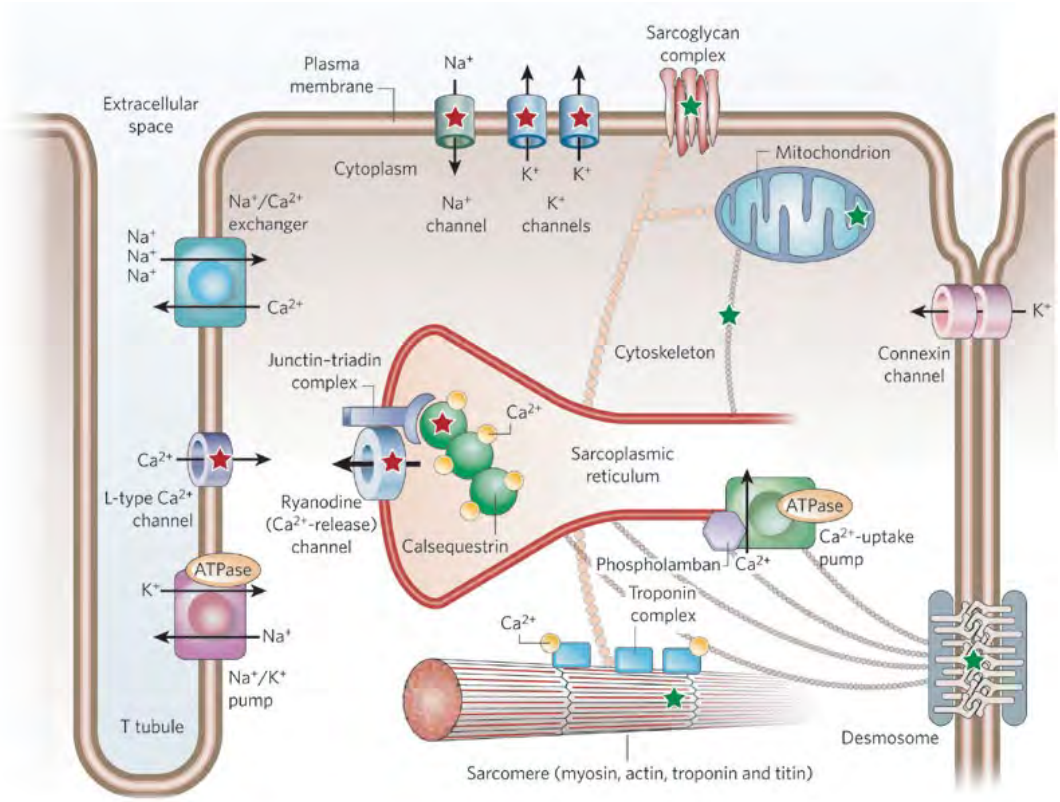
CANALOPATHIES



Dr Marie WILKIN / Dr Victor WALDMANN

Hôpital Européen Georges Pompidou — Hôpital Necker

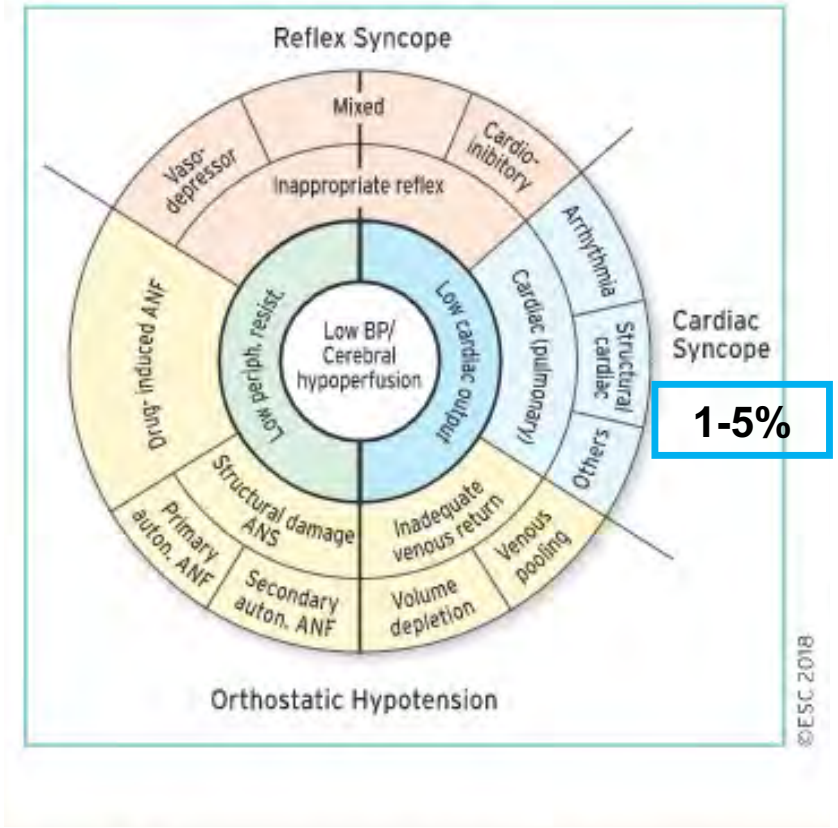
Spectre des Diagnostics

IVF: Idiopathic Ventricular Fibrillation

Evoquer le Diagnostic

Symptomes	**Syncope / Noyade / Comitialité**
Circonstances	**Effort / Emotion / Fièvre**
Famille	**Histoire familiale de mort subite** **—** **Surdité congénitale**

SYNCOPES



CARDIAQUES

- ***Bradycardies***

Dysfonction sinusale, BAV

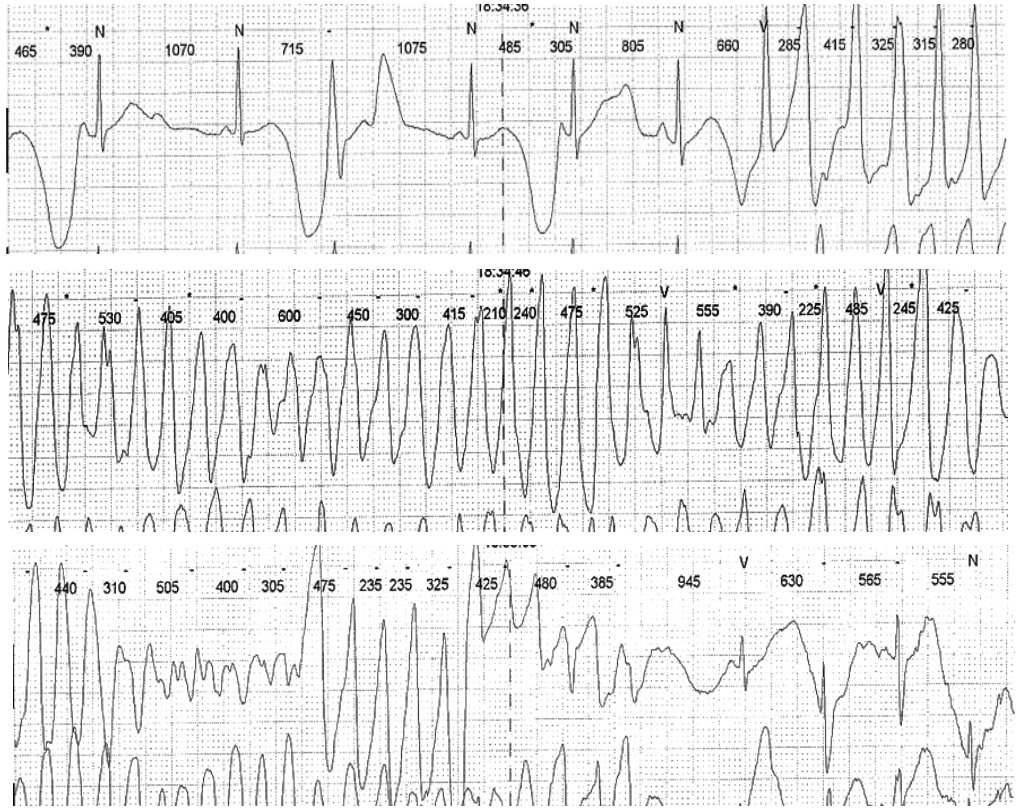
- ***Tachycardies***

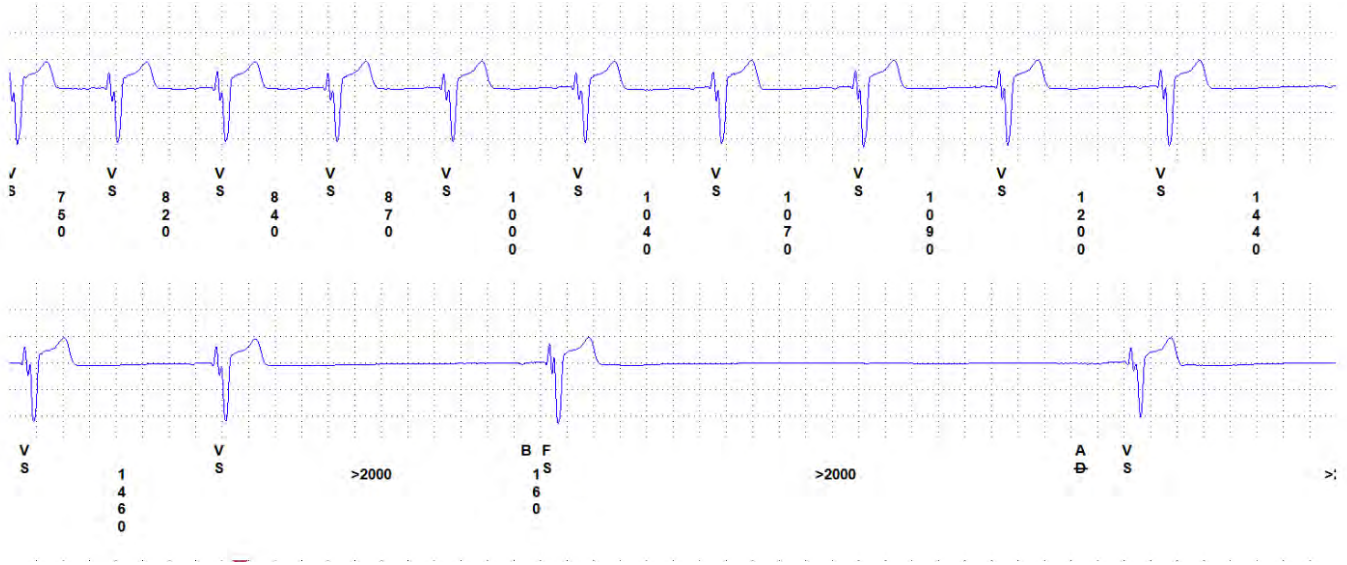
Ventriculaire >> supraventriculaire

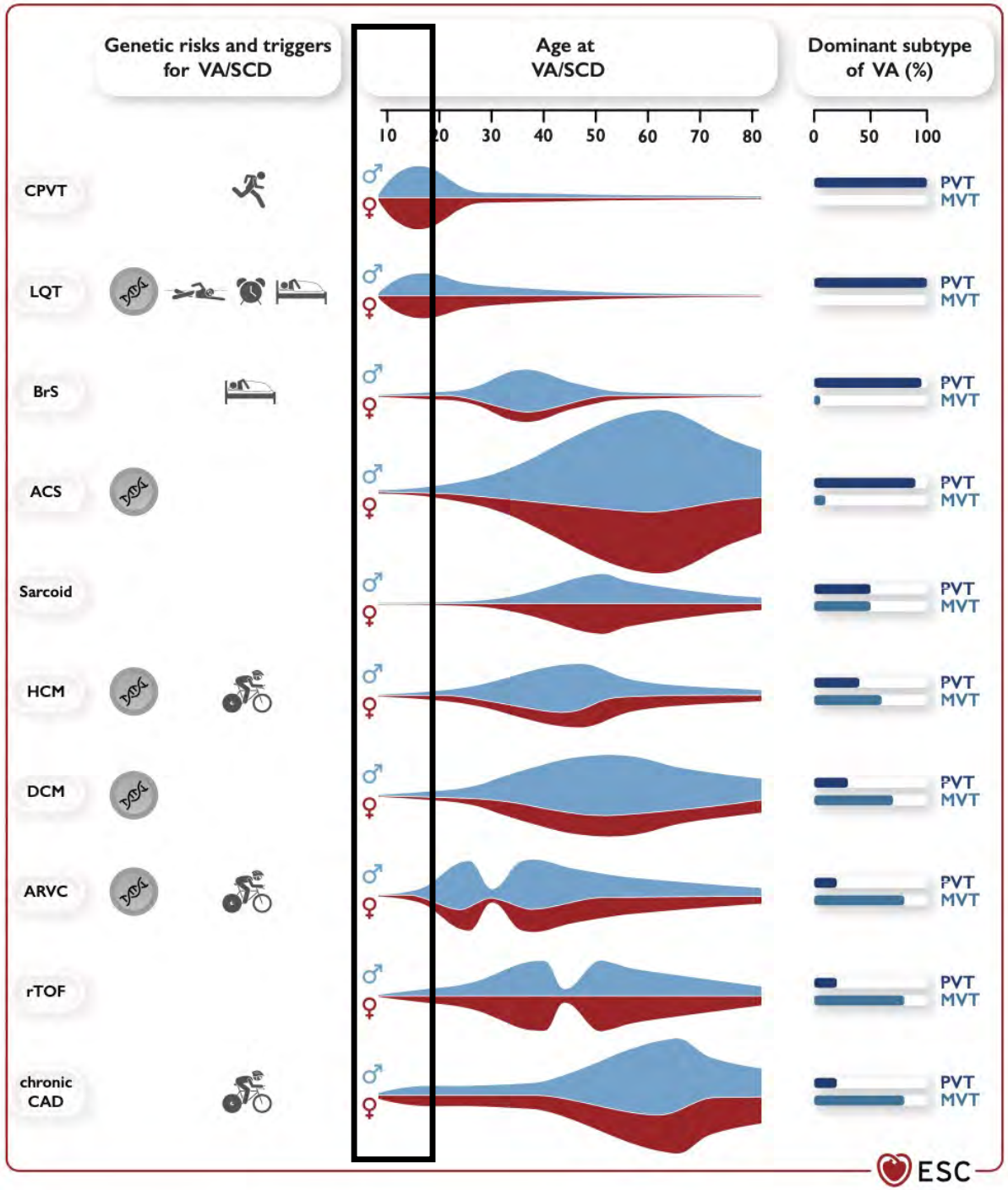
- ***« Mécaniques »***

Sténose aortique, EP, CMH…

TDR Ventriculaire vs. Syncope Vagale

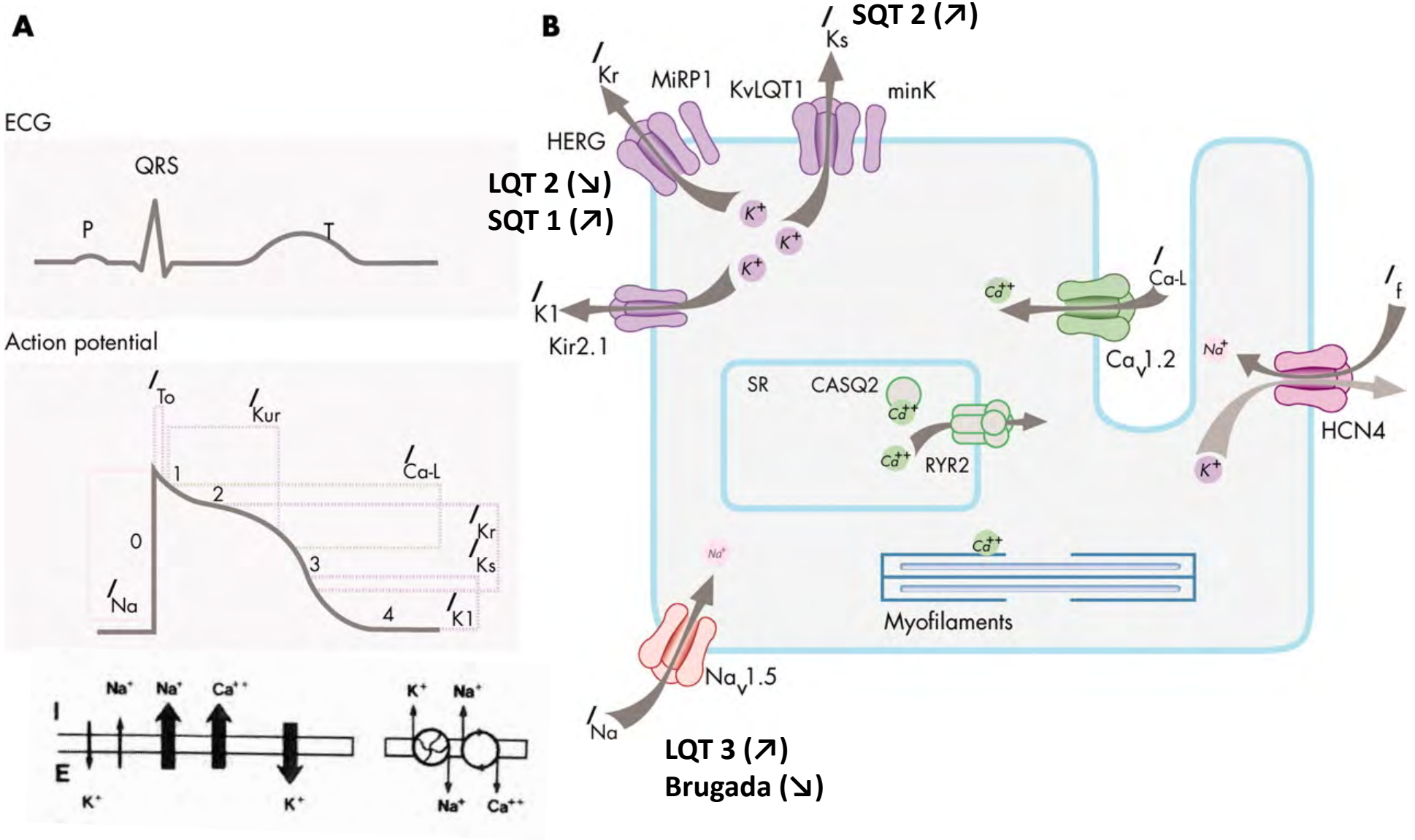






Canalopathies : **5-10%** des morts subites du nourrisson

Les Canalopathies



Channelopathy associated genes[2]

Disease	Affected channel	Gene	Protein
LQTS1	Potassium	KCNQ1	Kv7.1
LQTS2	Potassium	KCNH2	hERG Kv11.1
LQTS3	Sodium	SCN5A	Nav1.5
LQTS4	Calcium (related)	ANK2	Ank-B
LQTS5	Potassium	KCNE1	MinK
LQTS6	Potassium	KCNE2	MiRP1
LQTS7 (Anderson-Tawil syndrome)	Potassium	KCNJ2	Kv2.1 Kir2.1
LQTS8 (Timothy syndrome)	Calcium	CACNA1C	Cav1.2
LQTS9	Sodium (related)	CAV3	M-Caveolin
LQTS10	Sodium	SCN4B	Navβ4
LQTS11	Potassium (related)	AKAP9	Yotiao
LQTS12	Sodium (related)	SNTA1	Syntropin
LQTS13	Potassium	KCNJ5	Kv3.1 Kir3.4
LQTS14	Calcium	RYR2	Ryanodine receptor
CPVT1	Calcium	RYR2	Ryanodine receptor
CPVT2	Calcium	CASQ2	Calsequestrin
CPVT3	Potassium	KCNJ2	Kv2.1 Kir2.1
BS1	Sodium	SCN5A	Nav1.5
BS2	Sodium	GPD1-L	Glycerol-3-P-DH-1
BS3 (and SQTS4)	Calcium	CACNA1C	Cav1.2
BS4 (and SQTS5)	Calcium	CACNB2B	Voltage dependent β-2
BS5	Sodium	SCN1B	Navβ1
BS6	Potassium	KCNE3	MiRP2
BS7	Sodium	SCN3B	Navβ3
BS8	Potassium	KCNJ8	Kv6.1 Kir6.1
BS9	Potassium	HCN4	Hyperpolarisation cyclic nucleotide gated 4
BS10	Sodium (related)	MOG1	RAN-G-release factor
BS11	Potassium	KCNE5	Potassium voltage gated channel sub family E member1 like
BS12	Potassium	KCND3	Kv4.3 Kir4.3
BS13	Calcium	CACNA2D1	Voltage dependent α2/δ1
SQTS1	Potassium	KCNH2	hERG Kv11.1
SQTS2	Potassium	KCNQ1	Kv7.1
SQTS3	Potassium	KCNJ2	Kv2.1 Kir2.1
SQTS4 (and BS3)	Calcium	CACNA1C	Cav1.2
SQTS5 (and BS4)	Calcium	CACNB2B	Voltage dependent β-2

LQTS: Long QT syndrome, CPVT: Catecholaminergic polymorphic ventricular tachycardia, BS: Brugada syndrome, SQTS: Short QT syndrome

Inherited arrhythmias: The cardiac channelopathies

Ann Pediatr Cardiol. 2015 Sep-Dec; 8(3): 210–220.

Cas clinique 1

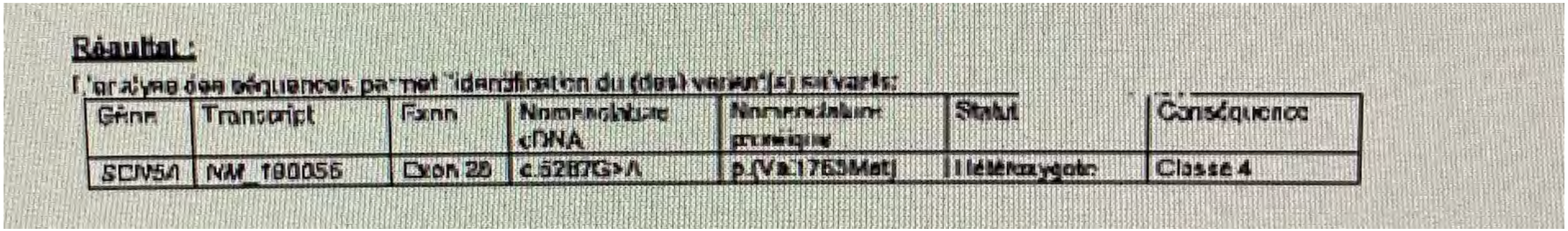
- Diagnostic anténatal d'un **BAV 2/1 à 34 SA** ; ETT normale
- Pas d'antécédent familiaux de maladie auto-immune / mort subite
- Naissance à 38 SA +2 jours :

➢QTc 500 ms

➢Rythme sinusal avec BAV 2/1

- **Pacemaker épicardique monochambre Medtronic le 08/12/2020**

- Traitement par Nadolol (50 mg/m^2)
- Bilan génétique :



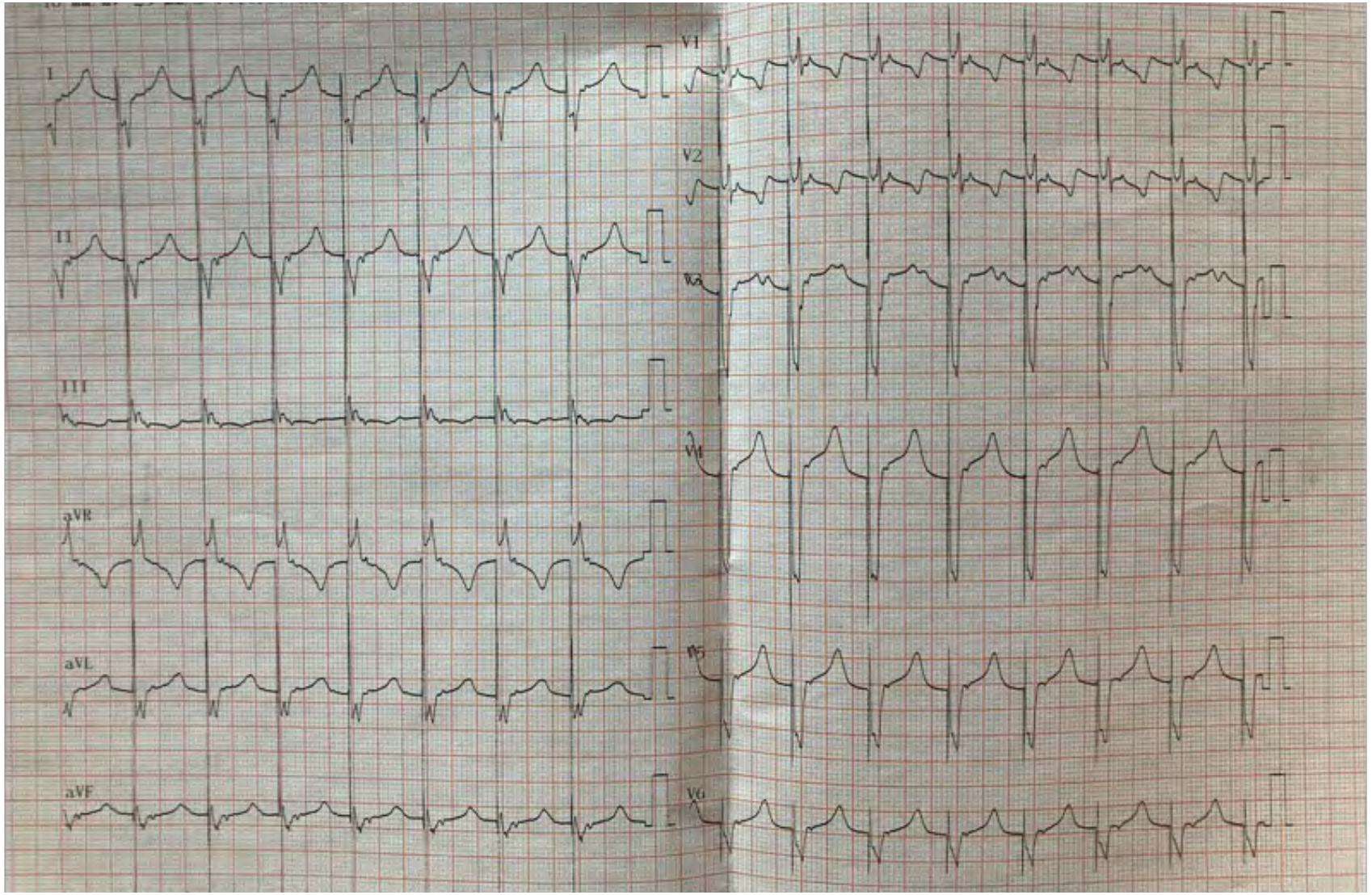
Résultat :

L'analyse des séquences par NGS permet l'identification du (des) variant(s) suivant :

Gène	Transcript	Exon	Nomenclature cDNA	Nomenclature protéique	Statut	Conséquence
SCN5A	NM_198056	Exon 20	c.5287G>A	p.(Val1763Met)	Hétérozygote	Classe 4

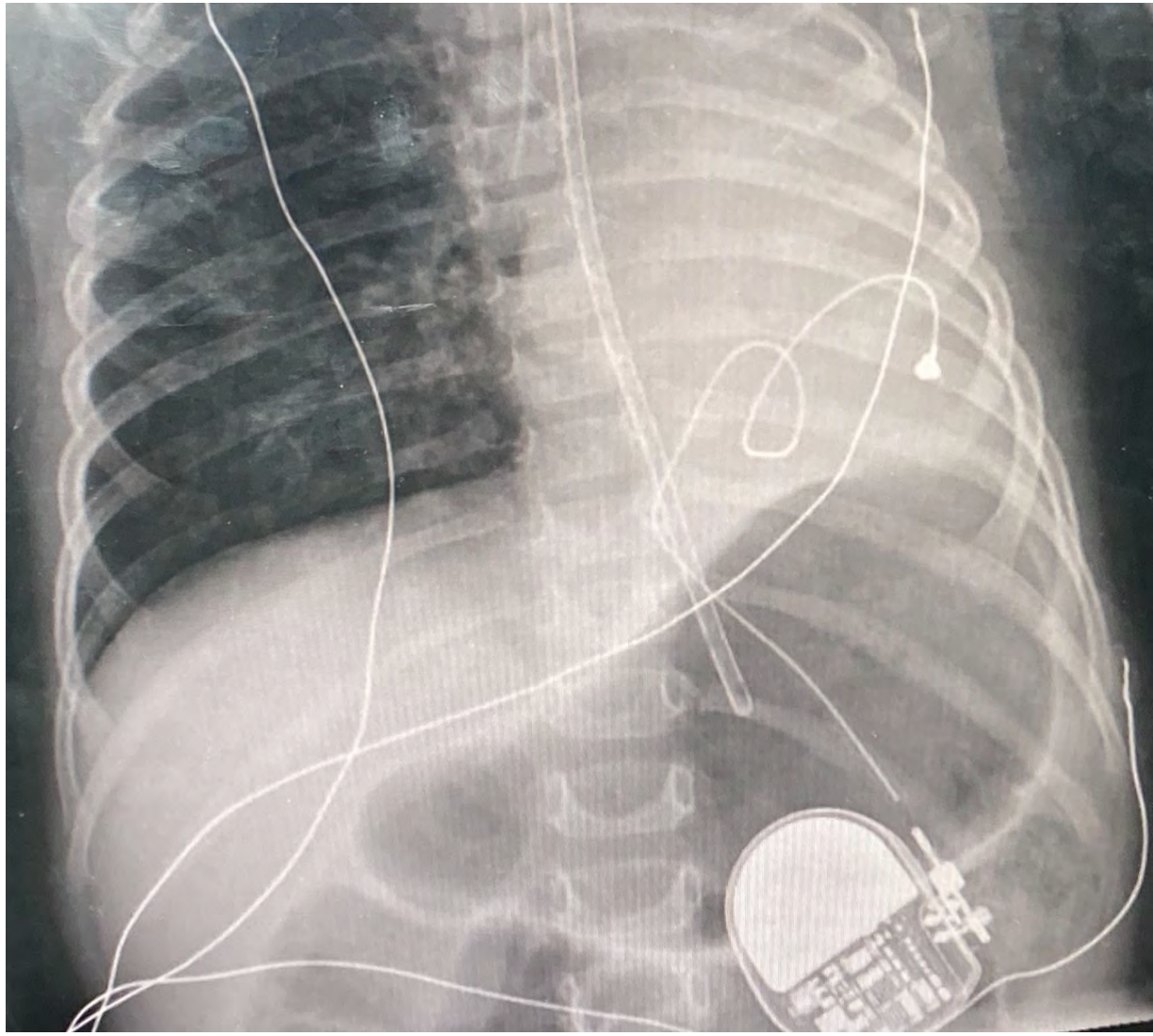
>>>> syndrome du QT long de type 3 : **mutation hétérozygote SCN5A** avec variant de classe 4

ECG juillet 2022 sous Corgard (posologie 50 mg/m2) : **560 ms**

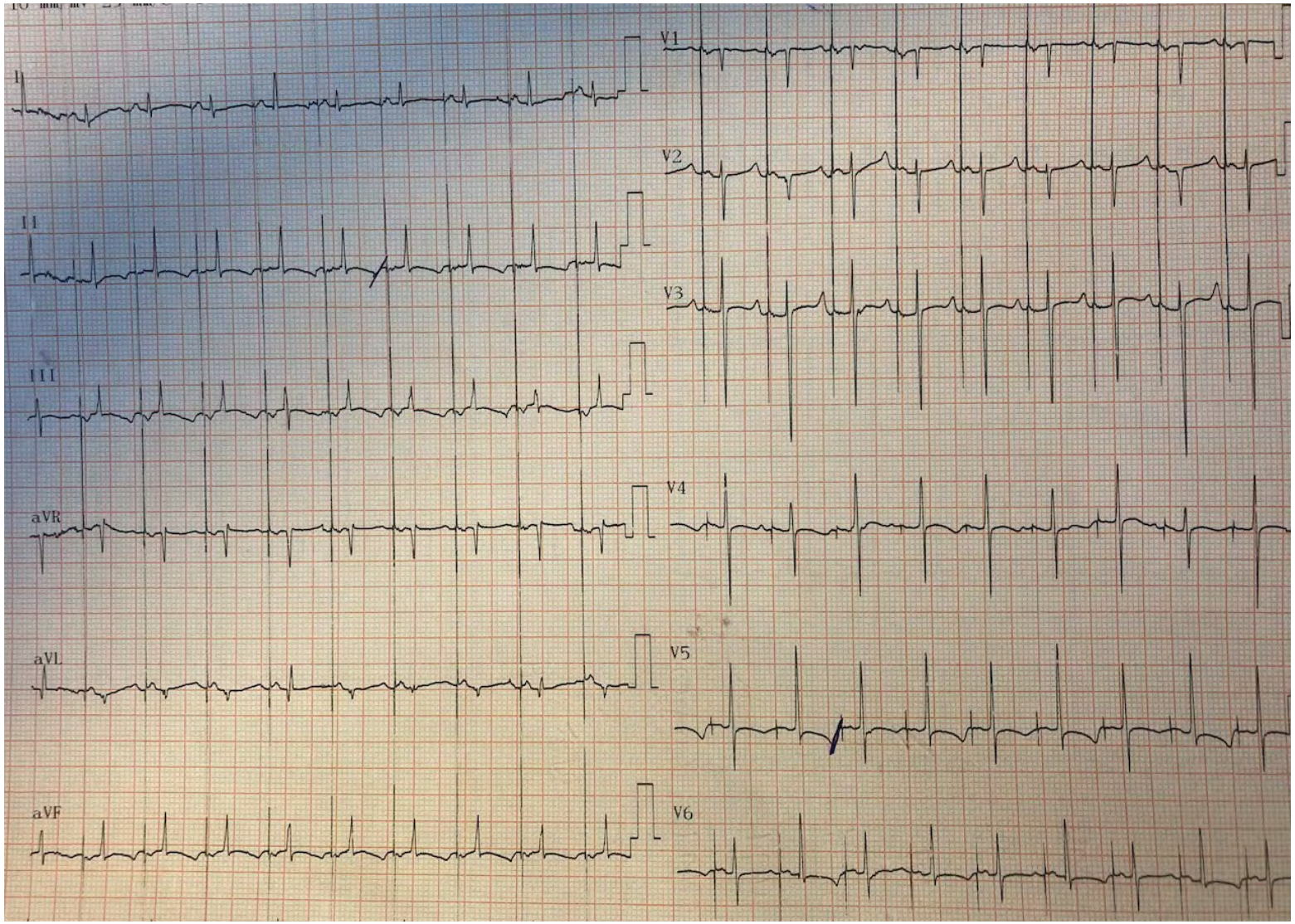


Octobre 2022

- ACR au domicile : low flow 30 minutes – 4 CEE
- Transfert en réanimation : **rupture sonde VD**
- **PM épicardique double chambre** Medtronic



- Introduction **Mexiletine 2 mg/kg/ jour** avec augmentation progressive de la posologie jusqu'à 6 mg/kg/jour
- Pas de récidive de troubles du rythme / **QTc à 460 ms**



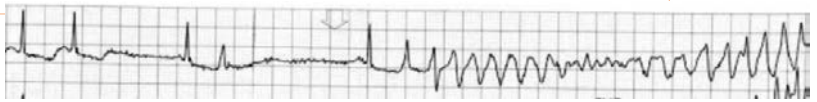
QT LONG

PHYSIOPATH

17 gènes connus – 1/2500
Mutation identifiée dans 75%:
LQTS1 KCNQ1 (effort ++, natation)
LQTS2 KNCH2 (émotion ou bruit)
LQTS3 SCN5A (repos ou sommeil)
Autosomique dominant (95%)

PRONOSTIC

Taux mort subite annuel 0.3-0.9%
5% par an si antécédent de syncope
LQT3 et QTc > 500 ms à haut risque



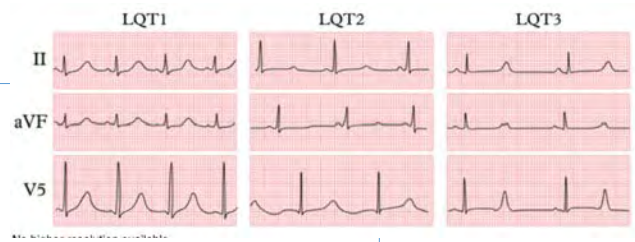
« Short-long-short »

DAI PREVENTION Iaire

ESC European Society of Cardiology
ESC GUIDELINES

TV malgré B-bloquants
Syncope

DIAGNOSTIC

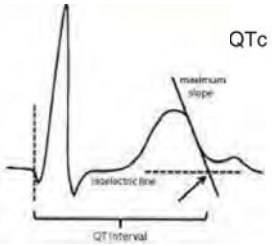


QTc ≥ 480 ms
Score > 3
Mutation positive
QTc > 460 ms + syncope inexpliquée

Variable	Points
Electrocardiogram	
QTc ms* ≥480	3
460-470	2
450 (males)	1
Torsade de pointes	2
T wave alternans	1
T wave notches in 3 leads	1
Bradycardia†	0.5
Clinical history	
Syncope	
With stress	2
Without stress	1
Congenital deafness	0.5
Family history‡	
Family members with confirmed LQTS§	1
Unexplained sudden death in first-order family members <30 years	0.5

Bazett

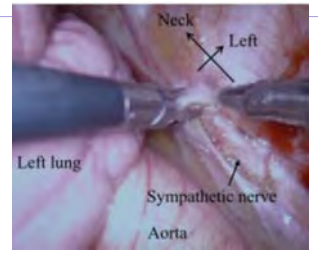
$$QTc\ interval = \frac{QT\ interval}{\sqrt{RR\ interval}}$$



D2 ++ ou V5

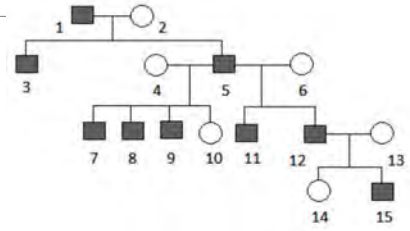
PERSPECTIVES

Dénervation
Flecaine ou mexiletine LQTS3
Stratification guidée par génétique



PRISE EN CHARGE

Médicaments contre indiqués
B-bloquants ++ (IIa pour porteur sain)
Restriction sportive
Dépistage familial
Orage rythmique: +/- SEES



CREDIBLEMEDS®
https://crediblemeds.org/

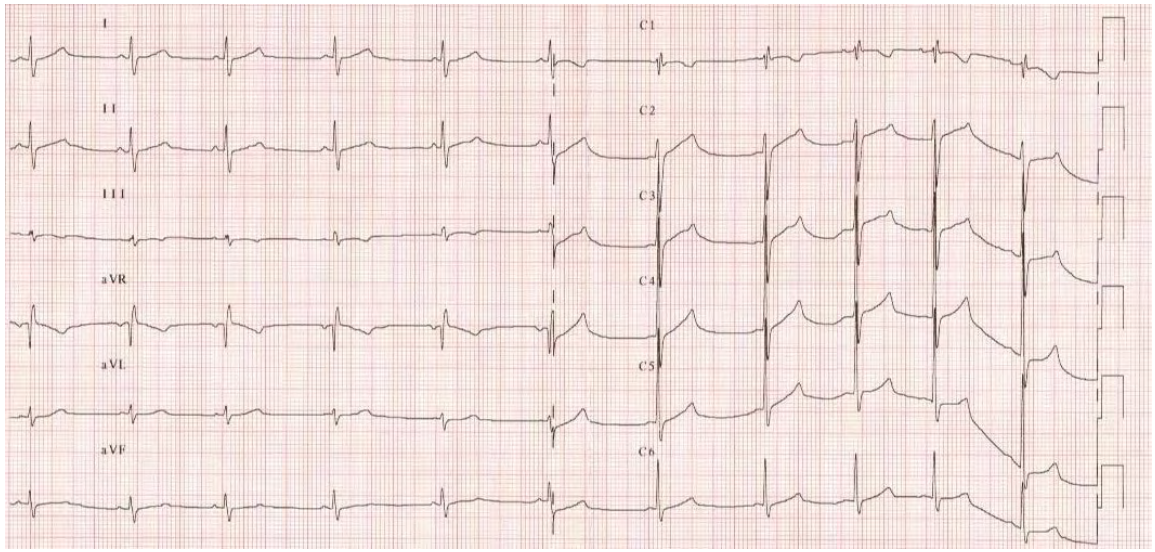
Schwartz score

Table 10 Modified long QT syndrome diagnostic score[243]

Findings			Points
ECG	QTc	≥480 ms	3.5
		=460–479 ms	2
		=450–459 ms (in males)	1
		≥480 ms during 4th minute of recovery from exercise stress test	1
	Torsade de pointes		2
	T wave alternans		1
	Notched T wave in 3 leads		1
	Low heart rate for age		0.5
Clinical history	Syncope	With stress	2
		Without stress	1
Family history	Family member(s) with definite LQTS		1
	Unexplained SCD at age <30 years in first-degree family		0.5
Genetic finding	Pathogenic mutation		3.5

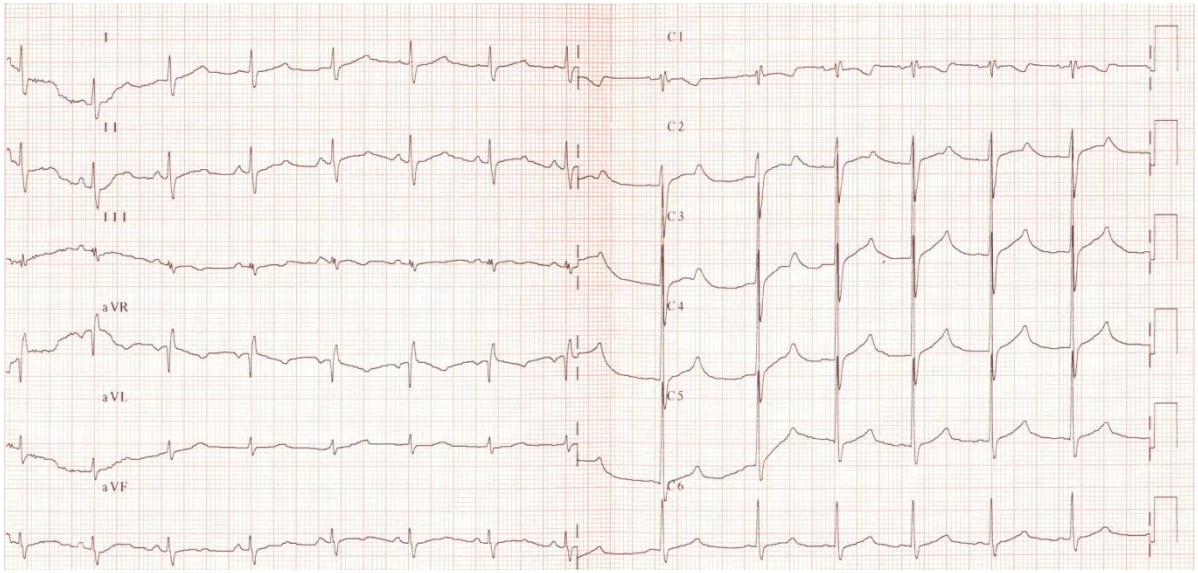
ECG, electrocardiogram; LQTS, long QT syndrome; SCD, sudden cardiac death.
Diagnosis of LQTS with a score >3.

QT LONG

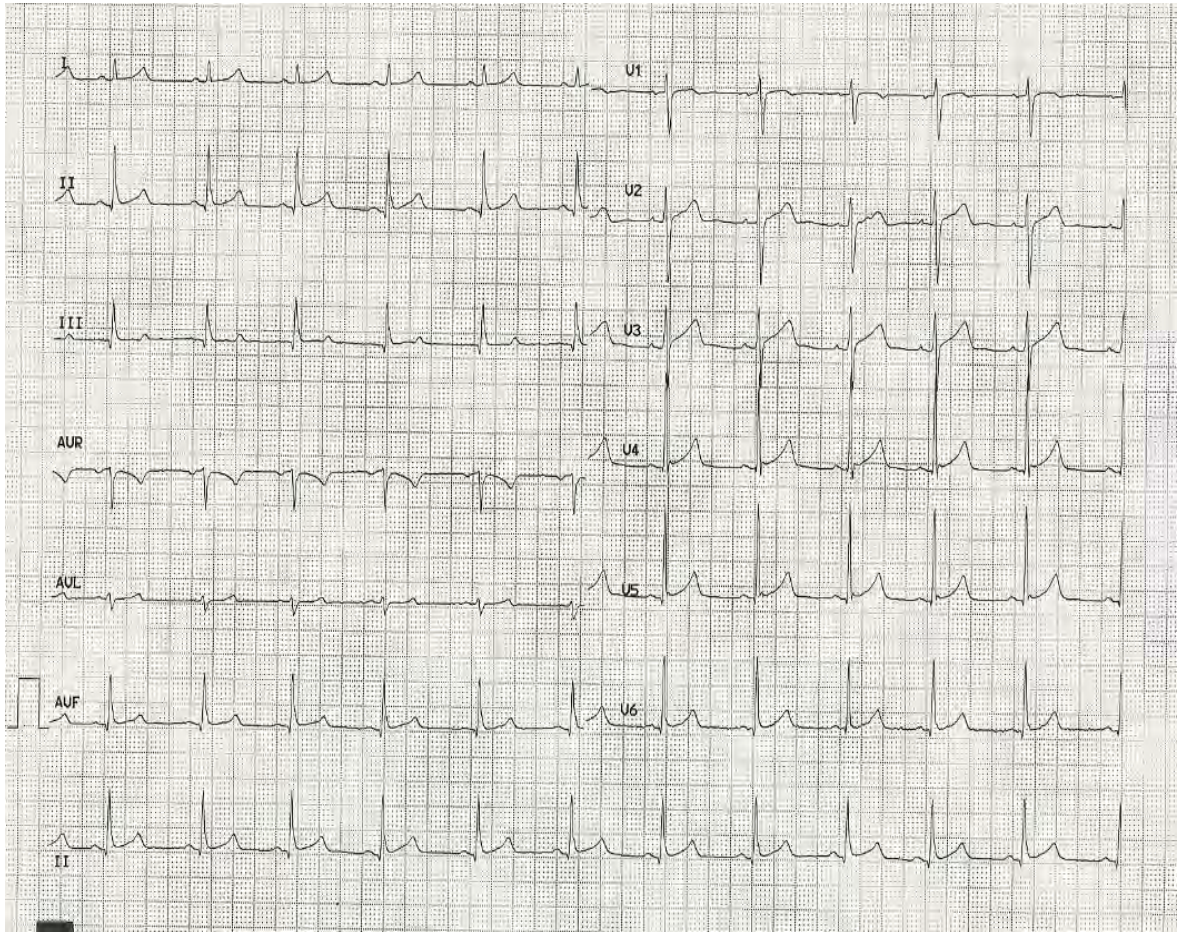


ECG allongé : QTc 440ms

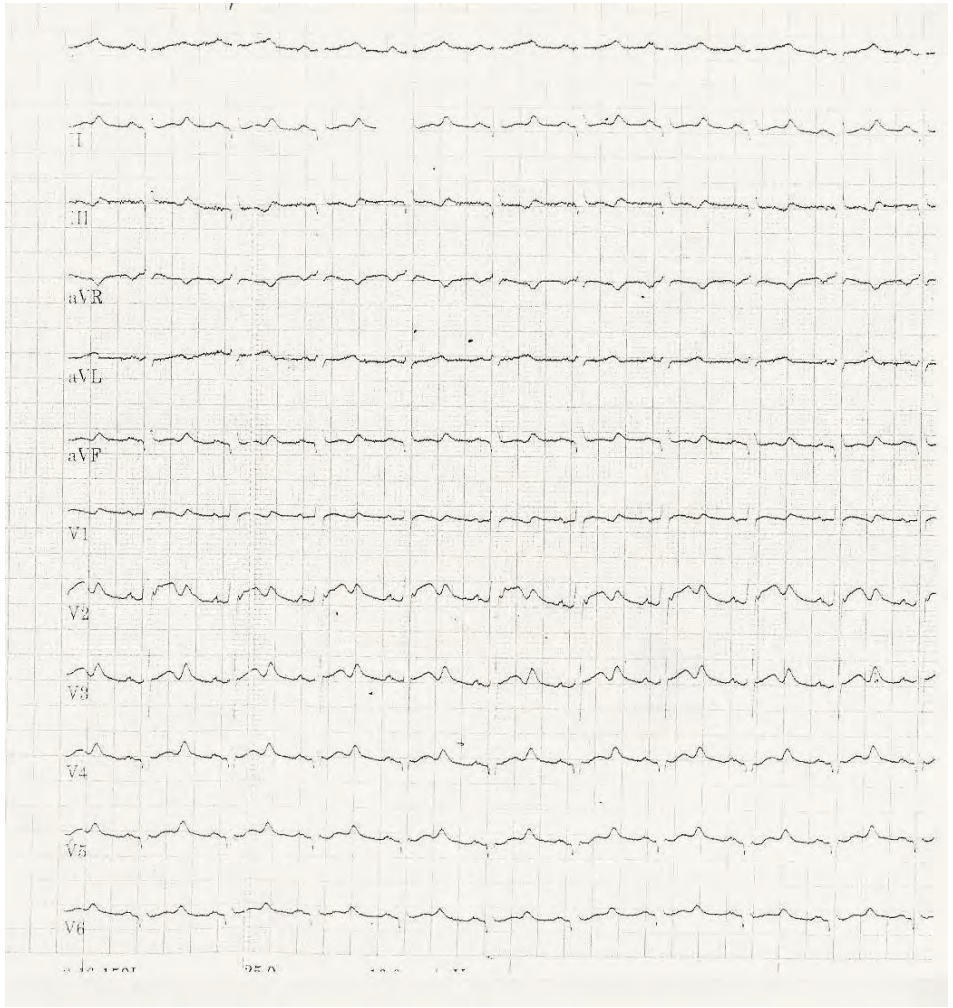
ECG debout : QTc 470ms



QT LONG

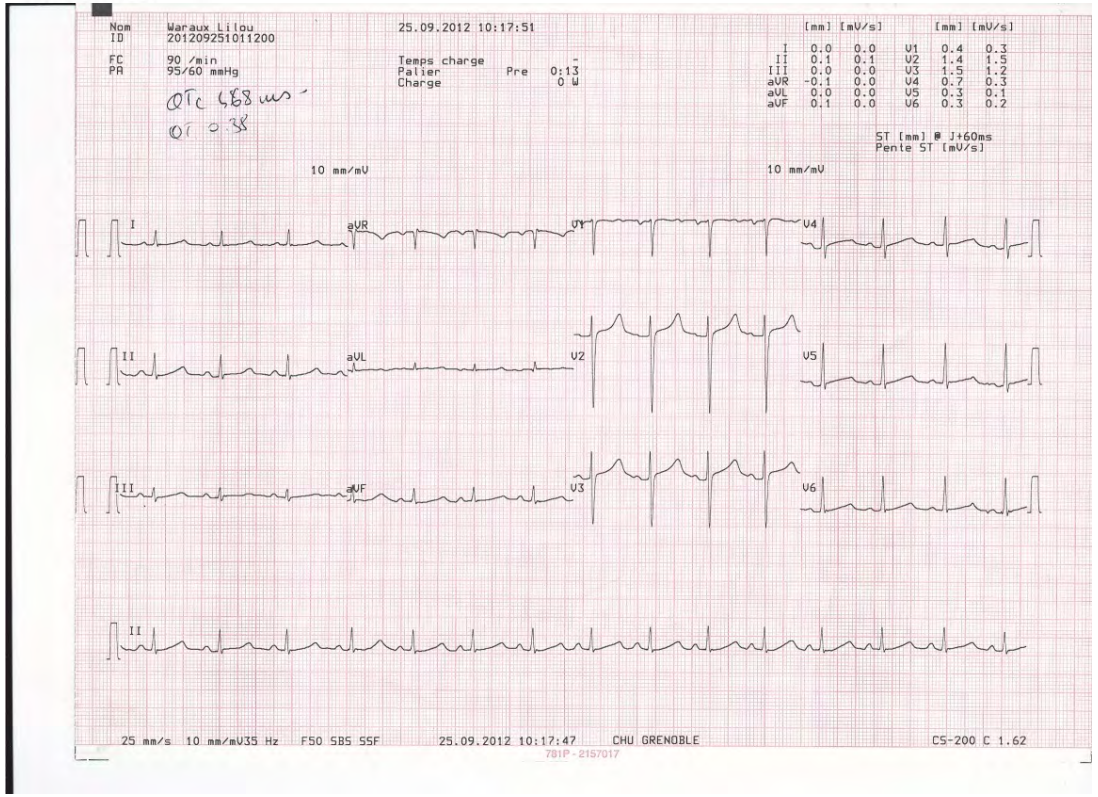


ECG de repos : QTc 450 ms

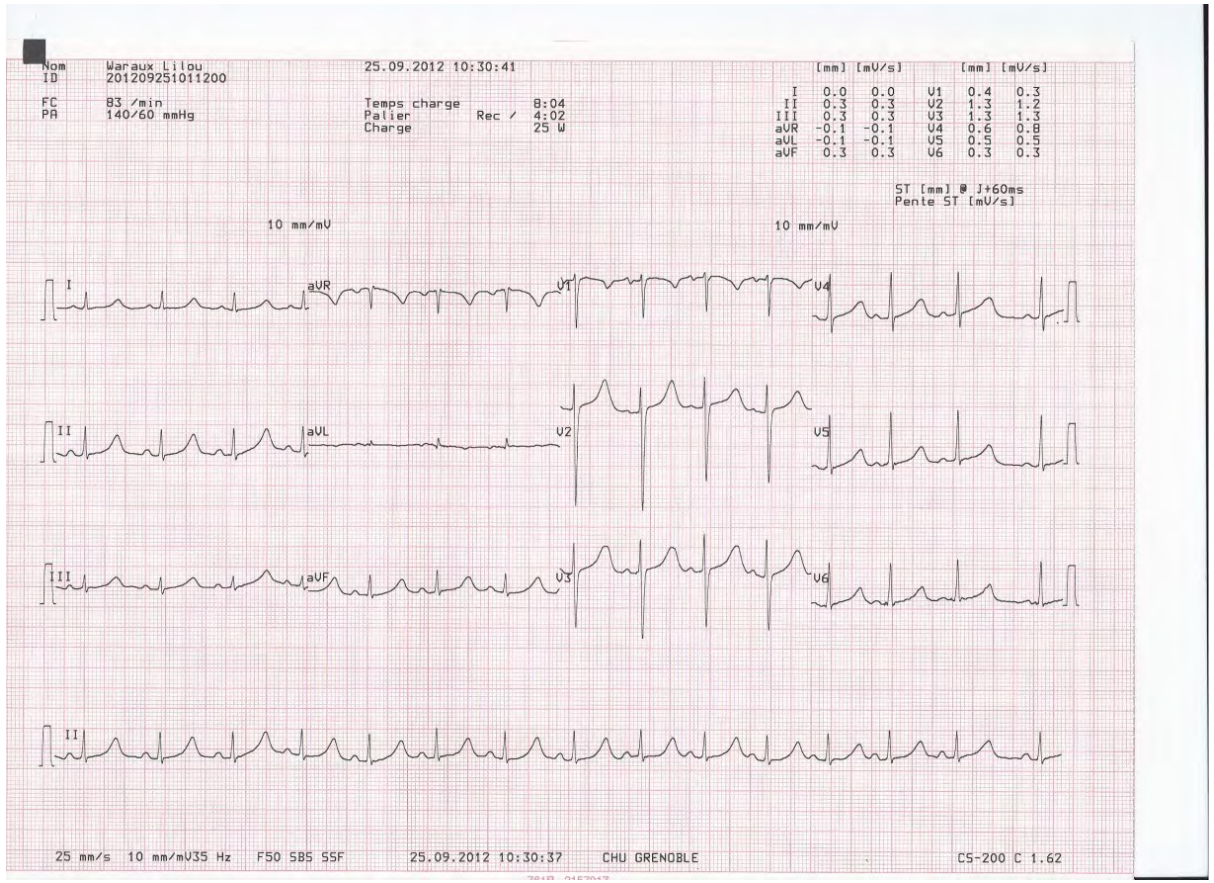


Test adrénaline : QTc 530 ms

QT LONG



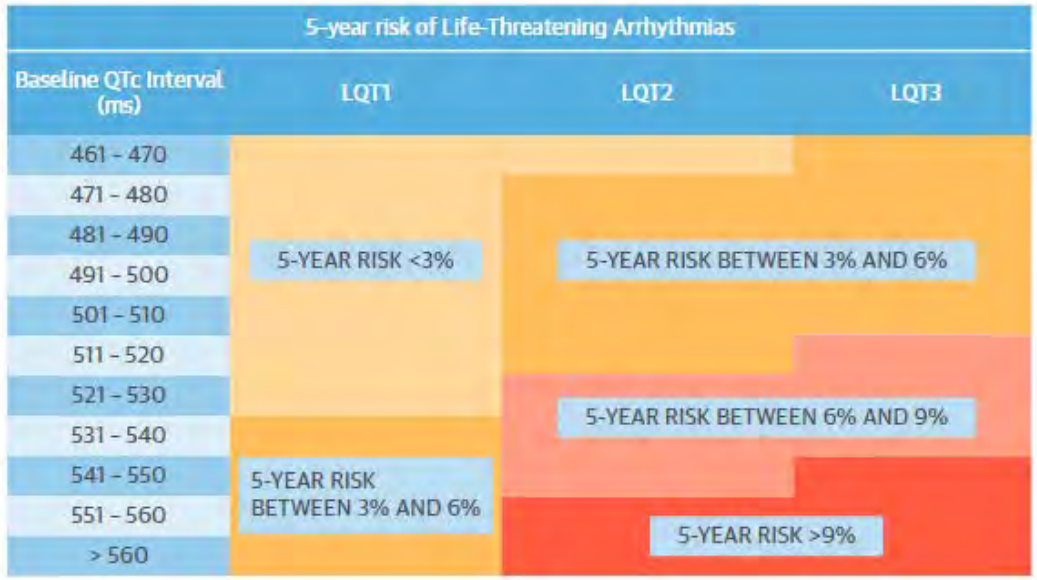
ECG de base



4ème mn de récupération

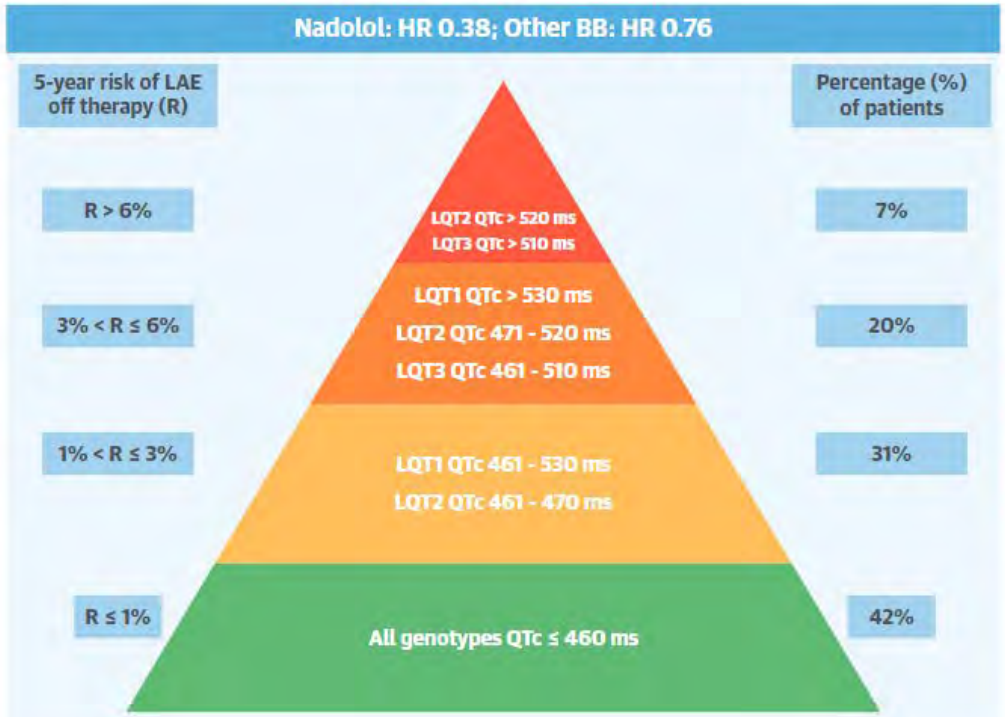
QT LONG

FIGURE 2 5-Year Risk of Life-Threatening Arrhythmic Events by Genotype for Each 10-ms Increment of QTc Duration for Patients Who Are Not Receiving Beta-Blockers



Visualization of the 5-year relative risk for patients with each genotype and for each QTc duration. The 4 colors group patients within the same 5-year risk of life-threatening arrhythmic events. This scheme can be used to personalize the risk estimate of patients at diagnosis in the absence of beta-blocker therapy and to estimate the risk of life-threatening arrhythmic events in patients who are not compliant with treatment. QTc = corrected QT interval.

CENTRAL ILLUSTRATION 5-Year Risk of LAEs by Genotype and QTc Interval Before Therapy and Effect of BBs



Mazzanti et al. JACC 2018

Diagnosis		
It is recommended that LQTS is diagnosed with either QTc ≥480 ms in repeated 12-lead ECGs with or without symptoms or LQTS diagnostic score >3.	I	C
In patients with clinically diagnosed LQTS, genetic testing and genetic counselling are recommended.	I	C
It is recommended that LQTS is diagnosed in the presence of a pathogenic mutation, irrespective of the QT duration.	I	C

Risk stratification, prevention of SCD and treatment of VA		
ICD implantation in addition to beta-blockers is recommended in LQTS patients with CA.[952,953,962,963]	I	B
ICD implantation is recommended in patients with LQTS who are symptomatic[d] while receiving beta-blockers and genotype-specific therapies.	I	C

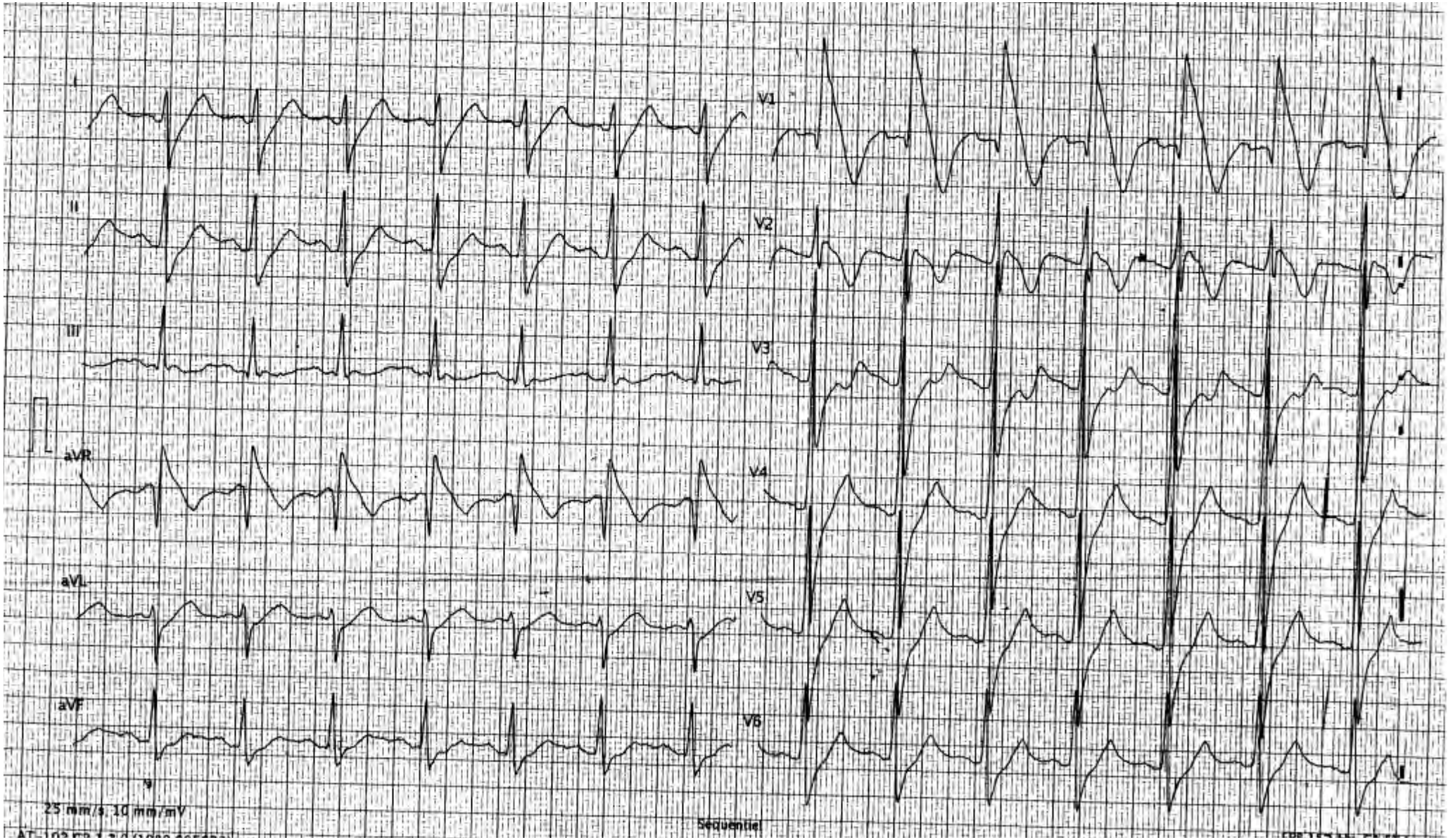
General recommendations to prevent SCD		
The following is recommended in LQTS: • Avoid QT-prolonging drugs.[c] • Avoid and correct electrolyte abnormalities. • Avoid genotype-specific triggers for arrhythmias.[943]	I	C
Beta-blockers, ideally non-selective beta-blockers (nadolol or propranolol), are recommended in LQTS patients with documented QT interval prolongation, to reduce risk of arrhythmic events.[940,945,946]	I	B
Mexiletine is indicated in LQT3 patients with a prolonged QT interval.[948]	I	C
Beta-blockers should be considered in patients with a pathogenic mutation and a normal QTc interval.[82]	IIa	B

Cas clinique 2

- Enfant âgé de 7 ans
- Mort subite chez un frère âgé de 4 ans dans son sommeil
- Dépistage familial : syndrome de Brugada
- Mutation SCN5A (mère / tante / fratrie)
- ECG : Brugada type 1

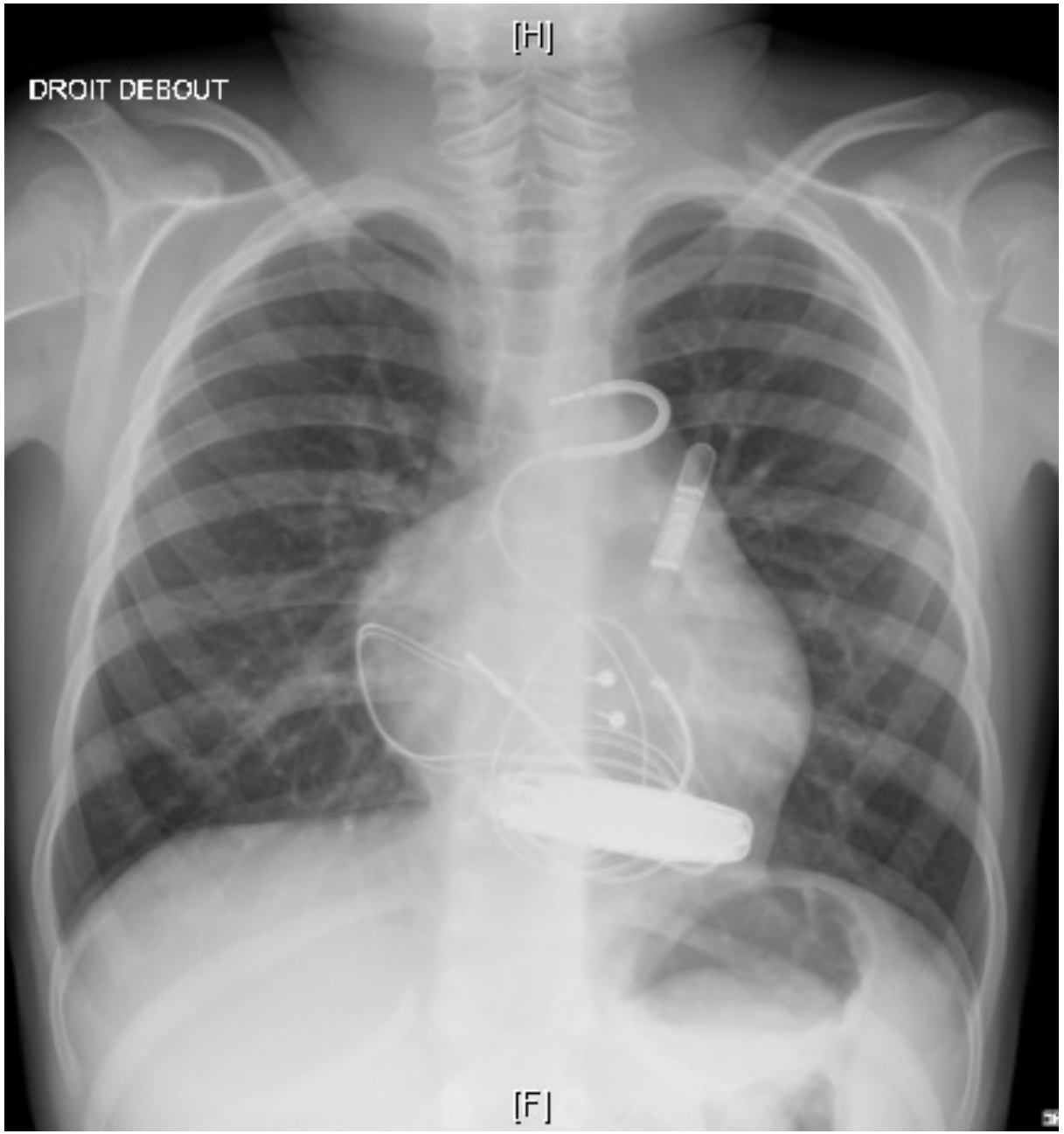
>>>> Moniteur implantable (Reveal)

>>>> Hydroquinidine


I
II
III
aVR
aVL
aVF
V1
V2
V3
V4
V5
V6
25 mm/s 10 mm/mV
Séquentiel

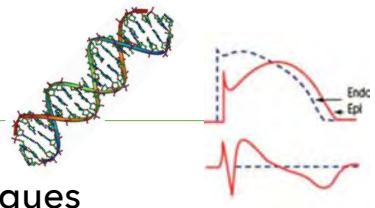
- Syncope sur terrain de foot
- Reveal : TV polymorphe

>>> Implantation d’un DAI épicardique



BRUGADA

PHYSIOPATH



Anomalie canaux sodiques
Prévalence 20 / 100 000
Mutation 20% (SCN5A ++, CACN1Ac)
Autosomique dominant
Pénétrance variable, H >> F

PRONOSTIC

Taux mort subite annuel
- 1% si type 1 spontané
- 3% si syncope
- 10% si ACR

Type 1 induit de meilleur pronostic

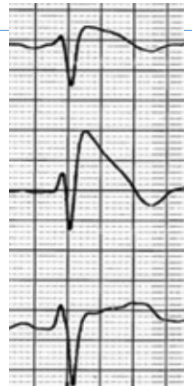
DAI PREVENTION I^aire

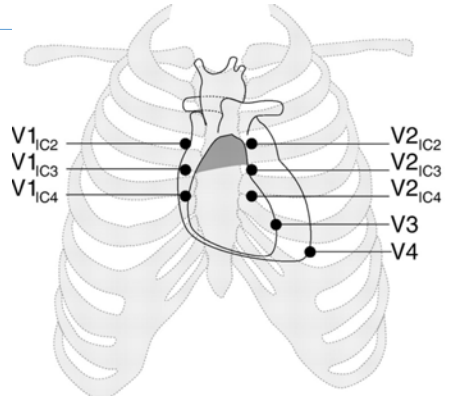
ESC European Society of Cardiology
ESC GUIDELINES

TV soutenue
Syncope rythmique

DIAGNOSTIC

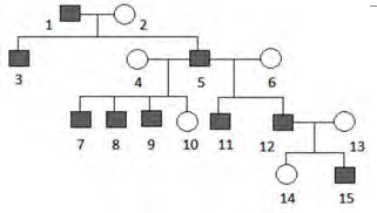
Type 1 ≥ 2 mm dans 1 dérivation
V1 ou V2 + dérivations hautes (2-3 EIC)
+/- ajmaline (1 mg/kg IV 5-10 min)





PRISE EN CHARGE

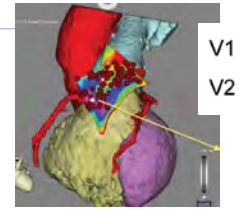
Médicaments contre indiqués
Traitement précoce fièvre
Eviter repas copieux ou alcool excessif
Restriction sportive
Dépistage familial
Orage rythmique: isuprel +/- quinidine

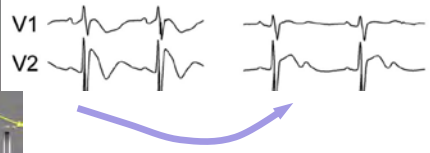


BrugadaDrugs.org
Safe drug use and the Brugada syndrome

PERSPECTIVES

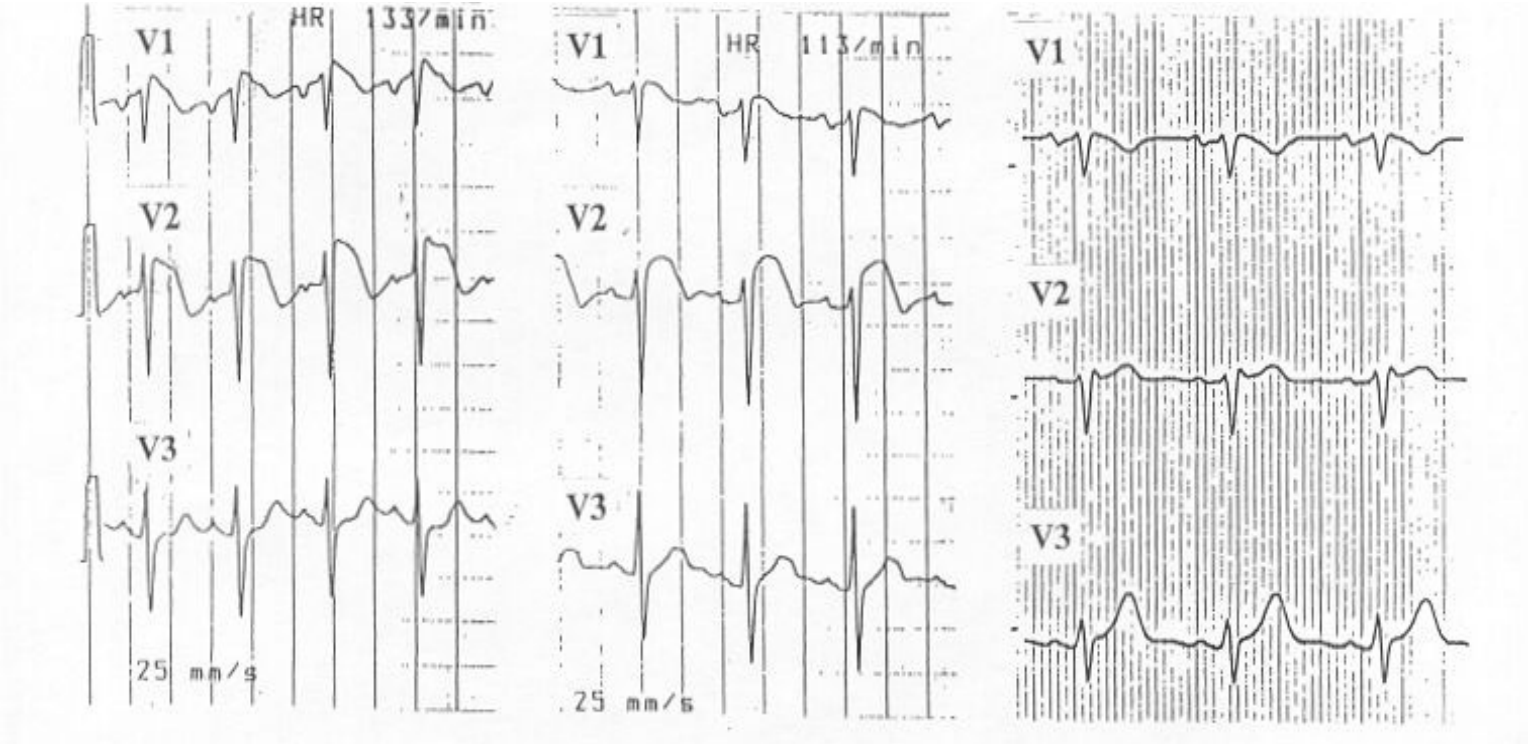
SVP controversée
Place de la quinidine
Indications élargies S-ICD ?
Ablations ?
(substrat épicardique VD)

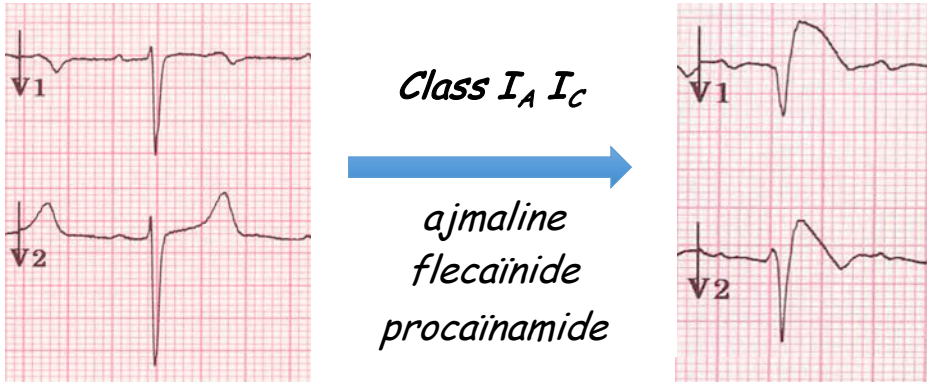




BRUGADA

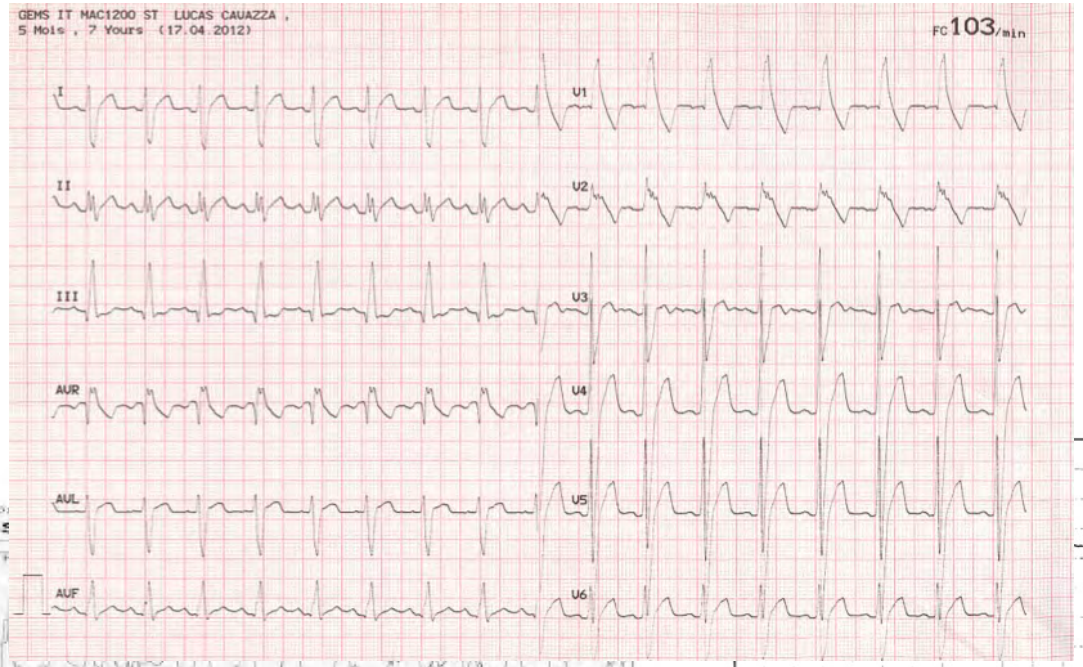
Temp. 41°C **Temp. 39°C** **Temp. 37°C**

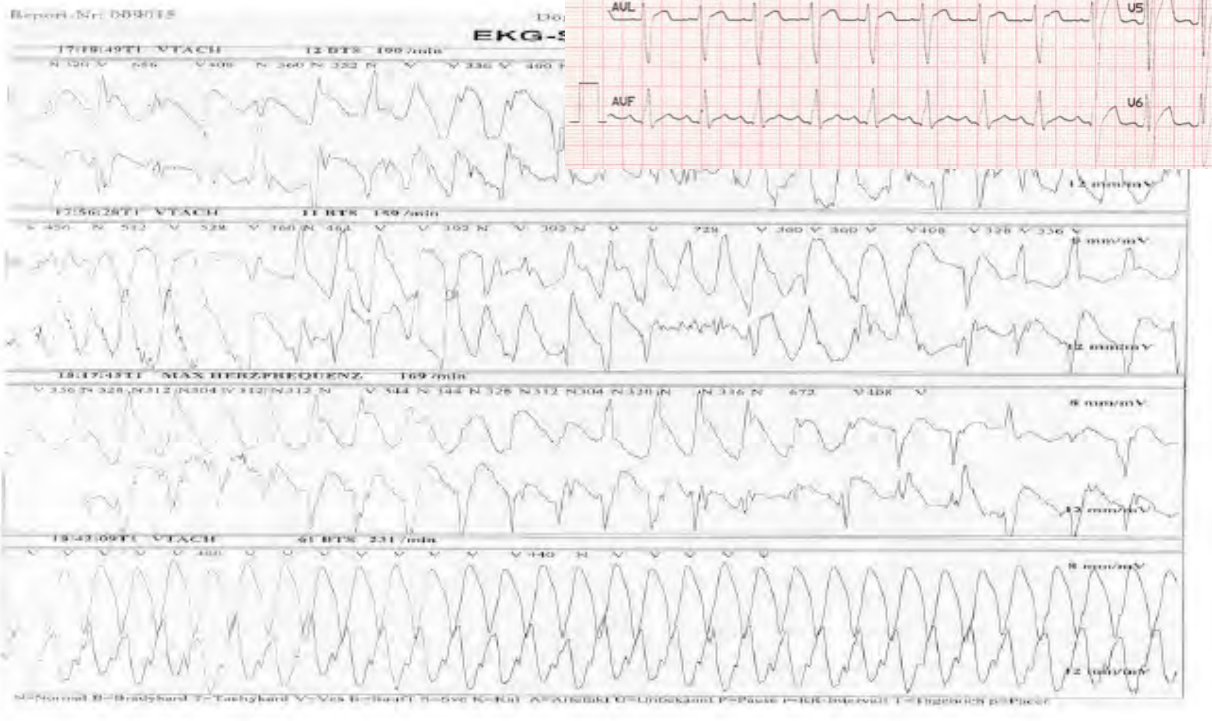


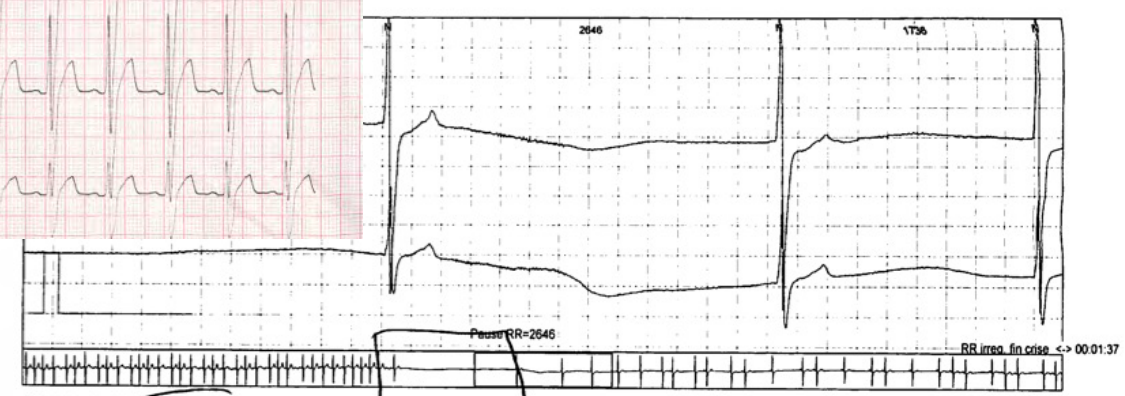


Test pharmaco après 15 ans ++
ou si morts subites pédiatriques

BRUGADA

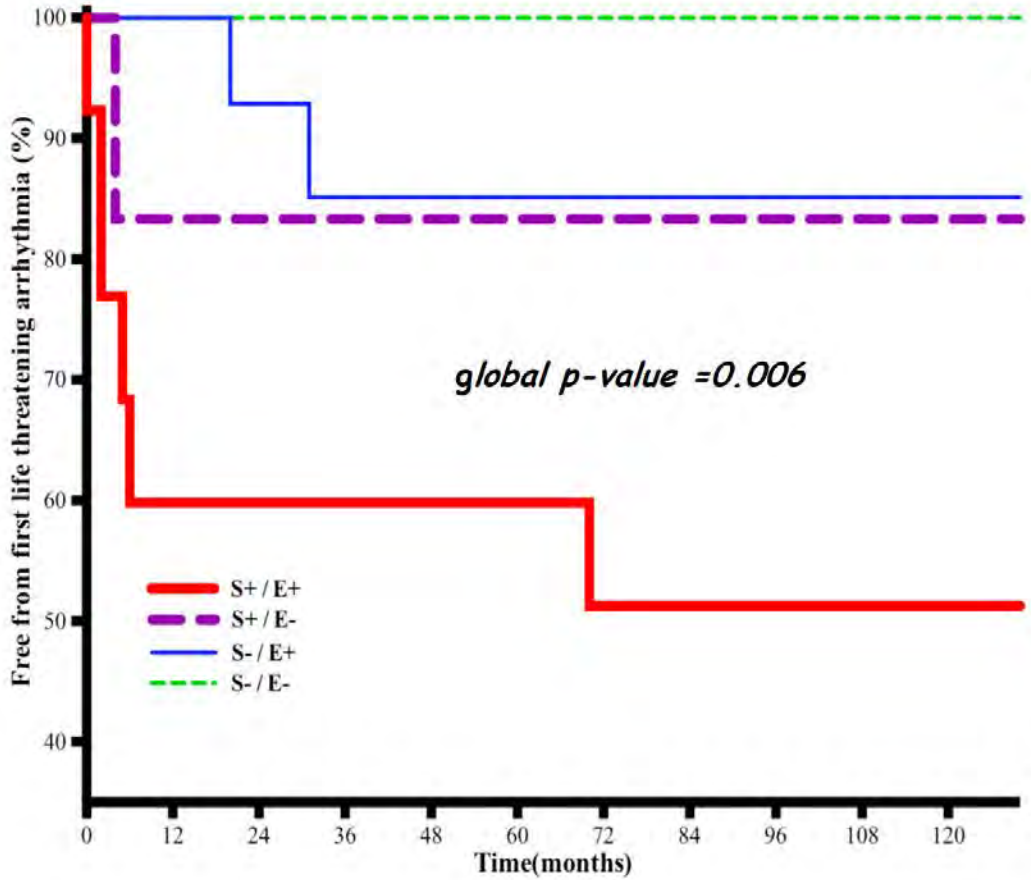

GEMS IT MAC1200 ST LUCAS CAUAZZA ,
5 Mois , 7 Yours (17.04.2012)
FC 103 /min
I
II
III
AUR
AUL
AUF
U1
U2
U3
U4
U5
U6


EKG-S


2646
1736
Pause RR=2646
RR irreg. fin crise <-> 00:01:37


22:38:16 16/12/2014 pause 3.8 sec
666
664
676
3776
680

BRUGADA



n=106 months	0	12	24	60	96	120
S+/E+ = Sympto. & Spont. Type 1	14	6	6	6	3	2
S+/E- = Sympto. & Drug induced	7	5	5	4	3	2
S-/E+ = Asympto. & Spont. Type 1	22	22	13	11	6	3
S-/E- = Asympto. & Drug induced	63	63	63	63	63	63

Asymptomatic AND drug-induced type 1 ECG pattern → LOW RISK

Others clinical situations → Intermediate risk ?

Symptoms AND Spontaneous type 1 ECG pattern → HIGH RISK

Andorin et al. Heart Rhythm 2016

BRUGADA

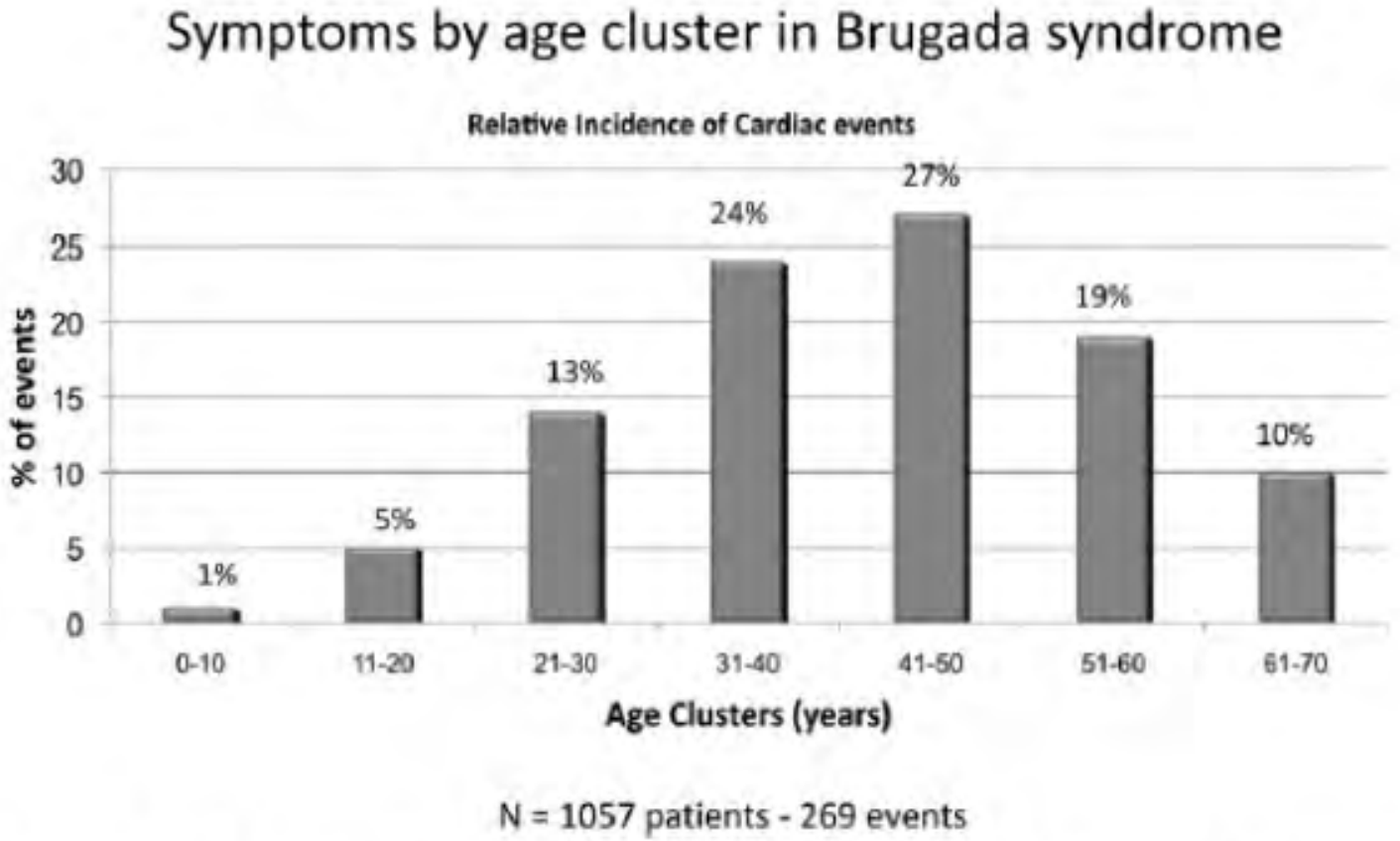
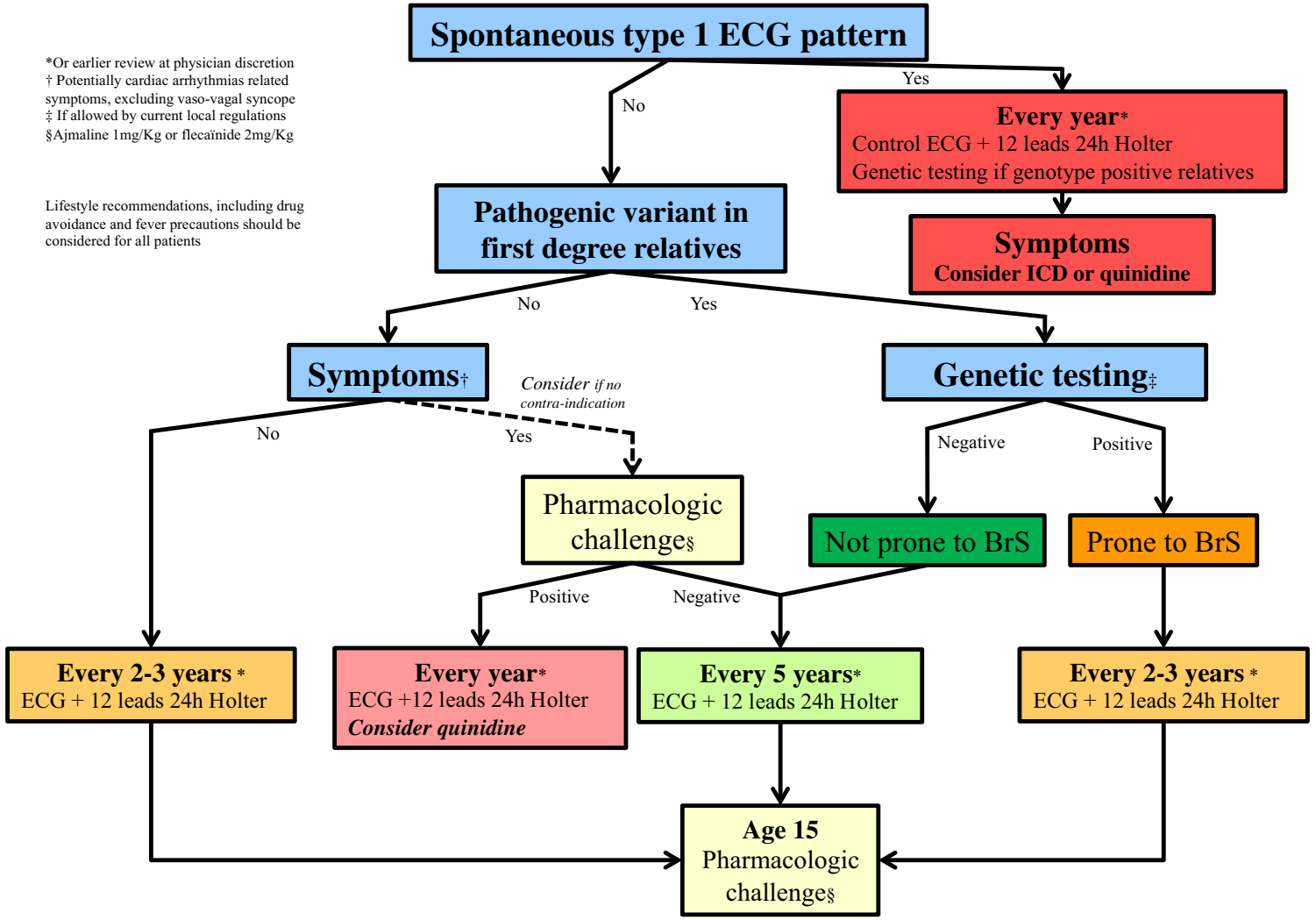


Figure 4. Relative percentage of symptomatic Brugada syndrome patients by age clusters showing a peak of incidence in the third and fourth decades of life (data from the Pavia Brugada syndrome registry).

BRUGADA



*Or earlier review at physician discretion
† Potentially cardiac arrhythmias related symptoms, excluding vaso-vagal syncope
‡ If allowed by current local regulations
§Ajmaline 1mg/Kg or flecaïnide 2mg/Kg

Lifestyle recommendations, including drug avoidance and fever precautions should be considered for all patients

Management of the Young with a known BrS in the Family

A Clinical Score Model to Predict Lethal Events in Young Patients (≤19 Years) With the Brugada Syndrome

M. Cecilia Gonzalez Corcia, MD[a,b,c,*], Juan Sieira, MD[a], Gudrun Pappaert, RN[a], Carlo de Asmundis, MD[a], Gian Battista Chierchia, MD[a], Andrea Sarkozy, MD[c], and Pedro Brugada, MD[a]

- Facteurs de risque proposés :

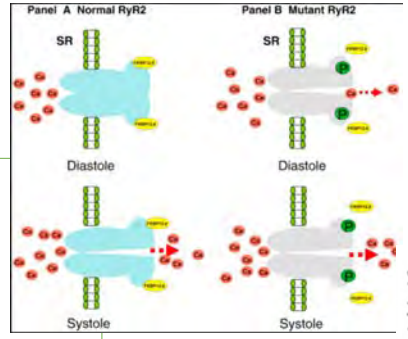
- ACR / syncope
- Pattern Brugada type 1 spontané
- Tachycardie atriale
- Troubles conductifs

Recommendations	Class[a]	Level[b]
Diagnosis		
It is recommended that BrS is diagnosed in patients with no other heart disease and a spontaneous type 1 Brugada ECG pattern.[974–976]	I	C
It is recommended that BrS is diagnosed in patients with no other heart disease who have survived a CA due to VF or PVT and exhibit a type 1 Brugada ECG induced by sodium channel blocker challenge or during fever.[135,136,975,981,982]	I	C
Genetic testing for *SCN5A* gene is recommended for probands with BrS.[164,1016]	I	C
ICD implantation is recommended in patients with BrS who: (a) Are survivors of an aborted CA and/or (b) Have documented spontaneous sustained VT.[980,990–992]	I	C
ICD implantation should be considered in patients with type 1 Brugada pattern and an arrhythmic syncope.[990,992,996]	IIa	C
Implantation of a loop recorder should be considered in BrS patients with an unexplained syncope.[997,999]	IIa	C
Quinidine should be considered in patients with BrS who qualify for an ICD but have a contraindication, decline, or have recurrent ICD shocks.[922,1006,1007]	IIa	C

CPVT

PHYSIOPATH

RyR2 ++ AD
CASQ2 AR
Mutation ~ 60%
Trigger effot/émotion ++



PRONOSTIC

1ères manifestations 10-20 ans

Mort subite à 8 ans → 11% sous B-bloquants
25% sans traitement

DAI PREVENTION Iaire

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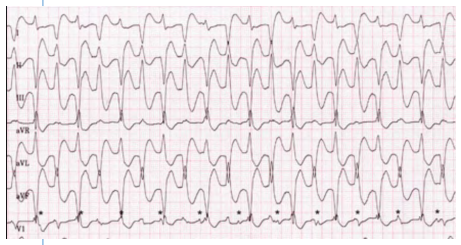
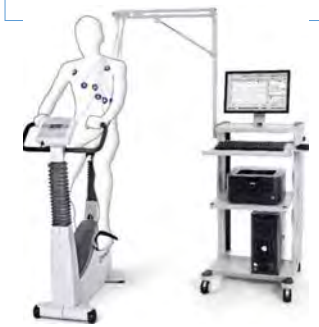
TV ou syncope sous B-bloquants

Programmation DAI → zones hautes
longue détection
Choc = stim Σ

DIAGNOSTIC

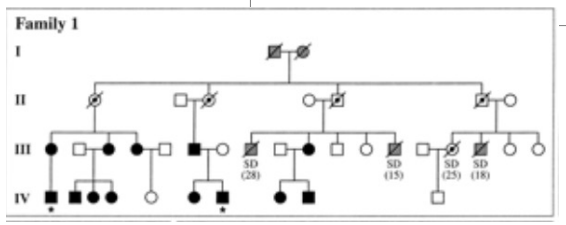
TV polymorphes
TV bidirectionnelles
Bilan morpho normal
Epreuve d'effort ++
Holter
Génétique





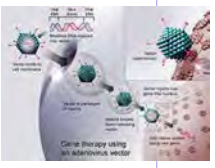
PRISE EN CHARGE

B-bloquants ++ (IIa pour porteur sain)
+/- Flecaine
Restriction sportive
Dépistage familial

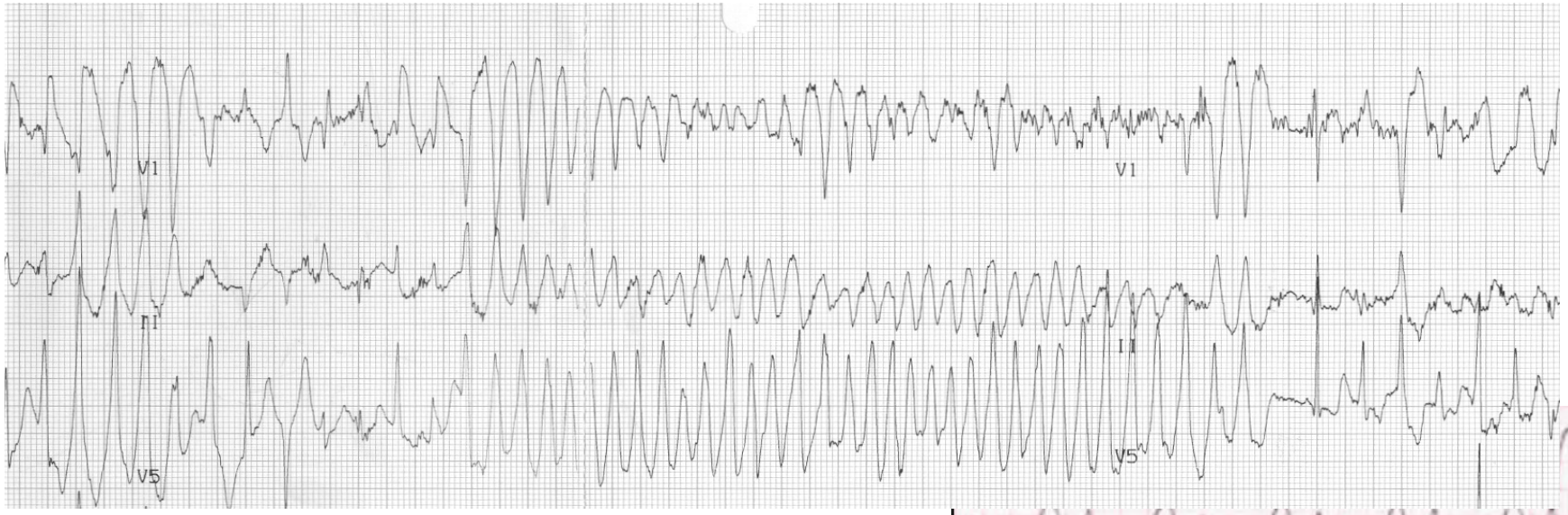


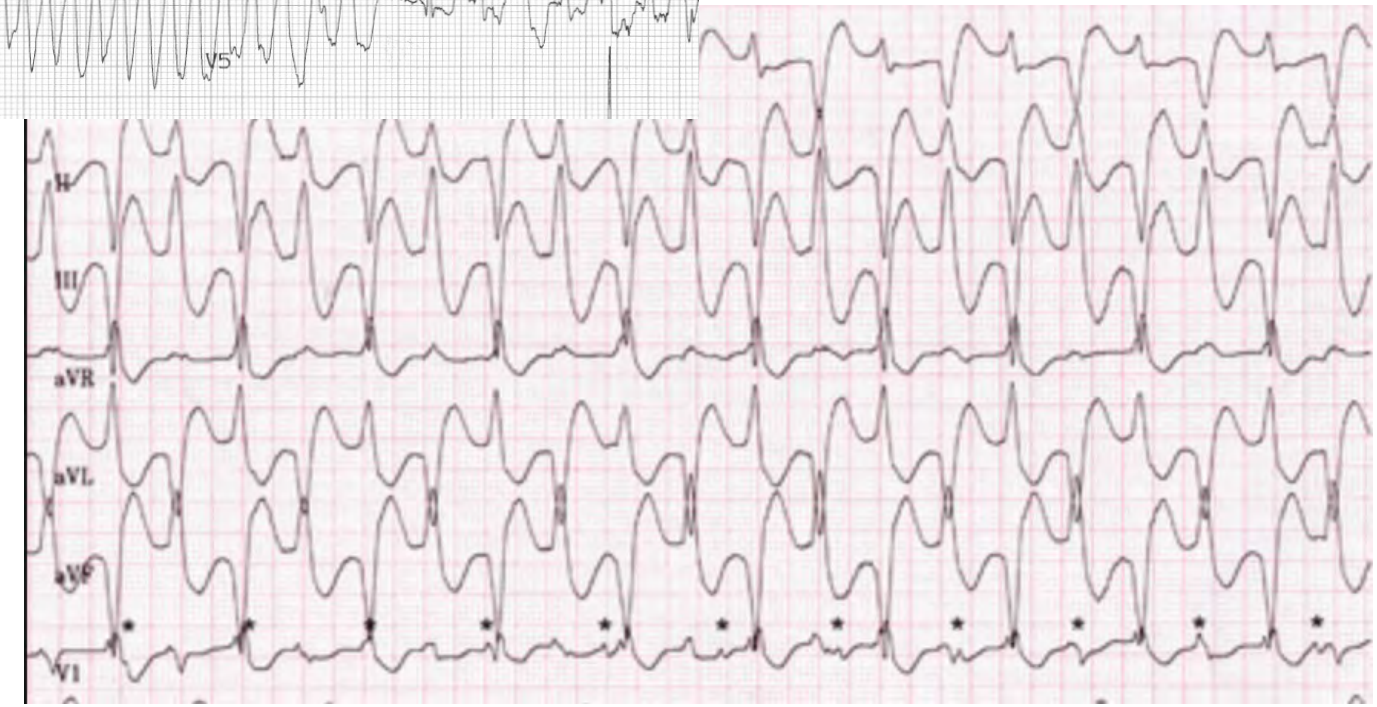
PERSPECTIVES

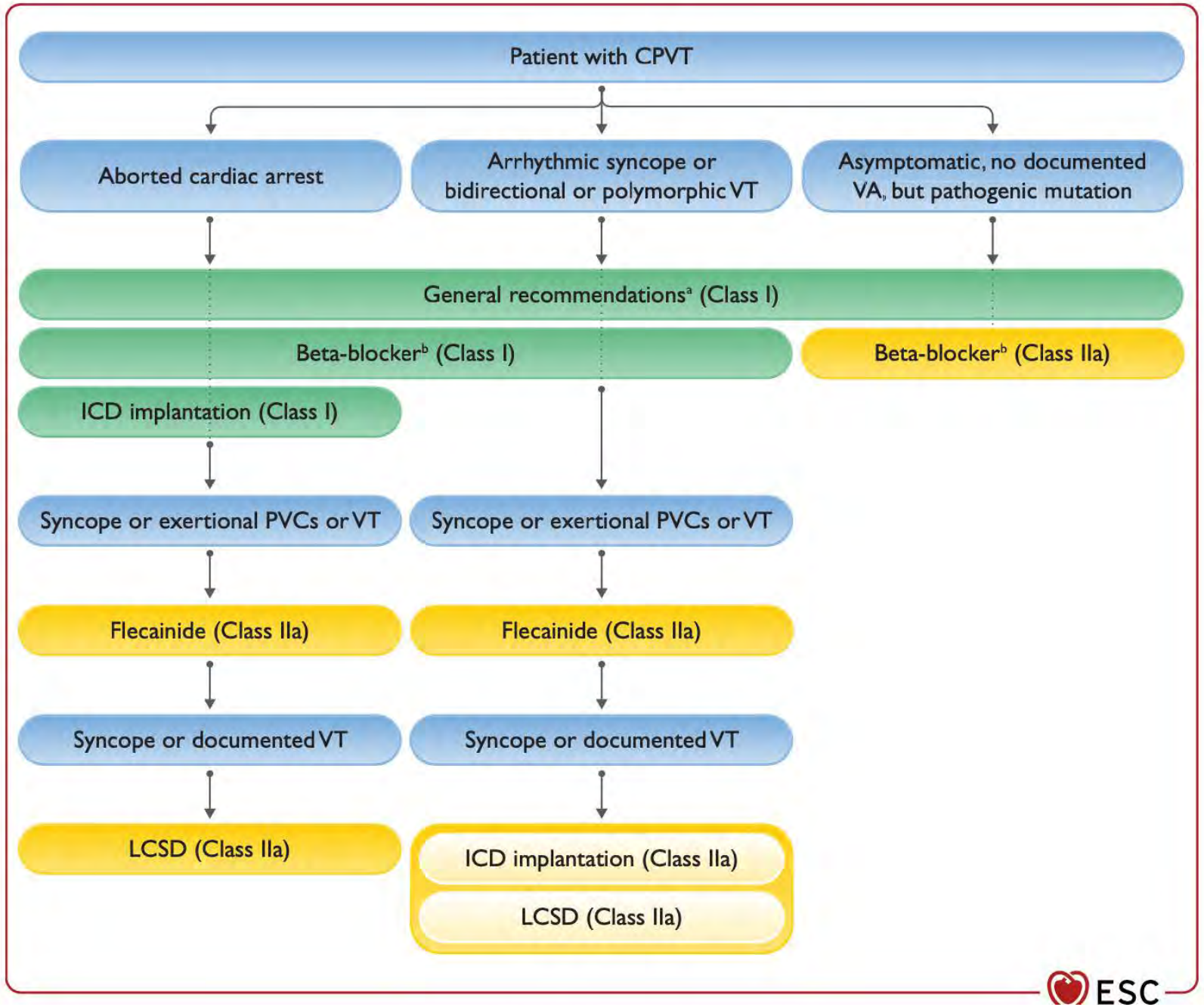
Dénervation
Verapamil
Test adrenaline
Ablation
Thérapie génique



CPVT



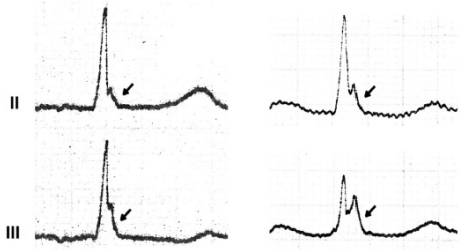



Patient with CPVT
Aborted cardiac arrest
Arrhythmic syncope or bidirectional or polymorphic VT
Asymptomatic, no documented VA, but pathogenic mutation
General recommendations[a] (Class I)
Beta-blocker[b] (Class I)
Beta-blocker[b] (Class IIa)
ICD implantation (Class I)
Syncope or exertional PVCs or VT
Syncope or exertional PVCs or VT
Flecainide (Class IIa)
Flecainide (Class IIa)
Syncope or documented VT
Syncope or documented VT
LCSD (Class IIa)
ICD implantation (Class IIa)
LCSD (Class IIa)
ESC

REPOLARISATON PRECOCE

PHYSIOPATH

??? H>F
Influence vagale
Génétique mal élucidée
Overlap Brugada
Pattern = 5% popu générale !



DIAGNOSTIC

Sus dec point J ≥1 mm dans 2 dérivations contiguës en inférieur ou latéral
+
TV polymorphe ou FV

PRISE EN CHARGE

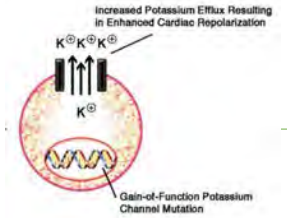
Isuprel orage rythmique
+/- Quinidine prévention IIaire

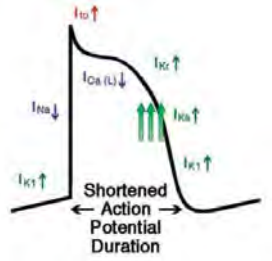
DAI PREVENTION Iaire

A priori pas d'indication
A discuter ++ centre expert (IIb)
(histoire familiale, syncope, pattern à risque)

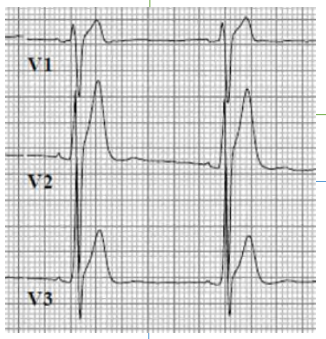
>2mm
ST horizontal ou descendant

QT COURT





5 gènes mais mutation 20%
Overlap gènes LQTS et Brugada
40% de mort subite à 40 ans



QTc ≤ 340
QTC ≤ 360 +:
Mutation
Histoire famille SQTS ou SCD < 40 ans
TV/FV idiopathique

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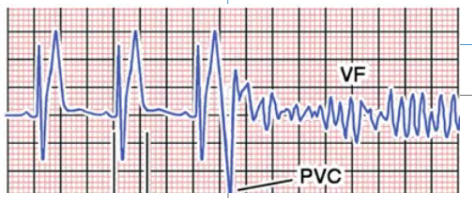


Quidine ? Sotalol ?
Si refus ou CI de DAI ou mort subite familiale (IIb)

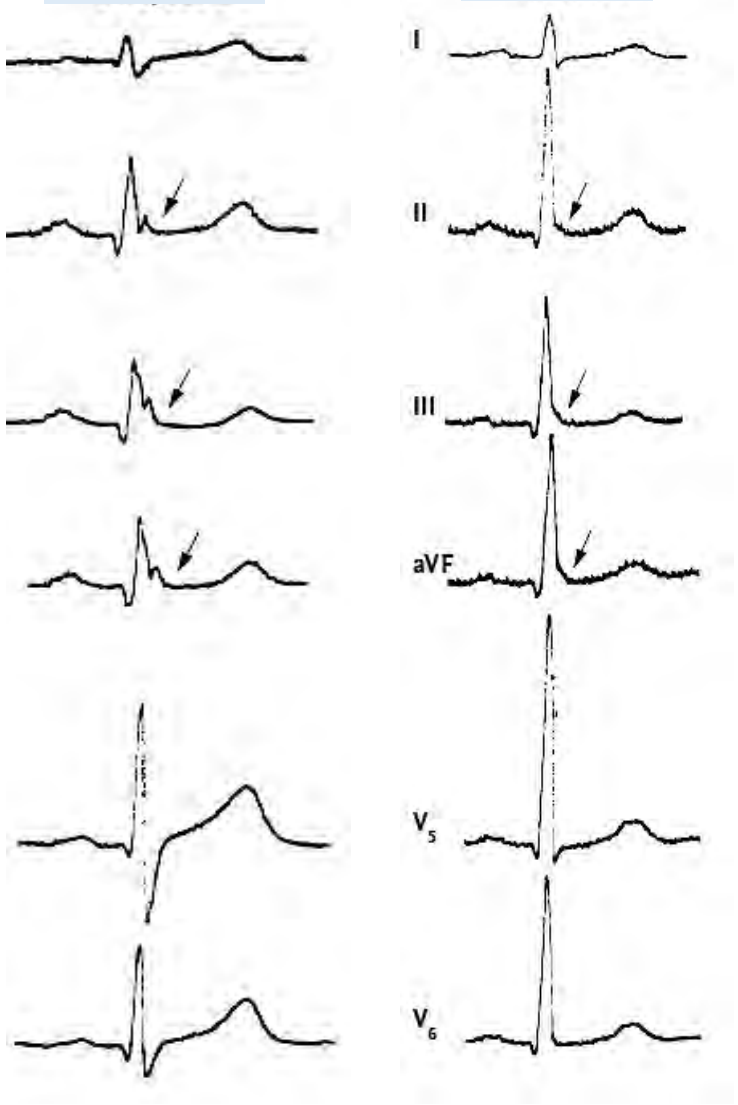
TV soutenue
Histoire familiale SCD ? (IIb)

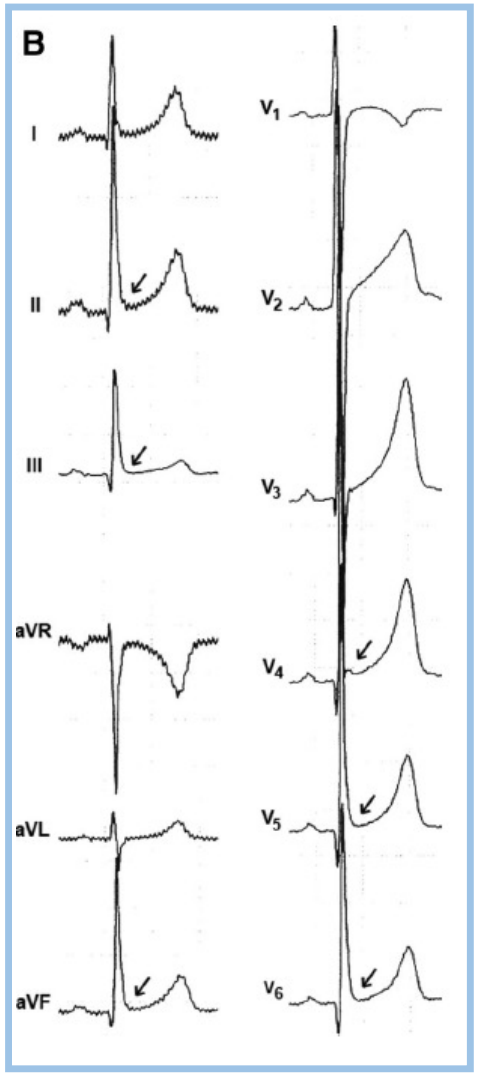
Eliminer cause 2aire:
hyperCa, hyperK, acidose, tachycardie, catécholamines…

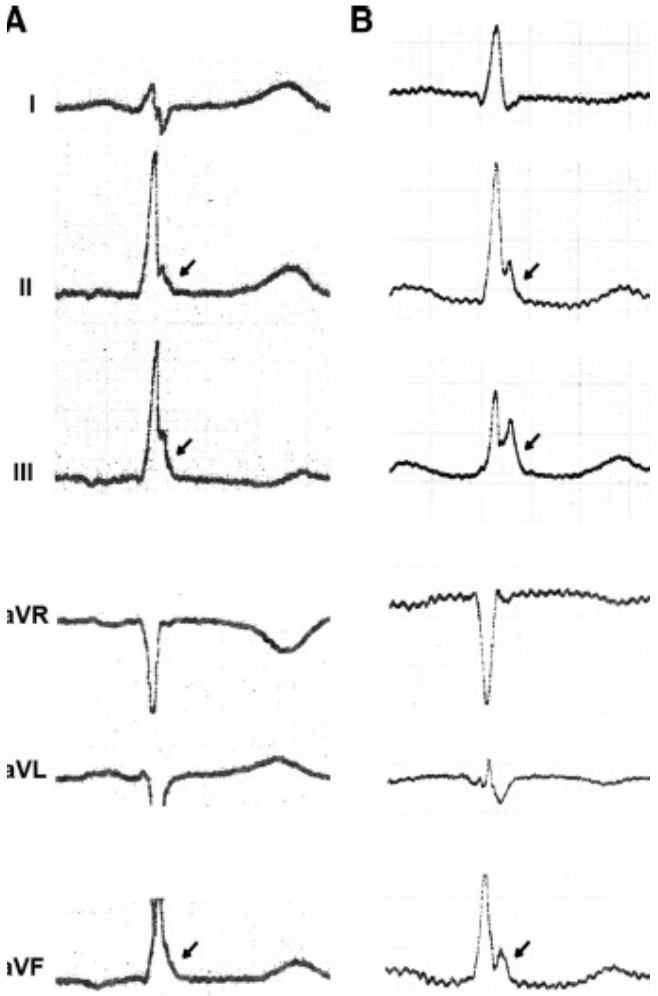
REPOLARISATON PRECOCE

Notching

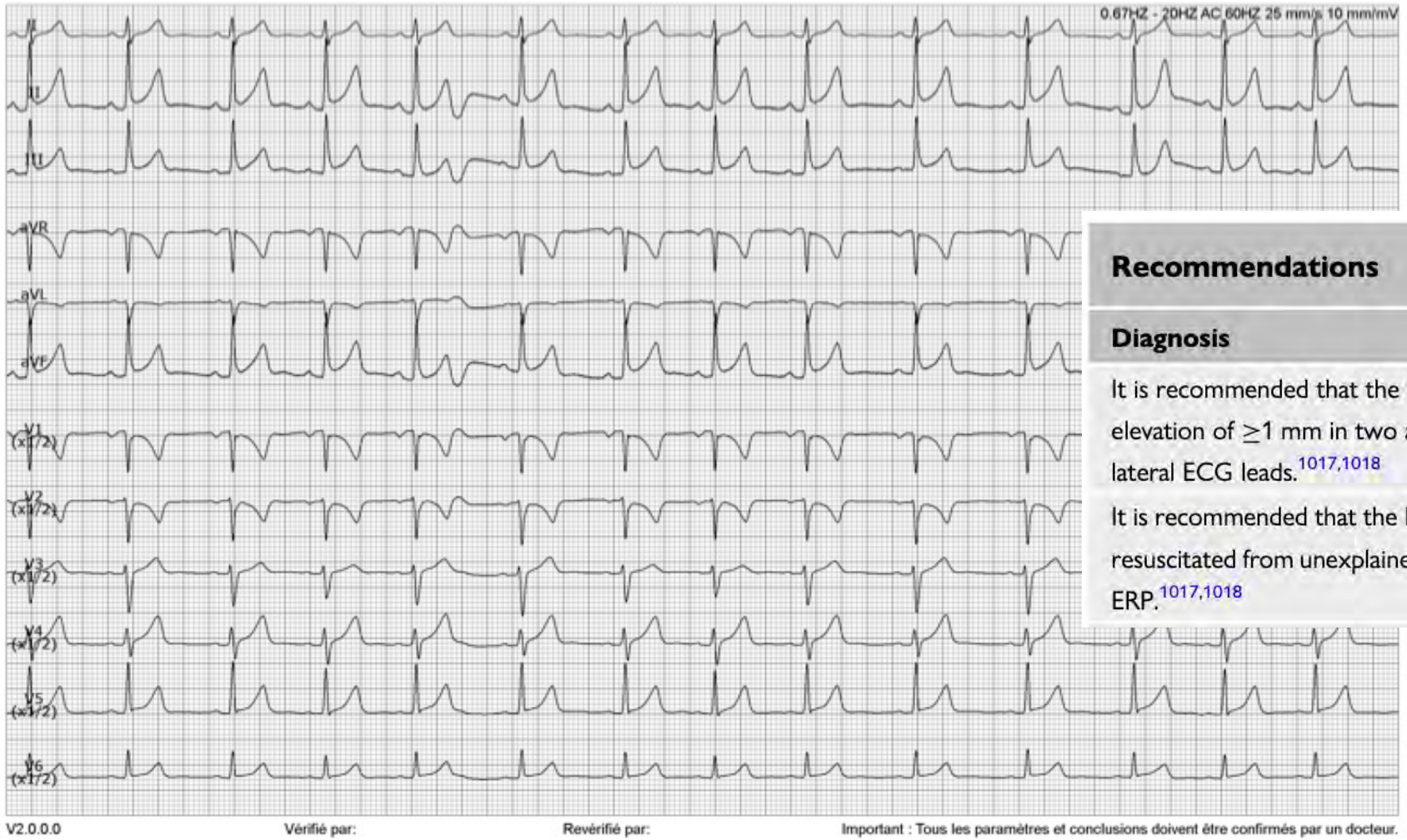
Slurring



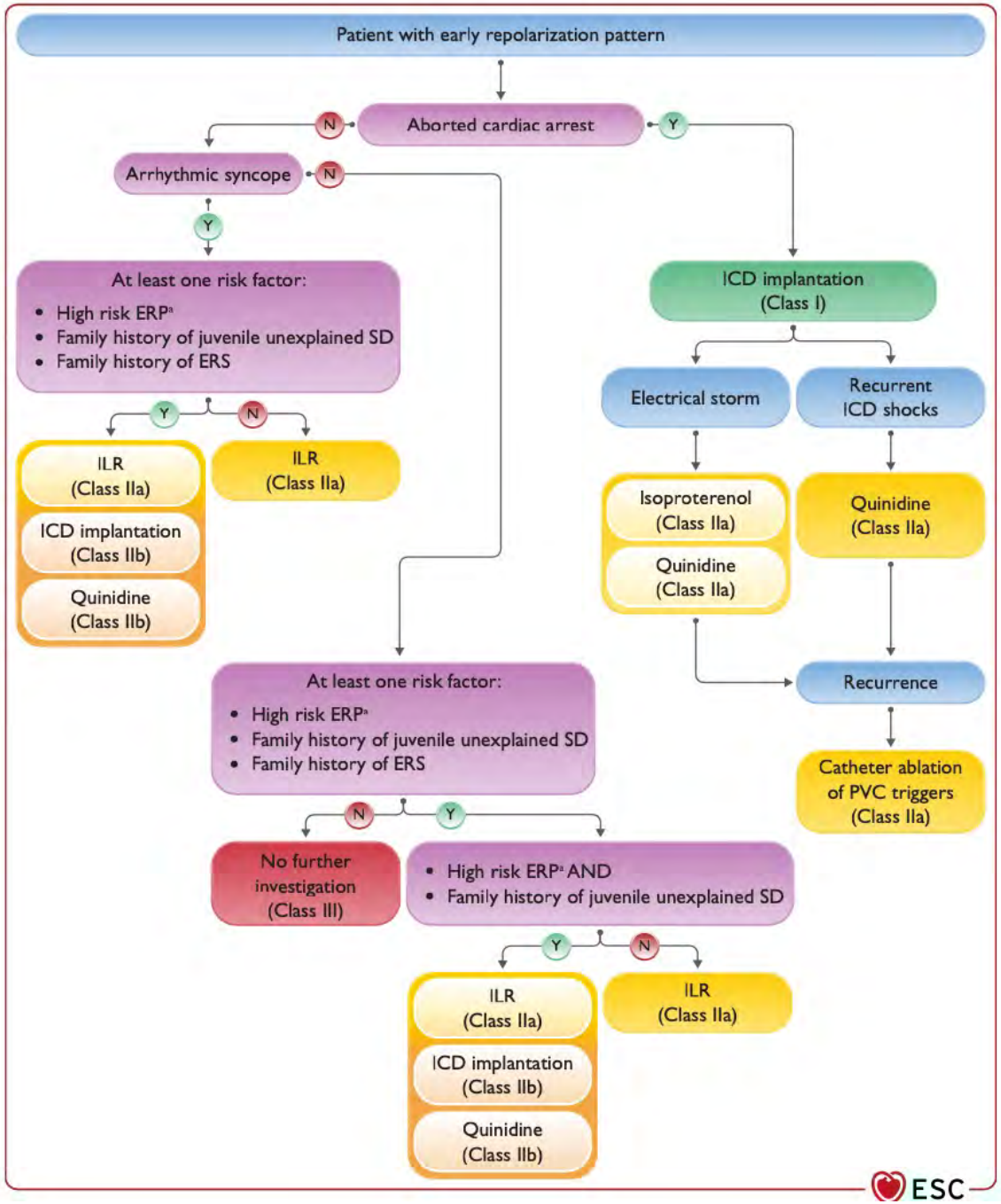


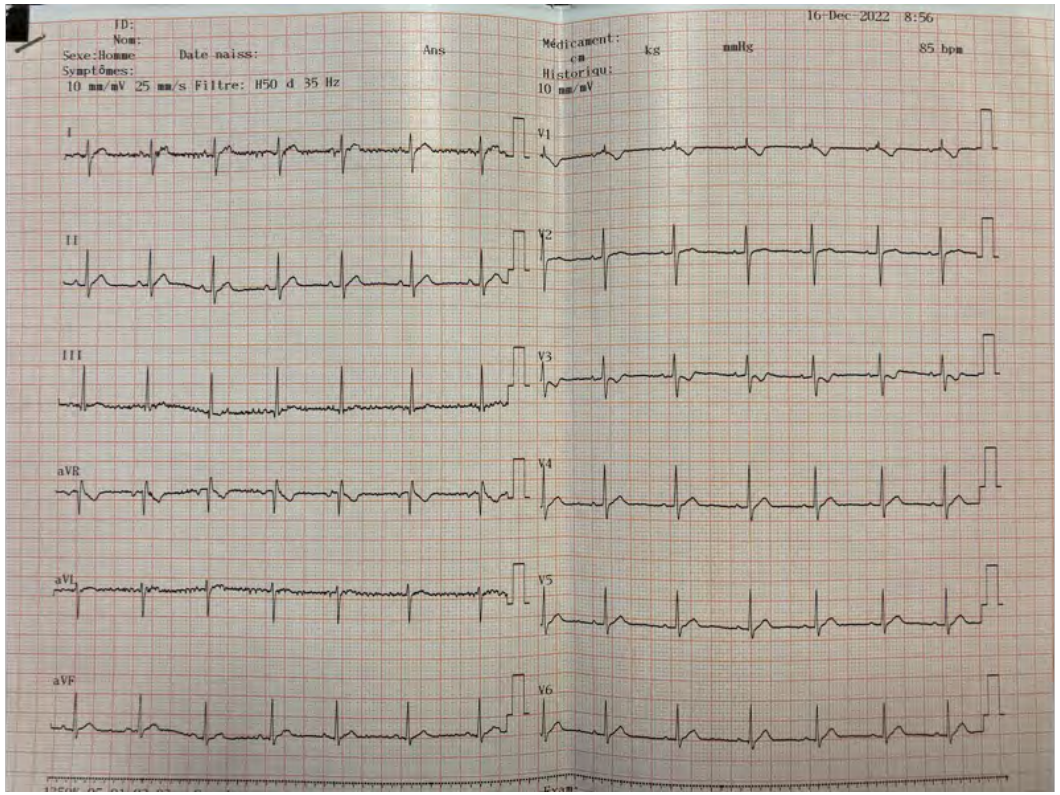


ST horizontal ou descendant = mauvais pronostic



Recommendations	Class[a]	Level[b]
Diagnosis		
It is recommended that the ERP is diagnosed as J-point elevation of ≥1 mm in two adjacent inferior and/or lateral ECG leads.[1017,1018]	I	C
It is recommended that the ERS is diagnosed in a patient resuscitated from unexplained VF/PVT in the presence of ERP.[1017,1018]	I	C

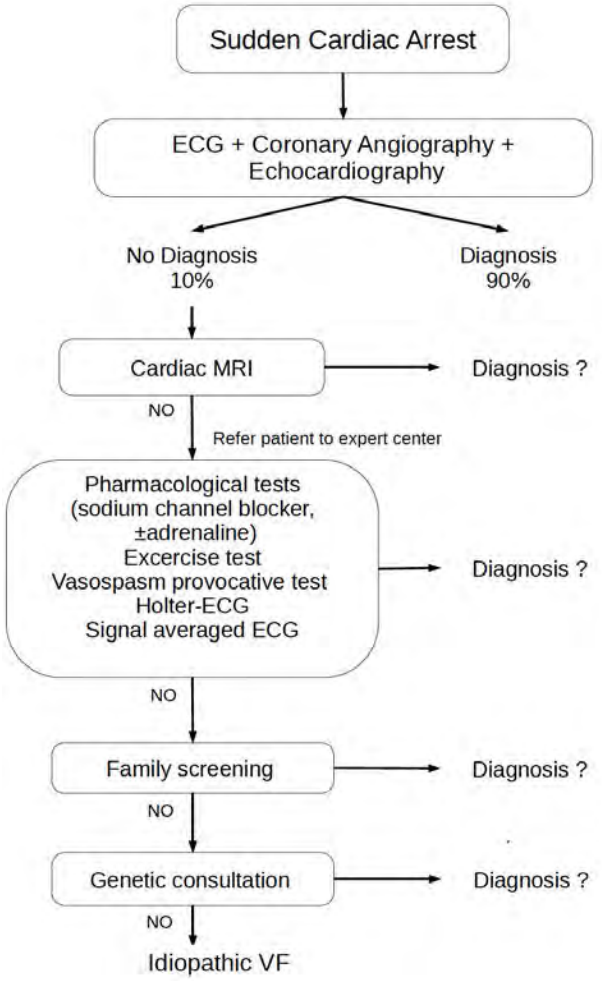

Patient with early repolarization pattern
Aborted cardiac arrest
N
Y
Arrhythmic syncope
N
Y
At least one risk factor:
• High risk ERP[a]
• Family history of juvenile unexplained SD
• Family history of ERS
Y
N
ILR (Class IIa)
ICD implantation (Class IIb)
Quinidine (Class IIb)
ILR (Class IIa)
ICD implantation (Class I)
Electrical storm
Recurrent ICD shocks
Isoproterenol (Class IIa)
Quinidine (Class IIa)
Quinidine (Class IIa)
Recurrence
Catheter ablation of PVC triggers (Class IIa)
At least one risk factor:
• High risk ERP[a]
• Family history of juvenile unexplained SD
• Family history of ERS
N
Y
No further investigation (Class III)
• High risk ERP[a] AND
• Family history of juvenile unexplained SD
Y
N
ILR (Class IIa)
ICD implantation (Class IIb)
Quinidine (Class IIb)
ILR (Class IIa)
ESC



Recommendations	Class[a]	Level[b]
Diagnosis		
It is recommended that SQTS is diagnosed in the presence of a QTc ≤360 ms and one or more of the following: (a) a pathogenic mutation, (b) a family history of SQTS, (c) survival from a VT/VF episode in the absence of heart disease.[1061,1068]	I	C
Genetic testing is indicated in patients diagnosed with SQTS.[1063]	I	C

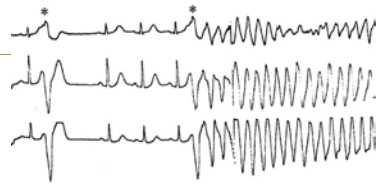
FV IDIOPATHIQUE

DIAGNOSTIC



PHYSIOPATH

???
ESV du Purkinje
5% ACR



PRONOSTIC

Risque de récidive significatif
20% à 4 ans registre francilien

PRISE EN CHARGE

Bilan étiologique exhaustif ++
Prise en charge psychologique
DAI en prévention II^{aire}
Enquête familiale:

Approach	Action*
History taking and physical examination	• Personal clinical history • Family history focused on cardiac diseases or sudden deaths
ECG	• Baseline 12-lead ECG with standard and high precordial leads • 24-hour ambulatory ECG • Exercise stress test • Signal-averaged ECG • Provocative test with ajmaline/flecainide (when Brugada syndrome is suspected)
Cardiac imaging	• Two-dimensional echocardiography and/or CMR (with or without contrast)
Genetic testing	• Targeted molecular testing and genetic counselling if there is the clinical suspicion of a specific disease • Referral to a tertiary centre specialized in evaluation of the genetics of arrhythmias

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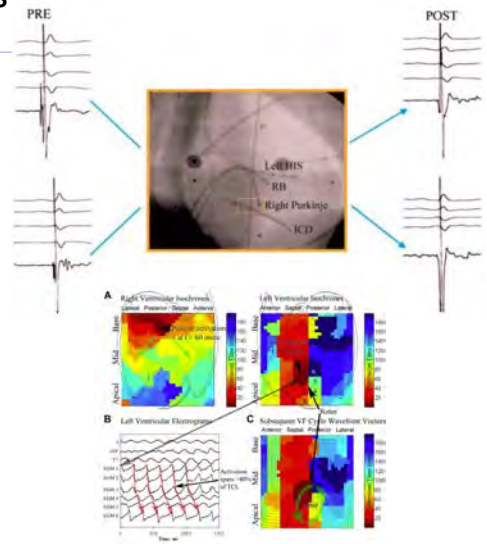
PERSPECTIVES

Nouveaux phénotypes ?
Ablation ESV initiatrice
Rotors



FV IDIOPATHIQUE

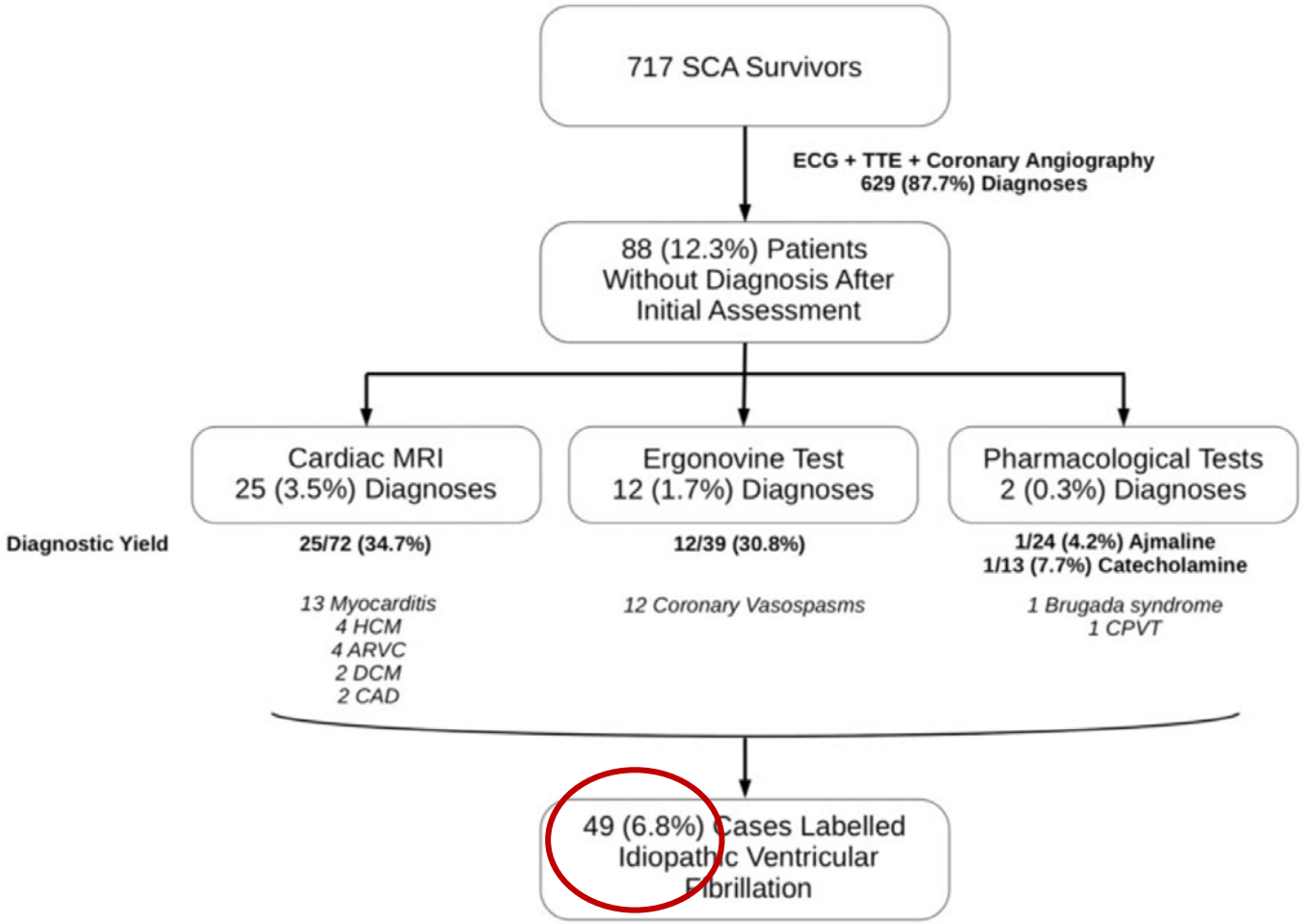


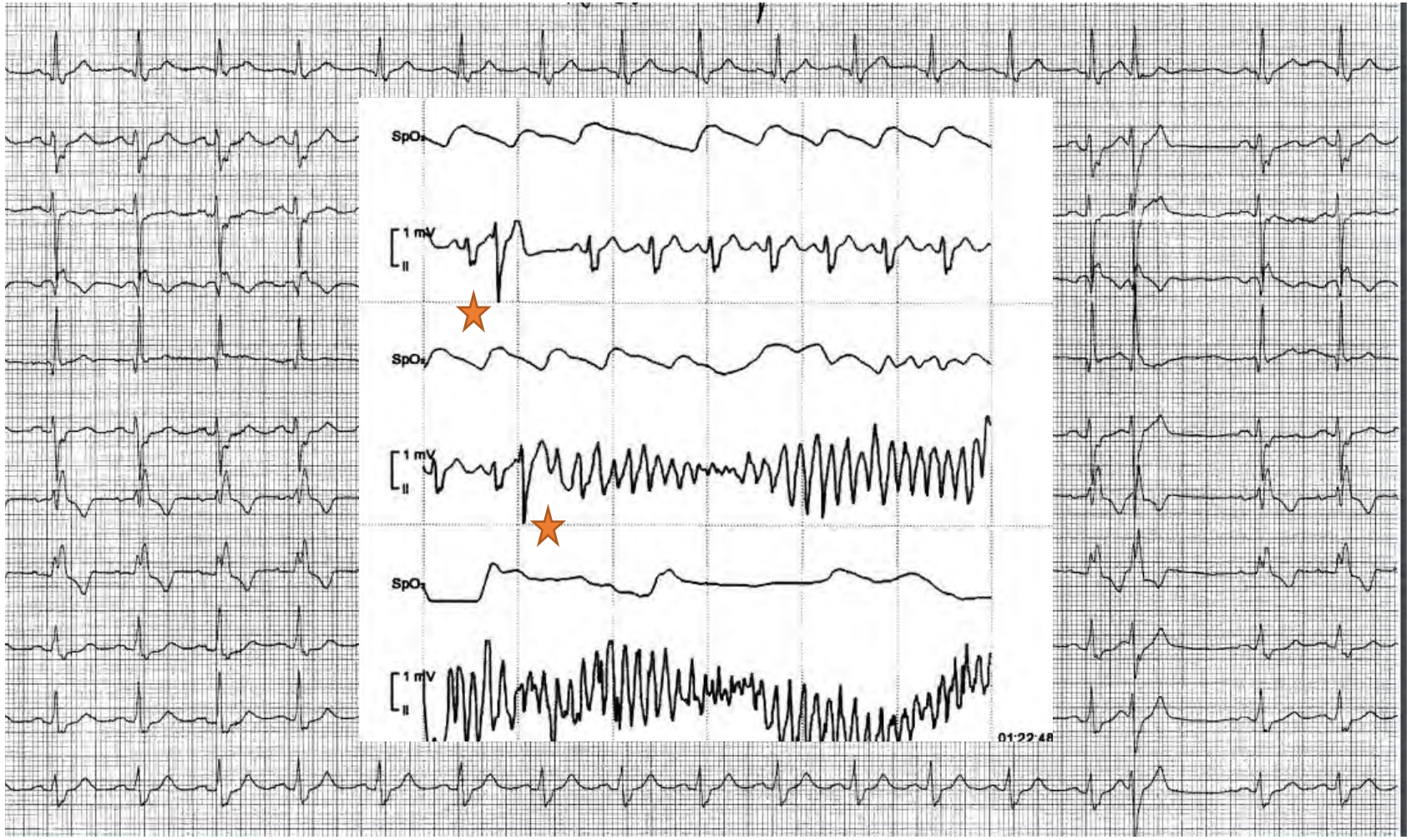
Table 2 Medical investigations of cases labelled idiopathic ventricular fibrillation (performed during the index hospitalization following the sudden cardiac arrest or planned subsequently after discharge)

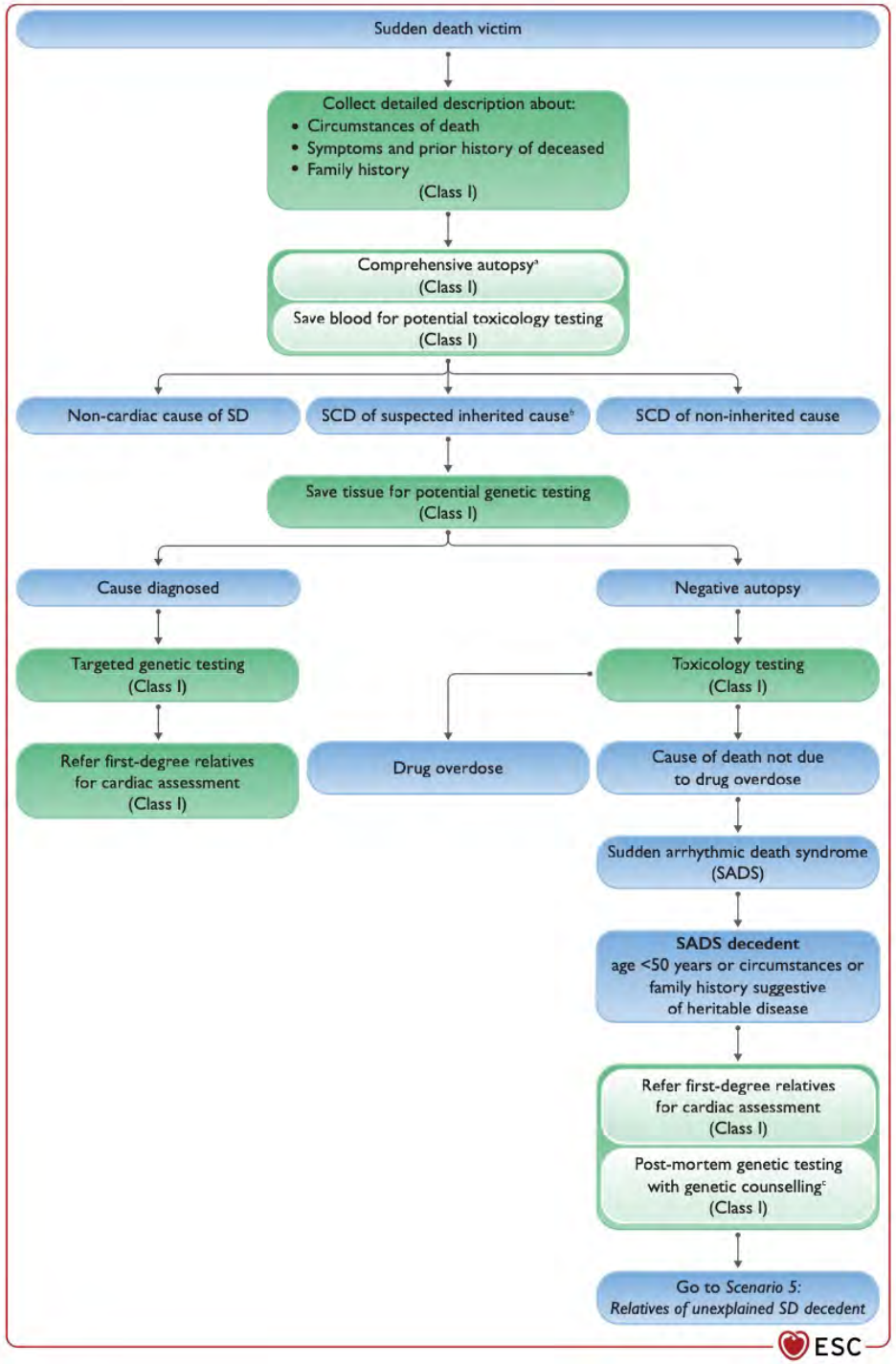
	IVFs (n = 49), n (%)
Coronary angiography	47 (95.9)
Cardiac MRI	40 (81.6)
Provocative testing	
Ergonovine	19 (38.8)
Ajmaline	21 (42.9)
Isoprenaline	10 (20.4)
Adenosine	2 (4.1)
Adrenaline	0 (0)
Electrophysiological study	12 (24.5)
Genetic testing	9 (18.4)
Holter-ECG	6 (12.2)
Right ventricular angiography	5 (10.2)
Exercise testing	4 (8.2)
Signal averaged ECG	2 (4.1)
Coronary CT	1 (2.0)
Cardiac scintigraphy (for ARVC)	1 (2.0)
Cardiac biopsy	0 (0)

Waldmann et al. European Heart Journal 2018

ESV du Purkinje

Aspect de BBD + hémi bloc
ESV à couplage court initiatrice de FV





- Proposer autopsie ++
- Prélèvements génétiques

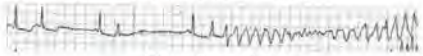
QT LONG

PHYSIOPATH

17 gènes connus – 1/2500
Mutation identifiée dans 75%:
LQTS1 KCNQ1 (effort ++, natation)
LQTS2 KNCH2 (émotion ou bruit)
LQTS3 SCN5A (repos ou sommeil)
Autosomique dominant (95%)

PRONOSTIC

Taux mort subite annuel 0.3-0.9%
5% par an si antécédent de syncope
LQT3 et QTc > 500 ms à haut risque



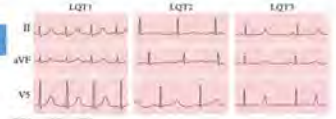
« Short-long-short »

DAI PREVENTION Iaire

ESC GUIDELINES

TV malgré B-bloquants
Syncope

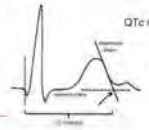
DIAGNOSTIC



QTc ≥ 480 ms
Score > 3
Mutation positive
QTc > 460 ms + syncope inexpliquée

Bazett

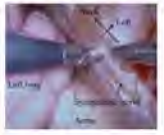
$$QTc\ interval = \frac{QT\ interval}{\sqrt{RR\ interval}}$$



D2 ++ ou V5

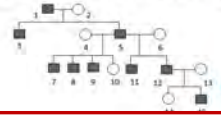
PERSPECTIVES

Dénervation
Flecaine ou mexiletine LQTS3
Stratification guidée par génétique



PRISE EN CHARGE

Médicaments contre indiqués
B-bloquants ++ (IIa pour porteur sain)
Restriction sportive
Dépistage familial
Orage rythmique: isuprel +/- SEES



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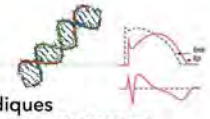
BRUGADA

PHYSIOPATH



Anomalie canaux sodiques
Mutation 20% (SCN5A ++, CACN1Ac)
Autosomique dominant
Pénétrance variable, H >> F

PRONOSTIC

Taux mort subite annuel
- 1% si type 1 spontané
- 3% si syncope
- 10% si ACR

Type 1 induit de meilleur pronostic

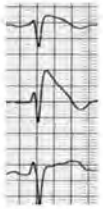
DAI PREVENTION Iaire

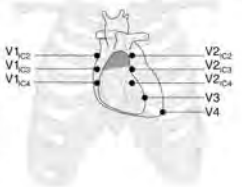
ESC GUIDELINES

TV soutenue
Syncope rythmique

DIAGNOSTIC

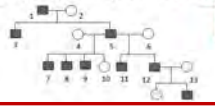
Type 1 ≥ 2 mm dans 1 dérivation
V1 ou V2 + dérivations hautes (2-3 EIC)
+/- ajmaline (1 mg/kg IV 5-10 min)





PRISE EN CHARGE

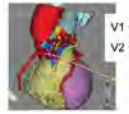
Médicaments contre indiqués
Traitement précoce fièvre
Eviter repas copieux ou alcool excessif
Restriction sportive
Dépistage familial
Orage rythmique: isuprel +/- quinidine

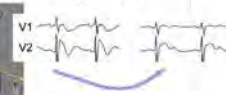


BrugadaDrugs.org

PERSPECTIVES

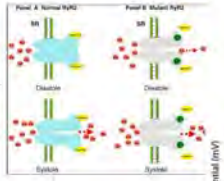
SVP controversée
Place de la quinidine
Indications élargies S-ICD ?
Ablations ?
(substrat épicardique VD)



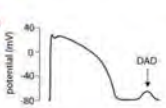


CPVT

PHYSIOPATH



RyR2 ++ AD
CASQ2 AR
Mutation ~ 60%
Trigger effot/émotion ++



PRONOSTIC

1ères manifestations 10-20 ans

Mort subite à 8 ans
- 11% sous B-bloquants
- 25% sans traitement

DAI PREVENTION Iaire

ESC GUIDELINES

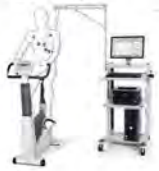
TV ou syncope sous B-bloquants

Programmation DAI
- zones hautes
- longue détection

Choc = stim Σ

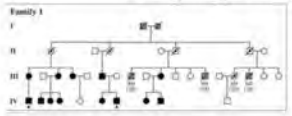
DIAGNOSTIC

TV polymorphes
TV bidirectionnelles
Bilan morpho normal
Epreuve d'effort ++
Holter
Génétique



PRISE EN CHARGE

B-bloquants ++ (IIa pour porteur sain)
+/- Flecaine
Restriction sportive
Dépistage familial



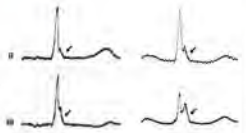
PERSPECTIVES

Dénervation
Verapamil
Test adrenaline
Ablation
Thérapie génique

REPOLARISATON PRECOCE

PHYSIOPATH

??? H>F
Influence vagale
Génétique mal élucidée
Overlap Brugada
Pattern = 5% popu générale !



DIAGNOSTIC

Sus dec point J ≥1 mm dans 2 dérivations contiguës en inférieur ou latéral
+
TV polymorphe ou FV

PRISE EN CHARGE

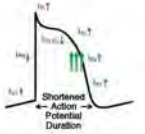
Isuprel orage rythmique
+/- Quinidine prévention IIaire

DAI PREVENTION Iaire

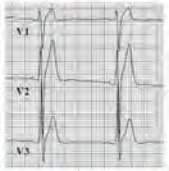
A priori pas d'indication
A discuter ++ centre expert (IIb)
(histoire familiale, syncope, pattern à risque)

>2mm
ST horizontal ou descendant

QT COURT

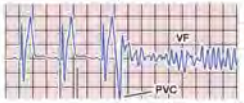


5 gènes mais mutation 20%
Overlap gènes LQTS et Brugada
40% de mort subite à 40 ans



QTc ≤ 340
QTC ≤ 360 +:
Mutation
Histoire famille SQTS ou SCD < 40 ans
TV/FV idiopathique

ESC GUIDELINES



Quidine ? Sotalol ?
Si refus ou CI de DAI ou mort subite familiale (IIb)

TV soutenue
Histoire familiale SCD ? (IIb)

Eliminer cause 2aire:
hyperCa, hyperK, acidose, tachycardie, catécholamines...

Merci pour votre attention !