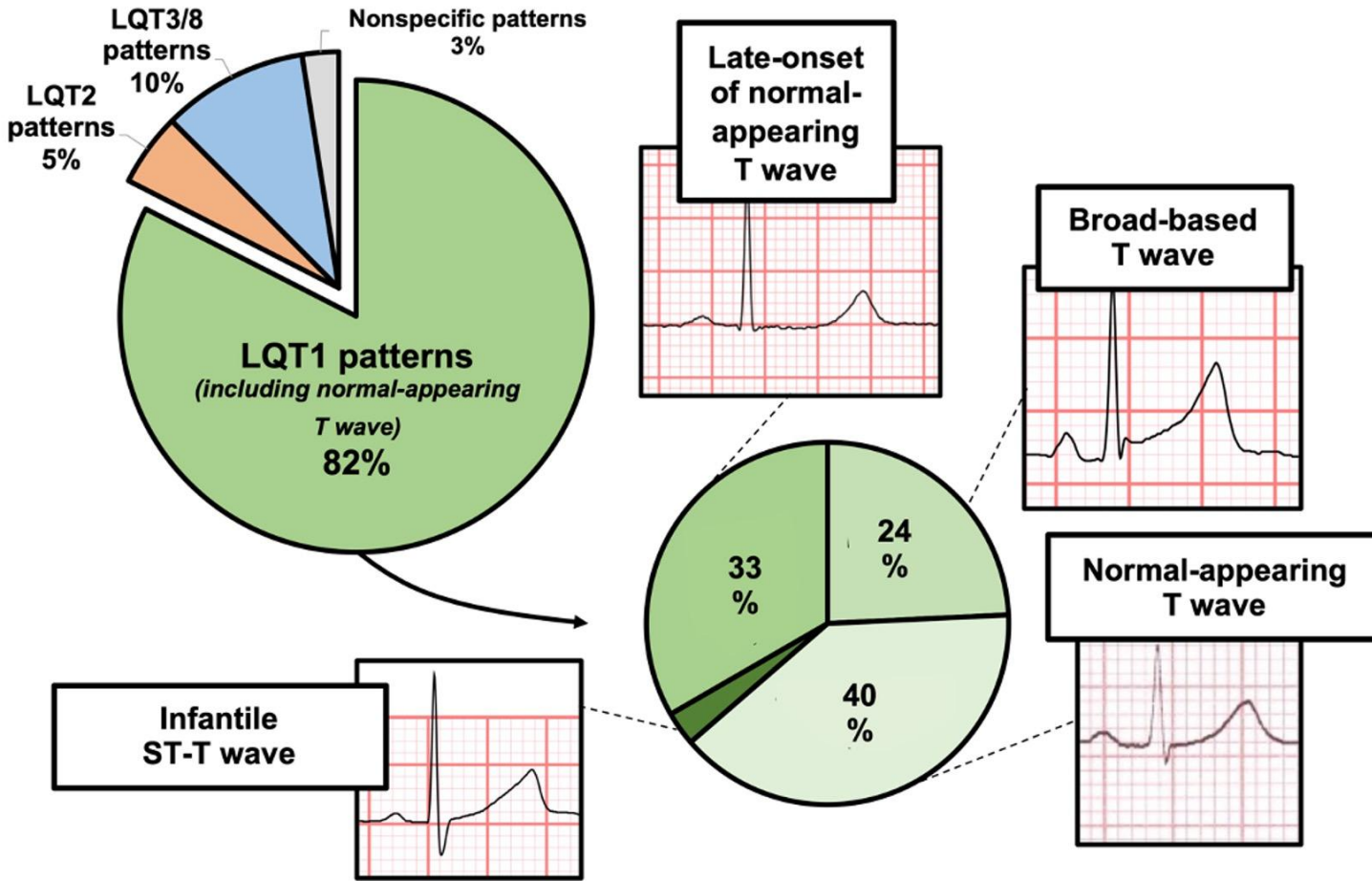
Gender most at risk



Wilde AAM et al. Heart (2022)  
 Zareba W et al. JACC (2003)

**LQTS type 1**

*KCNQ1*

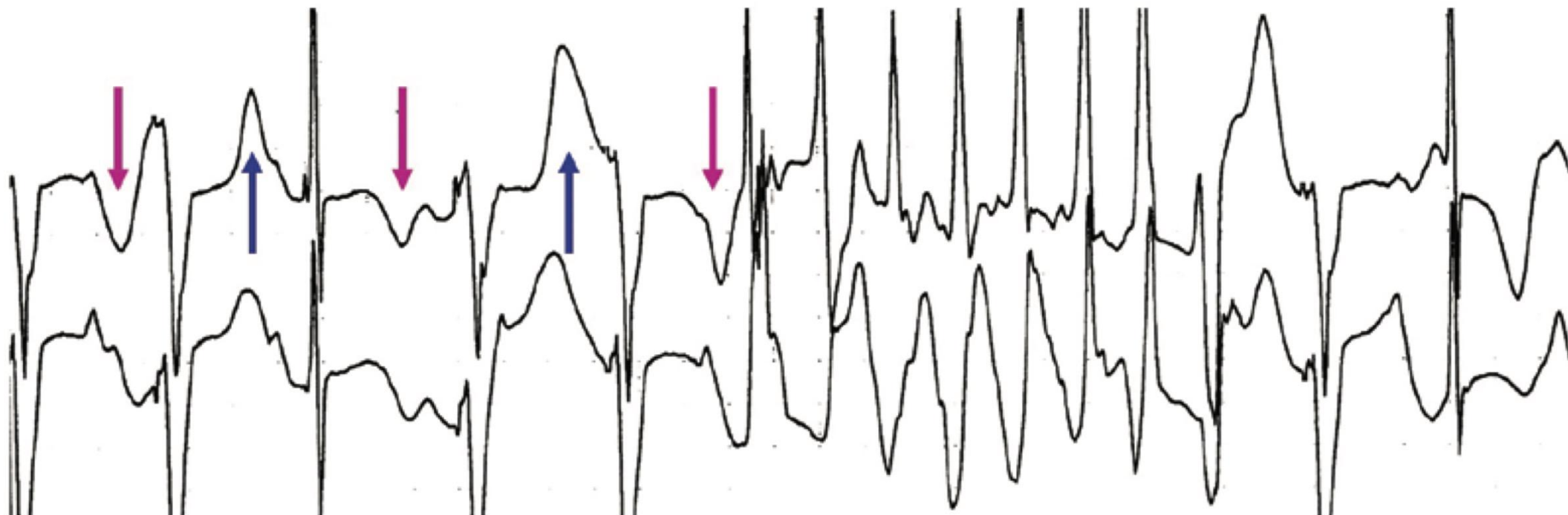


Delinière A et al. Arch Cardiovasc Dis (2024)

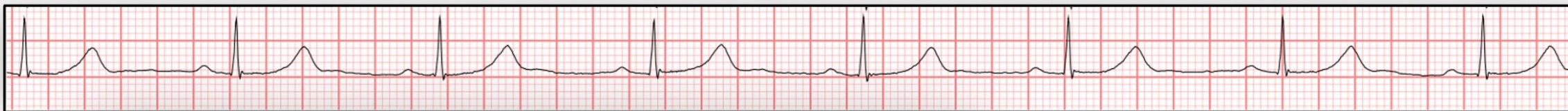
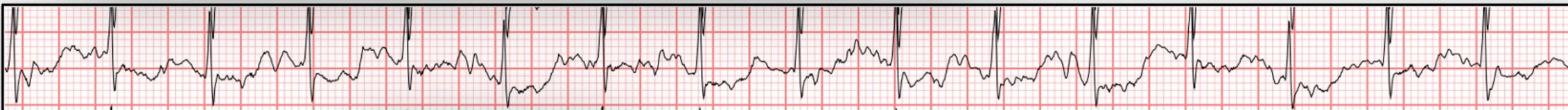
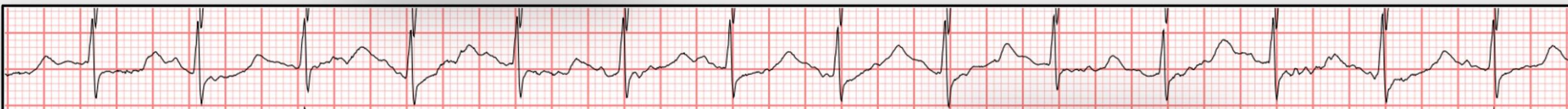
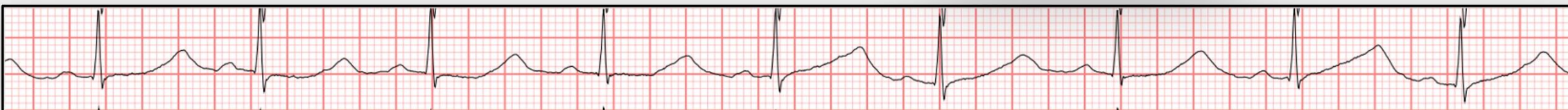
## LQTS type 1

KCNQ1

Les *Torsades de pointes* sont souvent **tachycardie-dépendentes** dans le LQT1



Viskin S et al. Circulation (2021)

**LQTS type 1***KCNQ1***Epreuve d'effort & LQT1****Repos****Pic****QTc allongé au pic (intérêt diagnostique limité & difficultés de mesure)****Récupération @ 1 min****QTc allongé +++****Récupération @ 4 min (7 min chez les enfants & adolescents)****QTc allongé +++**

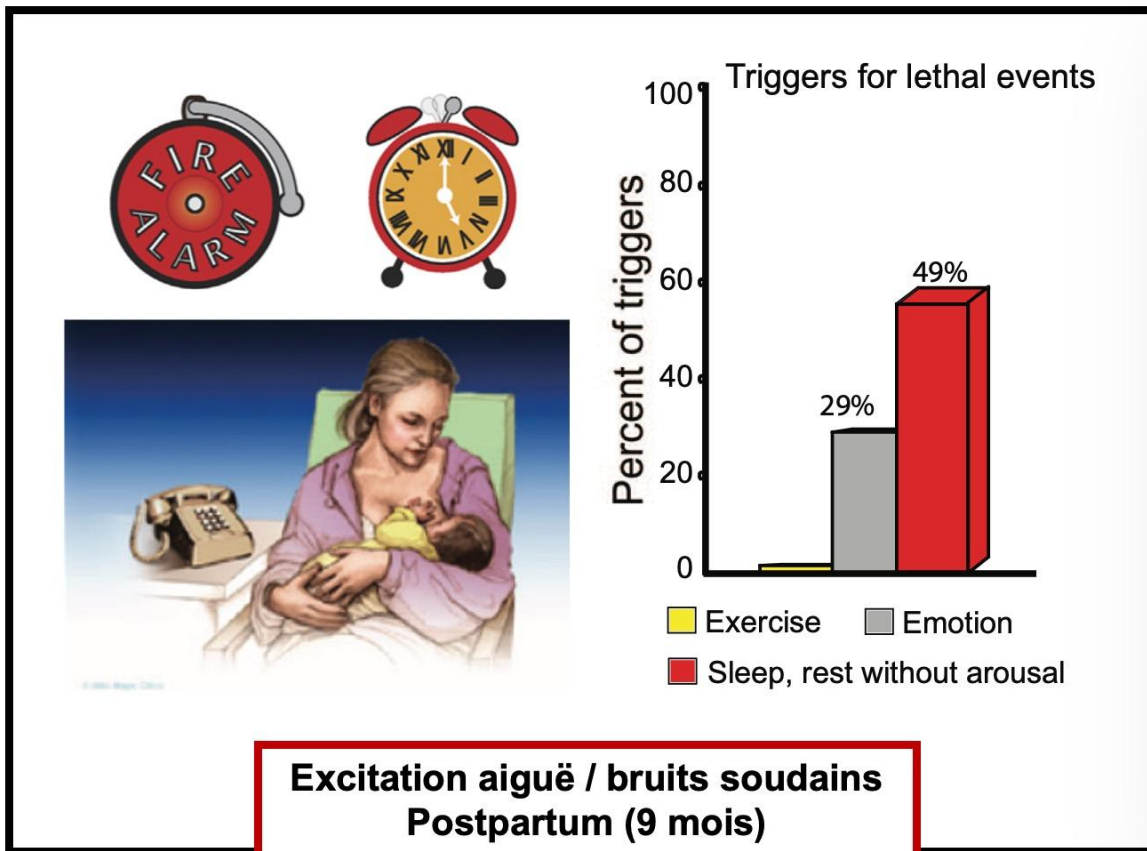
**LQTS type 2**

*KCNH2*

**LQT2 = 30%**



Wilde AAM et al. Heart (2022)

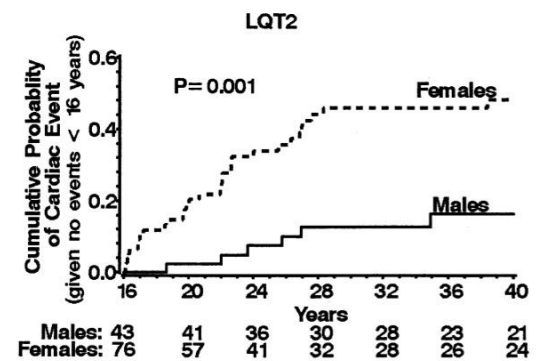
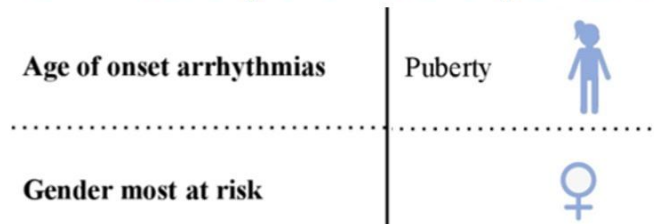


**Excitation aiguë / bruits soudains**  
**Postpartum (9 mois)**  
**Hypokaliémie**  
**Repos**  
**Fièvre**

Tester DJ & Ackerman MJ. Circulation (2011)  
 Ruan Y et al. Circ AE (2008)  
 Skinner JR et al. Heart Lung and Circulation (2019)  
 Schwartz PJ et al. Circulation (2001)

**RISQUE SUPERIEUR**

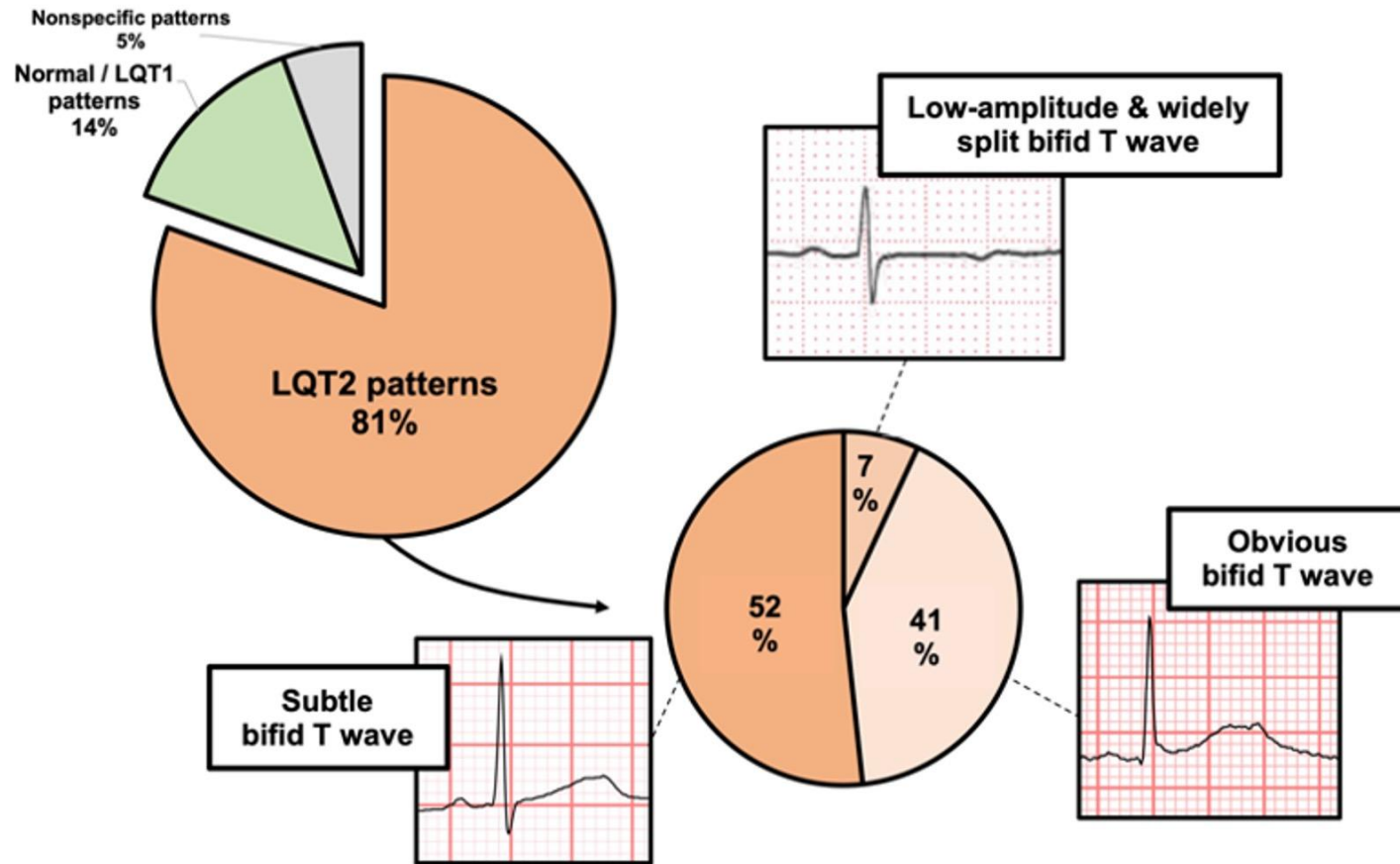
**Femmes à partir de la puberté**



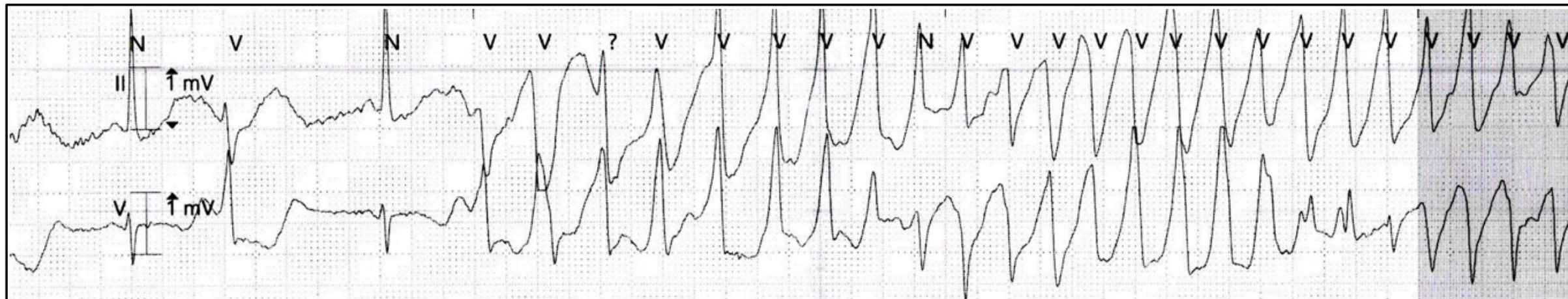
Wilde AAM et al. Heart (2022)  
 Zareba W et al. JACC (2003)

**LQTS type 2**

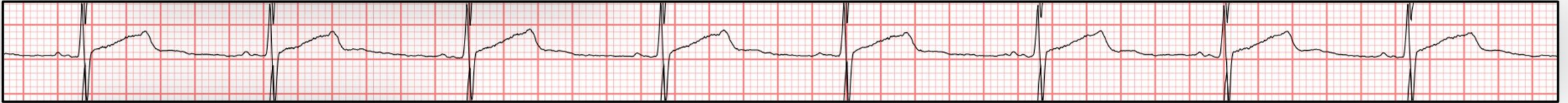
*KCNH2*



Delinière A et al. Arch Cardiovasc Dis (2024)

**LQTS type 2***KCNH2*

Les *Torsades de pointes* sont  
**pauses-dépendentes** dans le LQT**2**

**LQTS type 2***KCNH2***Epreuve d'effort & LQT2****Repos****Apparition des ondes T bifides au lever****Pic****Raccourcissement adapté****Récupération @ 1 min****Peu modifié en général****Récupération @ 4 min (7 min chez les enfants & adolescents)****QTc souvent allongé & Ondes T bifides ("stunning")**

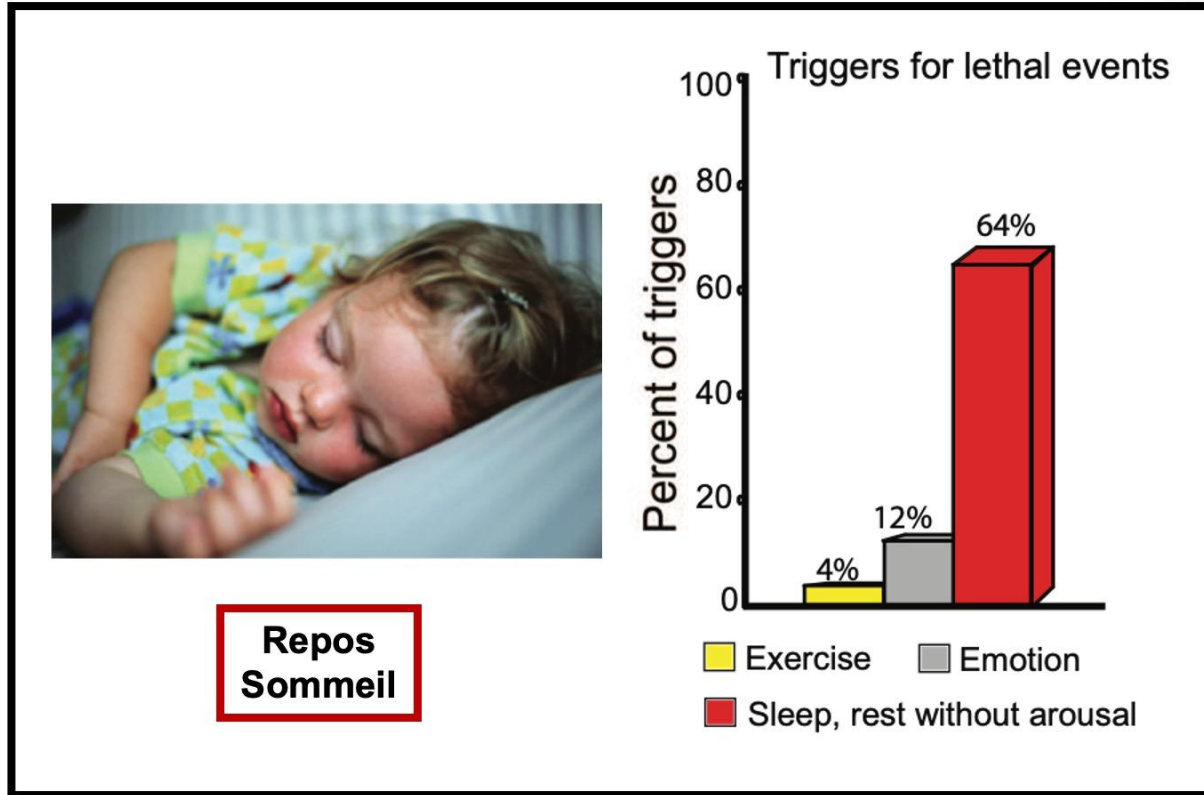
**LQTS type 3**

SCN5A

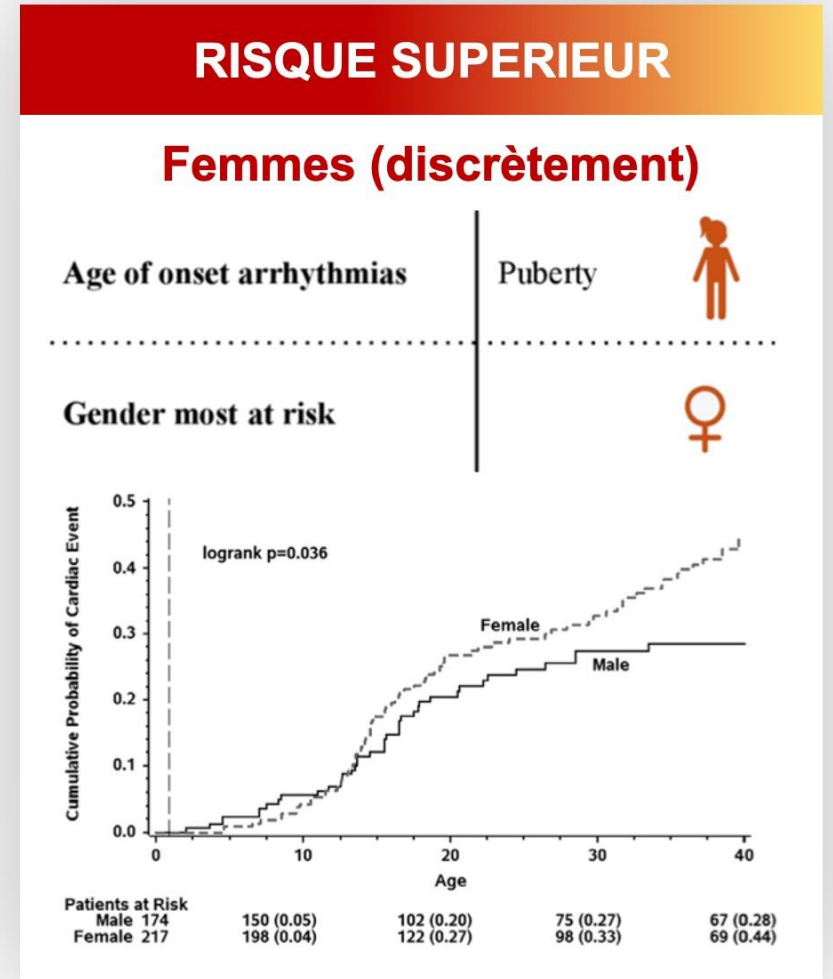
**LQT3 = 10%**



Wilde AAM et al. Heart (2022)



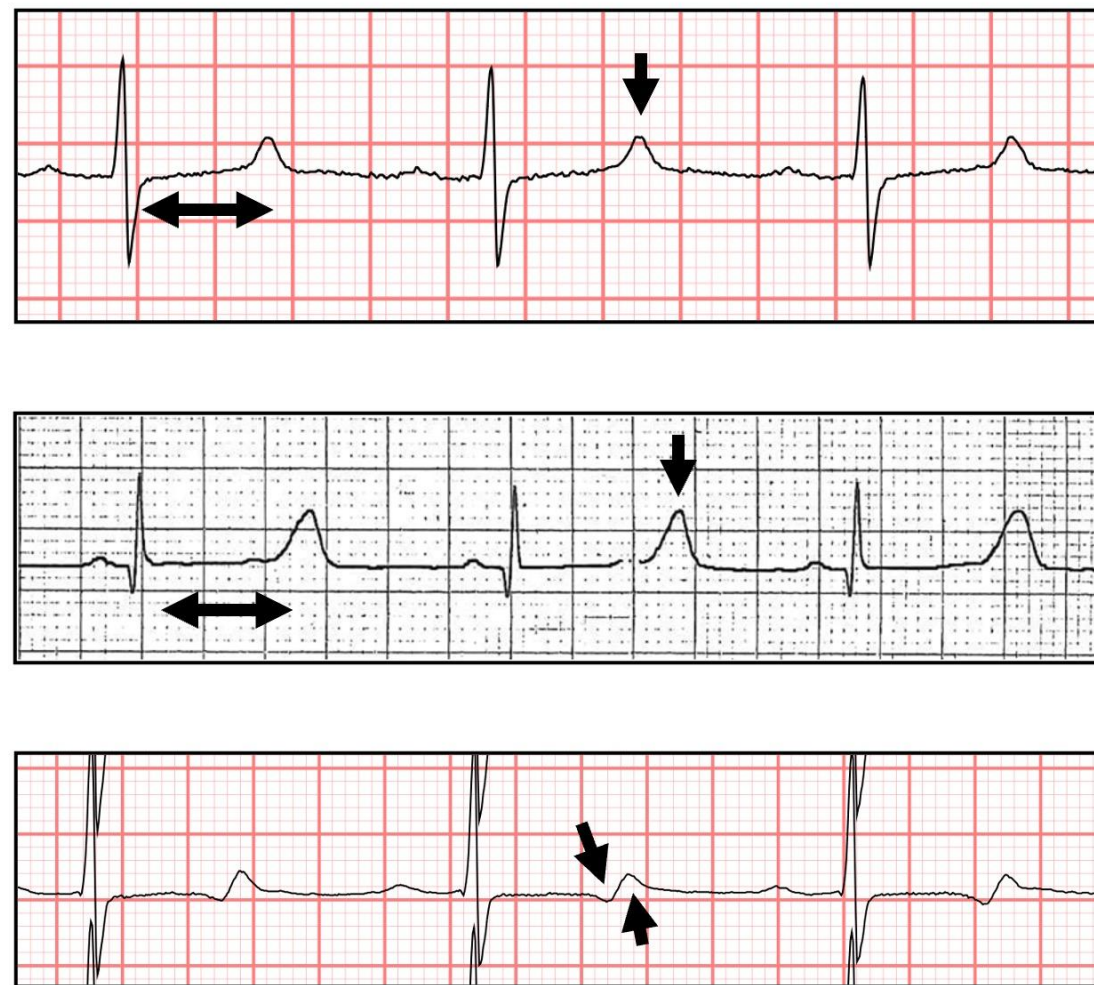
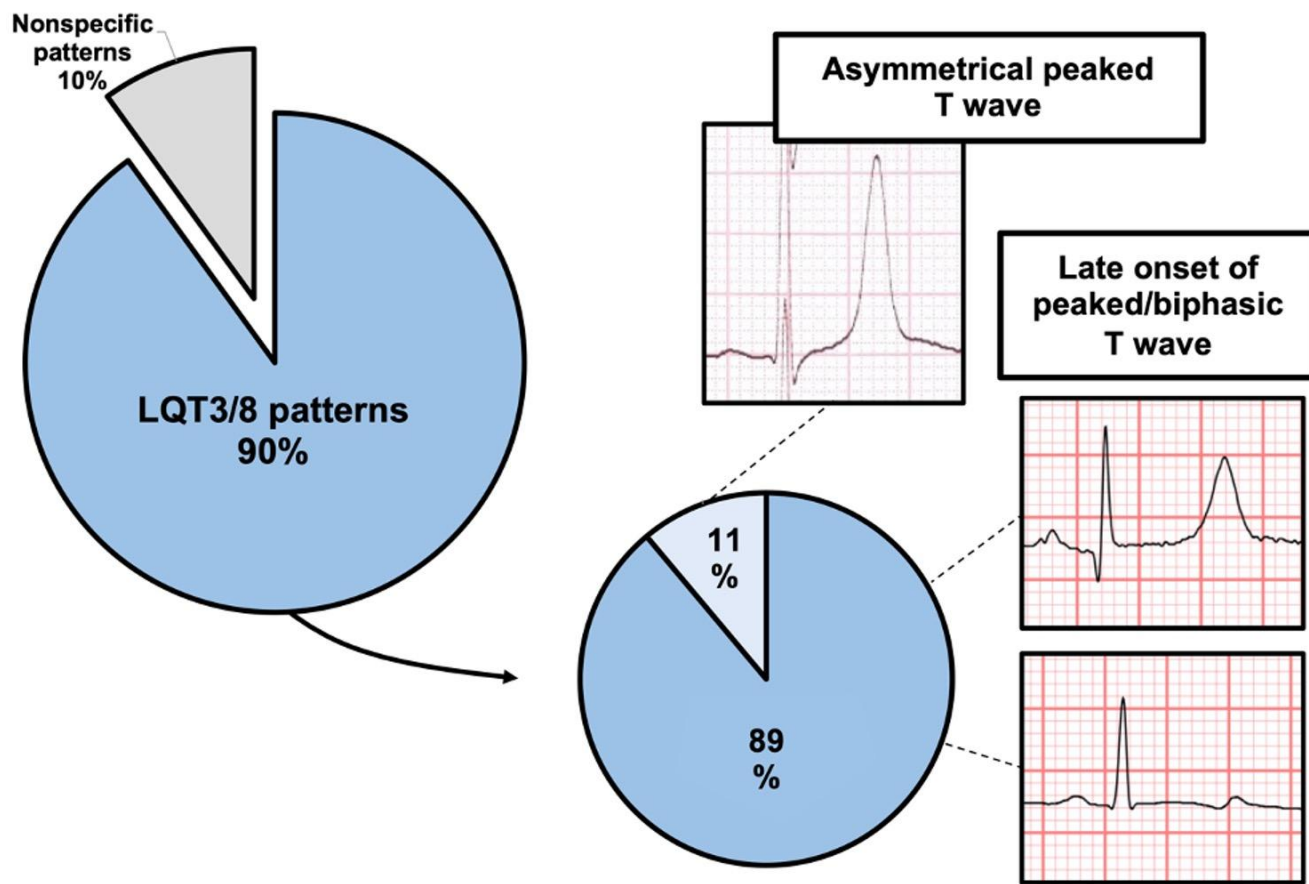
Tester DJ & Ackerman MJ. Circulation (2011)  
 Ruan Y et al. Circ AE (2008)  
 Schwartz PJ et al. Circulation (2001)



Wilde AAM et al. Heart (2022)  
 Zareba W et al. JACC (2003)

# LQTS type 3

SCN5A



Delinière A et al. Arch Cardiovasc Dis (2024)

## LQTS type 3

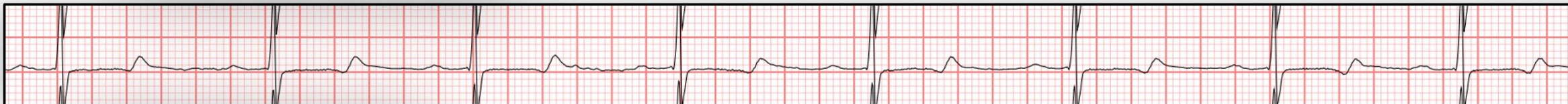
SCN5A

## Epreuve d'effort &amp; LQT3

Les manoeuvres dynamiques ne sont pas utiles dans le LQT3

Repos

Dysmorphie ST-T bien visible



Pic

Raccourcissement adapté



Récupération @ 1 min

Raccourcissement adapté



Récupération @ 4 min (7 min chez les enfants &amp; adolescents)

Amorce de réallongement encore discret



**S<sup>d</sup> de Jervell & Lange-Nielsen**

**KCNQ1, KCNE1**

(homozygote ou hétérozygote composite)

CONGENITAL DEAF-MUTISM, FUNCTIONAL HEART DISEASE WITH PROLONGATION OF THE Q-T INTERVAL, AND SUDDEN DEATH

ANTON JERVELL, M.D., AND FRED LANGE-NIELSEN, M.D.

TÖNSBERG, NORWAY

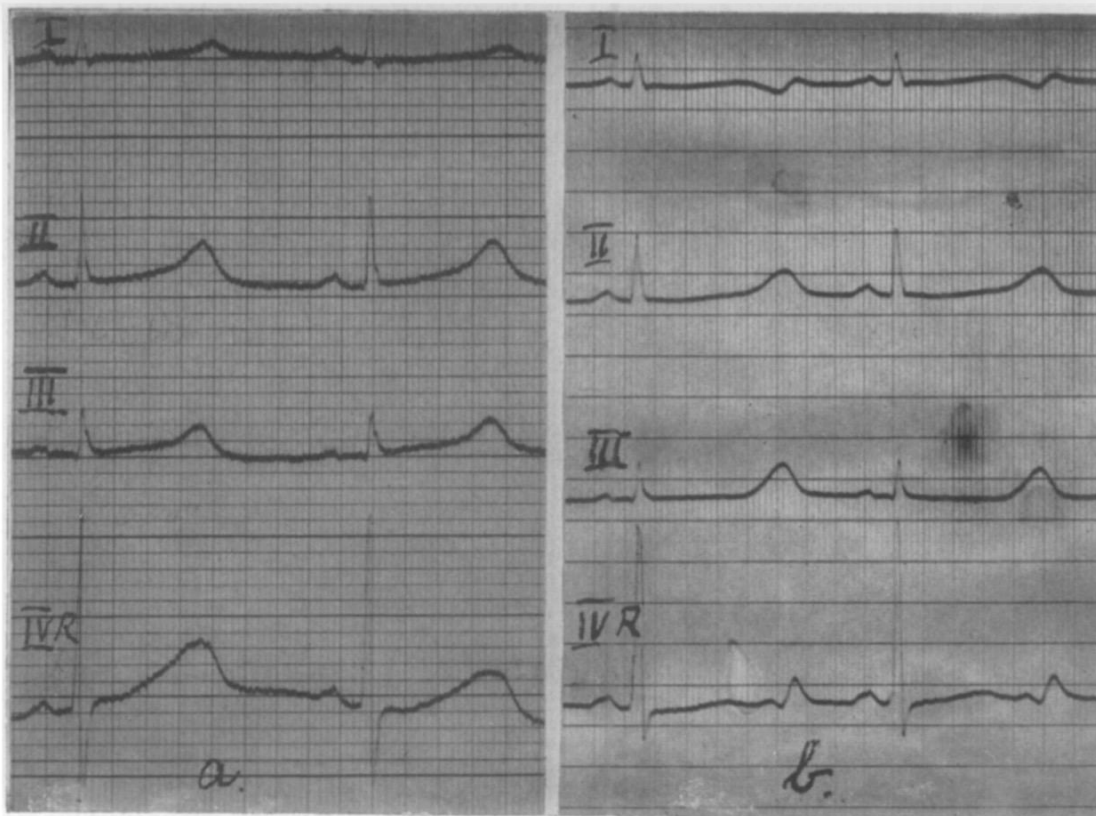
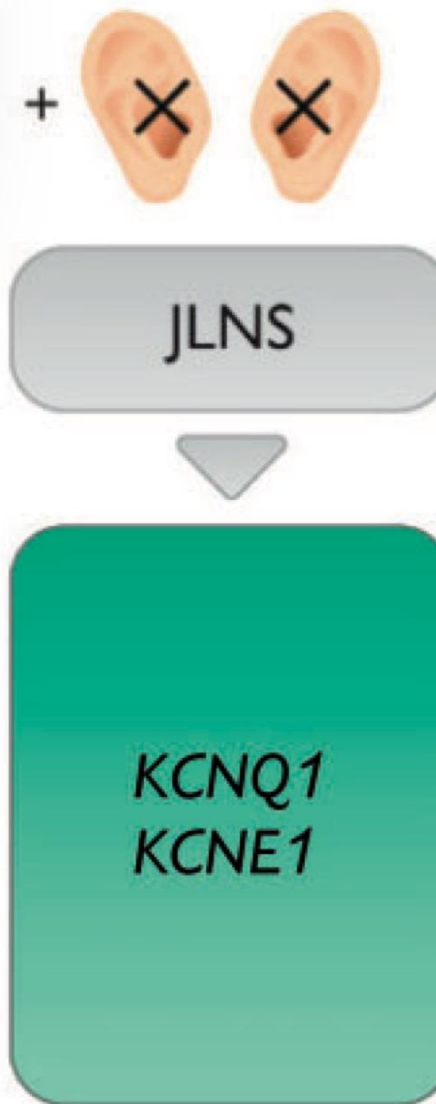


Fig. 1.—Tormod J. (a) ECG July 20, 1953, during rest. Leads I, II, III, IV R. Q-T = 0.50 sec. R-R = 0.88 sec. (b) ECG July 20, 1953, after stair-running. Leads I, II, III, IV R. Q-T = 0.60 sec. R-R = 0.86 sec.

**Jervell A & Lange-Nielsen F.**  
Am Heart J.  
(1957)



**Wilde AAM et al.**  
Europace (2022)

S<sup>d</sup> de Timothy  
& LQTS type 8

CACNA1C

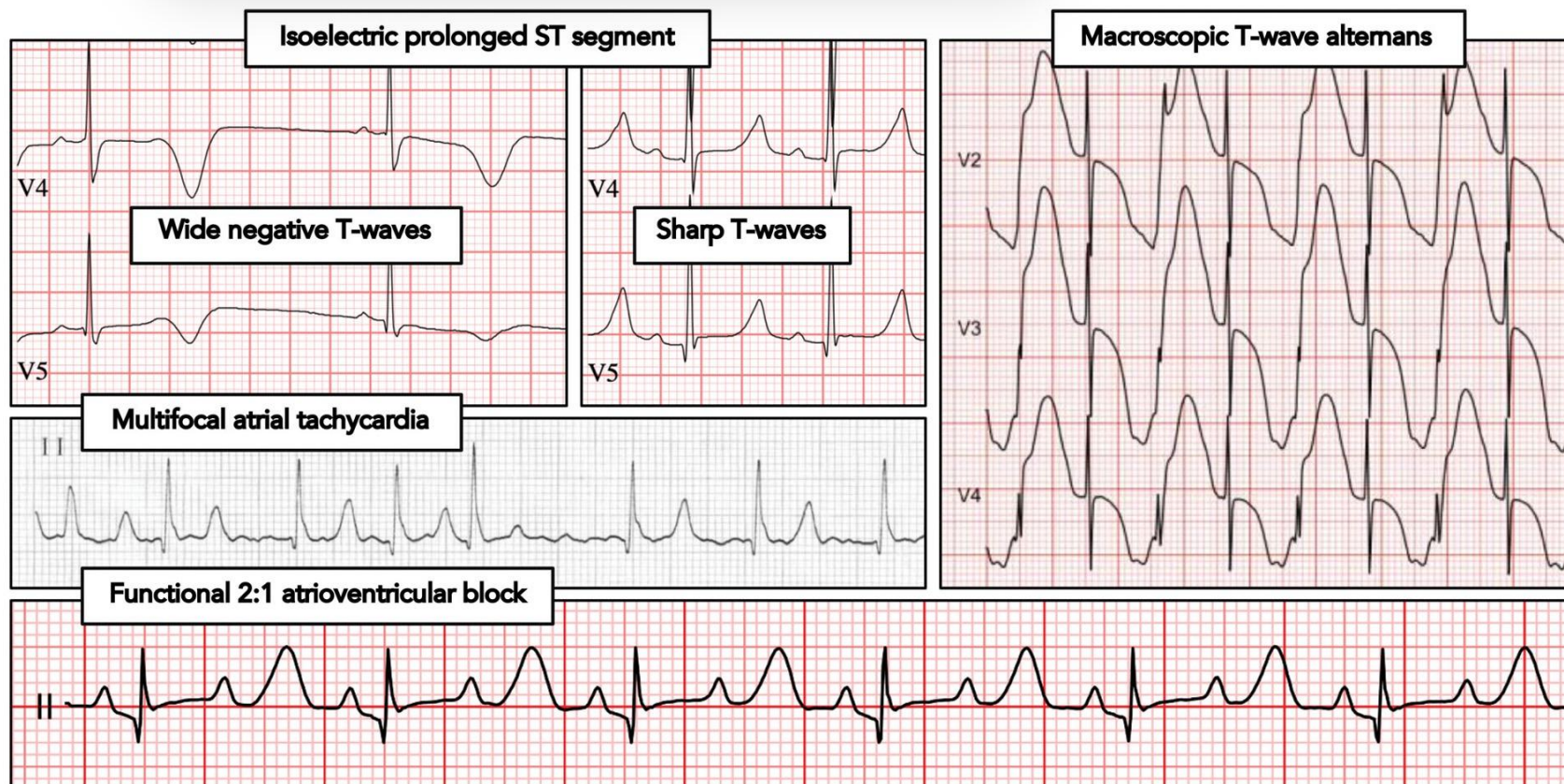
Circulation: Genomic and Precision Medicine

## RESEARCH LETTER

Phenotypic Characterization of Timothy Syndrome  
Caused by the CACNA1C p.Gly402Ser Variant

Antoine Delinière<sup>1</sup>, MD; Christelle Haddad, MD; Claudia Herrera-Siklody<sup>2</sup>, MD; Alexis Hermida<sup>3</sup>, MD; Etienne Pruvot<sup>4</sup>, MD; Sabrina Bressieux-Degueldre<sup>5</sup>, MD; Gilles Millat<sup>6</sup>, PhD; Alexandre Janin<sup>7</sup>, PharmD, PhD; Jean-Sylvain Hermida<sup>8</sup>, MD, PhD; Babken Asatryan<sup>9</sup>, MD, PhD; Philippe Chevalier<sup>10</sup>, MD, PhD

	Neurodevelopmental disorder	Syndactyly	Facial abnormalities	Baldness	CHD / LVH	Hypoglycaemia	Recurrent infections
Pt 1	Autism	No	No	No	No	No	Tonsillitis & otitis
Pt 2	Late psychomotor acquisition	No	Gingival & teeth abnormalities	No	No	No	No
Pt 3	No	No	No	No	No	No	No
Pt 4	Language delay	No	No	No	No	No	Tonsillitis



Delinière A et al. Circulation: Genomic and Precision Medicine (2023)

S<sup>d</sup> de Timothy  
& LQTS type 8

CACNA1C

Arrhythmogenic Cardiomyopathy Is a  
New Phenotype Associated With the  
CACNA1C p.Arg518Cys (R518C) Variant

Clément Boiteux, MD,<sup>a,b</sup> Samuel Chauveau, MD,<sup>c</sup> Kevin Gardey, MD,<sup>b</sup> Laurent Sebbag, MD, PhD,<sup>d,e</sup>  
Geoffroy Ditac, MD,<sup>b,f</sup> Cécile Cazeneuve, PHARM D, PhD,<sup>g</sup> Nathan Mewton, MD, PhD,<sup>d,e,h</sup>  
Alexandre Janin, PHARM D, PhD,<sup>g,i</sup> Philippe Chevalier, MD, PhD,<sup>a,b,j</sup> Antoine Delinière, MD, PhD<sup>a,b,j</sup>

## B A link between long-QT syndrome type 8 and arrhythmogenic cardiomyopathy

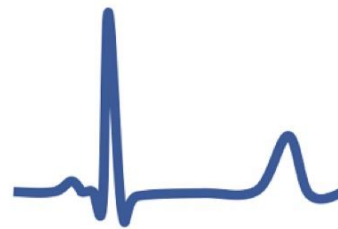
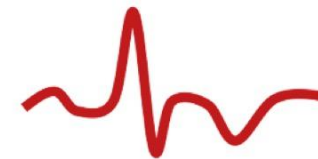
CACNA1C  
p.(Arg518Cys)

Novel phenotype identified in this study

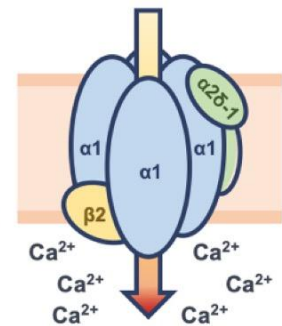
Arrhythmogenic cardiomyopathy

Previously described phenotypes

Long-QT syndrome type 8  
Cardiac-only Timothy syndrome  
Peripartum cardiomyopathy

LONG-QT SYNDROME  
TYPE 8ARRHYTHMOGENIC  
CARDIOMYOPATHY

IS THERE A CONTINUUM ?

Gain-of-function  
of  
Ca<sub>v</sub>1.2

Cardiotoxic  
effect of  
progressive Ca<sup>2+</sup>  
overload ?

**CALMODULINOPATHIES**

*CALM1-3*

140 subjects with variants in *CALM* genes

*CALM*-LQTS: 74 (53%)

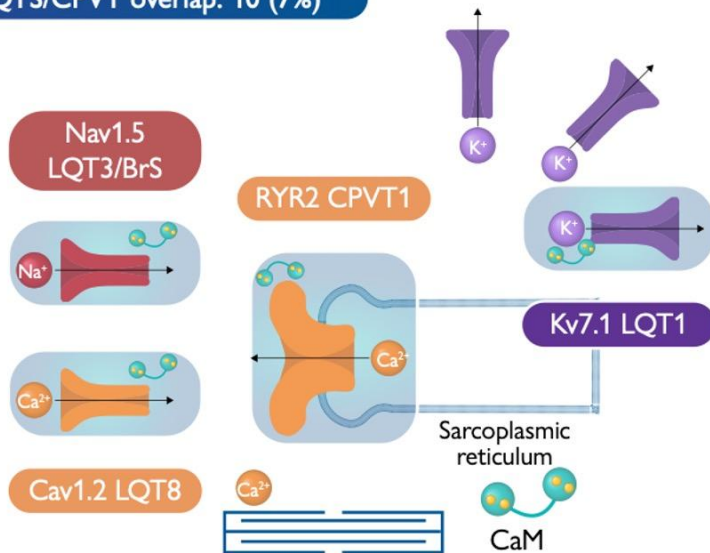


*CALM*-CPVT: 36 (26%)



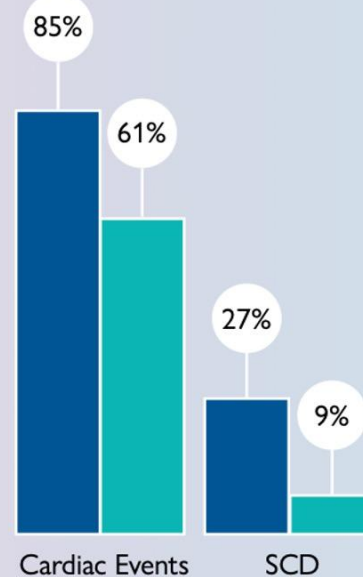
*CALM*-LQTS/CPVT overlap: 10 (7%)

- IVF 7 (5%)
- UD 11 (8%)
- Atypical 2 (1%)



Clinical Severity

Up to 2019  
2019-2023



Cytosol

Extracellular space



- Associated cardiomyopathies and/or congenital heart defects in 30% of *CALM* patients
- Neurological disorders in 35 patients: 15 post-anoxic sequelae, 20 primary neurological/neurodevelopmental features

PRISE EN CHARGE

**CredibleMeds Mobile Apps Available Now!**

Available for Apple IOS, Android and Windows Mobile devices

- Convenient mobile access to the QTdrugs database for healthcare providers and patients
- Instant access to latest revisions to the QTdrugs lists



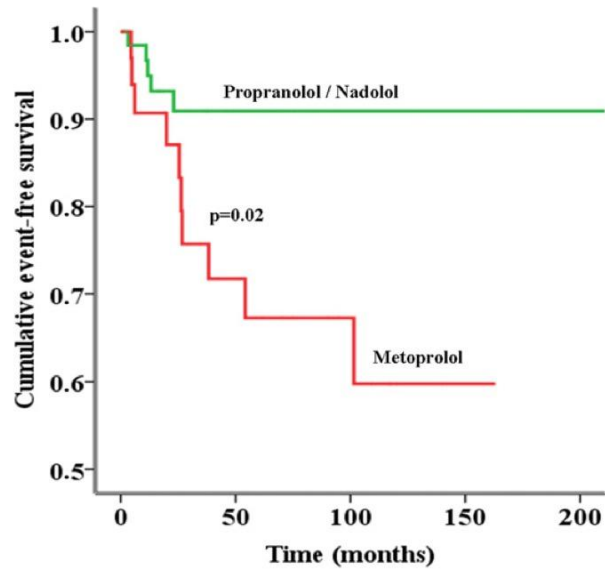
**EVICION DES TRAITEMENTS TORSADOGENES**

**LUTTE CONTRE LES TROUBLES ELECTROLYTIQUES**

- Hypokaliémie
- Hypomagnésémie
- Hypocalcémie

**EVICION DES TRIGGERS SELON LE GENOTYPE**

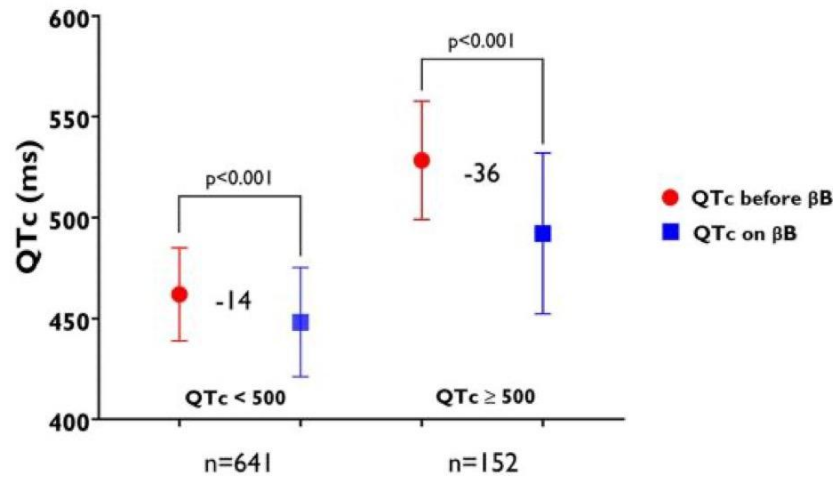
**NADOLOL ou PROPRANOLOL**



Patients at risk	0	50	100	150	200
Propranolol/Nadolol	66	23	7	5	2
Metoprolol	35	17	9	1	0

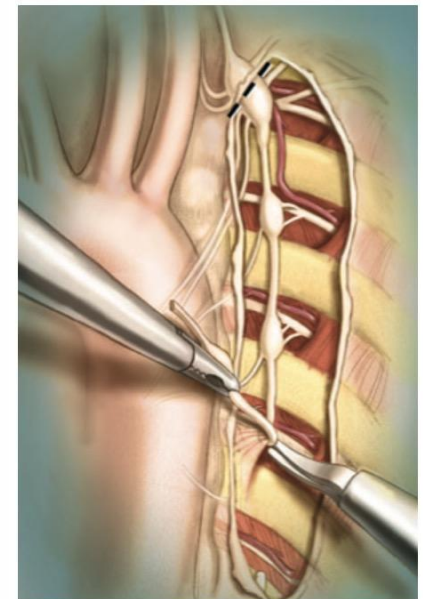
Chockalingam P et al. JACC. 2012;60(20):2092-9

**Impact of βB on Baseline QTc**



Dusi V et al. European Heart Journal (2024) 00, 1–10

**SYMPATHECTOMIE**

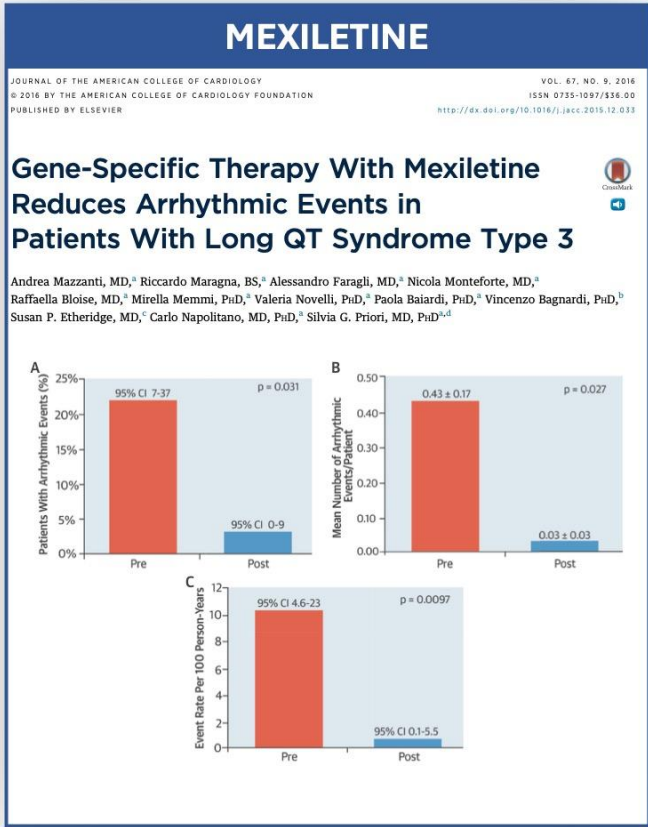
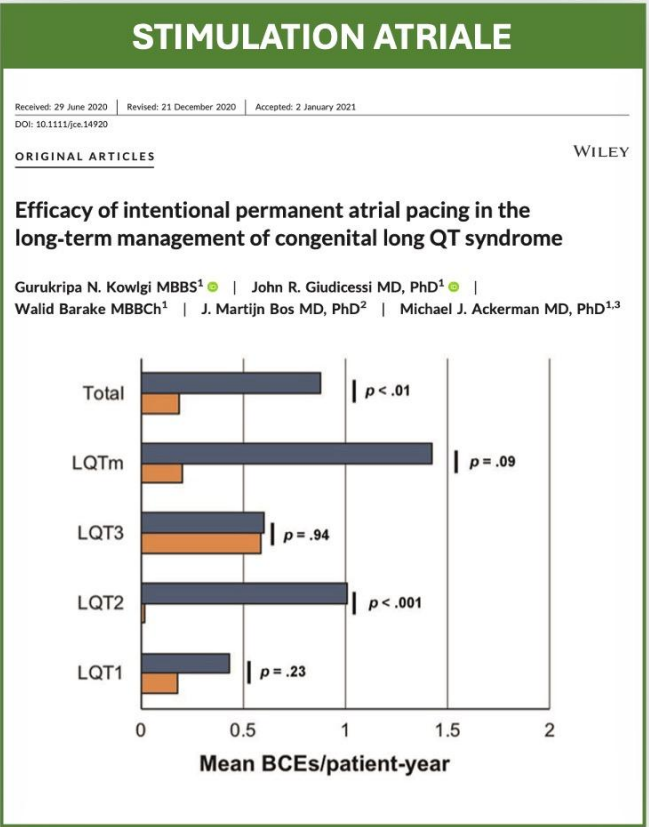
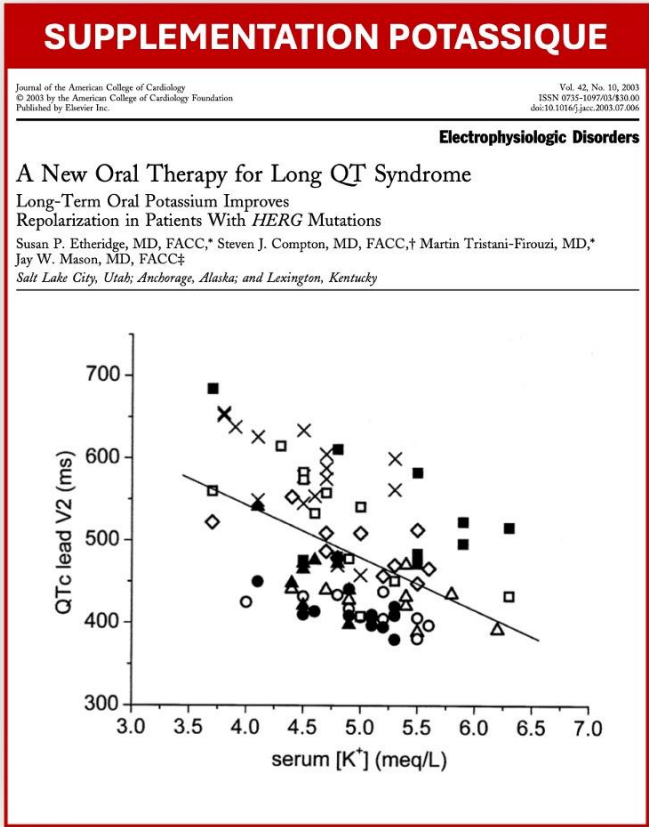


Witt CM et al. Europace (2017) 19, 1075–1083

PRISE EN CHARGE

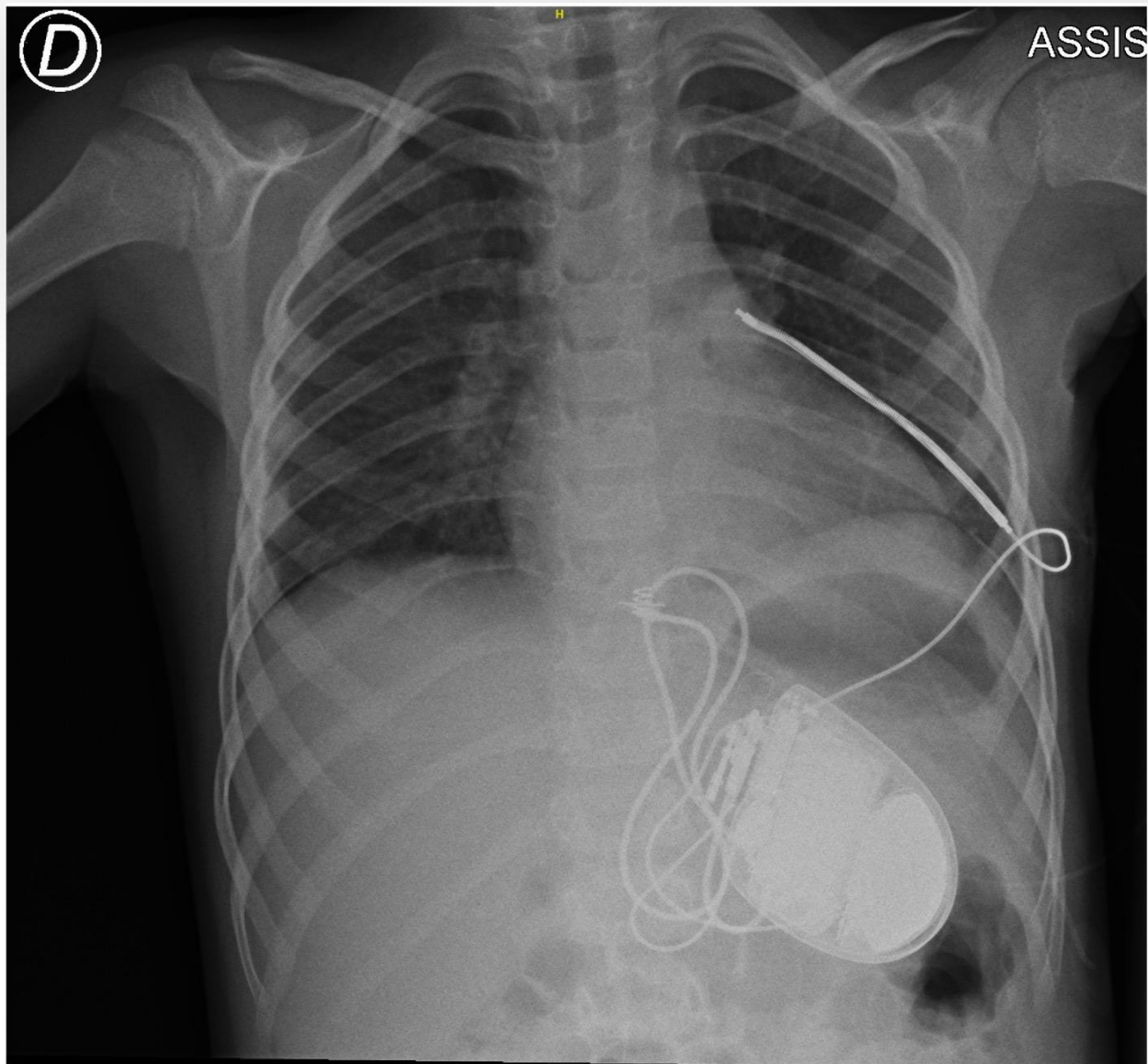
**LQT2  
(KCNH2)**

**LQT3  
(SCN5A)**



**LQT8  
/ Timothy  
(CACNA1C)**

## ▪ PRISE EN CHARGE



## Et le DAI ?

- Désormais exceptionnel en prévention primaire
- Reste indispensable en **prévention secondaire**

Avis centre expert **indispensable****Indications légitimes selon Pr Peter J. SCHWARTZ**  
*(situations exceptionnelles)*

- ♥ **Arrêt cardiaque (prévention secondaire)**
  - Sauf si cause réversible
  - Discussion si LQT1 qui n'était pas sous Bbl (nadolol très efficace)
- ♥ **Syncope malgré Bbl (nadolol ou propranolol) & dénervation sympathique (LCSD)**
- ♥ **Jervell et Lange-Nielsen avec syncope sous Bbl**
- ♥ **QTc > 550 ms & instabilité électrique** (alternance des ondes T, pauses nocturnes)
- ♥ **QTc > 600 ms**
- ♥ **Calmodulinopathies avec phénotype LQTS**

**■ QUE NOUS DISENT LES ETUDES ?**

## Return to play? Athletes with congenital long QT syndrome

Jonathan N Johnson,<sup>1</sup> Michael J Ackerman<sup>1,2,3</sup>

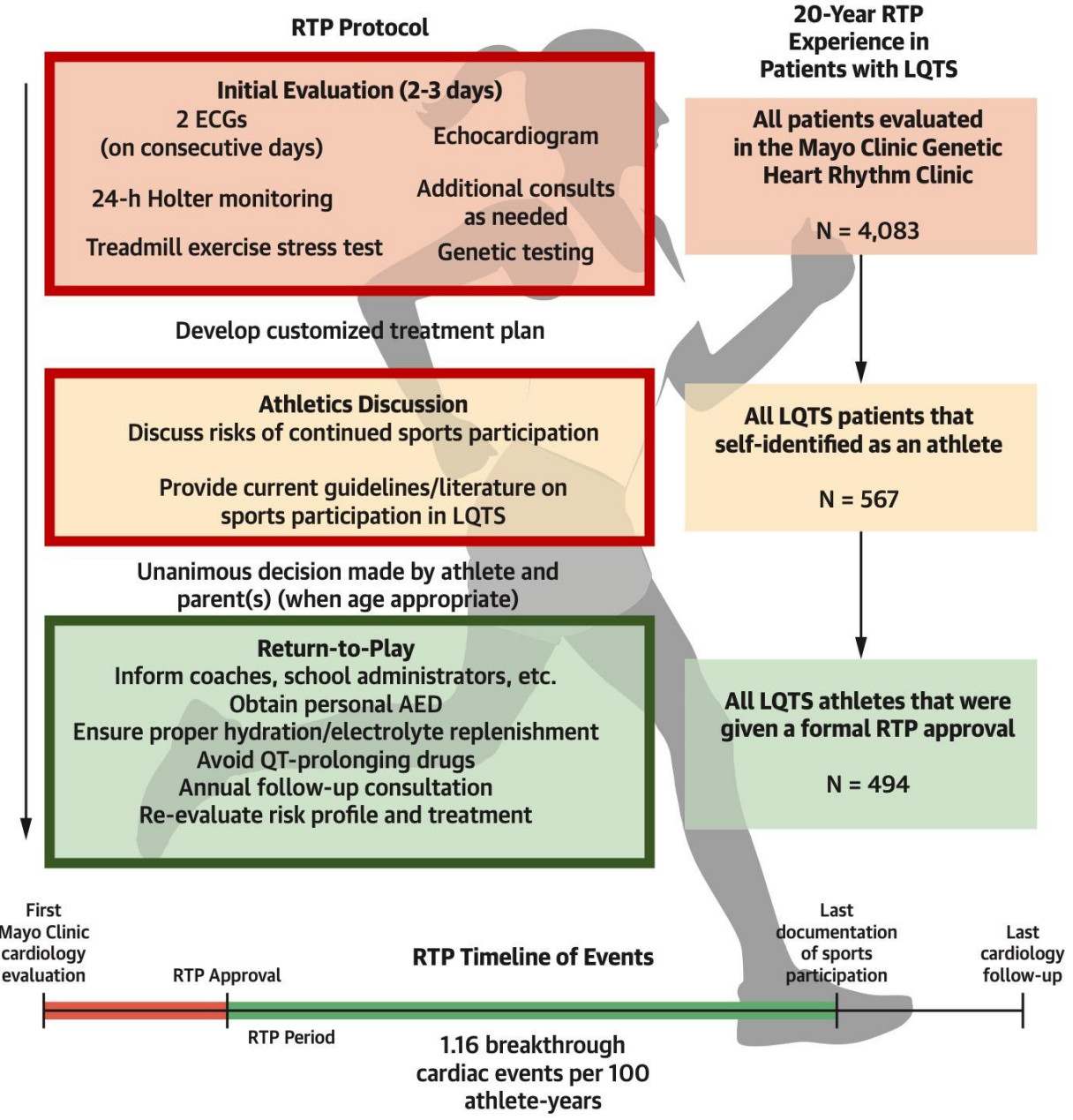
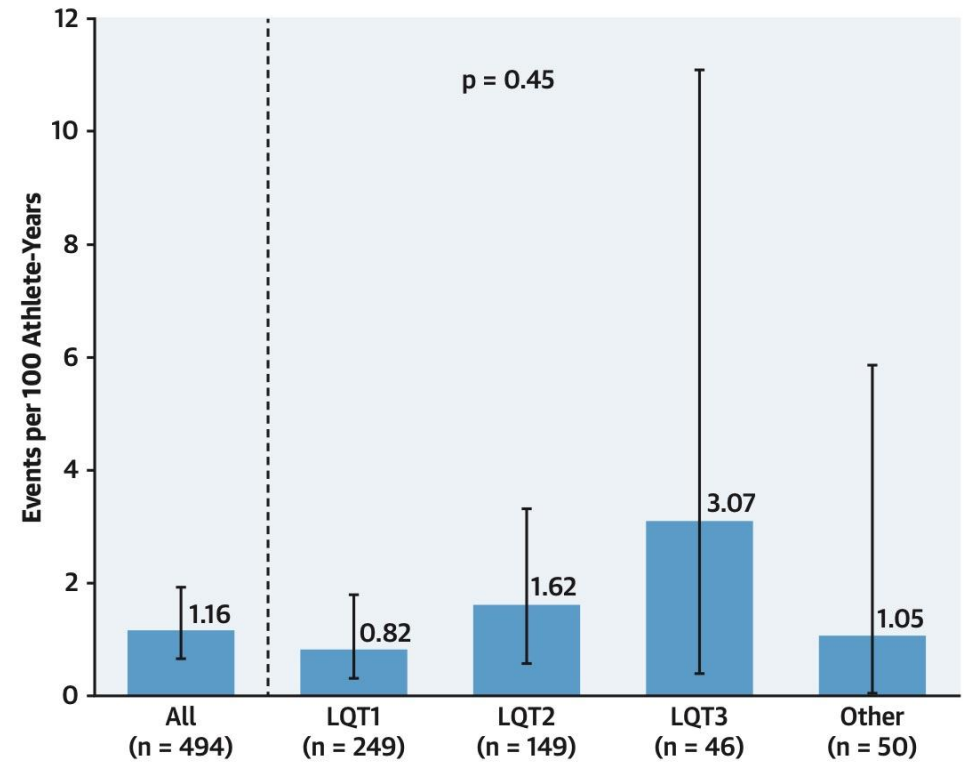
### What are the new findings?

- ▶ In a cohort of 130 athletes with long QT syndrome (LQTS) who continued to participate in competitive athletics following their diagnosis, only one had a cardiac event in over 650 athlete-years of follow-up.
- ▶ One patient with an event was a known high-risk 9-year-old LQT1 male with a history of cardiac arrest and implantable cardioverter-defibrillator (ICD) placement, who received two appropriate ICD therapies, both while warming up prior to practices, and both during admitted non-compliance with  $\beta$ -blocker medication.

QUE NOUS DISENT LES ETUDES ?

# Return-to-Play for Athletes With Long QT Syndrome or Genetic Heart Diseases Predisposing to Sudden Death

Kathryn E. Tobert, BA,<sup>a</sup> J. Martijn Bos, MD, PhD,<sup>a,b,c</sup> Ramin Garmany, BS,<sup>a,d</sup> Michael J. Ackerman, MD, PhD<sup>a,b,c</sup>



Tobert, K.E. et al. J Am Coll Cardiol. 2021;78(6):594-604.

QUE NOUS DISENT LES ETUDES ?

ESC European Society of Cardiology  
 Europeace (2022) 24, 1675-1683  
 https://doi.org/10.1093/europeace/eaac047

CLINICAL RESEARCH  
 Channelopathies and cardiomyopathies

Does sports participation increase risk in patients with long QT syndrome? Results from a large French cohort

Caroline Davydoff<sup>1</sup>, Antoine Andorin<sup>1\*</sup>, Damien Minois<sup>1</sup>, Marine Arnaud<sup>1</sup>, Mathilde Minier<sup>1</sup>, Frédéric Sacher<sup>2</sup>, Raphael Martins<sup>3</sup>, Nicolas Clementy<sup>4</sup>, Jean-Baptiste Gourraud<sup>1</sup>, and Vincent Probst<sup>1</sup>

Davydoff C et al. Europeace (2022)

What's new?

- In patients with long QT syndrome (LQTS) and a low-risk profile, European 'real-life' competitive and leisure sports practice is associated with a very low rate of cardiac arrhythmic event (CAE).
- Good compliance to beta-blocker therapy was associated with no CAE during sports practice in LQTS patients.

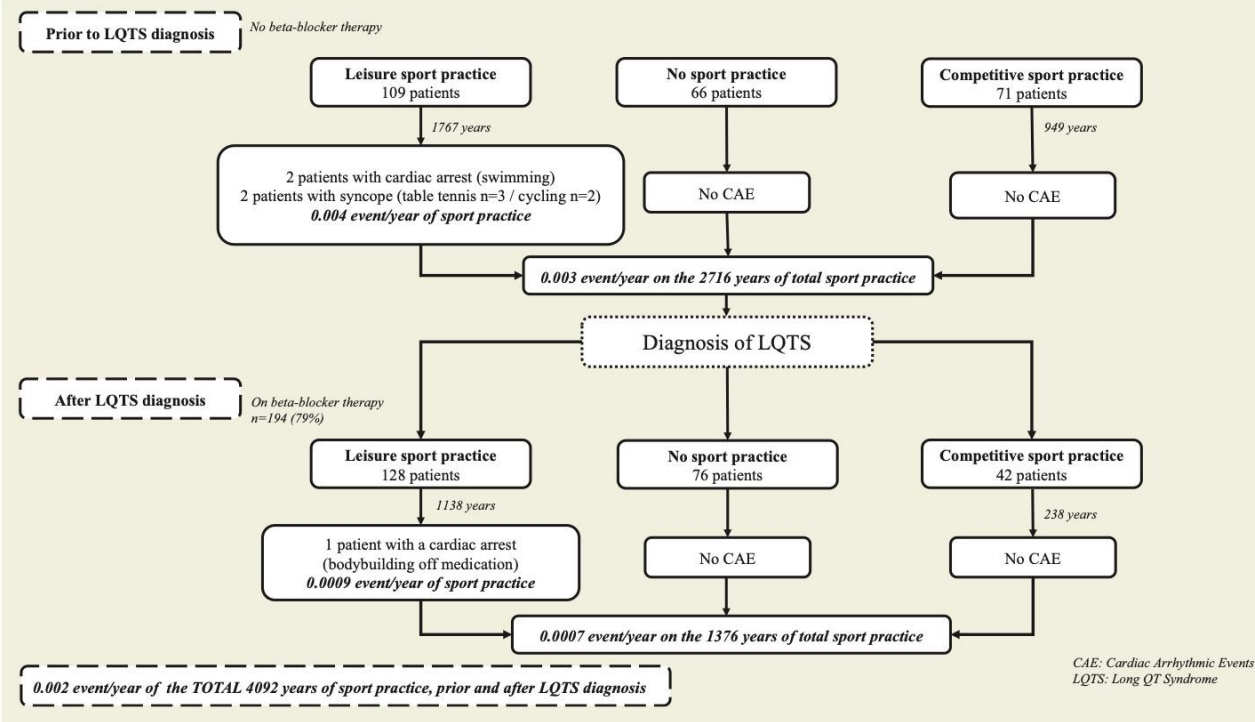


Table 6 Details of LQTS patients with CAE

Patient	Time of arrhythmic event	Type of event	Age	Sex	Mutation	QTc without/with beta-blocker (ms)	Type of sport (Mitchell's classification)	Medical therapy	Sports practice after CAE
1	Prior to LQTS diagnosis	Cardiac arrest	10	F	KCNQ1	540/490	Swimming (IIC) Leisure	None Nadolol after CAE	Yes Leisure
2	Prior to LQTS diagnosis	Cardiac arrest	11	M	None	455/465	Swimming (IIC) Leisure	None Nadolol after CAE	Yes Leisure
	After LQTS diagnosis	Cardiac arrest	16	M	None	455/465	Bodybuilding (IIIA) Leisure	Nadolol (non-compliance)	Yes Leisure
3	Prior to LQTS diagnosis	Syncope (n=2)	51	F	None	535/450	Cycling (IIIC) Leisure	Hydroxychloroquine Nadolol after CAE	Yes Leisure
4	Prior to LQTS diagnosis	Syncope (n=3)	6	M	KCNQ1	514/493	Table tennis (IB) Leisure	None Nadolol after CAE	Yes Leisure

CAE, cardiac arrhythmic event; LQTS, long QT syndrome.

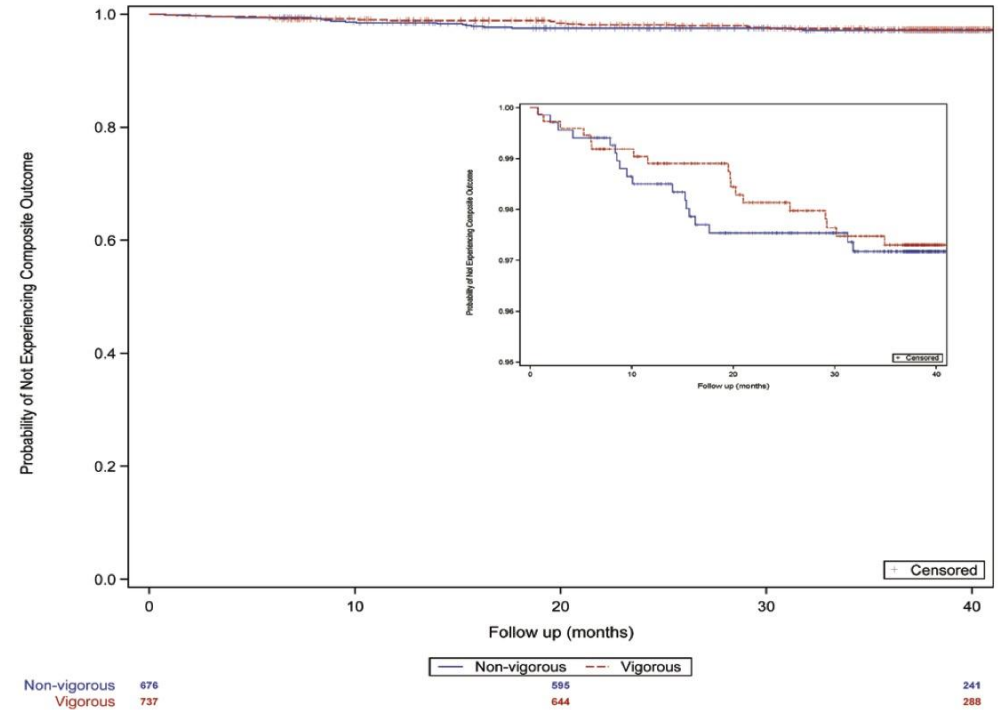
QUE NOUS DISENT LES ETUDES ?

**Circulation**

**ORIGINAL RESEARCH ARTICLE**

### Vigorous Exercise in Patients With Congenital Long QT Syndrome: Results of the Prospective, Observational, Multinational LIVE-LQTS Study

Rachel Lampert, MD; Sharlene Day, MD; Barbara Ainsworth, PhD, MPH; Matthew Burg, PhD; Bradley S. Marino, MD; Lisa Salberg, MD; Maria Teresa Tome Esteban, MD; Dominic J. Abrams, MD; Peter F. Aziz, MD; Cheryl Barth, BS; Elijah R. Behr, MD; Cheyanne Bell, MD; Charles I. Berul, MD; Johan M. Bos, MD; David Bradley, MD; David S. Cannom, MD; Bryan C. Cannon, MD; Maryann Anandi Concannon, MSW; Marina Cerrone, MD; Richard J. Czossek, MD; Anne M. Dubin, MD; James Dziura, PhD; Christopher C. Erickson, MD; N.A. Mark Estes III, MD; Susan P. Etheridge, MD; Ilan Goldenberg, MD; Belinda Gray, MD, MBBCh, PhD; Carla Haglund-Turnquist, MD; Kimberly Harmon, MD; Cynthia A. James, PhD; Christopher Johnsrude, MD; Prince Kannankeril, MD; Alice Lara, BSN; Ian H. Law, MD; Fangyong Li, MS; Mark S. Link, MD; Silvana M. Molossi, MD, PhD; Brian Olshansky, MD; Peter A. Noseworthy, MD; Elizabeth V. Saarel, MD; Shubhayan Sanatani, MD; Mauliy Shah, MD, MBBCh; Laura Simone, MS; Jonathan Skinner, MD, ChB; Gordon F. Tomaselli, MD; James Simon Ware, MD; Gregory Webster, MD; Wojciech Zareba, MD, PhD; Douglas P. Zipes, MD; Michael J. Ackerman, MD, PhD



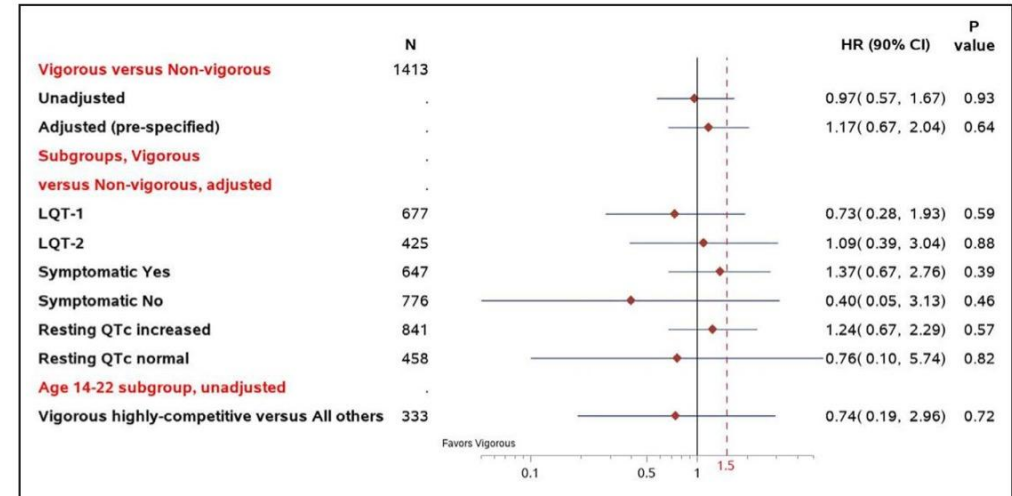
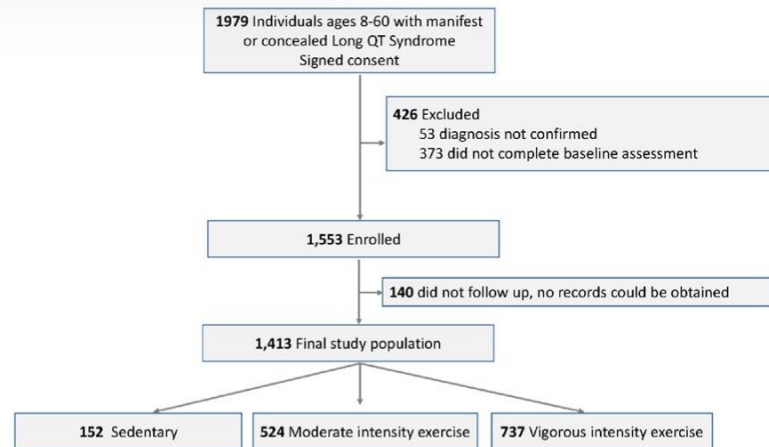
**Clinical Perspective**

**What Is New?**

- This is the first prospective study to investigate whether vigorous exercise increases risk of arrhythmic events in individuals with congenital long QT syndrome.
- In this study, long QT syndrome-triggered cardiac events were low in both those exercising vigorously and those exercising nonvigorously, and there was no statistical difference in event rate. CIs were wide, and noninferiority not demonstrated.
- Findings were similar in vigorous-competitive athletes 14 to 22 years of age.

**What Are the Clinical Implications?**

- These findings will further inform shared decision-making discussions between patient and physician about exercise and competitive sports participation.



Vigorous activity was categorized by participation in at least 1 activity at metabolic equivalents  $\geq 6.0$  for  $\geq 60$  hours per year.<sup>25</sup>

## ■ QUE NOUS DISENT LES ETUDES ?

JAMA  
Network | Open™

Original Investigation | Pediatrics

**Cardiopulmonary Fitness and Physical Activity Among Children and Adolescents With Inherited Cardiac Disease**

Luc Souilla, PhD; Oscar Werner, MD; Helena Huguet, MSc; Arthur Gavotto, MD, PhD; Marie Vincenti, MD; Jean-Luc Pasquie, MD, PhD; Gregoire De La Villeon, MD; Sophie Guillaumont, MD; Victor Pommier, MD; Stefan Matecki, MD, PhD; Alban-Elouen Baruteau, MD, PhD; Constance Beyler, MD; Christophe Delclaux, MD, PhD; Isabelle Denjoy, MD; Philippe Charron, MD, PhD; Philippe Chevalier, MD, PhD; Antoine Delinière, MD; Mathieu Andrianoely, BSc; Lauriane Cornuault, PharmD, PhD; Caroline Besnard-Neyraud, MD, PhD; Frederic Sacher, MD, PhD; Patricia Reant, MD, PhD; Denis Mottet, MD, PhD; Marie-Christine Picot, MD, PhD; Pascal Amedro, MD, PhD; for the Quality of Life in Children With Inherited Cardiomyopathy or Arrhythmia (QUALIMYORHYTHM) Study Group

**Question** What are the levels of cardiopulmonary fitness among children and adolescents with inherited cardiac disease compared with healthy controls and the factors associated with maximum oxygen uptake ( $\dot{V}O_2\text{max}$ ) in young patients?

**Findings** This cross-sectional study with 207 participants showed lower levels of cardiopulmonary fitness among youths with inherited cardiac disease. The multivariable model explained 80% of  $\dot{V}O_2\text{max}$ , including clinical, functional, sociodemographic, and behavioral parameters.

**Meaning** This study suggests that assessing cardiopulmonary fitness among children with inherited cardiac disease can be incorporated into the shared decision-making process for sports participation and may help identify eligible patients for early preventive cardiac rehabilitation programs.

Souilla L et al. JAMA Network Open (2025)

- **GUIDELINES**


**ESC**

 European Society  
of Cardiology

**2020**

Recommendations	Class	Level
It is recommended that all exercising individuals with LQTS with prior symptoms or prolonged QTc be on therapy with beta-blockers at target dose.	I	B
It is recommended that exercising individuals with LQTS should avoid QT prolonging drugs ( <a href="http://www.crediblemeds.org">www.crediblemeds.org</a> ) and electrolyte imbalance such as hypokalaemia and hypomagnesaemia.	I	B
Shared decision-making should be considered regarding sports participation in patients with genotype positive/phenotype negative LQTS (i.e. <470/480 ms in men/women). Type and setting of sports (individual vs. team), type of mutation, and extent of precautionary measures should be considered in this context.	IIa	C

- **GUIDELINES**


**ESC**

 European Society  
of Cardiology

**2020**

Recommendations	Class	Level
Participation in high intensity recreational and competitive sports, even when on beta-blockers, is not recommended in individuals with a QTc >500 ms or a genetically confirmed LQTS with a QTc $\geq$ 470 ms in men or $\geq$ 480 ms in women.	<b>III</b>	<b>B</b>
Participation in competitive sports (with or without ICD) is not recommended in individuals with LQTS and prior cardiac arrest or arrhythmic syncope.	<b>III</b>	<b>C</b>

- **GUIDELINES**



**EHRA**  
European Heart  
Rhythm Association

 European Society of Cardiology

2020

### Consensus statement—congenital LQTS

### Symbol

All LQTS athletes should avoid QT prolonging drugs ([www.crediblemeds.org](http://www.crediblemeds.org)) and electrolyte imbalance like hypokalaemia and hypomagnesemia (potassium supplementation is recommended before taking part to sports activity).



All LQTS athletes with prior symptoms or prolonged QTc should be on therapy with beta-blockers at target dose.



Athletes with LQTS and prior cardiac arrest or arrhythmic syncope should not be allowed to practice competitive sports (with or without ICD).



Athletes with a QTc >500 ms, a de novo disease-causing mutation (especially if LQT1), or genetically confirmed LQTS with a QTc  $\geq$ 470 ms in men or  $\geq$ 480 ms in women should not practice more than light- to moderate intensity recreational sports, even when on beta-blockers.



## ▪ GUIDELINES



**EHRA**  
European Heart  
Rhythm Association

 European Society of Cardiology

2020

Recommendations to sports participation require open discussion with the athlete and their entourage, finding a balance between life protection and quality of life during shared decision-making.



It is reasonable to allow individual sports at low to moderate intensity for asymptomatic athletes with an LQT1 mutation but QTc <470/480 ms and who are on prophylactic beta-blocker therapy, but team sports and high-intensity sports are discouraged.



It is reasonable to allow all types of sports participation for asymptomatic athletes with an LQT2 or LQT3 mutation but QTc <470/480 ms, and who are on prophylactic beta-blocker therapy.



For asymptomatic athletes with other LQTS mutations and QTc <470/480 ms, cardiogenetics consult and shared decision-making are required.



## GUIDELINES



2025

### General considerations

Competitive athletes with a cardiac channelopathy (including LQTS, CPVT, and BrS) should be assessed by a pediatric or adult cardiologist with expertise in cardiac channelopathies and with SDM.

In competitive athletes diagnosed with a cardiac channelopathy, an ICD should not be implanted for the sole purpose of competitive sports participation.

### Long QT syndrome

#### Specific clinical considerations

It is reasonable for competitive athletes with positive genetic test results for LQTS but who have a resting QTc <460 ms (ie, concealed variant positive LQTS) to participate in competitive sports.

In competitive athletes with LQTS (asymptomatic [QTc  $\geq$ 460 ms prepuberty,  $\geq$ 470 male,  $\geq$ 480 female] or previously symptomatic) but who are under expert assessment and supervision, competitive sports participation is reasonable with SDM after risk assessment, education, and implementation of guideline-directed therapies.

In competitive athletes with LQTS (including LQT1), competitive swimming and diving can be considered with appropriate precautions.\*

- \* Precautions include swimming with supervision by an individual who is trained in cardiopulmonary resuscitation, with preference for pools rather than open water, and access to an automatic external defibrillator.

Kim JH et al. Circulation (2025)

## CENTRE EXPERT DECISION MEDICALE PARTAGEE



**NADOLOL**  
(ou **PROPRANOLOL**)  
± MEXILETINE



± **SYMPATHECTOMIE**



**EMERGENCY ACTION  
PLAN**



**DEFIBRILLATEUR  
AUTO EXTERNE**

**$\geq$  3 MOIS SANS SYMPTÔME**

Circulation**ON MY MIND**

## Return to Play With Genetic Heart Disease: The Importance of Developing a Personalized Emergency Action Plan

Belinda Gray , MBBS, PhD; Rachel Lampert , MD; Michael Papadakis , MD

Gray B et al. Circulation 2025



American  
Heart  
Association®

### PERSONALISED EMERGENCY ACTION PLAN

Name : XXX  Weight: 89kg  
DOB: XXX Height: 191cm

**Medical Condition:** Hypertrophic Cardiomyopathy (HCM)  
Implantable cardioverter-defibrillator (ICD) - pacemaker

**Medication:** Bisoprolol, dosage 2.5mg daily – beta blocker

### MANAGEMENT

- Support crew to include trained First Aiders.
- AED and first aid kit on the boat.
- All support crew to complete online AED training.
- Two dedicated spotters maintaining visual contact with the swimmer.
- Monitor swimmer for degradation of stroke/ swimming action.
- Periodical verbal communication with swimmer.
- Monitor food and fluid intake per feeding plan.
- Remove swimmer from water if deemed unsafe to continue.
- Swimmer to go to medical recovery area at completion of race.

### INCIDENT ACTION PLAN

***IF conscious**, but experiencing symptoms like chest pain, dizziness, unrelieved breathlessness, temporarily blacked out, ICD discharged.*

#### ACTION:

- **[Support Crew 1, Support Crew 2]** Remove XX from the water and sit him up on deck of boat.
- **[Support Crew 1, Support Crew 2]** Keep warm, monitor, and reassure him.
- **[Support Crew 3]** If symptoms persist for more than a few minutes call for help; radio call Channel Swim Base.
  - VHF Channel 16 - **“Pan-Pan, Pan-Pan, Pan-Pan! Our position is [GPS coordinates], our emergency is swimmer with pre-existing heart condition experiencing chest pain that requires immediate medical attention.”**
  - Race Number: xxx

- Boat Rego Number: **XXXXXX**
- Follow instructions from Channel Swim Base

#### IF unconscious

#### ACTION:

- **[Support Crew 1, Support Crew 2]** Remove XX from the water. Paddler may need to enter water and roll XX onto his back to bring him to the boat (note, leave kayak if required – it can be picked up by another boat).
- **[Driver]** Ensure motor is off/ in neutral.
- **[Support Crew 1, 2 and 3]** Bring XX aboard via ladder/door at stern of boat. Might require multiple people to lift onto boat.
- **[Support Crew 3]** Prep area on boat to receive XX, grab defib and towels (dry towel on bottom and towels to dry off chest)
- **[Support crew 1]** Lie XX on back commence ABC (already done D&R, if unsure commence CPR).
- **[Support Crew 2]** Simultaneously with above, dry down and apply defib pads (this is a priority and should start at the same time as the ABC assessment). Pause to rhythm check as soon as pads applied and follow instructions on AED.
- **[Support Crew 3]** Call for help as soon as available to do so; radio call Channel Swim Base.
  - VHF Channel 16
  - **“Pan-Pan, Pan-Pan, Pan-Pan! Our position is [GPS coordinates], our emergency is unconscious swimmer, suspected Cardiac Arrest, +/- CPR in progress, we require immediate medical assistance.”**
  - Race Number: xxx
  - Boat Rego Number: xxx
  - Follow instructions from Channel Swim Base
  - Further information: Swimmer has pre-existing heart condition. Has an ICD and we have an AED.

Protocole National de Diagnostic et de Soins - Syndrome du QT long

## Protocole National de Diagnostic et de Soins (PNDS)

### Syndrome du QT Long

#### Centre de référence constitutif (CRMR)

Centre de Référence des Maladies cardiaques héréditaires  
Hôpital Bichat APHP, Dr Isabelle DENJOY

#### Centre de référence associé (CRMA)

Centre de référence pour la prise en charge des maladies rythmiques héréditaires de l'Ouest, CHU Nantes, Hôpital Nord, Pr Vincent PROBST

Mai 2021

## Protocole National de Diagnostic et de Soins - Syndrome du QT long

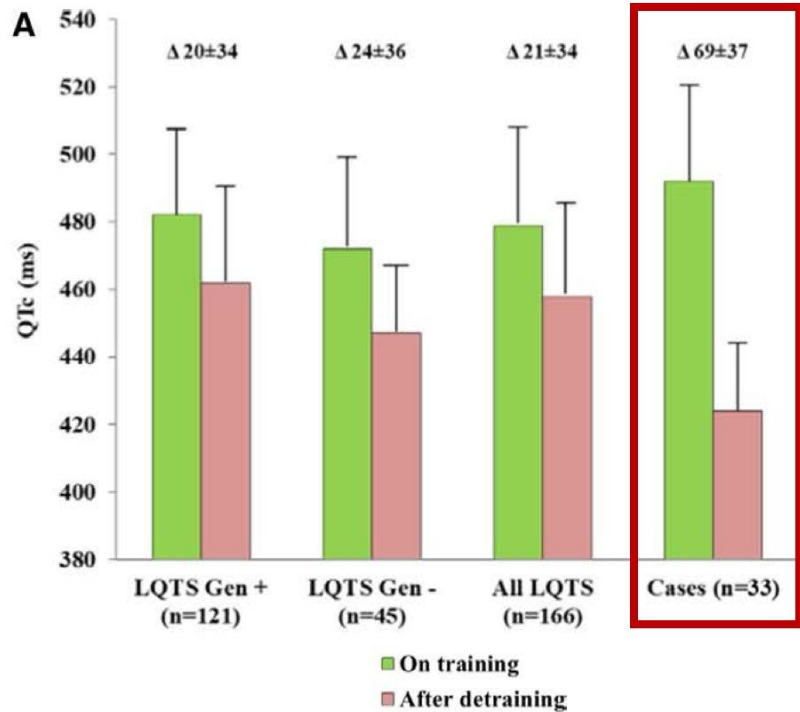
La contre-indication aux médicaments allongeant l'intervalle QT est formelle. Les sports de compétition sont contre-indiqués, sauf avis au cas par cas d'un centre spécialisé.

Circulation

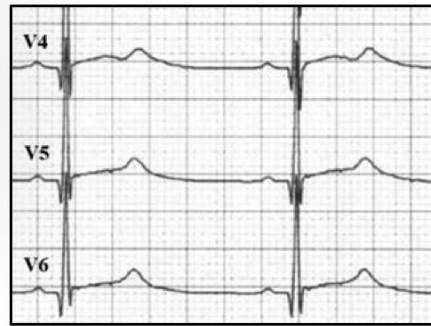
ORIGINAL RESEARCH ARTICLE

**Exercise Training-Induced Repolarization Abnormalities Masquerading as Congenital Long QT Syndrome**

Federica Dagradi, MD\*  
 Carla Spazzolini, DVM, MS\*  
 Silvia Castelletti, MD  
 Matteo Pedrazzini, BSc  
 Maria-Christina Kotta, MSc, PhD  
 Lia Crotti, MD, PhD  
 Peter J. Schwartz, MD



Dragadi F et al. Circulation (2020)



Désentrainement



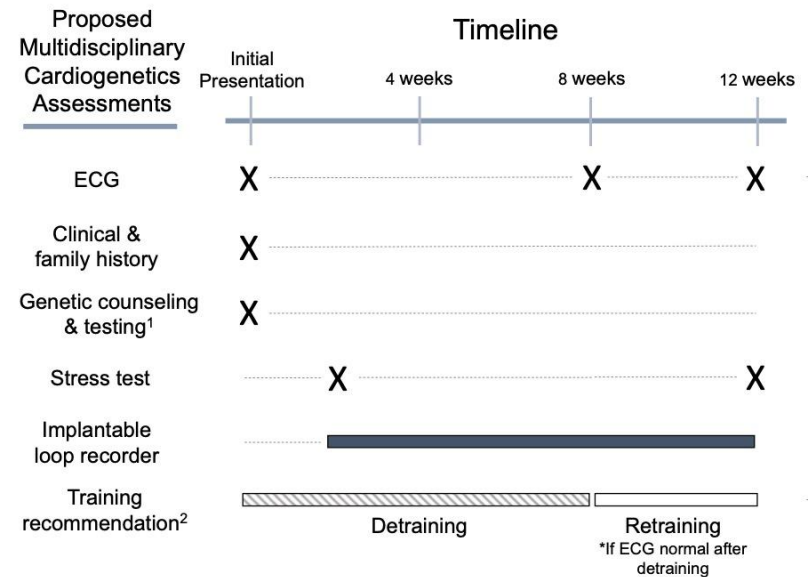
Réentrainement



**Diagnostic différentiel :  
 QT long acquis de l'athlète**

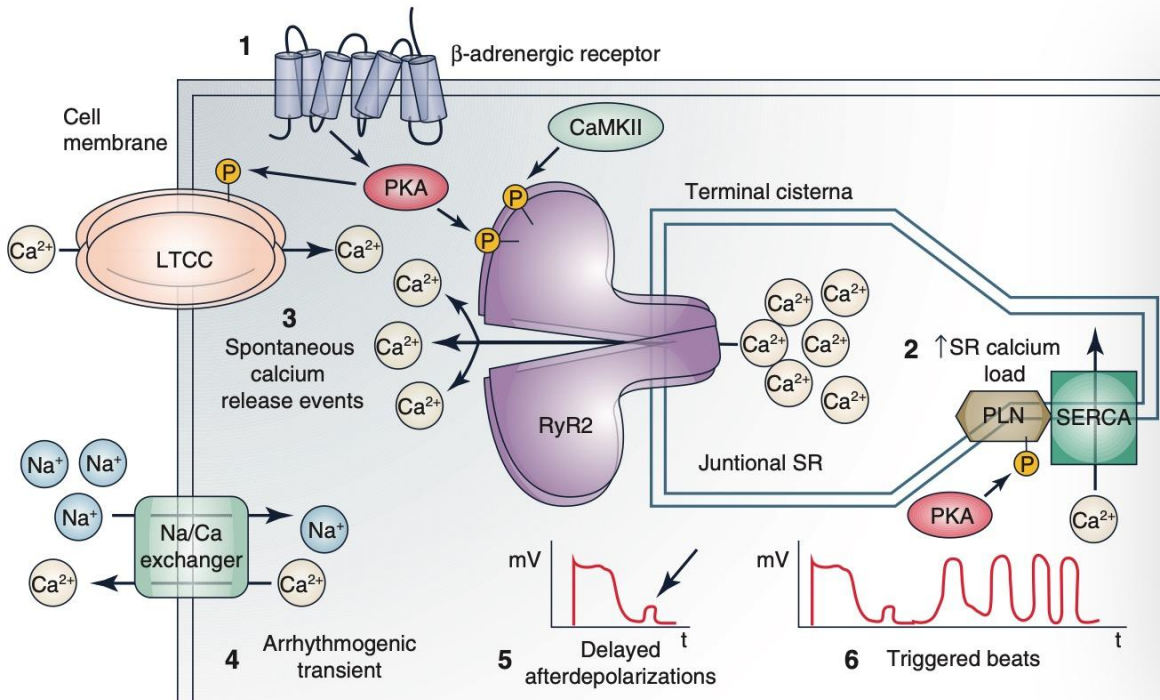
**Exercise-induced QTc prolongation and implications for military service members: A case series**

Austin Pagani, MGC, CGC,\*† Lydia D. Hellwig, PhD, ScM, CGC,\*†† Craig P. Dobson, MD,§  
 Brian N. Hughes, DO,‡ John P. Schacht, DO,§ Mark Haigney, MD||



Pagani A et al. HeartRhythm Case Reports (2023)

## GENERALITES



Wiekliniski MJ et al. J Physiol. 2020

- Canalopathie génétique **rare** (prévalence : 1/10 000)
- Dérégulation du **cycle calcique du cardiomyocyte**
- **Arythmies ventriculaires polymorphes en contexte adrénargique**
- **Pronostic sévère en l'absence de prise en charge**
- **Nombreuses possibilités thérapeutiques**

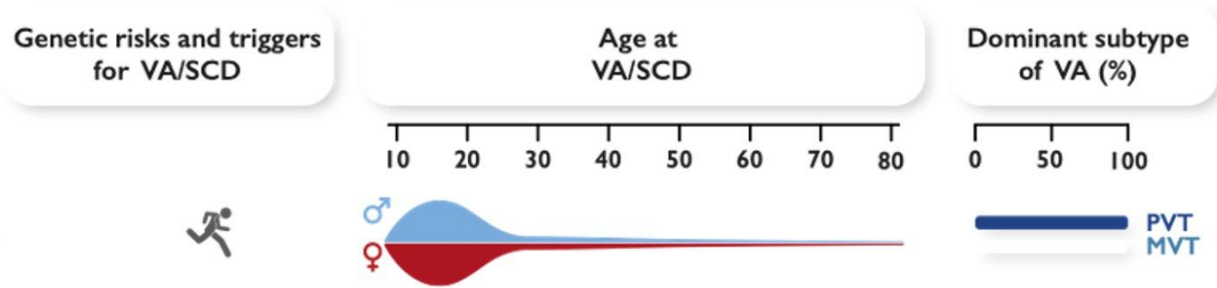
GENERALITES

**Tachycardie ventriculaire bidirectionnelle**  
(pas toujours objectivée)

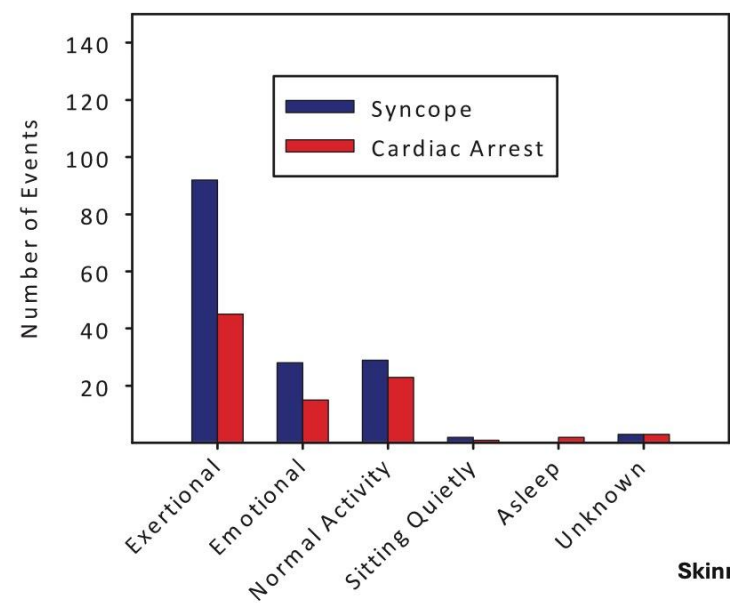


Autres TV polymorphes, fibrillation ventriculaire, ...

- Premiers épisodes dans l'enfance ou l'adolescence
- Principalement en contexte **adrénergique**



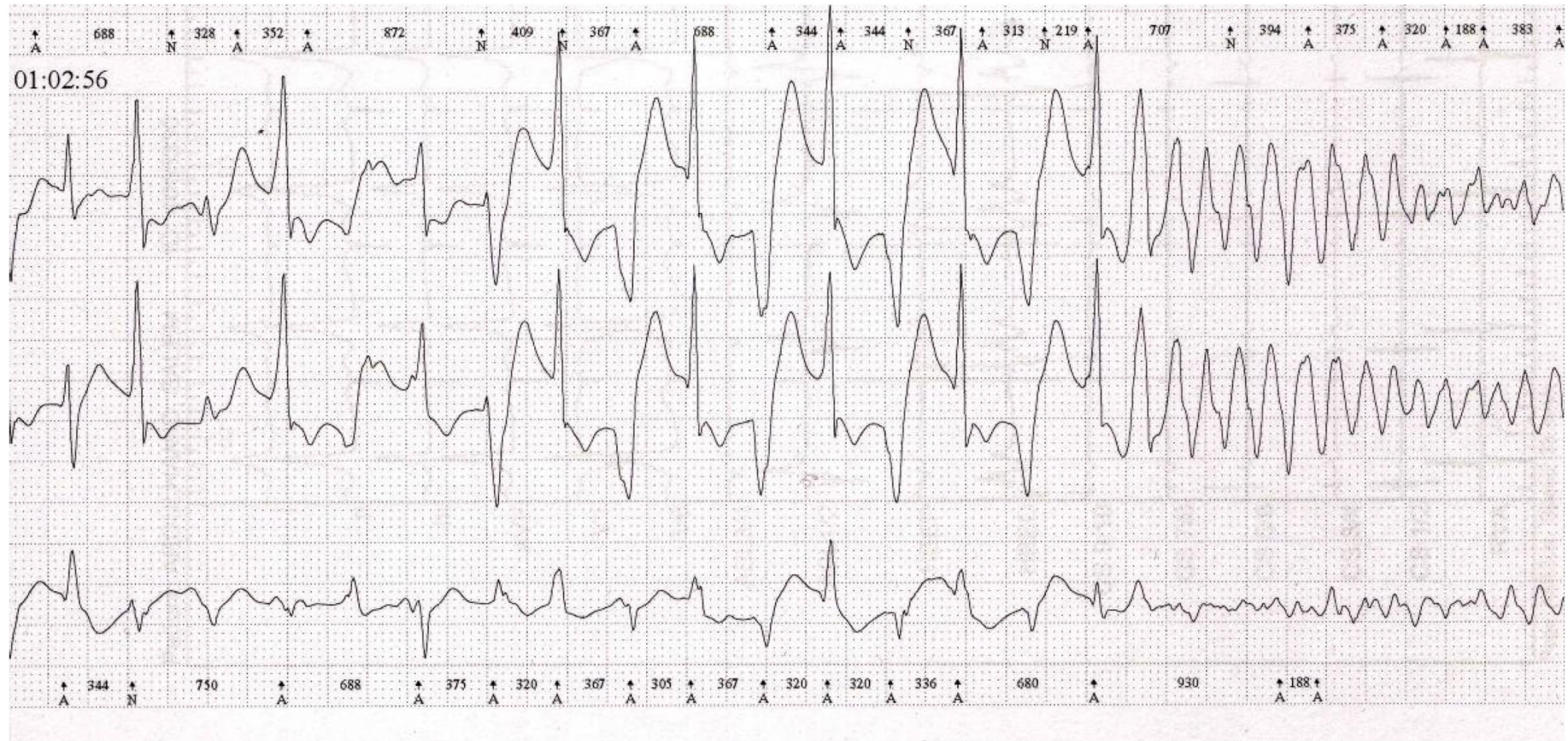
Typical event triggers: Exercise (swimming), emotion



**50% de mortalité avant 20 ans en l'absence de prise en charge**

Leenhardt et al. Circulation (1995)

## GENERALITES

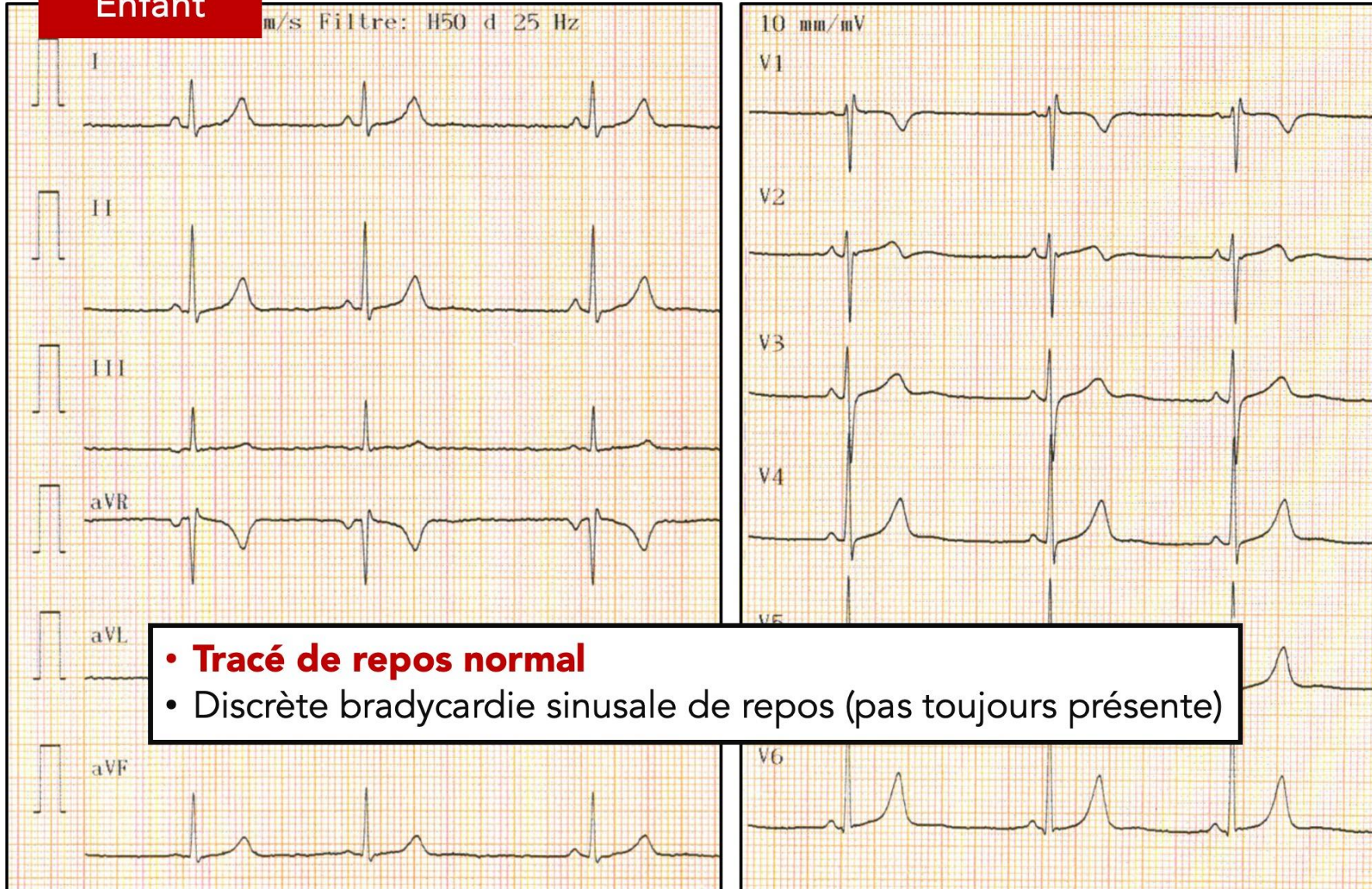


The Cardiac Society of Australia and New Zealand – CSANZ (2011)

## ▪ DIAGNOSTIC

Enfant

m/s Filtre: H50 d 25 Hz

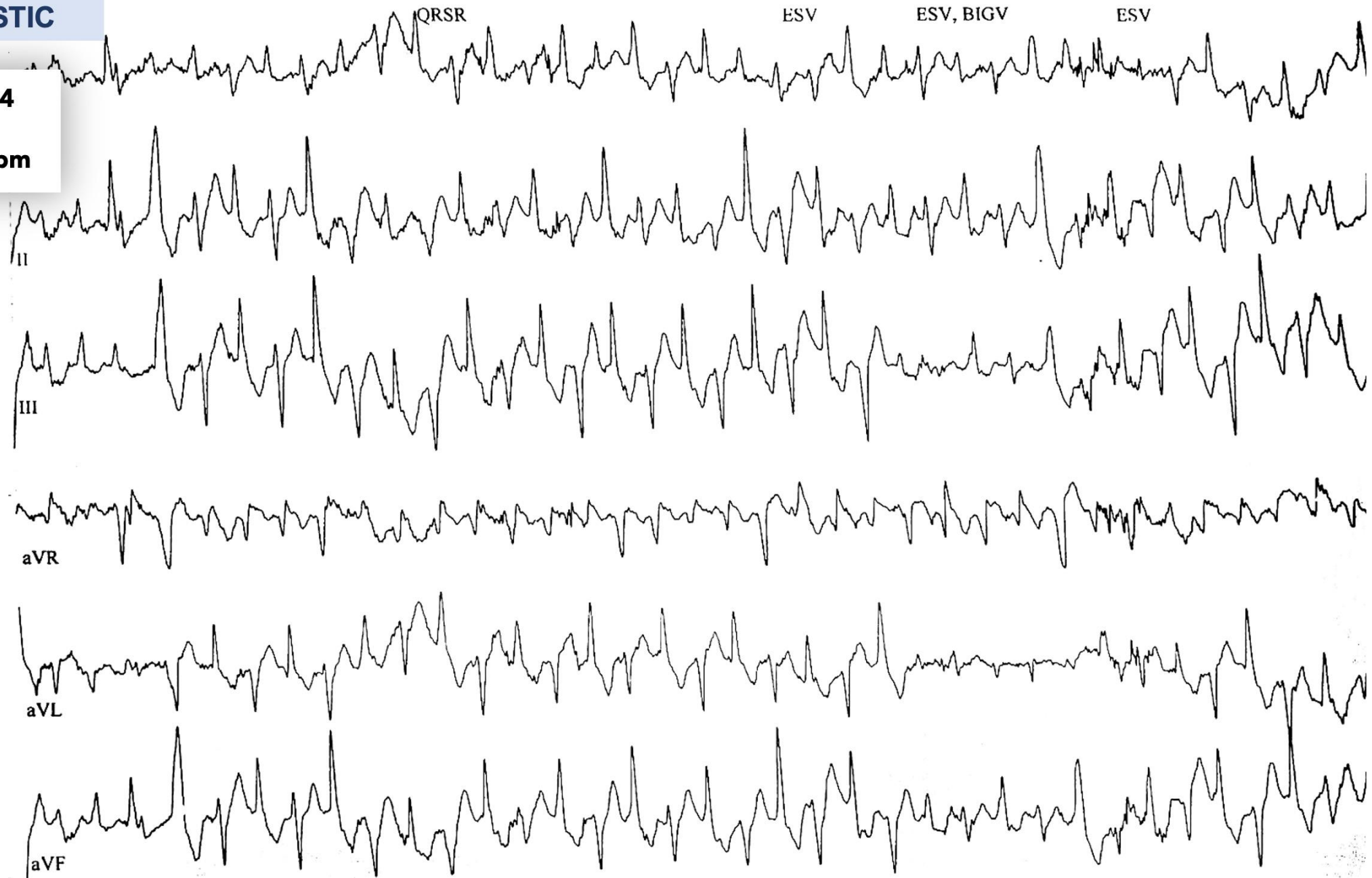


- **Tracé de repos normal**
- Discrète bradycardie sinusale de repos (pas toujours présente)

Les examens de  
**repos** sont  
**normaux**

## ■ DIAGNOSTIC

- Palier 4
- 55 W
- 129 bpm

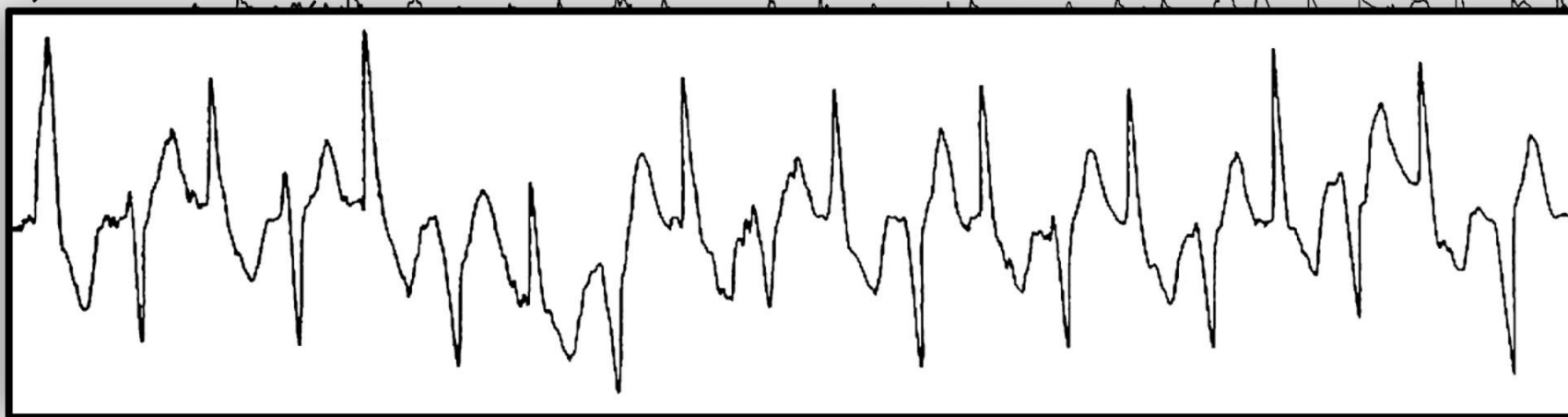


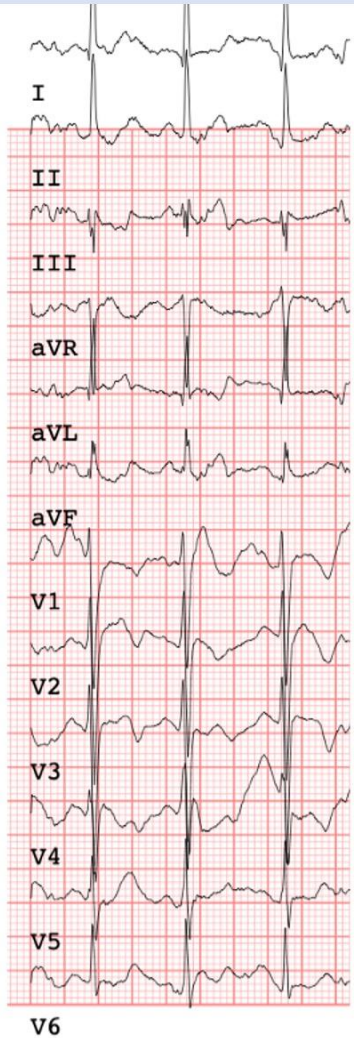
## ■ DIAGNOSTIC

- Palier 4
- 55 W
- 129 bpm

**TACHYCARDIE VENTRICULAIRE  
POLYMORPHE  
CATECHOLERGIQUE**

- ✓ TV **polymorphe** ou **bidirectionnelle**
- ✓ Contexte **adrénergique** (effort ou émotion)
- ✓ **Absence de cardiopathie** structurelle
- ✓ (Après 40 ans : *exclure coronaropathie*)

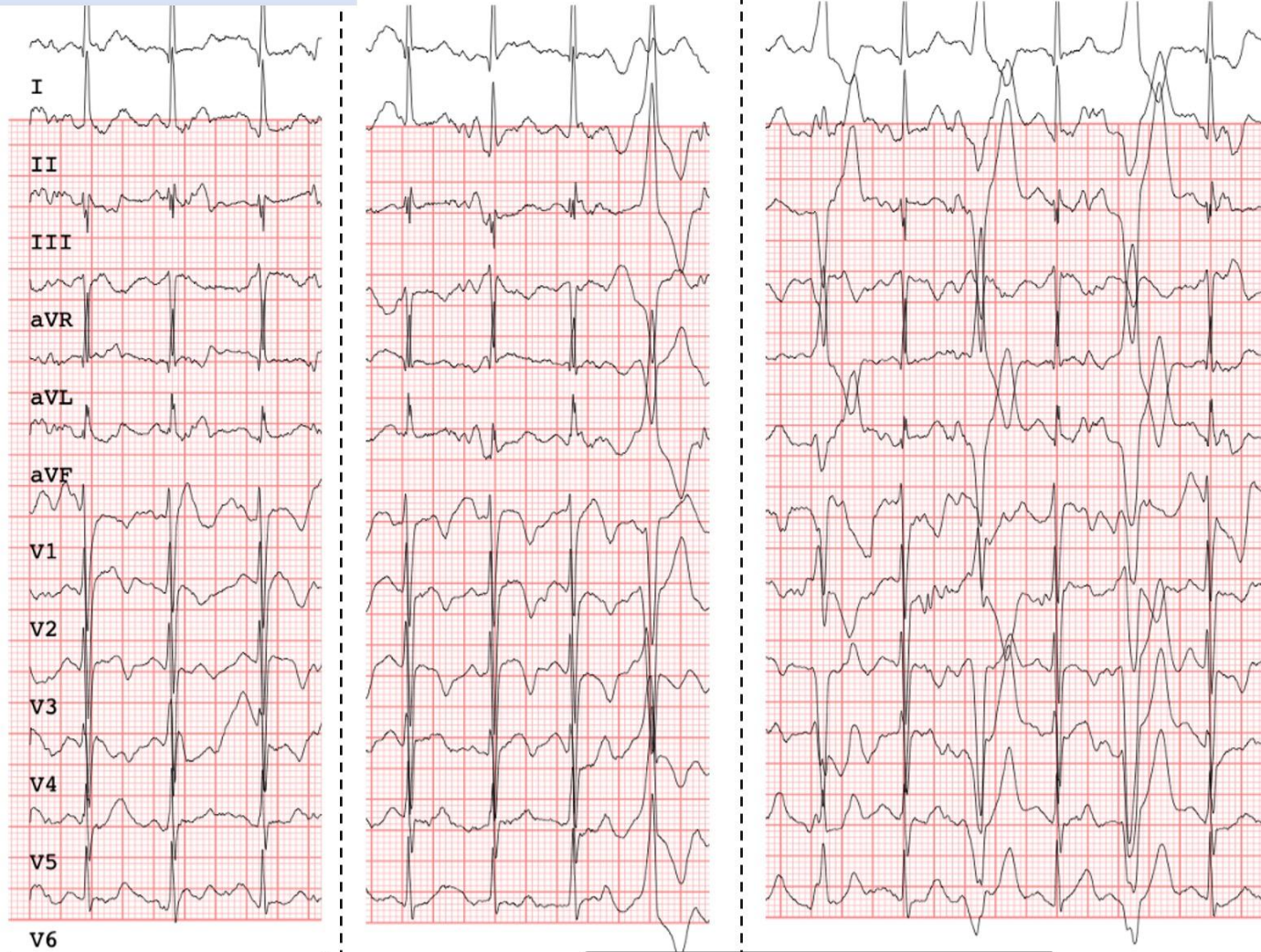


**DIAGNOSTIC**

Palier 3 - 105 bpm 40W

- Pas de modification

## ■ DIAGNOSTIC



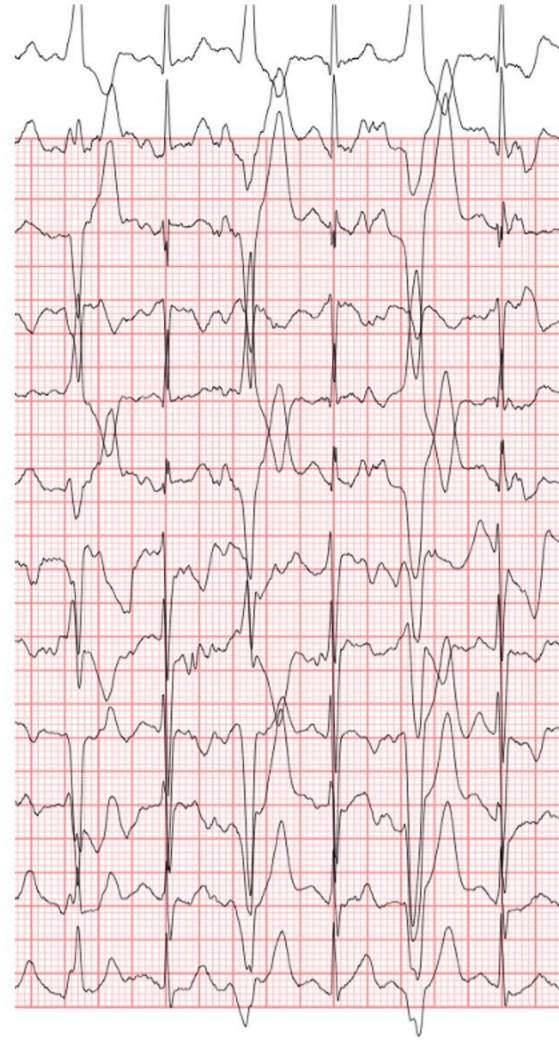
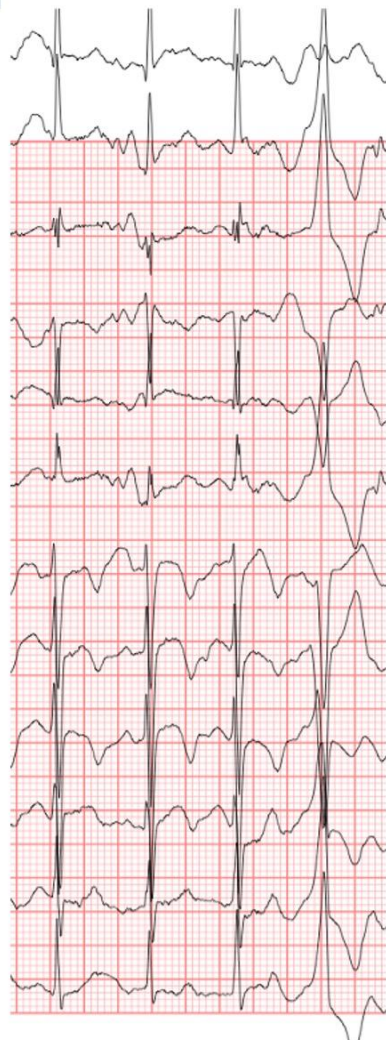
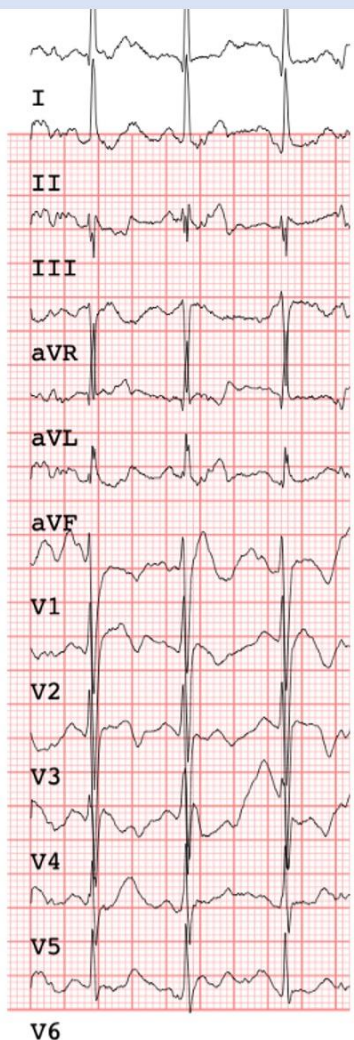
Palier 3 - 105 bpm 40W

- Pas de modification

Palier 4 – 110 puis 120 bpm 50W

- Fréquence seuil d'apparition des ESV (variable)
- ESV d'abord isolées (retard gauche en général)
- Puis polymorphes, bi/trigémínées

## DIAGNOSTIC



Palier 3 - 105 bpm 40W

Palier 4 - 110 puis 120 bpm 50W

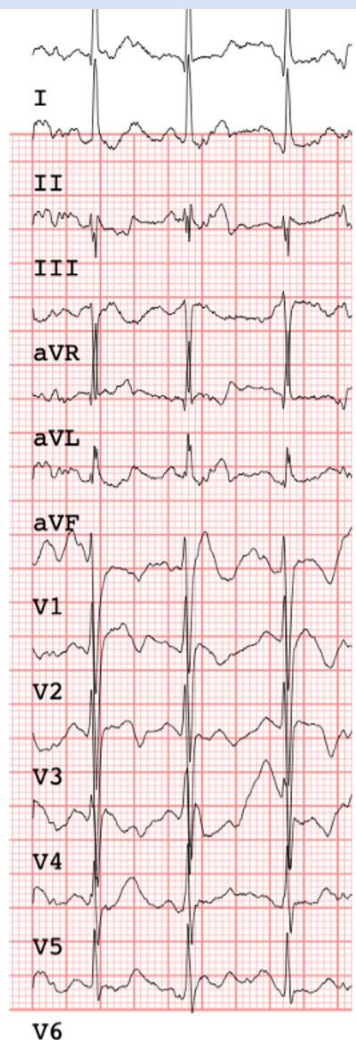
Palier 5 - 116 bpm 60W

- Pas de modification

- Fréquence seuil d'apparition des ESV (variable)
- ESV d'abord isolées (retard gauche en général)
- Puis polymorphes, bi/trigémínées

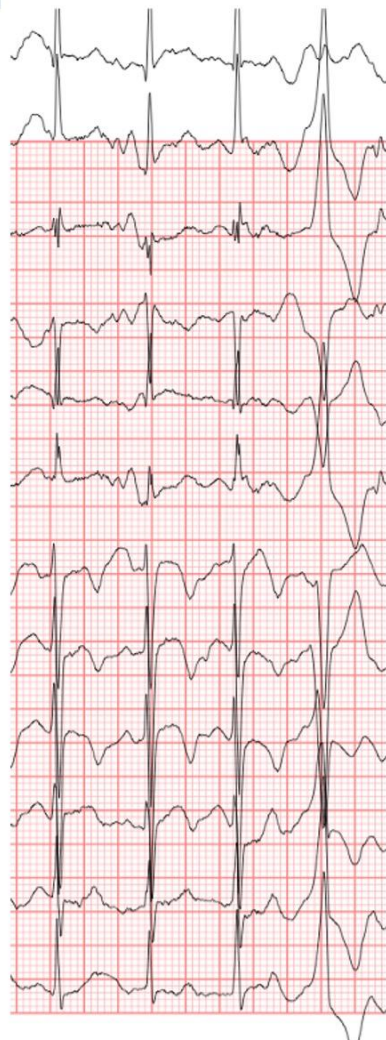
- Doublets bidirectionnels

## DIAGNOSTIC



Palier 3 - 105 bpm 40W

- Pas de modification



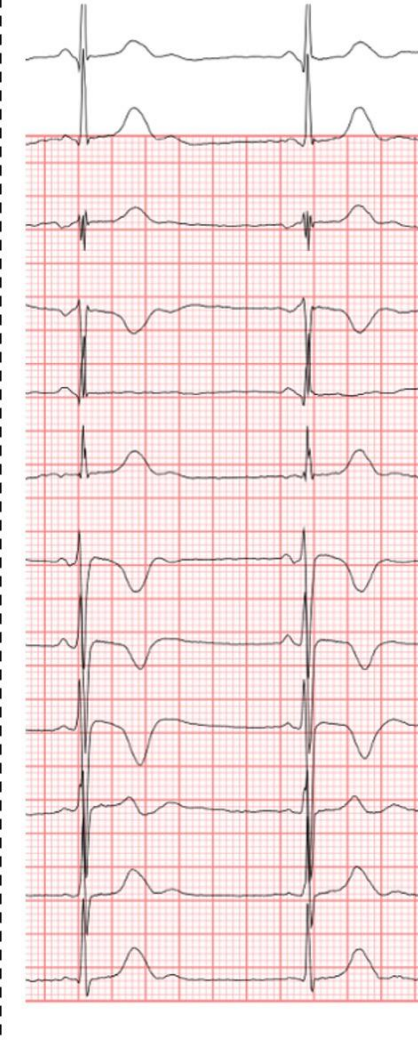
Palier 4 - 110 puis 120 bpm 50W

- Fréquence seuil d'apparition des ESV (variable)
- ESV d'abord isolées (retard gauche en général)
- Puis polymorphes, bi/trigémées



Palier 5 - 116 bpm 60W

- Doublets bidirectionnels



Récupération

- Disparition rapide des ESV à l'arrêt de l'effort

DIAGNOSTIC

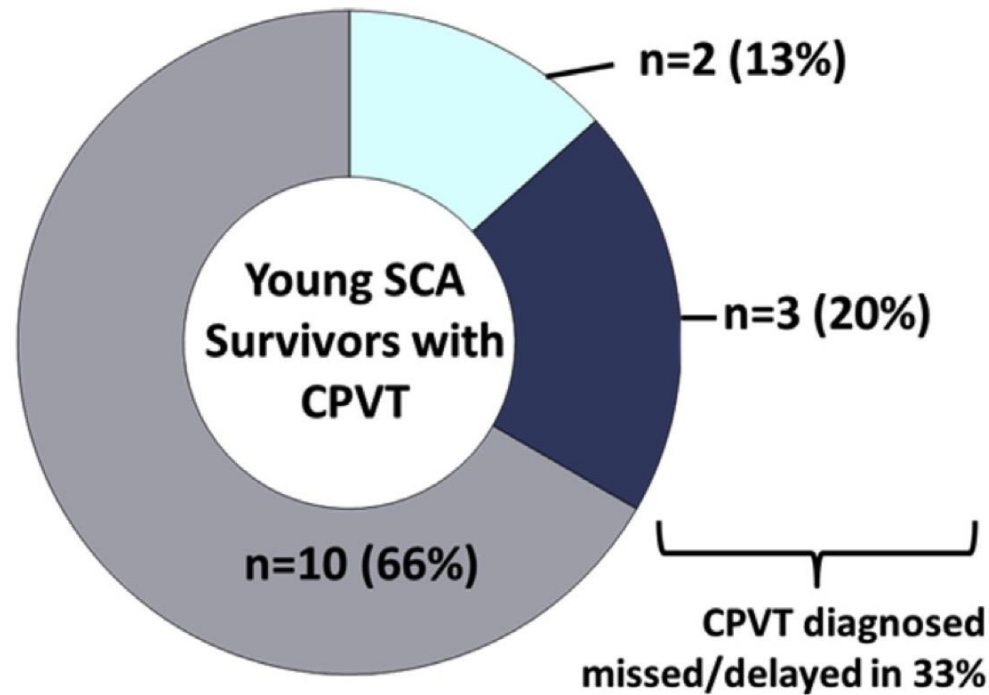
It is recommended that CPVT is diagnosed in the presence of a structurally normal heart, normal ECG, and exercise- or emotion-induced bidirectional, or PVT.	I	C
It is recommended that CPVT is diagnosed in patients who are carriers of a mutation in disease-causing genes.	I	C

Zeppenfeld K et al. EHJ. 2022

Exercise testing oversights underlie missed and delayed diagnosis of catecholaminergic polymorphic ventricular tachycardia in young sudden cardiac arrest survivors

John R. Giudicessi, MD, PhD,\* Michael J. Ackerman, MD, PhD<sup>†‡§</sup>

L'épreuve d'effort est un examen **indispensable** après un arrêt cardiaque ou une syncope en contexte **adrénergique**



- Delayed diagnosis (no post-SCA EST)
- Missed diagnosis (overlooked EST finding)
- CPVT diagnosed < 6 months post-SCA

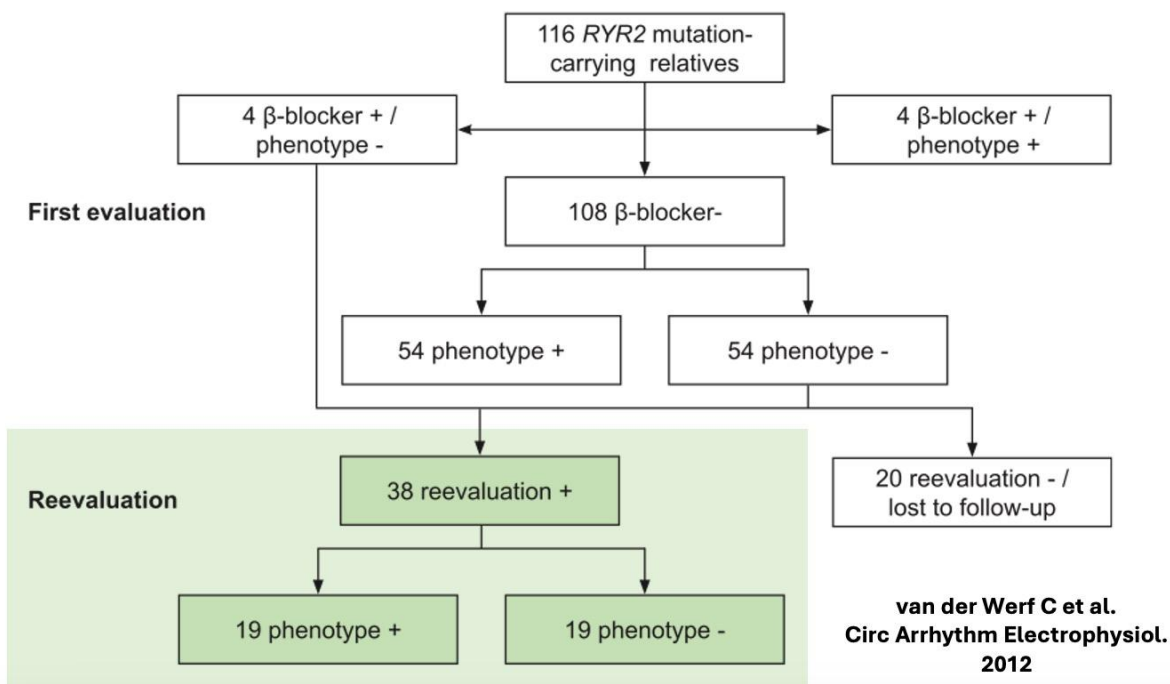
Giudicessi JR & Ackerman MJ. Heart Rhythm. 2019

DIAGNOSTIC

**Familial Evaluation in Catecholaminergic Polymorphic Ventricular Tachycardia**

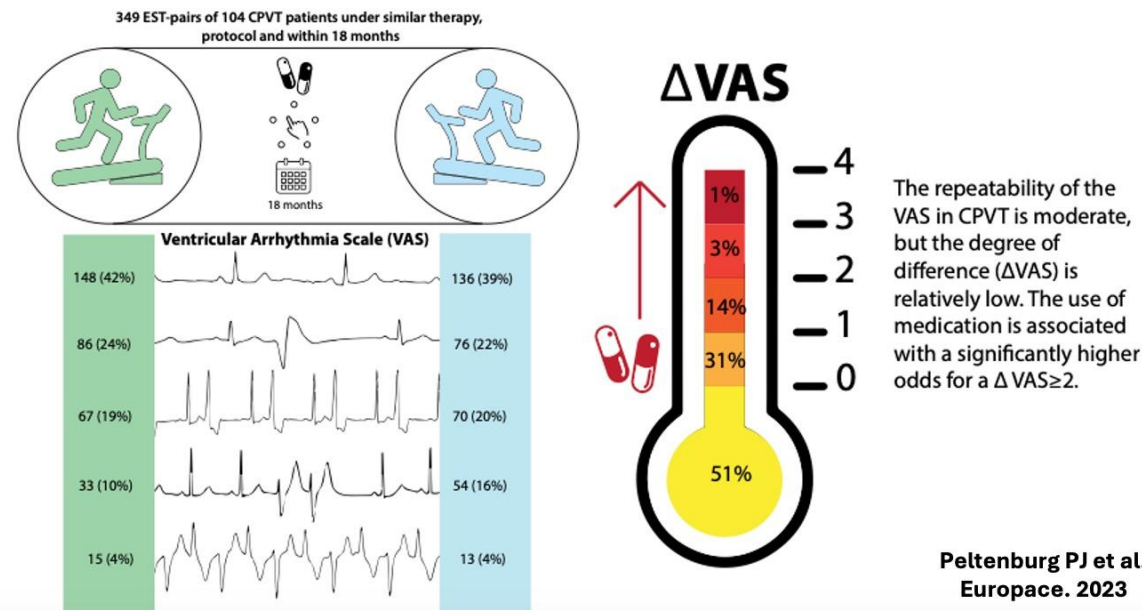
**Disease Penetrance and Expression in Cardiac Ryanodine Receptor Mutation-Carrying Relatives**

Christian van der Werf, MD; Ineke Nederend, MSc; Nynke Hofman, MSc; Nan van Geloven, MSc; Corné Ebink, MD; Ingrid M.E. Frohn-Mulder, MD; A. Marco W. Alings, MD, PhD; Hans A. Bosker, MD, PhD; Frank A. Bracke, MD, PhD; Freek van den Heuvel, MD, PhD; Reinier A. Waalewijn, MD, PhD; Hennie Bikker, PhD; J. Peter van Tintelen, MD, PhD; Zahurul A. Bhuiyan, MD, PhD; Maarten P. van den Berg, MD, PhD; Arthur A.M. Wilde, MD, PhD



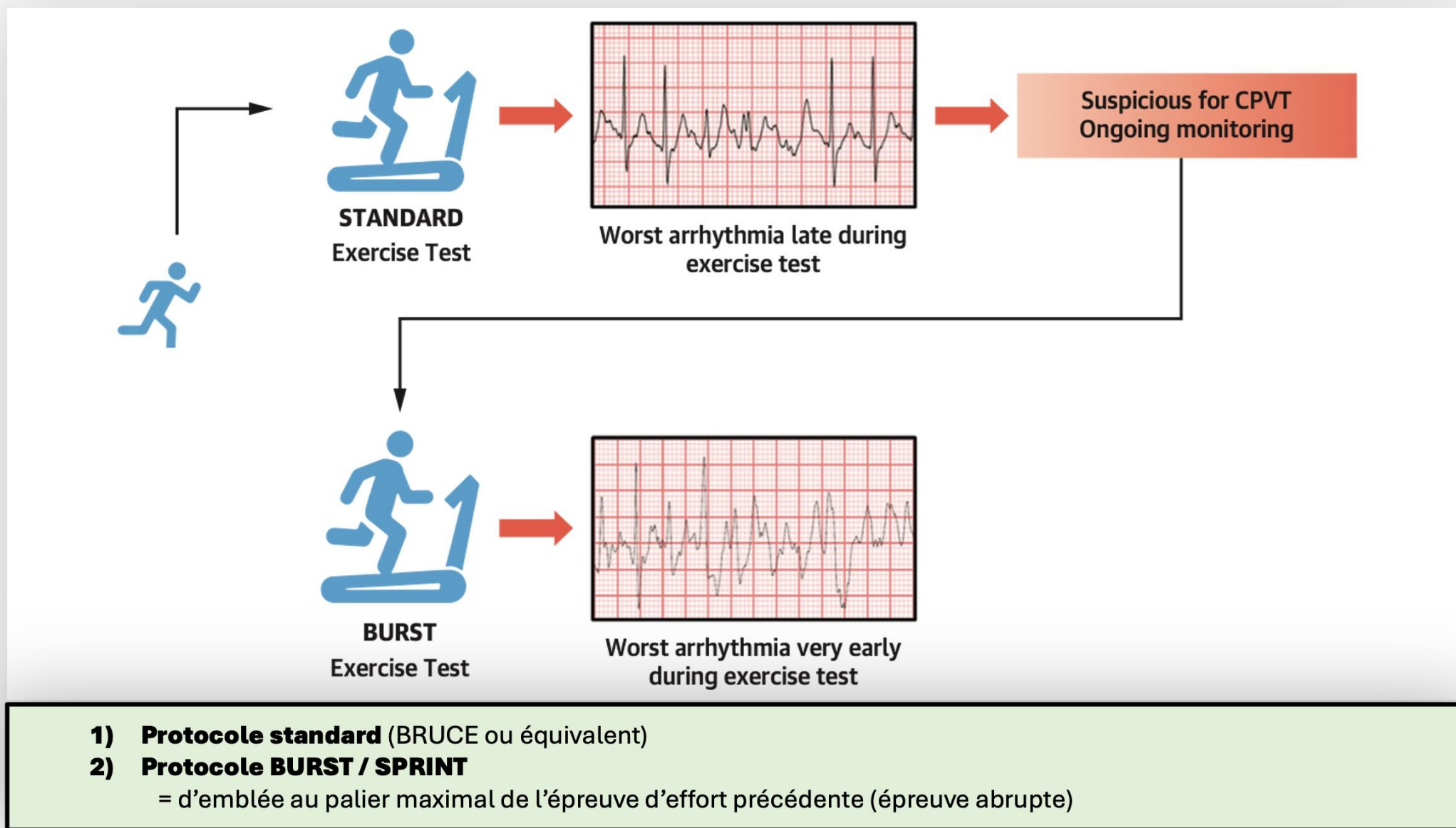
**Repeatability of ventricular arrhythmia characteristics on the exercise-stress test in RYR2-mediated catecholaminergic polymorphic ventricular tachycardia**

Puck J. Peltenburg<sup>1,2</sup>, Sanjeev N.J. Pultoo<sup>1</sup>, Kathryn E. Tobert<sup>3</sup>, J. Martijn Bos<sup>3</sup>, Krystien V.V. Lieve<sup>1</sup>, Michael Tanck<sup>1</sup>, Sally-Ann B. Clur<sup>2</sup>, Nico A. Blom<sup>2,4</sup>, Michael J. Ackerman<sup>3†</sup>, Arthur A.M. Wilde<sup>1†</sup>, and Christian van der Werf<sup>1\*†</sup>

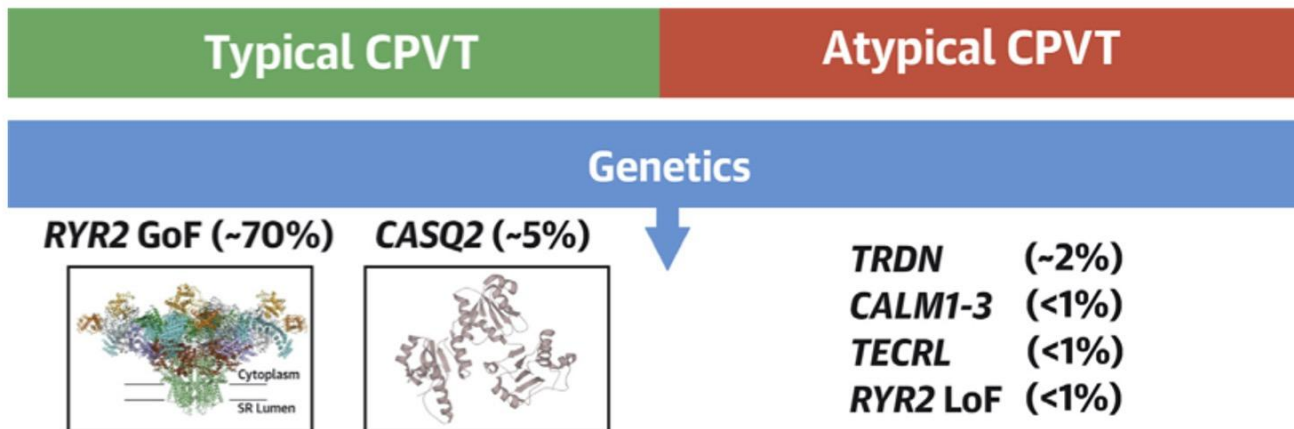


- **Reproductibilité modérée** du test = sa négativité ne permet pas d'exclure le diagnostic
- Ne pas hésiter à le répéter avec protocole **BURST / SPRINT**

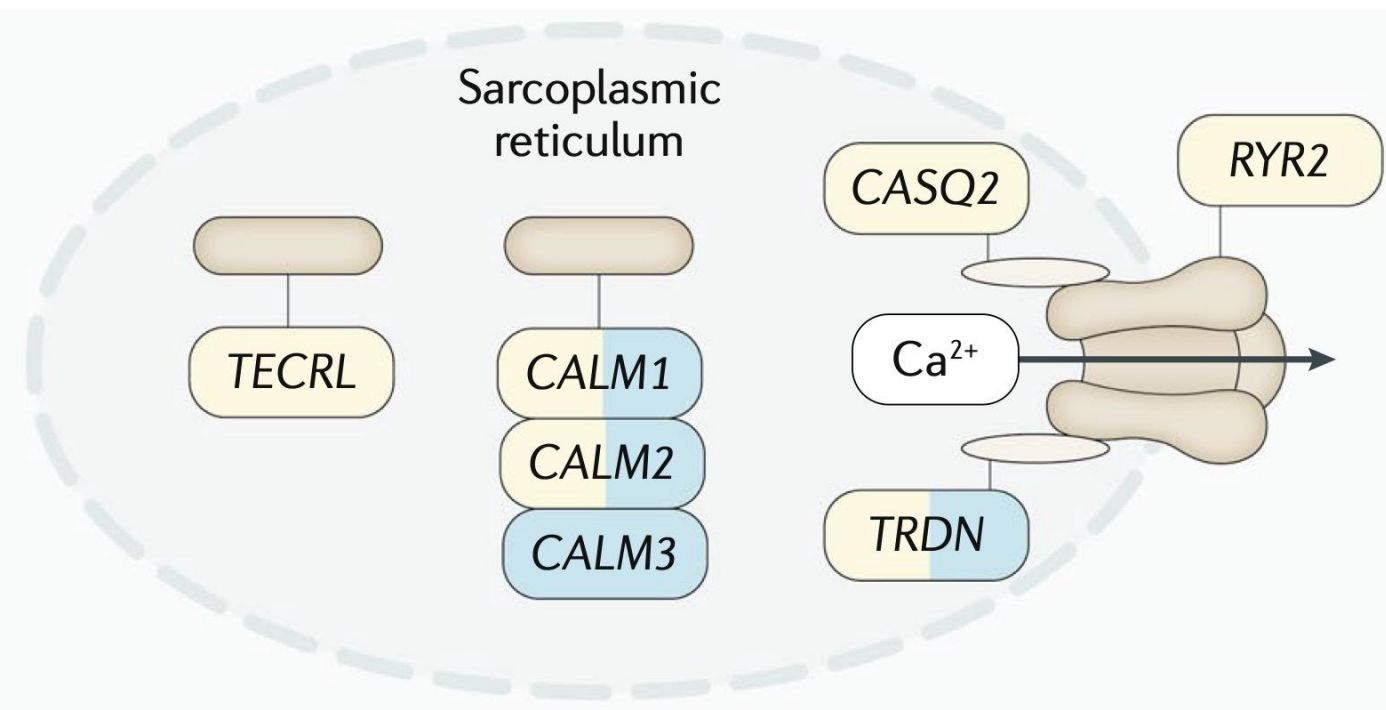
## ▪ DIAGNOSTIC



GENETIQUE



Priori SG et al. JACC 2021



Schwartz PJ et al. Nature Reviews Disease Primers 2020

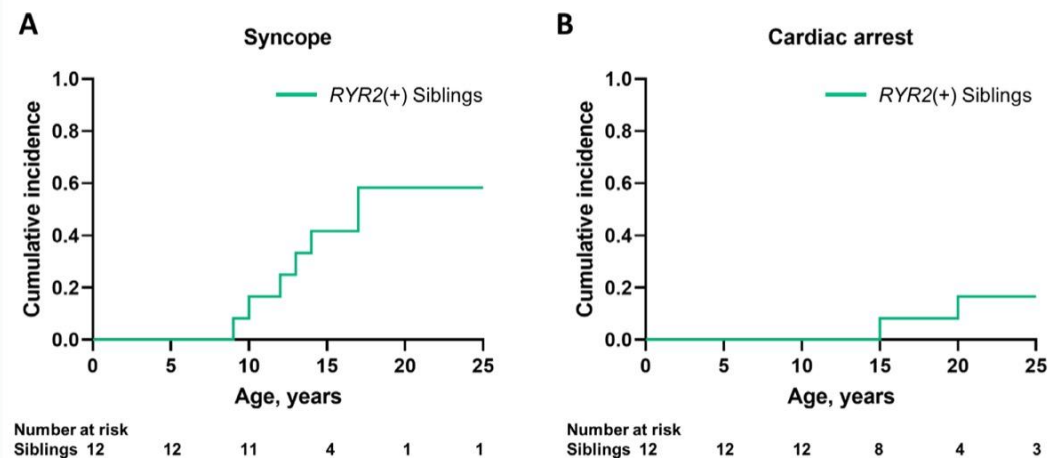
**En pratique**

**RYR2, CASQ2, CALM1-3, TRDN, TECRL**

**Phénotopies : KCNJ2, PKP2, SCN5A**

**Dépistage des apparentés au 1<sup>er</sup> degré**

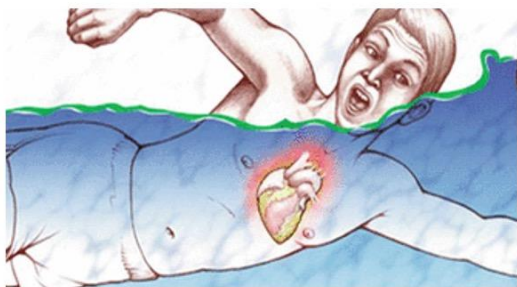
- **Indispensable**
- **La présence du variant classe 4 ou 5 permet de poser le diagnostic (instauration traitement et suivi)**



Shimamoto K et al. Heart 2022

PRISE EN CHARGE

Eviction des triggers adrénergiques



Typical event triggers: Exercise (swimming), emotion



MAYO CLINIC  
Skinner JR et al.  
Heart, Lung and Circulation 2019

**NADOLOL**

PROPRANOLOL possible en alternative au NADOLOL

**FLECAINIDE**

**Tout faire (NADOLOL, FLECAINE, sympathectomie,...) pour essayer d'éviter d'implanter un DAI en prévention primaire dans la CPVT**

**SYMPATHECTOMIE**

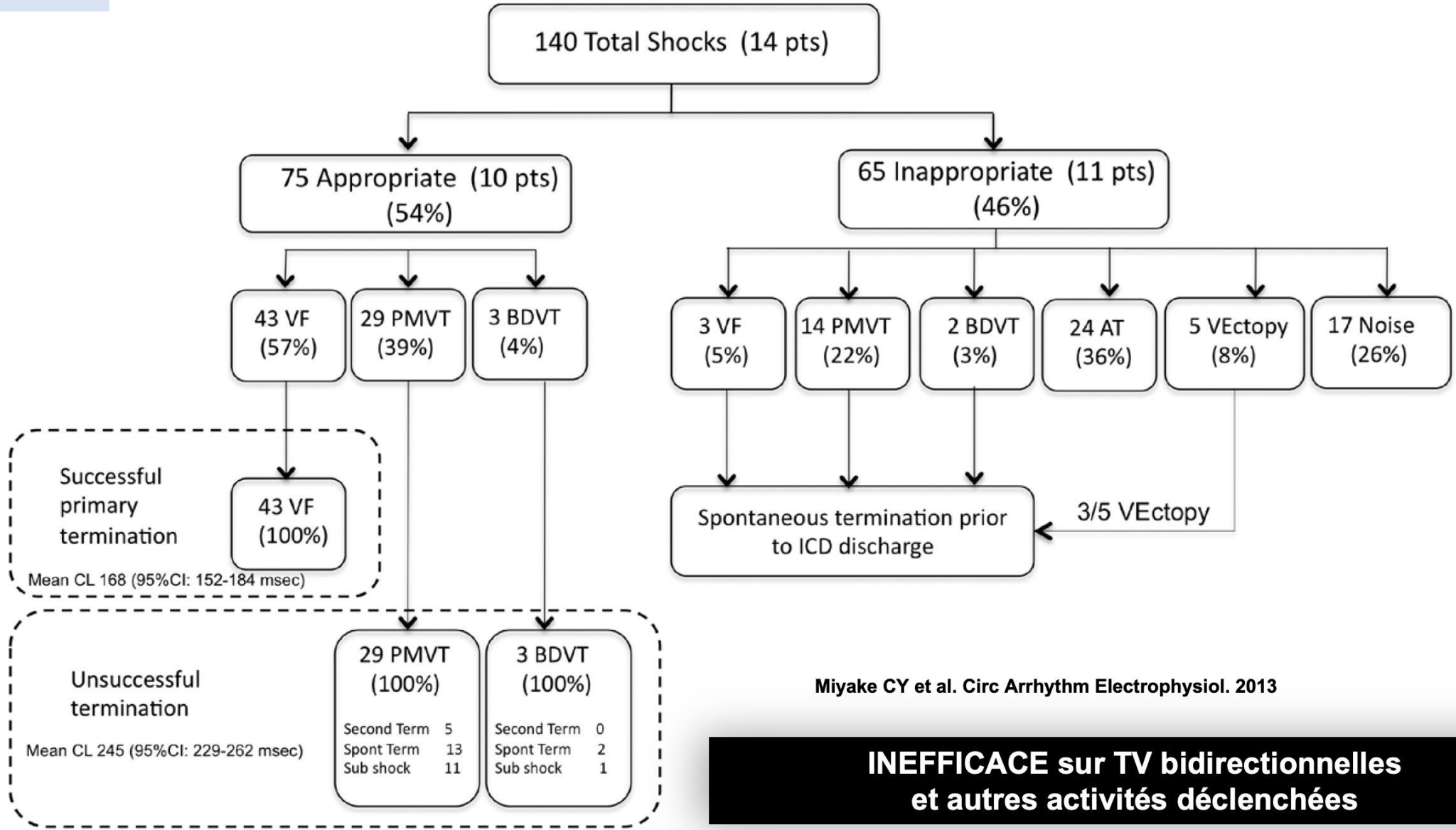
Witt CM et al.  
Europace (2017) 19, 1075–1083

Contrôle de l'efficacité via **EPREUVE D'EFFORT**

**OBJECTIFS** (les deux objectifs doivent être atteints) :

- 1. FC maximale d'effort < 70% de la FMT**
- 2. Absence d'ESV polymorphes, doublets, triplets ou TVNS d'effort**  
*(dans l'idéal absence d'ESV d'effort)*

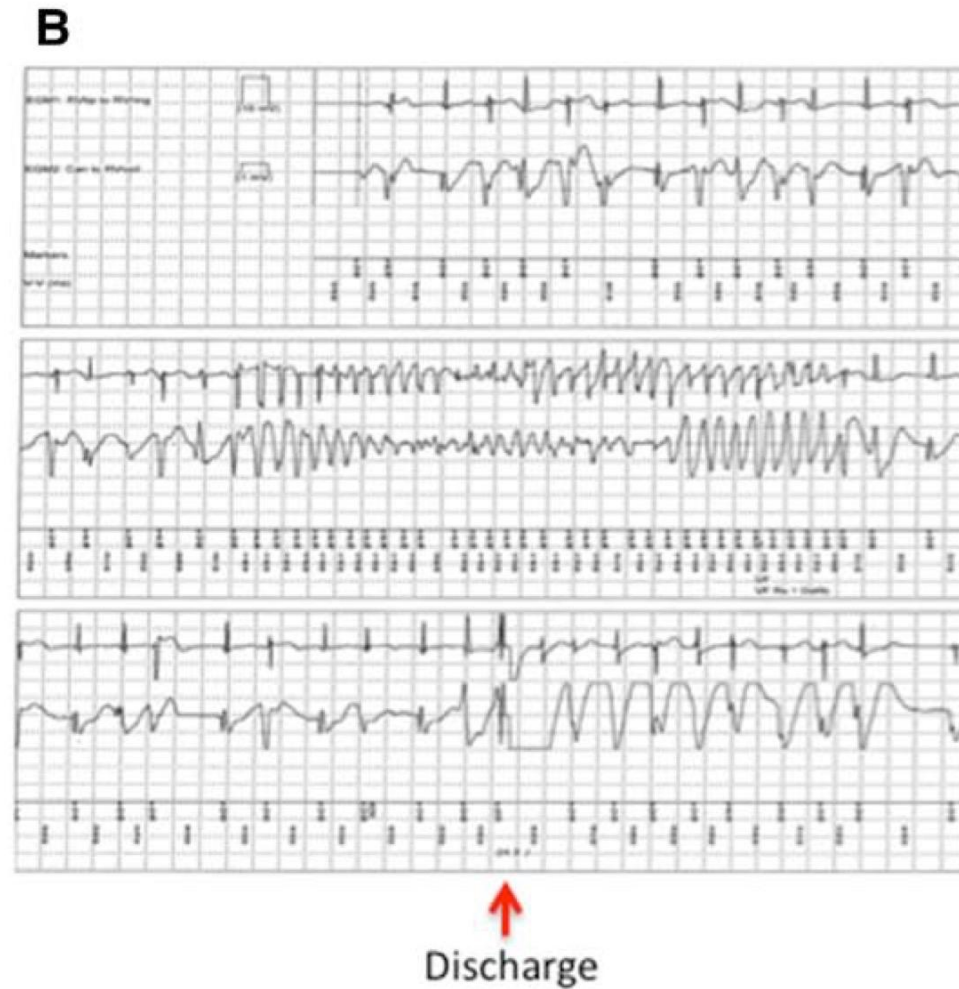
ET LE DAI ?



Miyake CY et al. Circ Arrhythm Electrophysiol. 2013

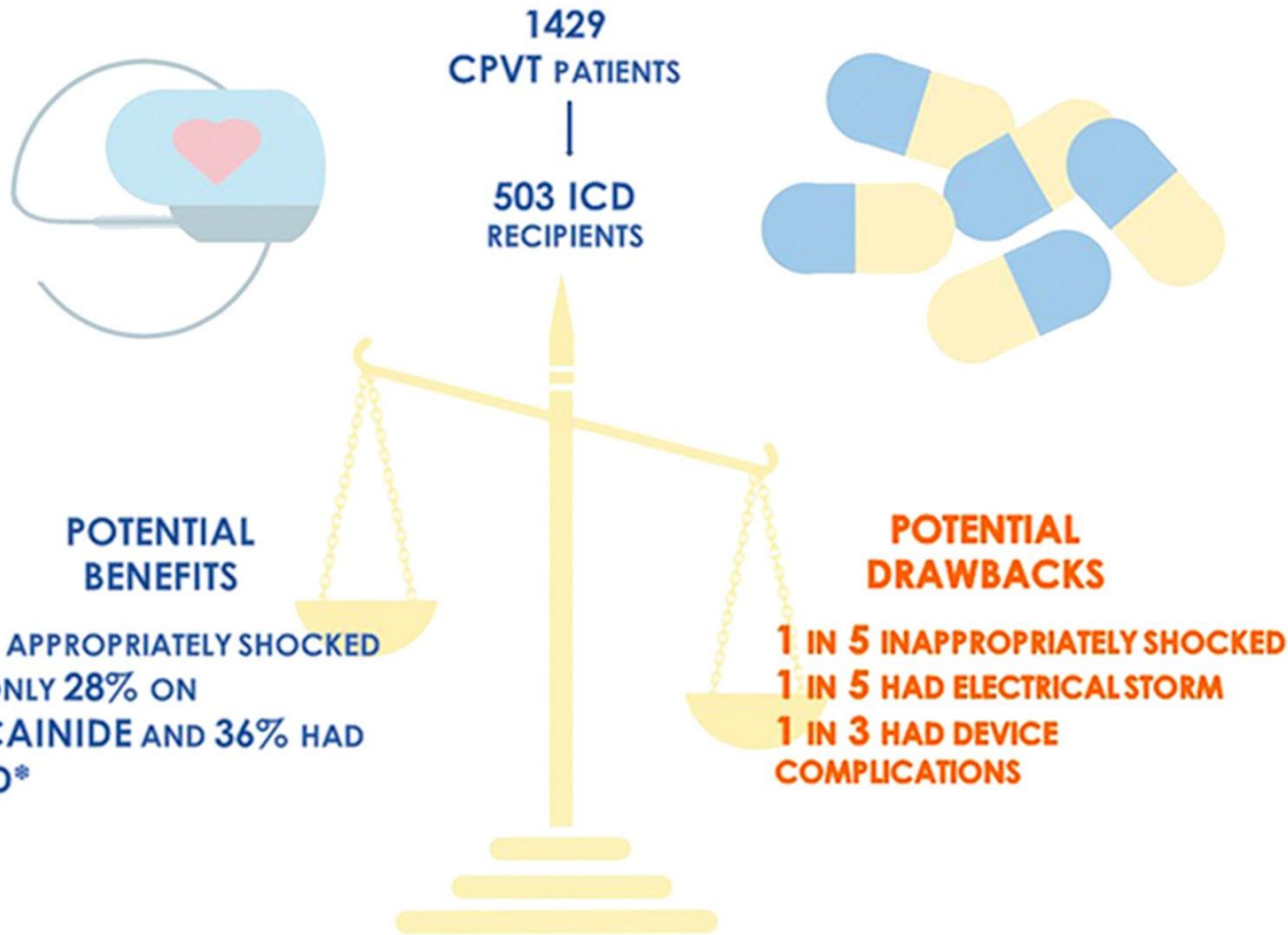
**INEFFICACE sur TV bidirectionnelles et autres activités déclenchées**

- ET LE DAI ?

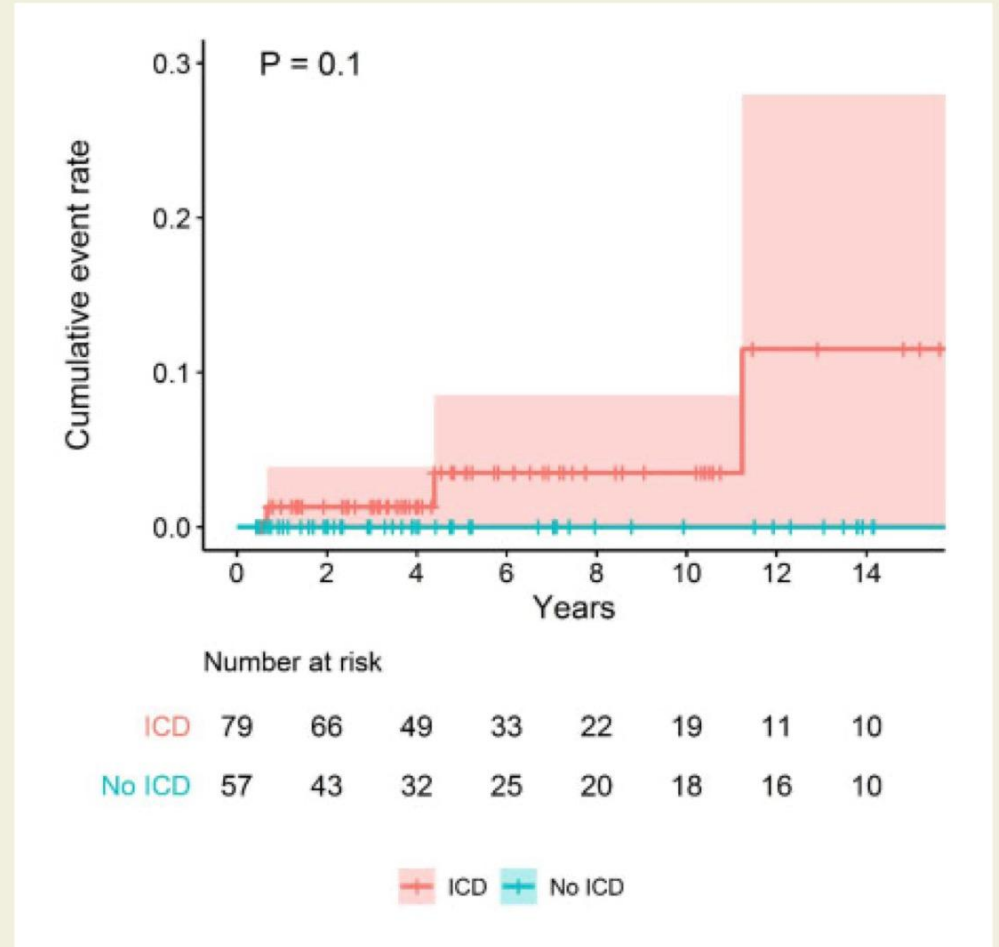


**Figure 1. A**, Inappropriate shock caused by atrial tachycardia resulting in a more malignant ventricular arrhythmia. **Top** electrogram (EGM): RVtip to RVring, **bottom** EGM: HVA to RVring. **B**, Inappropriate shock caused by spontaneous termination before implantable cardioverter-defibrillator discharge. **Top** EGM: RVtip to RVring, **bottom** EGM: Can to RV coil. **C**, Appropriate but unsuccessful shock for polymorphic ventricular tachycardia. **Top** EGM: RVtip to RVring, **bottom** EGM: Can to RVcoil.

ET LE DAI ?



Roston TM et al. Heart Rhythm 2018



**Take home figure** Time-to-event curve for sudden cardiac death. In previously undiagnosed patients with catecholaminergic polymorphic ventricular tachycardia who presented with sudden cardiac arrest, an implantable cardioverter-defibrillator was not associated with improved survival.

## ET LE DAI ?

HeartRhythm



The Official Journal of the Heart Rhythm Society, The Cardiac Electrophysiology Society, and The Pediatric &amp; Congenital Electrophysiology Society

An international multicenter cohort study on implantable cardioverter-defibrillators for the treatment of symptomatic children with catecholaminergic polymorphic ventricular tachycardia

**CONCLUSION** SCD events occurred only in the no ICD group and in those not on optimal medical therapy. Patients with ICD had a high risk of appropriate and inappropriate shocks, which may be reduced with appropriate device programming. Severe ICD complications were common, and risks vs benefits of ICDs need to be considered.

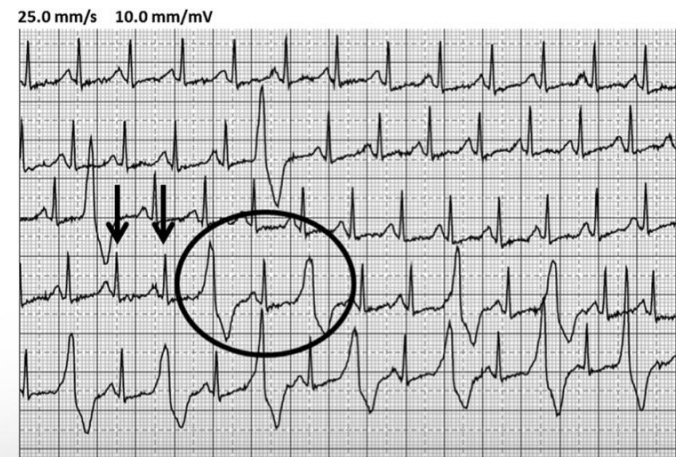
Patient no.	Circumstance during the event	Age at the event (y)	Proband status	<i>RYR2</i> variant classification	Medications/therapies during the event (daily dosage)	Compliance
1	The patient was found unresponsive after sports training. The patients forgot to take medications on the day of the event	17	Proband	VUS	Metoprolol (0.86 mg/kg)	Noncompliant
2	The event occurred immediately after the patient left work. The patient was stressed immediately before the event. The patient discontinued taking medications ~13 mo before the event	36	Proband	Pathogenic	None	Noncompliant
3	The event occurred at home while at rest. The patient was an avid sports player. Compliance at the time of the event is unknown; however, the patient had a history of being difficult to instruct regarding medications	16	Proband	Pathogenic	Propranolol (1.59 mg/kg)	Unknown
4	The event occurred during sports training. The patient had discontinued taking flecainide before the event	22	Proband	Pathogenic	Nadolol (2 mg/kg); flecainide (2.5 mg/kg)	Noncompliant
5	The patient experienced severe VAs during the hospital stay before the event. Verapamil boluses were tried during VT episodes without success. Before initiating oral propranolol, the patient was treated with esmolol intravenous therapy and labetalol intravenous therapy. During hospitalization, the patient was under sedation with pentobarbital. LCSD was clinically ineffective, and VA persisted. Right-sided CSD was planned 6 d after LCSD; however, the patient died before planned RCSD	8	Proband	Pathogenic	Propranolol (dose unknown); flecainide (2.4 mg/kg). LCSD performed 5 d before death to manage VT. Extent of denervation not known	Compliant
6	The event occurred during sports training. The patient had not been taking medications as prescribed. The patient previously had an ICD; however, the ICD was explanted 7 mo before the event	18	Proband	VUS	Nadolol (0.25 mg/kg); propafenone (2.8 mg/kg). LCSD (complete denervation) performed 8 y before death to manage VT	Noncompliant
7	The patient was found unresponsive after an emotionally stressful event. The patient had a history of being compliant with medications	27	Nonproband	Pathogenic	Nadolol (1.27 mg/kg); flecainide (1.59 mg/kg)	Probably compliant

## QUE NOUS DIT LA LITTERATURE ?

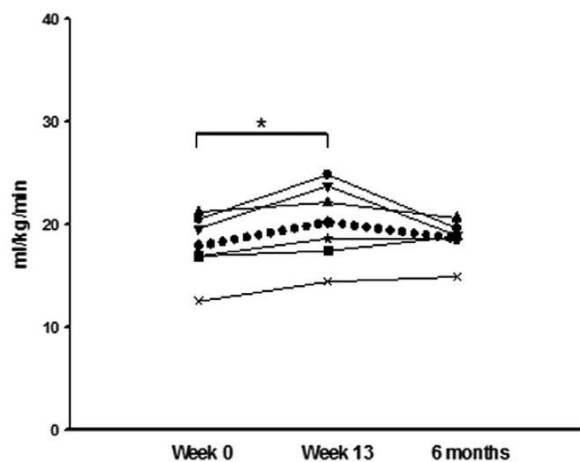
### Effects of Individualized Exercise Training in Patients With Catecholaminergic Polymorphic Ventricular Tachycardia Type 1

Ravinea Manotheepan, MD<sup>a,b,\*</sup>, Jørg Saberniak, MD<sup>c,d</sup>, Tore K. Danielsen, MD<sup>a,b</sup>, Thor Edvardsen, MD, Dr Med<sup>c,d</sup>, Ivar Sjaastad, MD, Dr Med<sup>a,b</sup>, Kristina H. Haugaa, MD, PhD<sup>c,d</sup>, and Mathis K. Stokke, MD, PhD<sup>a,b,e</sup>

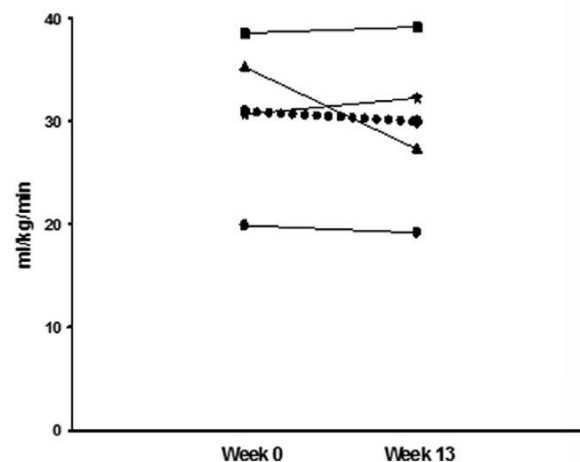
Am J Cardiol (2014)



**A** ET patients - individual and mean  $VO_{2max}$  Values



**B** SED patients - individual and mean  $VO_{2max}$  Values



Changes in threshold HR

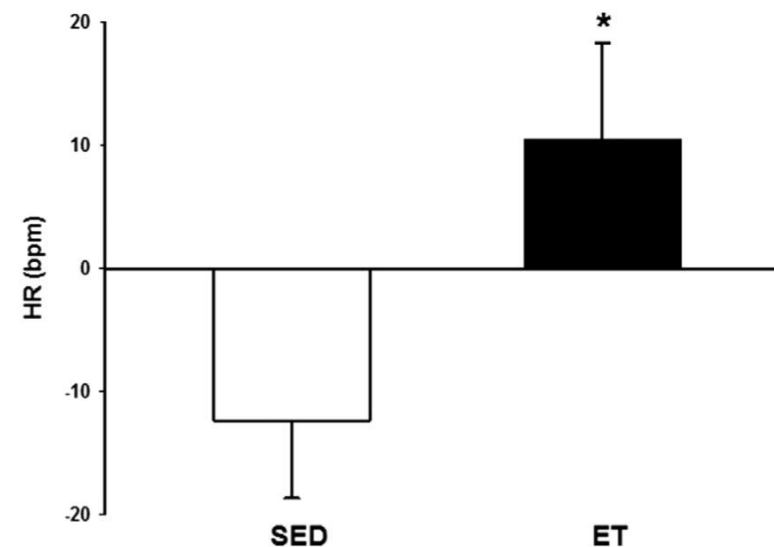


Figure 4. Comparison of the development in threshold HR for VA during the study period in ET and SED patients. \* $p < 0.05$ .

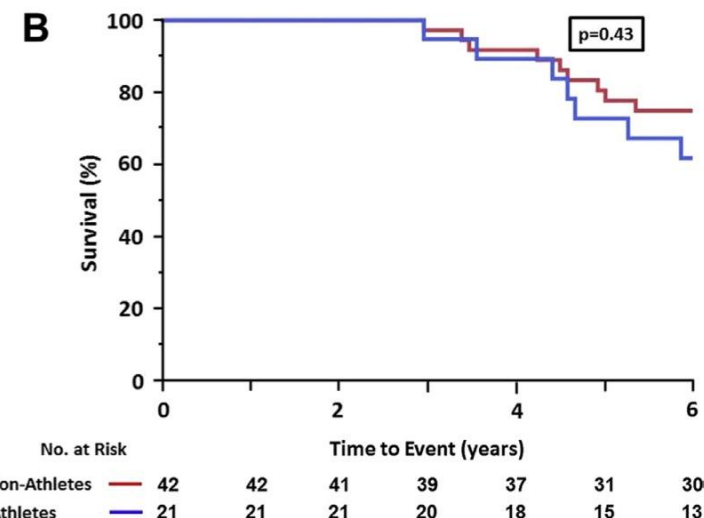
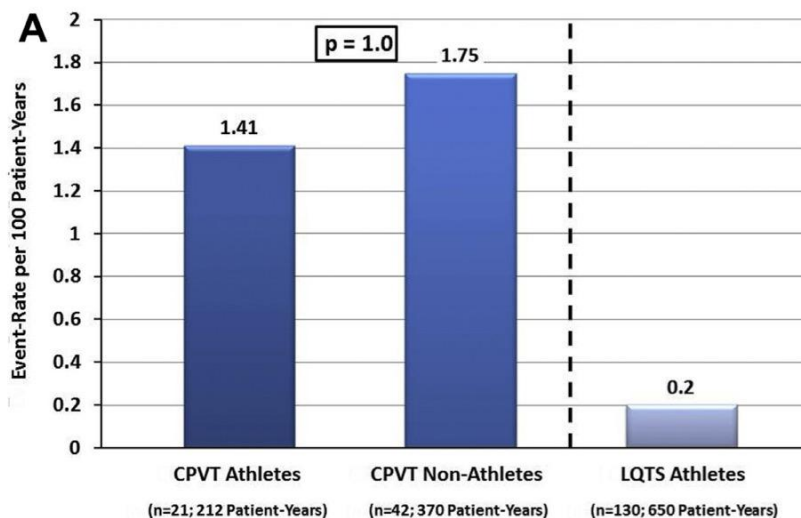
QUE NOUS DIT LA LITTERATURE ?

# Competitive Sports Participation in Patients With Catecholaminergic Polymorphic Ventricular Tachycardia



## A Single Center's Early Experience

Stuart A. Ostby, BS,<sup>a</sup> J. Martijn Bos, MD, PhD,<sup>b,c</sup> Heidi J. Owen, RN,<sup>b</sup> Philip L. Wackel, MD,<sup>b</sup> Bryan C. Cannon, MD,<sup>b</sup> Michael J. Ackerman, MD, PhD<sup>a,b,c,d</sup>








- GUIDELINES



**EHRA**  
European Heart  
Rhythm Association

 European Society of Cardiology

2020

Consensus statement—catecholaminergic polymorphic ventricular tachycardia	Symbol
In CPVT patients, competitive and intensive leisure-time sports are NOT recommended.	
Under appropriate treatment, if stress-test shows absence of any type of ventricular ectopy/arrhythmia and if the patient is asymptomatic for a minimum of 3 months, low-intensity to moderate leisure-time sports may be considered, including those with an ICD. <sup>a</sup>	
Gene carriers of a pathogenic CPVT mutation without an overt phenotype should be managed as patients with manifest CPVT (i.e. only allowing low-intensity sports). A beta-blocker should be considered.	
Follow-up should include stress tests and/or continuous ECG monitoring (Holter) during leisure-time low-intensity sports activities to ensure control of exercise-induced ventricular arrhythmias.	
Avoidance of stressful/emotional situations, dehydration, electrolyte disturbances, or hyperthermia is recommended.	
<sup>a</sup> Avoid contact sports in case of ICD implanted.	

## GUIDELINES



American  
Heart  
Association.



AMERICAN  
COLLEGE of  
CARDIOLOGY.

2025

### General considerations

Competitive athletes with a cardiac channelopathy (including LQTS, CPVT, and BrS) should be assessed by a pediatric or adult cardiologist with expertise in cardiac channelopathies and with SDM.

In competitive athletes diagnosed with a cardiac channelopathy, an ICD should not be implanted for the sole purpose of competitive sports participation.

### Catecholaminergic polymorphic ventricular tachycardia

#### Specific clinical considerations

In an asymptomatic competitive athlete with positive genetic test results for CPVT but no exercise-induced ventricular ectopy on exercise stress testing (ie, genotype-positive and phenotype-negative), competitive sports participation is reasonable with discussion about prophylactic CPVT-directed medical therapy.

In competitive athletes with asymptomatic CPVT who have a positive stress test with evidence of exercise-induced ventricular ectopy, competitive sports participation can be considered with SDM and after optimization of therapies and normalization of the stress test.†

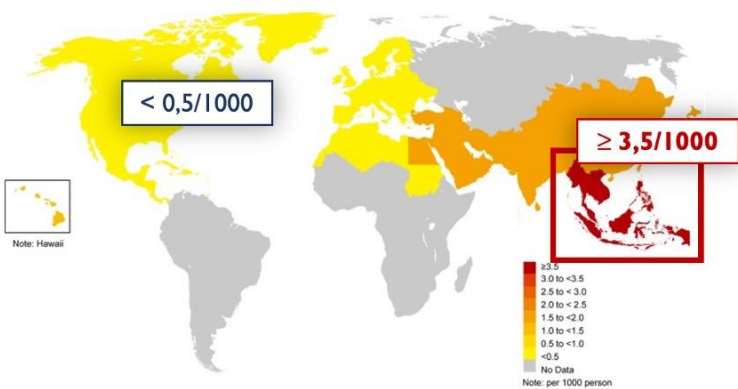
In competitive athletes with previously symptomatic CPVT for whom competitive sports participation are being considered, combination therapy with  $\beta$ -blocker and flecainide, and possibly the addition of LCSD, is required before resumption of competitive sports participation. Such CPVT therapies should be optimized with normalization of the stress test before participation in competitive sports.†



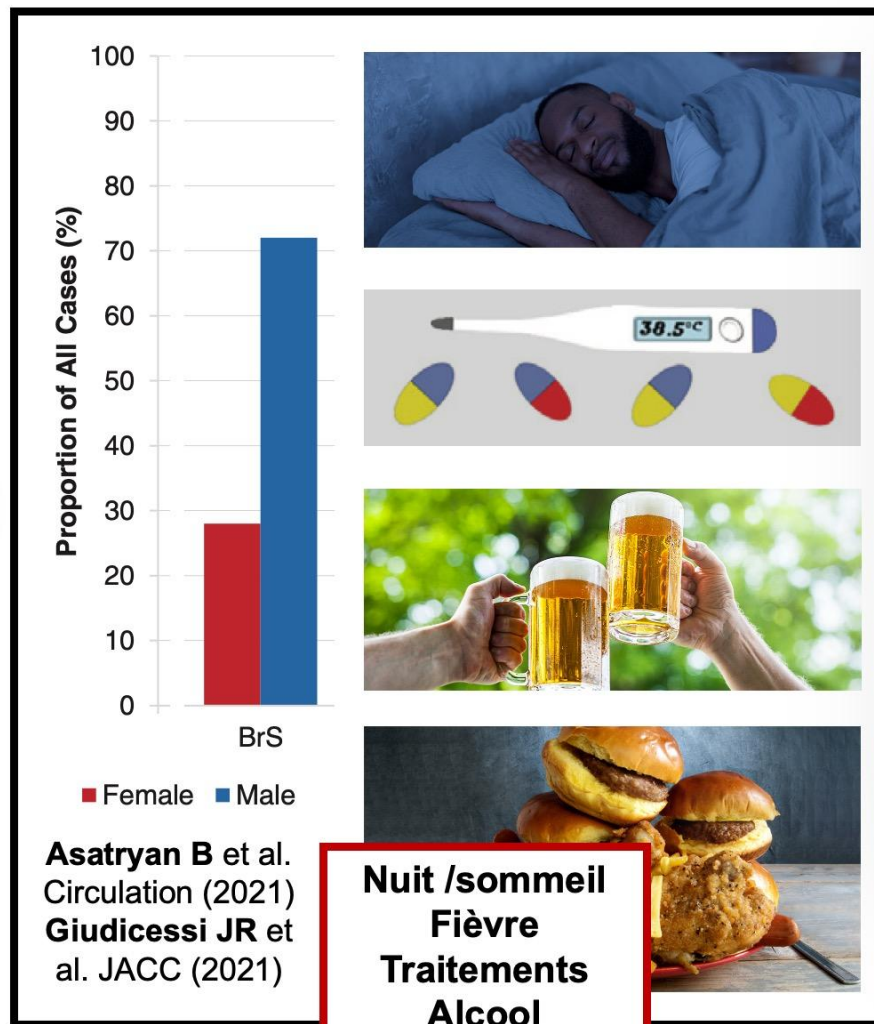
† Normalization of the stress test ideally encompasses absence of ectopy; bigeminal premature ventricular contractions may be acceptable but ventricular couplets or more extensive nonsustained ventricular tachycardia require continued treatment intensification.

GENERALITES

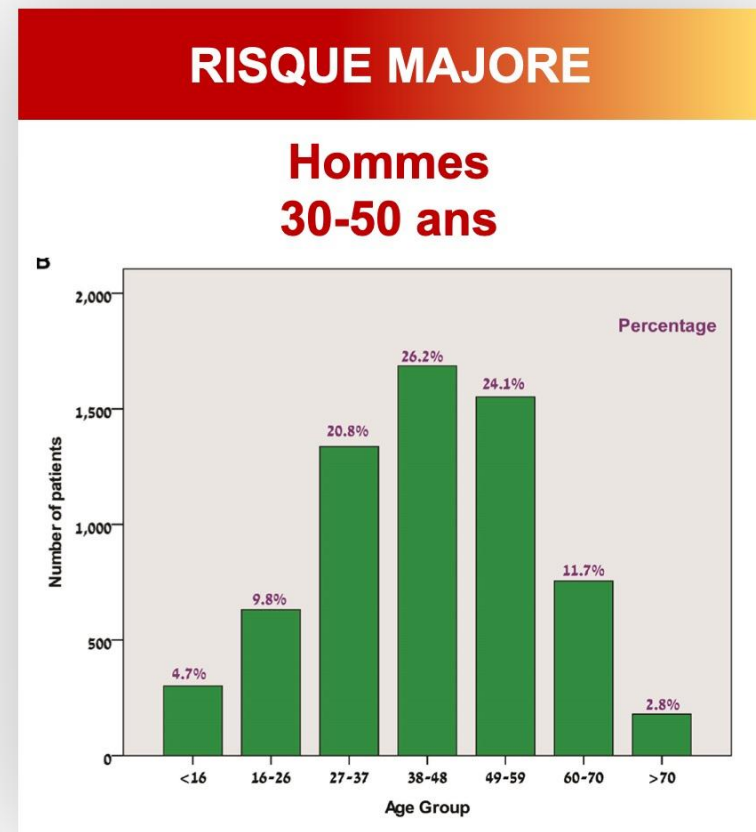
Prevalence  
1:2000



Vutthikraivit W et al.  
Acta Cardiol Sin (2018)

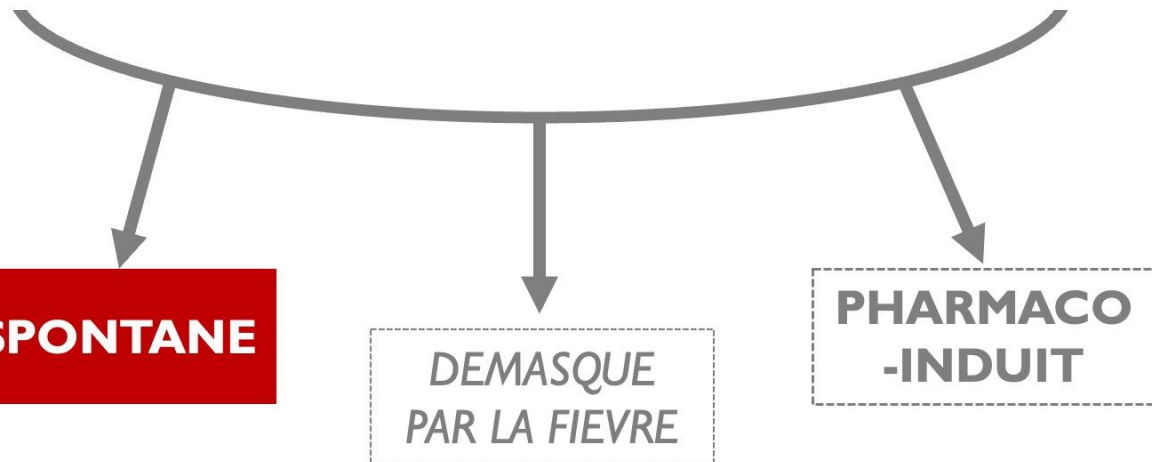
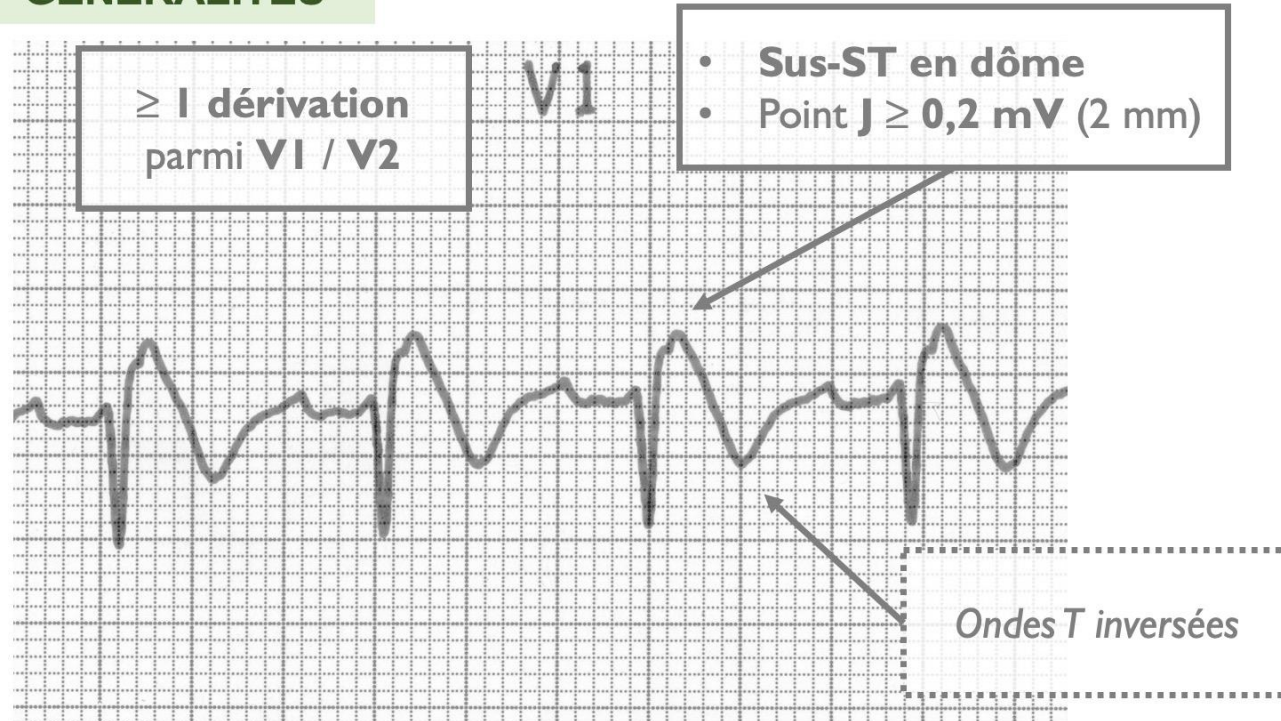


Nuit /sommeil  
Fièvre  
Traitements  
Alcool  
Repas riches



Milman A et al. Circ AE (2017)

## GENERALITES



## SPONTANE

- Permet d'affirmer le diagnostic  
→ Mais bilan pour éliminer une phéno-copie

## INDUIT

- Prudence
- 2-4% des sujets sains
- Spécificité inconnue du test à l'ajmaline
- Nécessité d'un **contexte** pour évoquer le diagnostic

BrS should be considered in patients with no other heart disease and induced type 1 Brugada pattern who have at least one of:

- Arrhythmic syncope or nocturnal agonal respiration
- A family history of BrS
- A family history of SD (<45 years old) with a negative autopsy and circumstance suspicious for BrS.

IIa

C

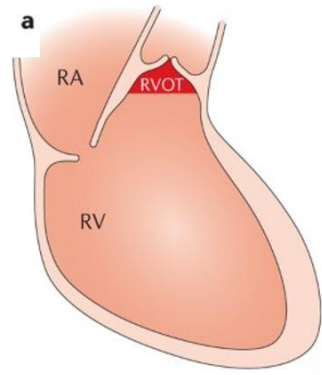
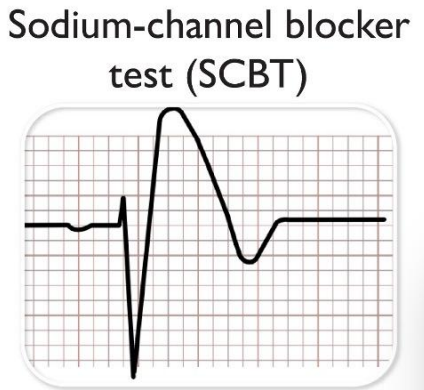
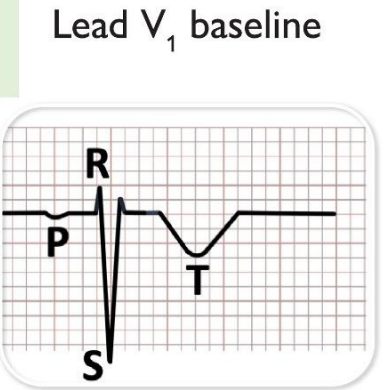
BrS may be considered as a diagnosis in patients with no other heart disease who exhibit an induced type 1 Brugada

ECG. [136,973,975,978,984,985](#)

IIb

C

CHANGEMENT DE PARADIGME



Reduced conduction reserve in RVOT (heterogeneous)

Pas automatiquement synonyme de **Syndrôme de Brugada**

**INDICATION**

% with Brugada syndrome depends on indication for SCBT

Brugada syndrome

BrS should be considered in patients with no other heart disease and induced type 1 Brugada pattern who have at least one of:

- Arrhythmic syncope or nocturnal agonal respiration
- A family history of BrS
- A family history of SD (<45 years old) with a negative autopsy and circumstance suspicious for BrS.

IIa	C
-----	---

Wilde AAM et al. European Heart J. 2023  
 Sieira J et al. Nature Reviews Cardiology 2016  
 Zeppenfeld et al. Eur Heart J. 2022

## CHANGEMENT DE PARADIGME

### Suspected Brugada syndrome (BrS) considered for SCB testing in the context of at least one of the following

- Cardiac arrest or syncope
- Family history of BrS
- Family history of sudden unexplained death
- Type 2/3 Brugada ECG pattern with other ECG features and/or one of the above

ESC European Society of Cardiology  
Europace (2025) 27, eua067  
https://doi.org/10.1093/eurpace/eua067

EHRA DOCUMENT  
EHRA European Heart Rhythm Association

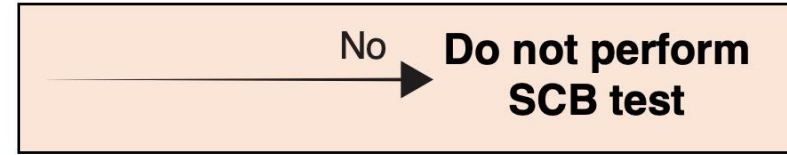
**The diagnostic role of pharmacological provocation testing in cardiac electrophysiology: a clinical consensus statement of the European Heart Rhythm Association and the European Association of Percutaneous Cardiovascular Interventions (EAPCI) of the ESC, the ESC Working Group on Cardiovascular Pharmacotherapy, the Association of European Paediatric and Congenital Cardiology (AEPC), the Paediatric & Congenital Electrophysiology Society (PACES), the Heart Rhythm Society (HRS), the Asia Pacific Heart Rhythm Society (APHRS), and the Latin American Heart Rhythm Society (LAHRS)**

Elijah R. Behr (Chair)<sup>1,2,3\*</sup>, Bo Gregers Winkel<sup>4,5†</sup>, Bode Ensam<sup>1,6†</sup>, Alberto Alfie (LAHRS)<sup>7</sup>, Elena Arbelo<sup>5,8,9,10</sup>, Colin Berry (EAPCI)<sup>11</sup>, Marina Cerrone (HRS)<sup>12</sup>, Giulio Conte<sup>13</sup>, Lia Crotti<sup>14,15</sup>, Cecilia M. Gonzalez Corcia (PACES)<sup>16</sup>, Juan Carlos Kaski (Cardio Pharma WG)<sup>1</sup>, Koonlawee Nademanee (APHRS)<sup>17</sup>, Pieter G. Postema<sup>5,18</sup>, Silvia Priori<sup>5,19,20</sup>, Vincent Probst<sup>5,21</sup>, Georgia Sarquella-Brugada (AEPC)<sup>5,22</sup>, Eric Schulze-Bahr<sup>5,23</sup>, Rafik Tadros<sup>24</sup>, Arthur Wilde<sup>5,21</sup>, and Jacob Tfelt-Hansen (Co-Chair)<sup>4,5,25</sup>

Behr ER et al. Europace 2025



Yes  
Patient counselling of pros and cons before taking consent for SCB test



### Advantages of performing SCB testing

- Excludes BrS in presence of a negative test, especially when using ajmaline
- Avoids diagnostic ambiguity
- Guides extended family screening
- Informs on safety of sodium channel blocker use in patients who require such drugs
- Informs of need for suppressing fever



### Disadvantages of performing SCB testing

- Limited specificity (e.g. ajmaline) and sensitivity (e.g. procainamide)
- A positive test can generate anxiety and unnecessary interventions despite favourable prognosis in asymptomatic patients
- Potential negative impact on insurability
- Procedural risk especially for patients with a pathogenic *SCN5A* variant

CHALEUR CORPORELLE



The American Journal of Emergency Medicine

Volume 27, Issue 5, June 2009, Pages 634.e1-634.e3



Case Report

# Heat stroke, an unusual trigger of Brugada electrocardiogram

Javier Lacunza MD, Irene San Román MD, Sara Moreno MD, Esperanza García-Molina MD, Juan Gimeno MD, Mariano Valdés MD



Revista Española de Cardiología (English Edition)

Volume 69, Issue 12, December 2016, Pages 1228-1230

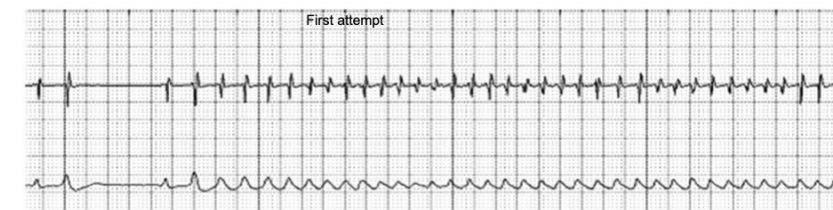
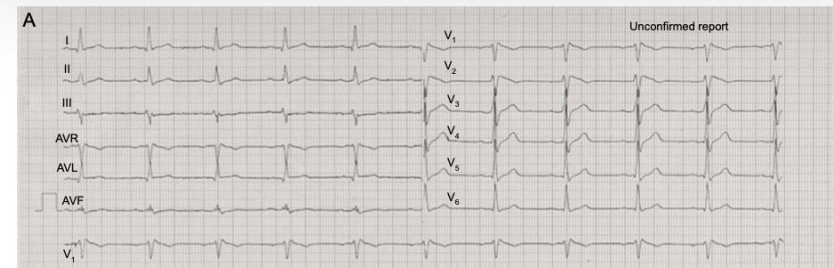


Scientific letter

# Heat Wave: A Trigger of Electrical Storm in a Patient With Brugada Syndrome

## Ola de calor: desencadenante de tormenta arrítmica en un paciente con síndrome de Brugada

Isabel Molina Borao, Naiara Calvo Galiano, Javier Urmeneta Ulloa, Elena Rivero Fernández, Teresa Olóriz Sanjuan, Antonio Asso Abadía



VS 585 VF 263 VS 925 VF 265 VF 243 VF 215 VF 213 VF 200 VF 183 VF 163 VF 158 VF 153 VF 153 VF 170 VF 155 VF 173 VF 165 VF 200 VF 185 VS 325 VF 170 VF 165 VF 163 VF 173 VF 16

V-Epsd V-Detect Chrg

## ▪ GUIDELINES

**ESC**European Society  
of Cardiology

2020

Recommendations	Class	Level
ICD implantation is recommended in patients with BrS with episodes of arrhythmic syncope and/or aborted SCD	I	C
Following implantation of an ICD, resumption of leisure or competitive sports should be considered after shared-decision making in individuals who have not experienced recurrent arrhythmias over 3 months after ICD implantation.	IIa	C

- GUIDELINES


**ESC**

 European Society  
of Cardiology

**2020**
**Recommendations**
**Class**
**Level**

In asymptomatic individuals with BrS, asymptomatic mutation carriers and asymptomatic athletes with only an inducible ECG pattern, participation in sports activities that are not associated with an increase in core temperature  $>39^{\circ}\text{C}$  (e.g. endurance events under extremely hot and/or humid conditions) may be considered.

**IIb**
**C**

Prescription of drugs that may aggravate BrS\*, electrolyte abnormalities and sports practice that increases core temperature  $> 39^{\circ}\text{C}$  are not recommended in individuals with overt BrS or phenotypically-negative mutation carriers.

**III**
**C**

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**EHRA**  
European Heart  
Rhythm Association

 European Society of Cardiology

2020

Consensus statement—Brugada syndrome	Symbol
In BrS patients with episodes of suspected arrhythmic syncope and/or aborted SCD, ICD implantation is recommended.	
In all patients with overt BrS and in all phenotypically-negative mutation carriers, avoidance of drugs that may aggravate the BrS ( <a href="http://www.brugadadrugs.org">www.brugadadrugs.org</a> ), of electrolyte imbalance, and of increases in core temperature are recommended. In case of febrile illness, fever should be treated aggressively.	
If there is no recurrent event during 3 months in symptomatic BrS patients after ICD implantation, leisure or competitive sports may be resumed based on shared decision-making. <sup>a</sup>	
Asymptomatic BrS patients, asymptomatic mutation carriers, and asymptomatic athletes with only an inducible ECG pattern, may participate in all sports that are not associated with an increase in core temperature >39°C (e.g. endurance events under extremely hot and/or humid conditions).	
<sup>a</sup> Avoid contact sports in case of ICD implanted.	

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American  
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2025

**General considerations**

Competitive athletes with a cardiac channelopathy (including LQTS, CPVT, and BrS) should be assessed by a pediatric or adult cardiologist with expertise in cardiac channelopathies and with SDM.

In competitive athletes diagnosed with a cardiac channelopathy, an ICD should not be implanted for the sole purpose of competitive sports participation.

**Brugada syndrome**

## Specific clinical considerations

It is reasonable for competitive athletes with BrS to participate in competitive sports after expert assessment and management.

There are no data to support competitive sports participation restrictions with Brugada syndrome. These individuals should avoid known arrhythmic triggers, including heat exhaustion and exercise during febrile illnesses, and hydration during exercise should be prioritized.<sup>224,225</sup>

# MERCI DE VOTRE ATTENTION

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