Cardiomyopathies:
de A à Z

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WIKIPÉDIA
L'encyclopédie libre

Cardiomyopathie

Une **cardiomyopathie** ou *myocardiopathie* (littéralement, *maladie du muscle cardiaque*) correspond à un groupe hétérogène de maladies touchant le myocarde, responsable d'un dysfonctionnement de ce muscle.

Les cardiomyopathies peuvent avoir une origine extrinsèque (secondaire à une pathologie extérieure au muscle cardiaque) ou intrinsèque (par atteinte primitive du muscle cardiaque).

À terme, les sujets atteints risquent insuffisance cardiaque, troubles du rythme et mort subite.

Extrinsèque: = cardiopathies

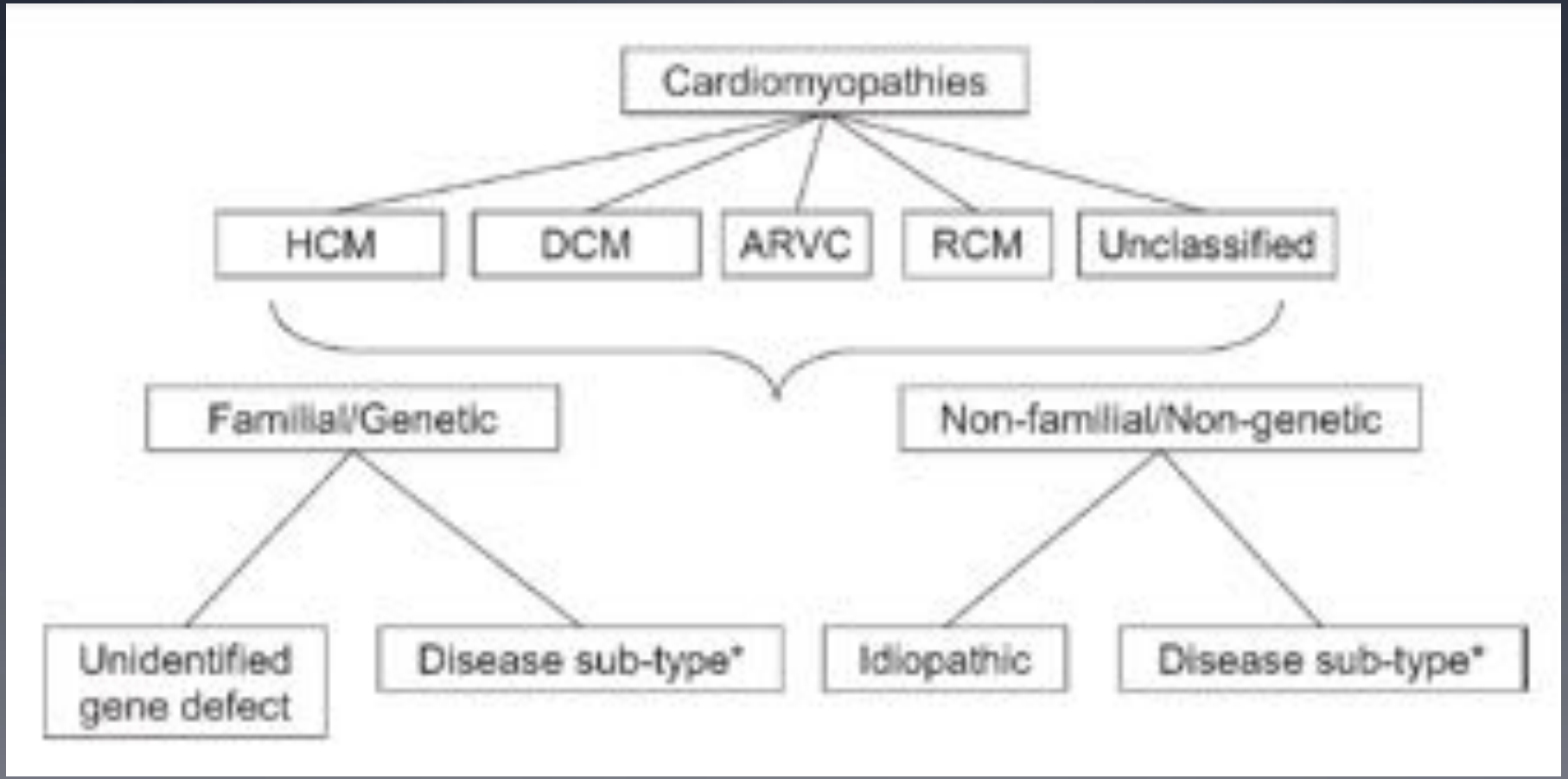
- Ischémique
- Congénitale
- Hypertensive
- Valvulaire
- Post myocardite
- Toxique:
  - Alcool
  - Chimiothérapie (anthracyclines)
- Métabolique:
  - surcharge en fer
  - Amylose (portugais)
  - Hyperthyroïdie
  - Diabète
- Auto-immune
  - Lupus
  - Sclérodermie
  - Périartétite noueuse
- Malnutrition

Cardiomyopathie (CM): Définition

In this statement we define a cardiomyopathy as: A myocardial disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.

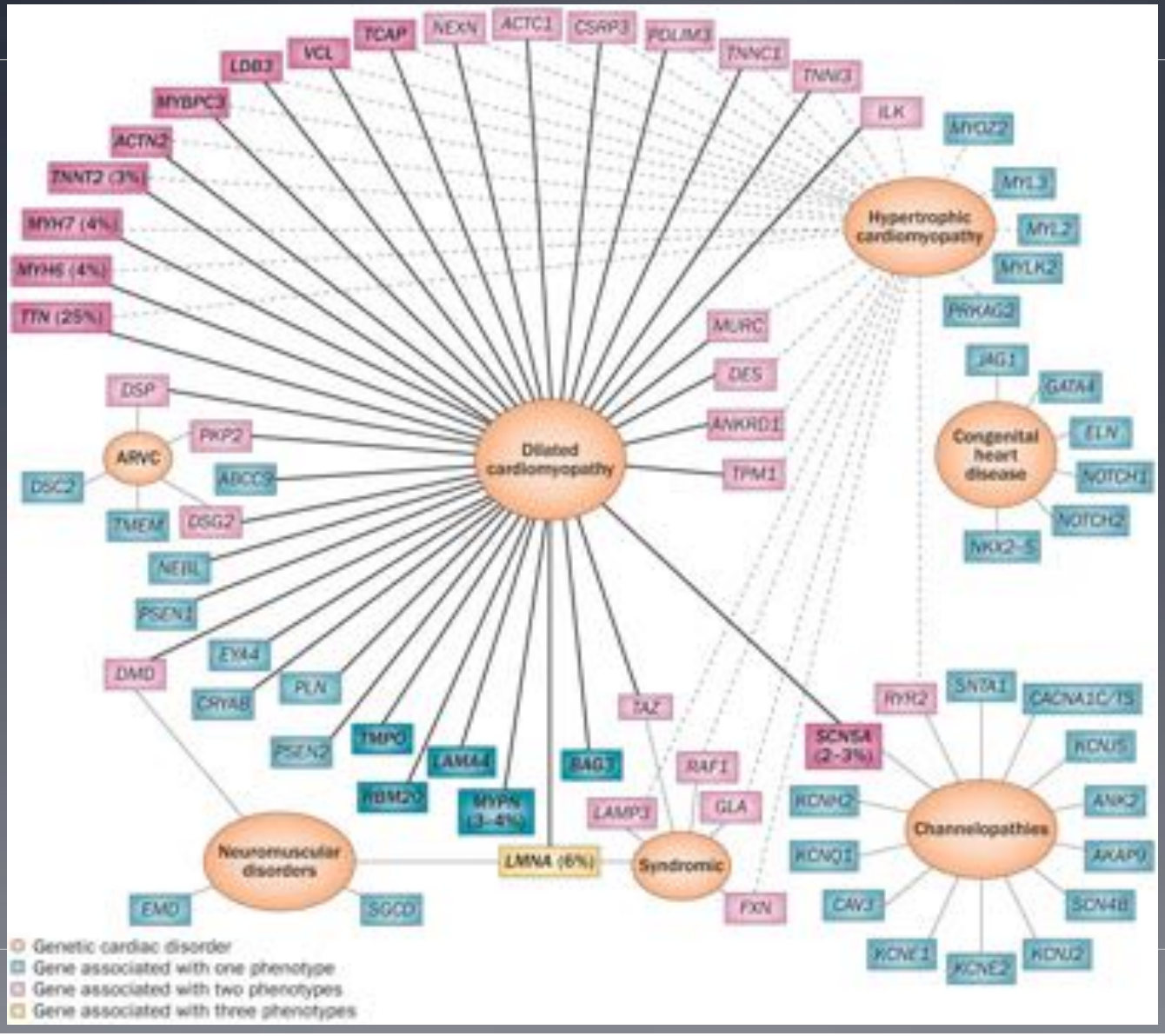
European Heart Journal (2008) 29, 270

CM: Classification



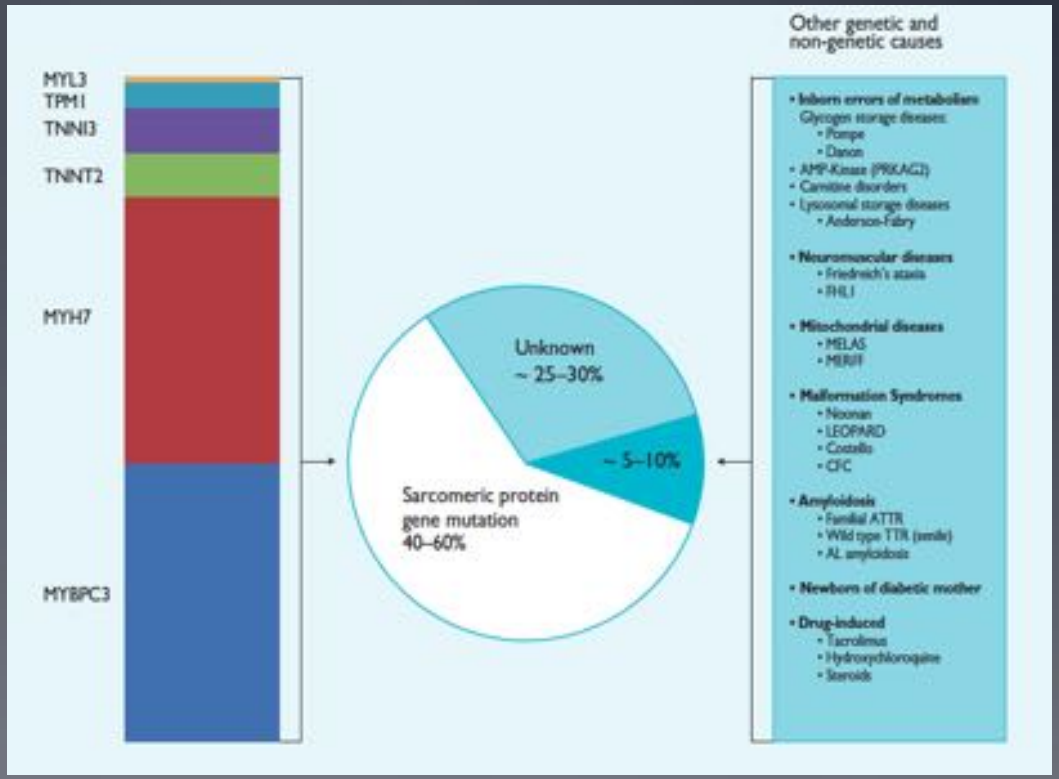
European Heart Journal (2008) 29, 270

Les gènes


TCAP
NEXN
ACTC1
CSRP3
PDLIM3
TNNC1
TNNI3
LDB3
VCL
MYBPC3
ACTN2
TNNT2 (3%)
MYH7 (4%)
MYH6 (4%)
TTN (25%)
ILK
MYOZ2
MYL3
MYL2
MYLK2
PRKAG2
Hypertrophic cardiomyopathy
MURC
DES
ANKRD1
TPM1
Dilated cardiomyopathy
JAG1
GATA4
ELN
NOTCH1
NOTCH2
NKX2-5
Congenital heart disease
DSP
PKP2
ABCC9
DSC2
TMEM
DSG2
NEBL
PSEN1
ARVC
EYA4
CRYAB
PLN
DMD
PSEN2
TMPO
LAMA4
RBM20
MYPN (3–4%)
BAG3
TAZ
RAF1
LAMP2
GLA
FXN
LMNA (6%)
Syndromic
SCN5A (2–3%)
RYR2
SNTA1
CACNA1C/TS
KCNJ5
ANK2
AKAP9
SCN4B
KCNJ2
Channelopathies
KCNQ1
CAV3
KCNE1
KCNE2
KCNJ2
Neuromuscular disorders
EMD
SGCD
Genetic cardiac disorder
Gene associated with one phenotype
Gene associated with two phenotypes
Gene associated with three phenotypes

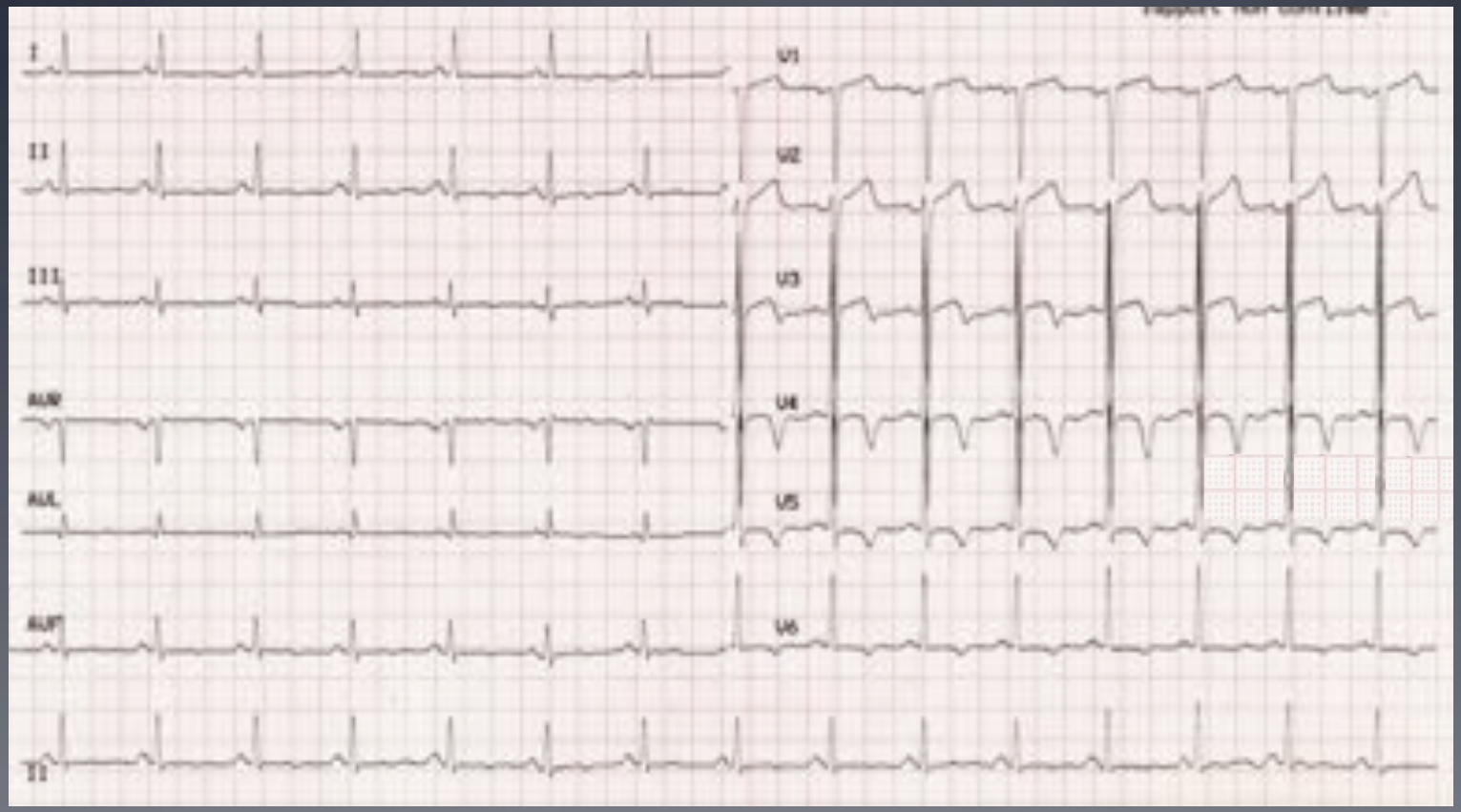
CMH

- ≠ post HTA
- 0,02 à 0,23% adultes
- 3 à 5/10^6 enfants
- Localisation:
  - Septale
  - Concentrique
  - Apicale
- Obstructive dans 20%

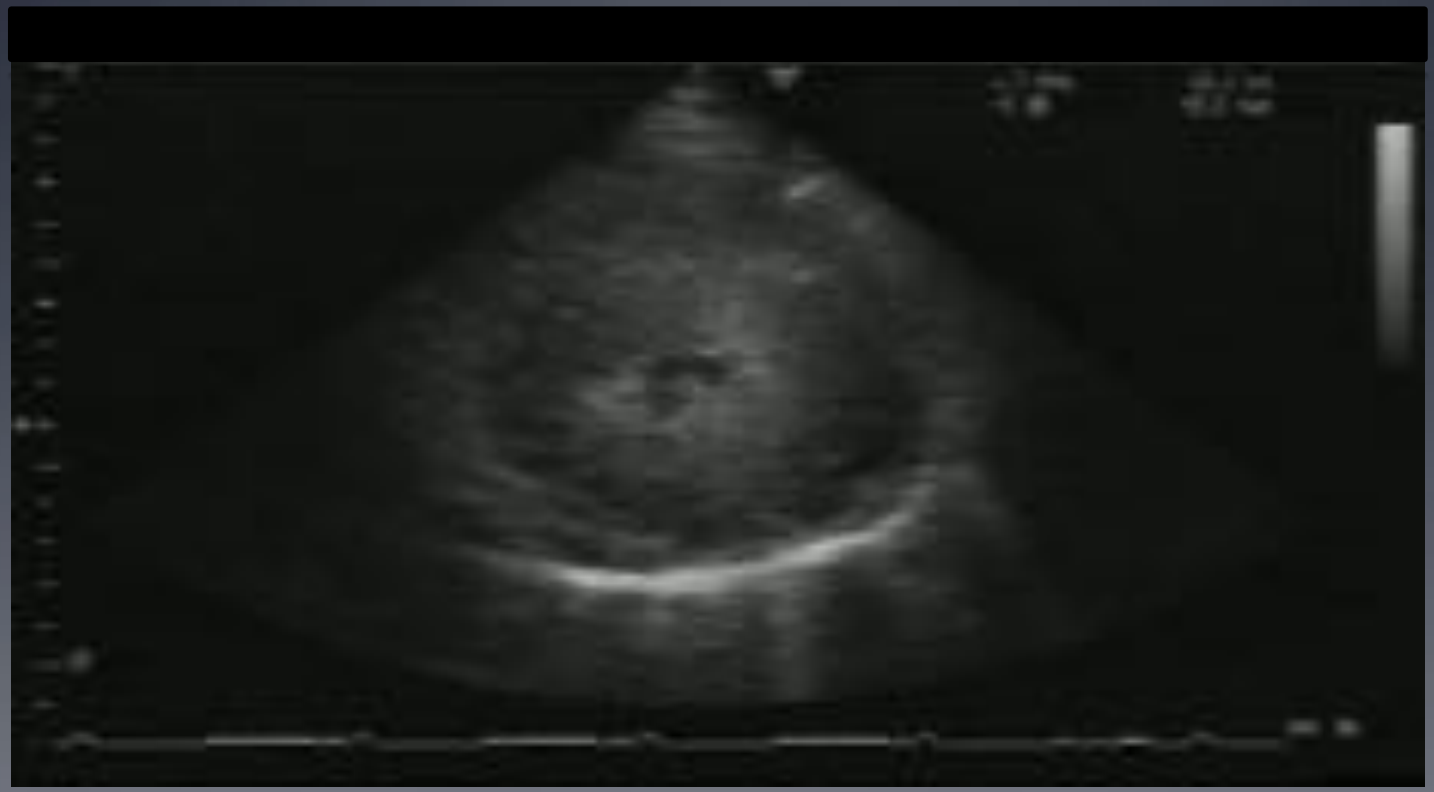


European Heart Journal 2014;35:2733

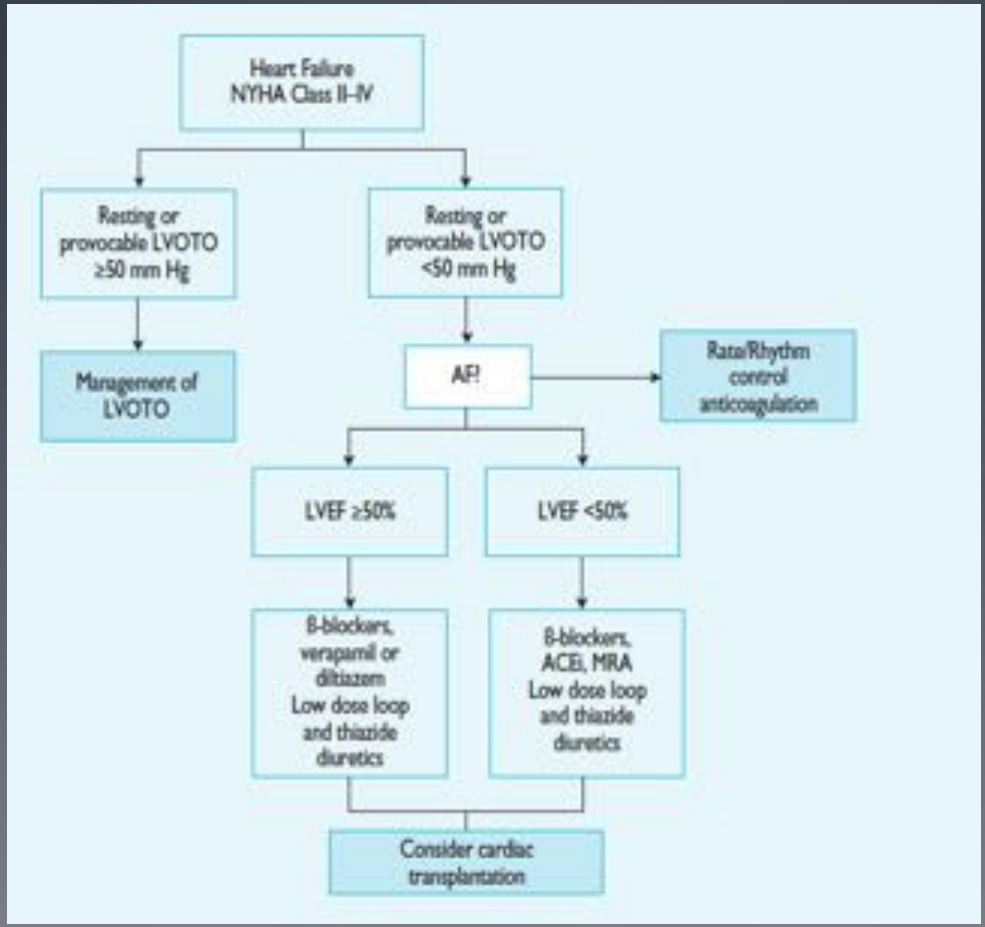
CMH:
diagnostic ECG



CMH: diagnostic écho



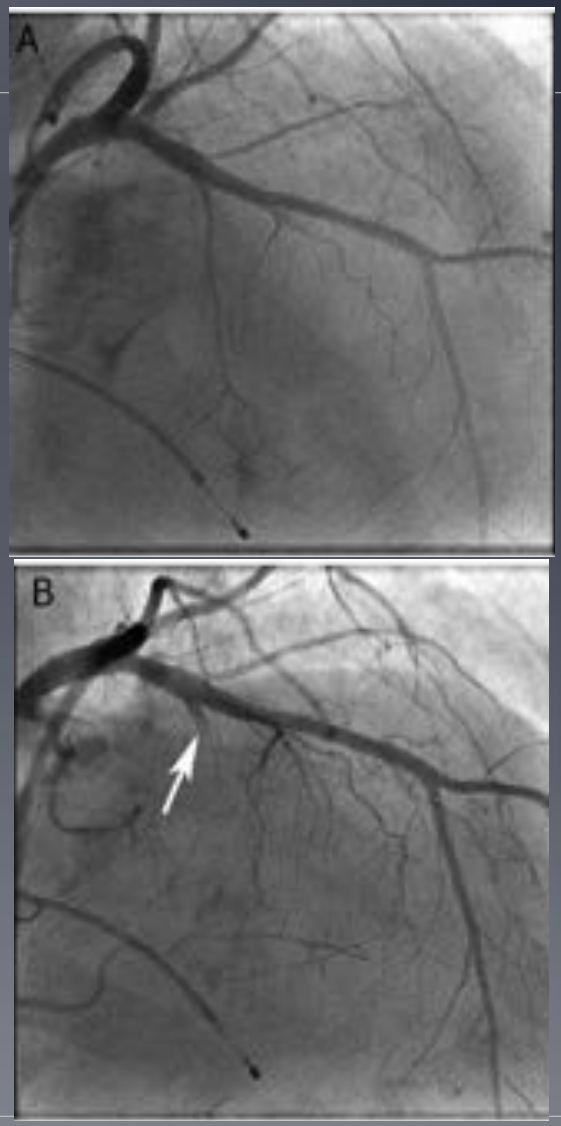
CMH: algorythme



European Heart Journal 2014;35:2733

CMHO: TT

- Medical
  - b-, verapamil
- Pace (asynchronisme)
- Alcoolisation septale
- Myomectomie chirurgicale
  - Si maladie mitrale associée



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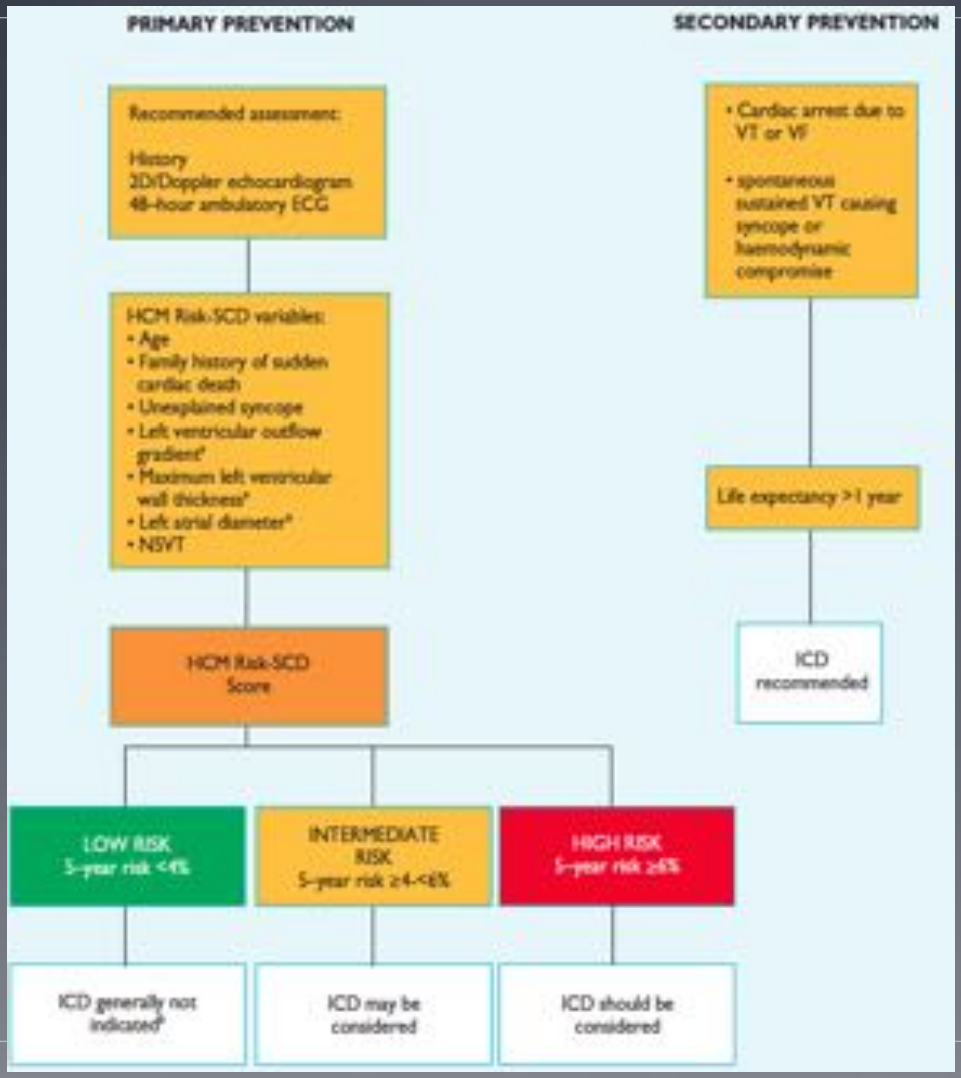
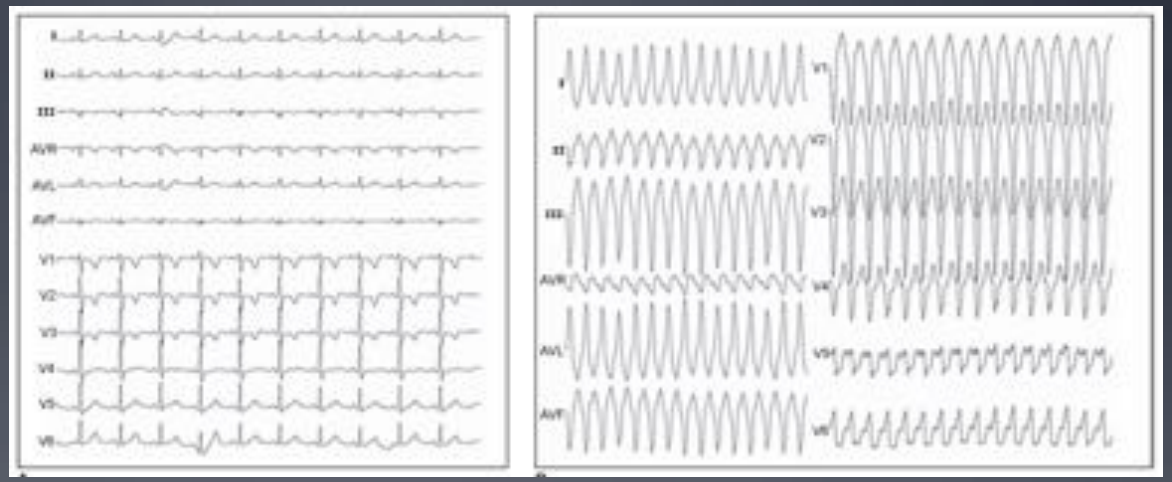
CMH et ICD


PRIMARY PREVENTION
SECONDARY PREVENTION
Recommended assessment:
History
2D/Doppler echocardiogram
48-hour ambulatory ECG
HCM Risk-SCD variables:
• Age
• Family history of sudden cardiac death
• Unexplained syncope
• Left ventricular outflow gradient*
• Maximum left ventricular wall thickness*
• Left atrial diameter*
• NSVT
HCM Risk-SCD Score
LOW RISK
5-year risk <4%
INTERMEDIATE RISK
5-year risk ≥4-<6%
HIGH RISK
5-year risk ≥6%
ICD generally not indicated*
ICD may be considered
ICD should be considered
• Cardiac arrest due to VT or VF
• spontaneous sustained VT causing syncope or haemodynamic compromise
Life expectancy >1 year
ICD recommended

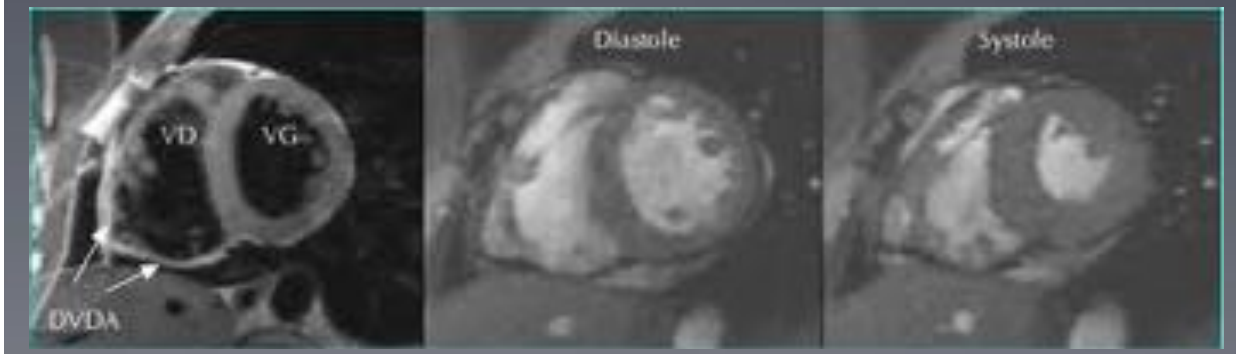
DAVD

- 1/5000
- Homme jeune
- 80% avant 40 ans
- Etiologie:



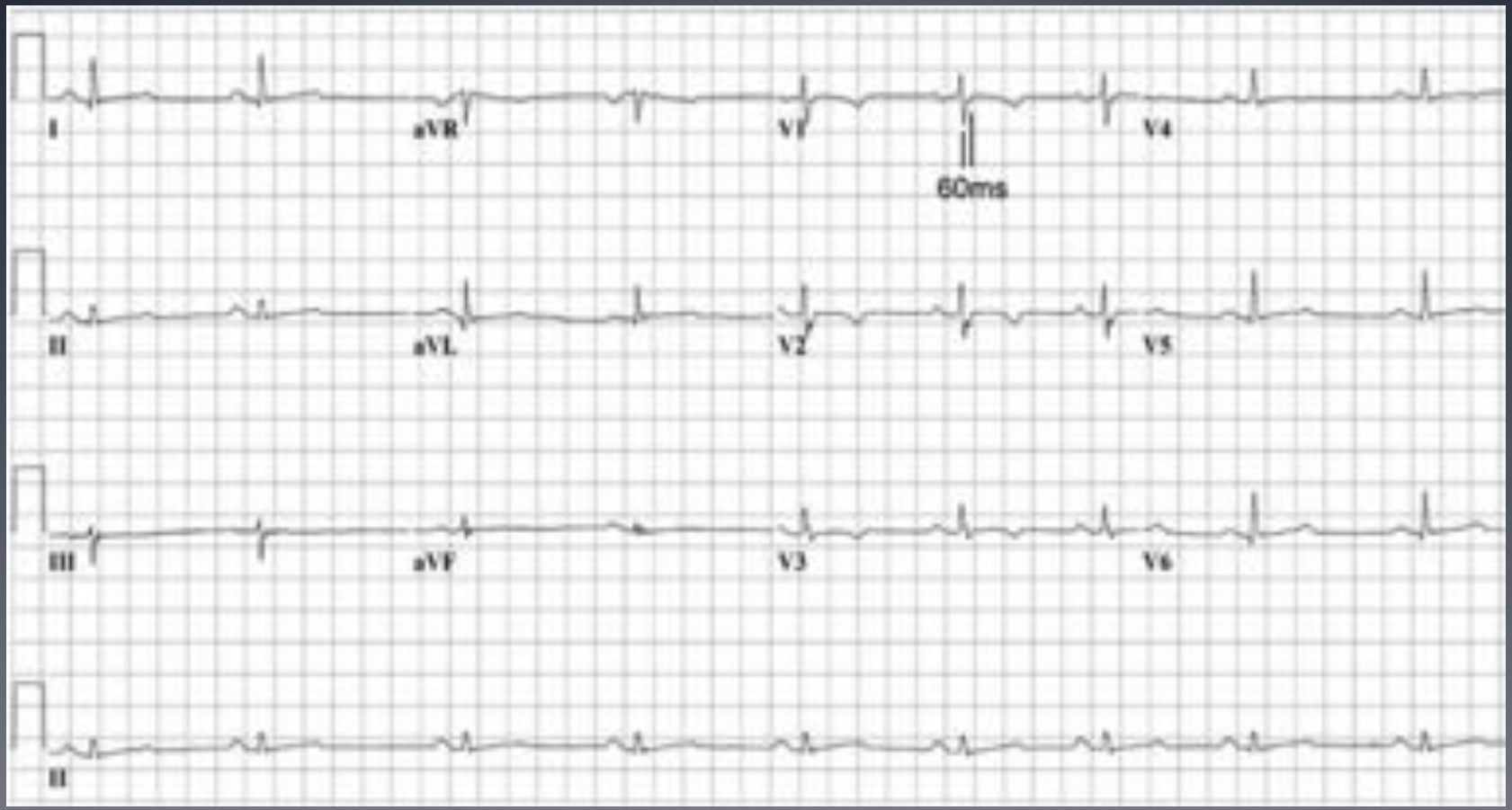
Familial, unknown gene
Intercalated disc protein mutations
- Plakoglobin
- Desmoplakin
- Plakophilin 2
- Desmoglein 2
- Desmocollin 2

Cardiac ryanodine receptor (RyR2)
Transforming growth factor-β3 (TGFβ3)



Transformation du VD en tissu fibreux et adipeux

DAVD:
ECG

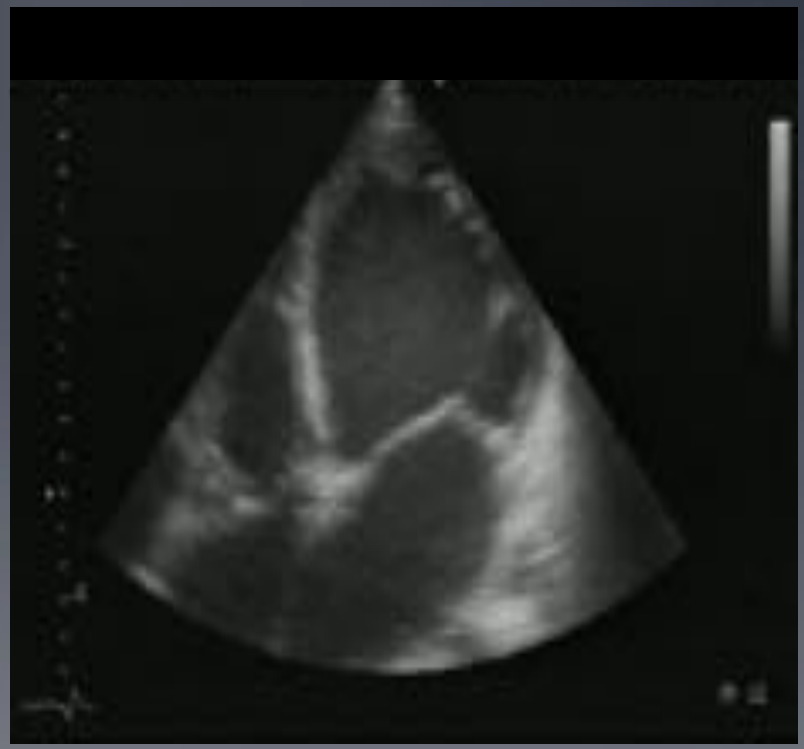


DAVD:
TT

- ICD
  - Primaire
  - Secondaire
- TT de l'IC

CMD

- 1/2500
- 50% familiale
- H>F



CMD : étiologie

- familiale

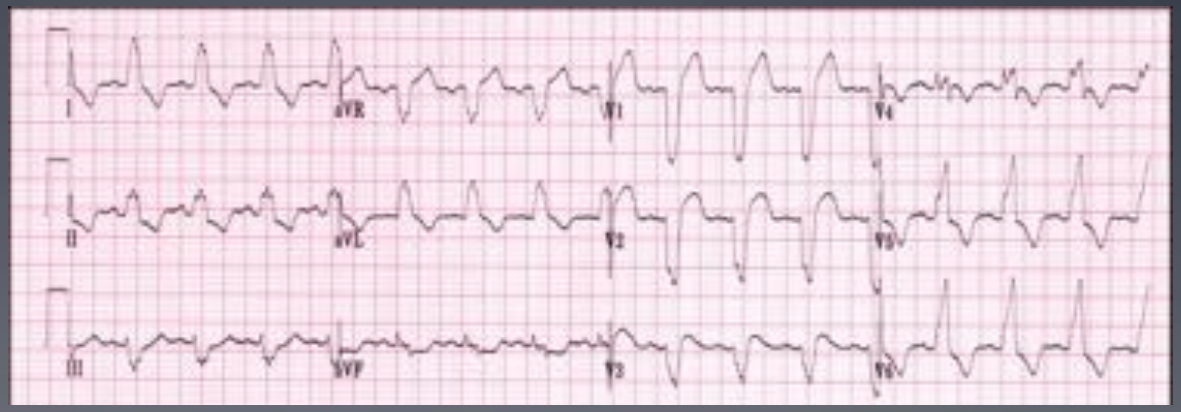
Familial, unknown gene
Sarcomeric protein mutations (see HCM)
Z-band
  Muscle LIM protein
  TCAP
Cytoskeletal genes
  Dystrophin
  Desmin
  Metavinculin
  Sarcoglycan complex
  CRYAB
  Epicardin
Nuclear membrane
  Lamin A/C
  Emerin
Mildly dilated CM
Intercalated disc protein mutations (see ARVC)
Mitochondrial cytopathy

- Non familiale

Myocarditis (infective/toxic/immune)
Kawasaki disease
Eosinophilic (Churg Strauss syndrome)
Viral persistence
Drugs
Pregnancy
Endocrine
Nutritional — thiamine, carnitine, selenium, hypophosphataemia, hypocalcaemia
Alcohol
Tachycardiomyopathy

CMD:
TT

- IC
  - (b-, IEC, diurétiques, anti-aldostérone…)
  - Règles hygiéno-diététiques
- CRT (si BBGc)
- transplantation



CM peu dilatée

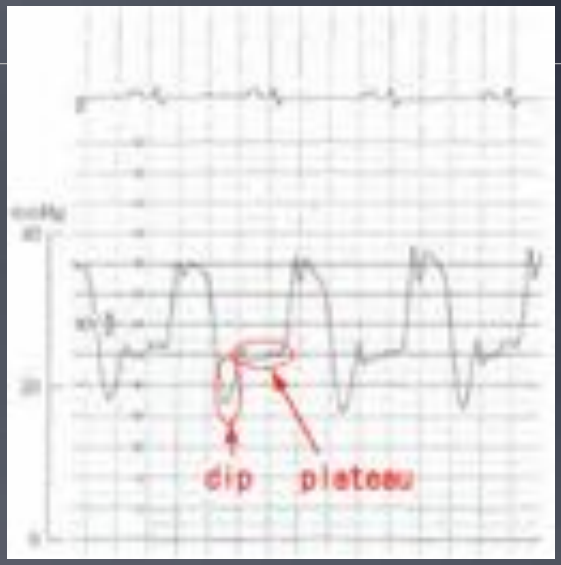
- FE altérée
- Peu ou pas de dilatation
- Pronostic identique à la CMD

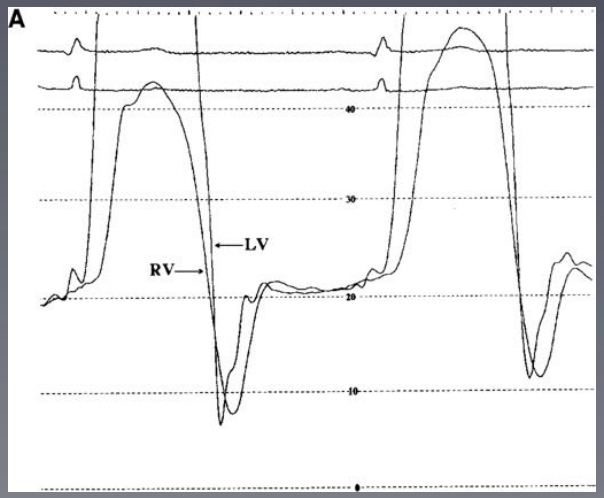
CM du péripartum

- Entre le dernier mois de grossesse et les 5 mois postpartum
- Surtout > 30ans
- Associé:
  - Tocolyse
  - G Gemellaire
  - HTA gravidique
- Etiologie:
  - Myocardite
  - Auto-immun
  - Prolactine / Stress oxidatif

CMR

- FE normale
- Troubles du remplissage/ relaxation
- Volumes N ou ↘
- Épaisseur N





CM R

- Familiale

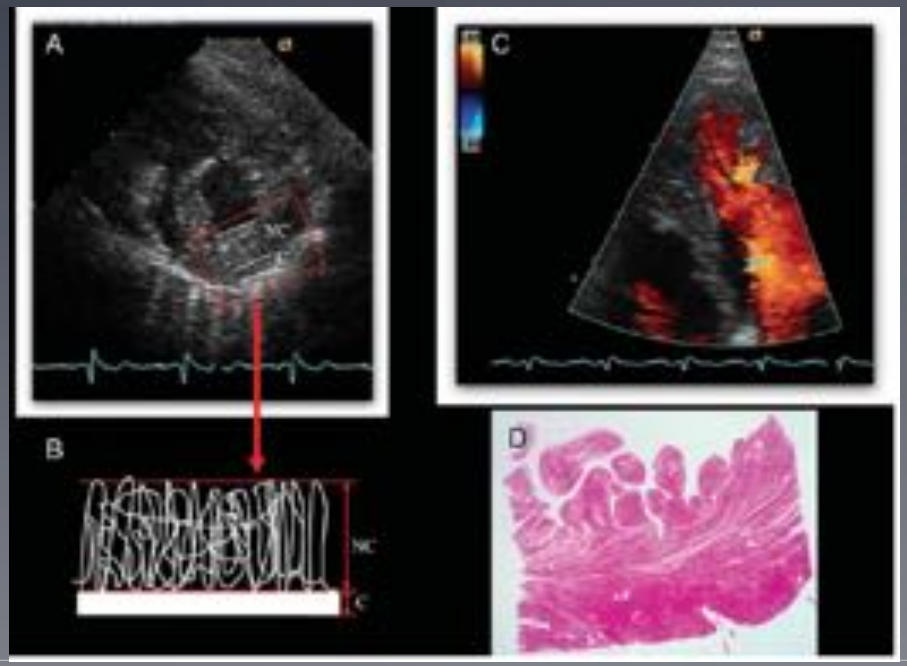
Familial, unknown gene
Sarcomeric protein mutations
  Troponin I (RCM +/− HCM)
  Essential light chain of myosin
Familial amyloidosis
  Transthyretin (RCM + neuropathy)
  Apolipoprotein (RCM + nephropathy)
Desminopathy
Pseuxanthoma elasticum
Haemochromatosis
Anderson–Fabry disease
Glycogen storage disease

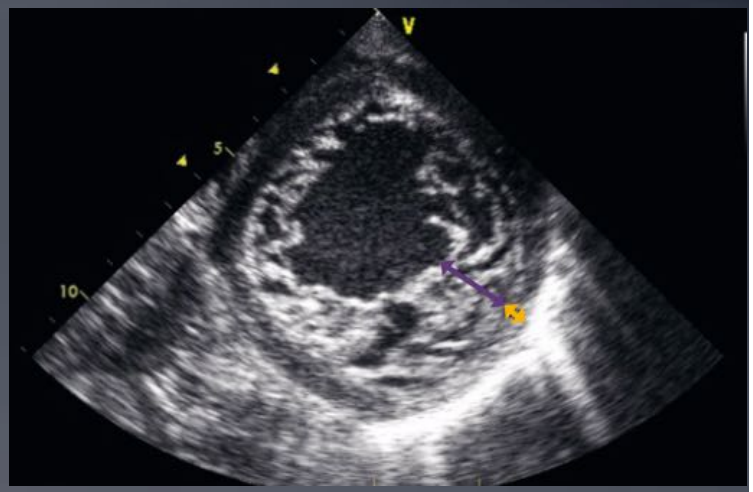
- Non familiale

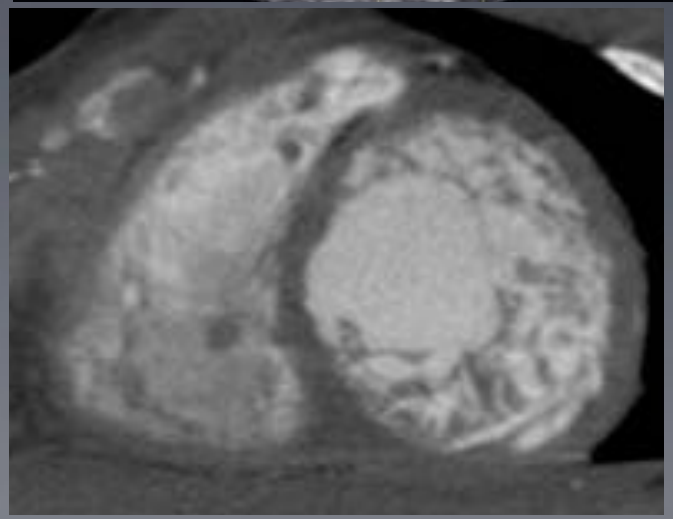
Amyloid (AL/prealbumin)
Scleroderma
Endomyocardial fibrosis
  Hypereosinophilic syndrome
  Idiopathic
  Chromosomal cause
  Drugs (serotonin, methysergide, ergotamine, mercurial agents, busulfan)
Carcinoid heart disease
Metastatic cancers
Radiation
Drugs (anthracyclines)

Non classés:
Non compaction du VG

- = trabéculations + récessus
- 1/2000
- 0,014% à 1,3% des echo cœur



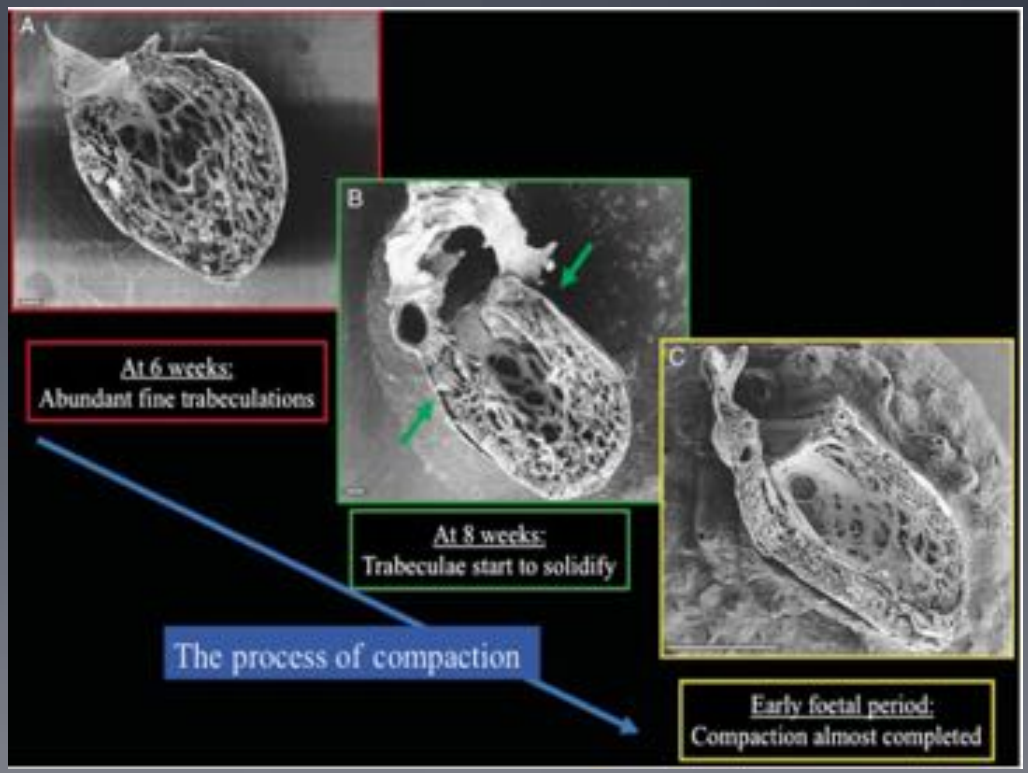




European Heart Journal (2011) 32, 1446

Non compaction: Génotype

- Gènes:
  - G 4.5 encoding taffazin (X-linked)
  - alpha dystrobrevin
  - ZASP
  - actin
  - lamin A/C
  - chromosome 11 p 15



European Heart Journal (2011) 32, 1446

Non compaction: pronostic

- Evolution
  - IC
  - TV
  - Mort subite

Table 4 Predictors of death or heart transplantation

Clinical predictors[38,51,59]
Age at initial presentation
Functional capacity, NYHA class III–IV
Sustained ventricular arrhythmias
Echocardiographic parameters[34,38,51,57,63]
Ratio of non-compacted to compacted layers
Number of affected segments
LV end-diastolic diameter
Abnormal lateral mitral tissue Doppler Ea velocity

Therapeutic strategies	
Normal LV size/systolic function	Regular follow-up (every 2 years)
Heart failure therapy	As per guidelines for heart failure
Anticoagulation	LVEF <40%
ICD	Secondary prevention/(primary prevention?)
Biventricular pacing	Advanced heart failure/LVEF <35%/ dyssynchrony (as per guidelines)

Non classés:
TAKO TSUBO

- Femme post-ménopausique
- 1,7 à 2,2% STEMI
- Dysfonction microcirculation
- Stress +++
- TT symptomatique