Diagnosi descrittive

CARDIOMYOPATHIES ARE *"MYOCARDIAL DISORDERS CHARACTERIZED BY STRUCTURALLY AND FUNCTIONALLY ABNORMAL HEART MUSCLE AND ABSENCE OF OTHER DISEASES SUFFICIENT TO CAUSE THE OBSERVED MYOCARDIAL ABNORMALITY".*
(EUR HEART J, ESC POSITION PAPER, 2008)

CHE COS'E' LA CLASSIFICAZIONE MOGE(S)

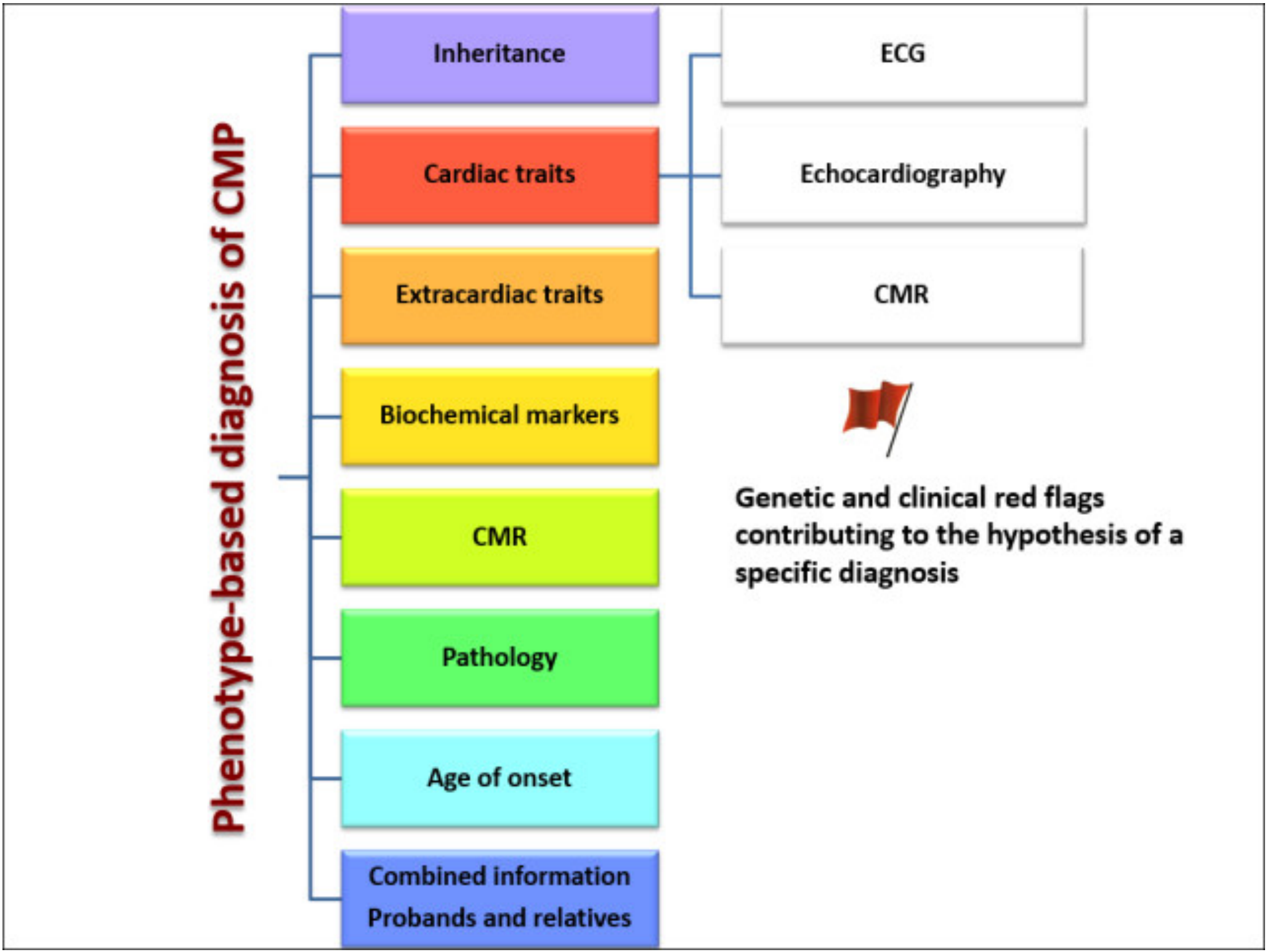
MOGE(S) system/nomenclature - WHF

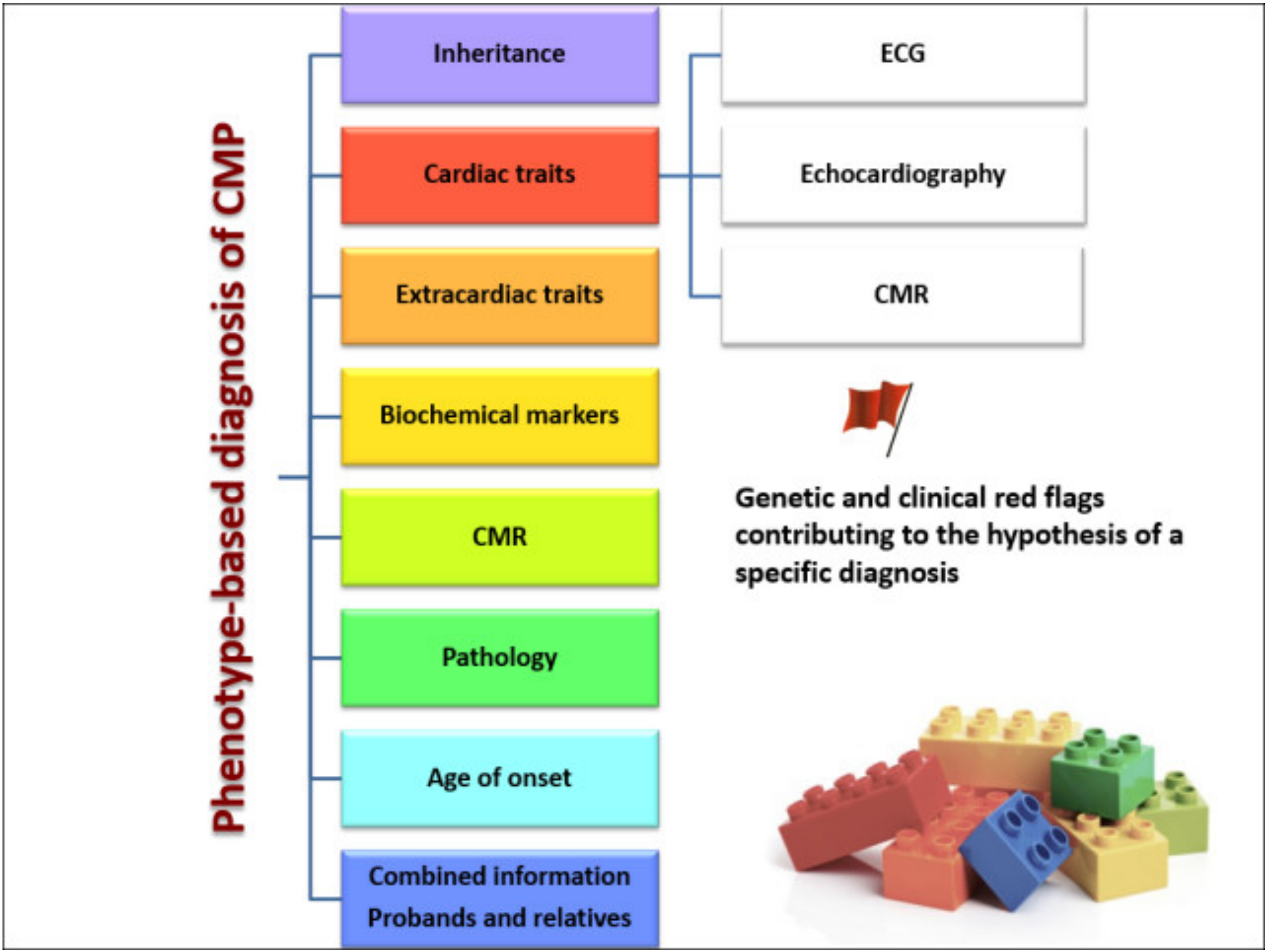
- M = morphofunctional phenotype
- O = organ involvement
- G = genetic/familial
- E = etiology
- (S) = AHA/NYHA class
- Further → future → A = arrhythmias

The MOGE(S) Classification for a Phenotype–Genotype Nomenclature of Cardiomyopathy Endorsed by the World Heart Federation

Elena Arbustini, Norrett Narula, G. William Dec, Kathryn Tuchen, K. Srinath Reddy, Barry Greenberg, MD, Sudhir Kushwaha, Thomas Marwick, Sean Pinney, Valentina Favalli, Christopher Kramer, Robert Roberts, MD, William A. Zoghbi, Robert Bonow, Luigi Tavazzi, Valentin Fuster, Jagat Narula, MD, PhD

Italy; New York, New York; Switzerland; India; Australia; and Canada


Phenotype-based diagnosis of CMP
Inheritance
Cardiac traits
Extracardiac traits
Biochemical markers
CMR
Pathology
Age of onset
Combined information
Probands and relatives
ECG
Echocardiography
CMR
Genetic and clinical red flags
contributing to the hypothesis of a
specific diagnosis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CMR
Genetic and clinical red flags
contributing to the hypothesis of a
specific diagnosis


moges.biomeris.com/moges.html
MOGES
Morpho-functional
Organ/system involvement
Genetic
Etiological Annotation
Stage
$M_0\ O_0\ G_0\ E_0\ S$
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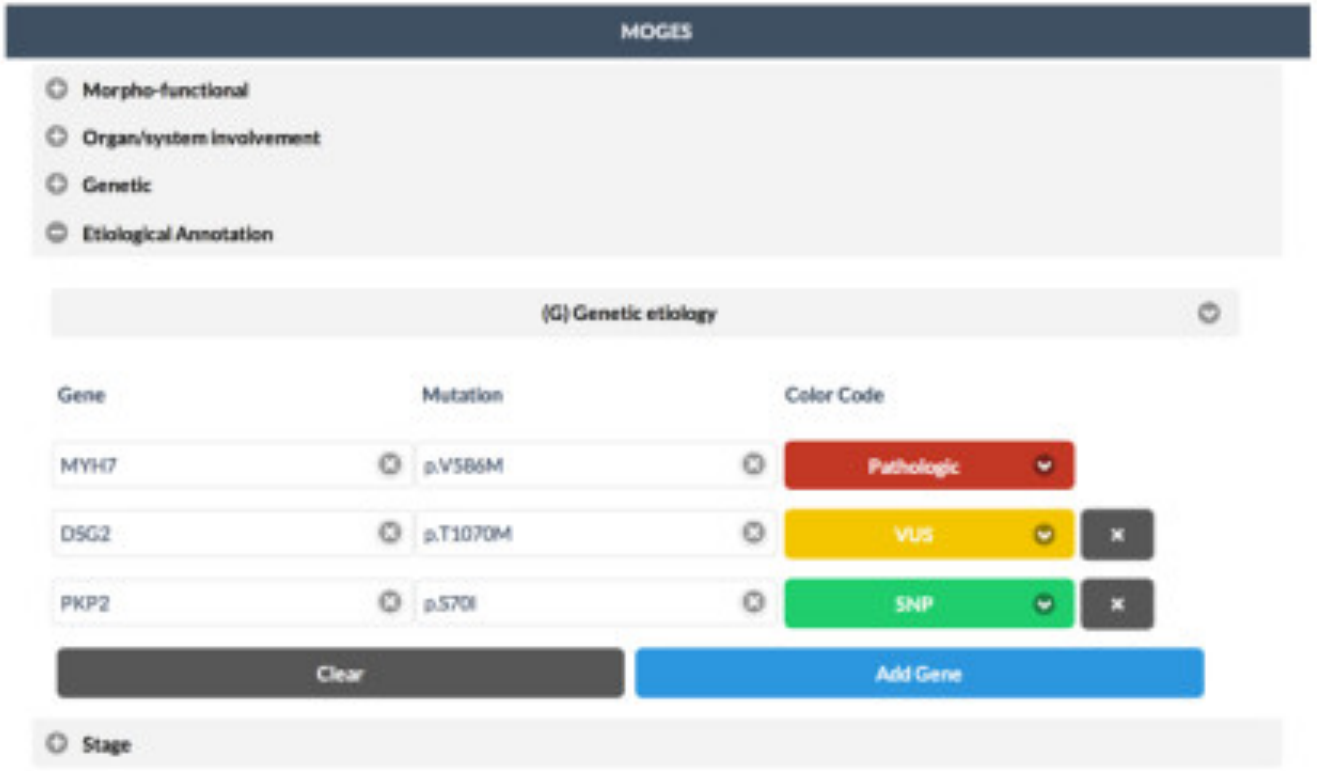
MOGES Nosology describing the phenotype and genotype data: example

http://moges.biomeris.com/moges.html



$M_D\ O_H\ G_{AD}\ E_{G\text{-}MYH7[p.V586M]+DSG2[p.T1070M]+PKP2[p.S70I]}\ S$

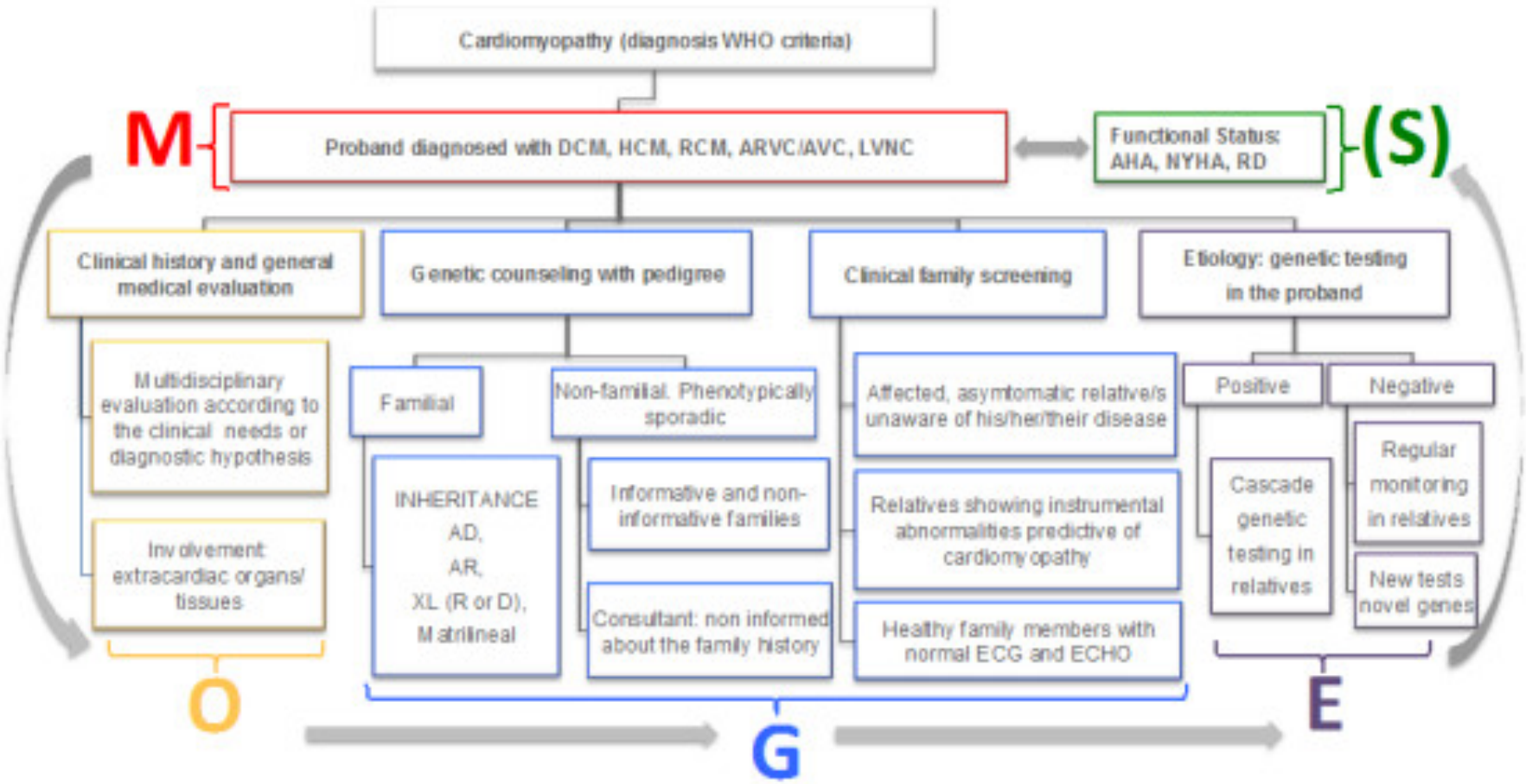
$M_H\ O_H\ G_{AD}\ E_{G\text{-}MYH7[p.Val586Met]+DSG2[p.Thr1070Met]+PKP2[p.Ser70Ile]}\ S$

Describes a patient with DCM morphofunctional phenotype (M), heart as unique involved organ (O), genetic (G) autosomal dominant, with etiology (E) definied by the genetic testing demonstrating a missense mutation in MYH7, a missense variant of unknown significance in DSG2 and a missense variant, likely a polymorphism, in PKP2. The stage (S) is optional and includes the possibility of adding AHA stage and functional NYHA class.
The color code helps immediate perception of the role of the mutations/variants.

Braunwald - Table 1 MOGE(S) system: the compilation is supported by the app at http://moges.bioinfo.cnio.es/moges.html

MOGE(S)	Description	Specific information (app)
M	Morpho-functional Phenotype	**(D)** Dilated → M_D **(H)** Hypertrophic → M_H • Obstructive (Obs)→ M_{HObst} • Non-obstructive (N-Obs) → $M_{HNonObst}$ **(R)** Restrictive → M_R **(R)** (EM F) Endomyocardial fibrosis [LV= left ventricle; RV=right ventricle; RLV=biventricular] → $M_{RestrictiveEndomyocardialFibrosis}$ **(A)** ARVC [M= major; m=minor; c=category] [LV= left ventricle; RV=right ventricle; RLV=biventricular] **(NC)** LVNC Overlapping **(H+R), (D+H), (NC+H)** , **(H+D), (D+NC)** or more complex combinations such as **(H+R+NC)** **(E)** Early, with type in parentheses $M_{E.G.}$ **E[H], E[D],** **(N.S)** Non-specified phenotype **(N/A)** information non available **(0)** Unaffected
O	Organ/system Involvement	**(H)** Heart [LV= left ventricle; RV=right ventricle; RLV=biventricular] **(M)** Muscle, skeletal **(N)** Nervous **(C)** Cutaneous **(E)** Eye, Ocular **(A)** Auditory **(K)** Kidney **(G)** Gastrointestinal **(S)** Skeletal **(Lu)** Lung **(Li)** Liver **(0)** absence of organ/system involvement
G	Genetics/Inheritance of the phenotype	**(N)** Family History Negative **(U)** Family History Unknown **(AD)** Autosomal dominant **(AR)** Autosomal recessive **(XLR)** X-linked recessive **(XLD)** X-linked dominant **(XL)** X-linked **(M)** Matrilineal **(0)** Family history not investigated **(Undet)** Inheritance of the phenotype is undetermined **(S)** Phenotypically Sporadic (apparent or real)
E	Etiological Annotation	**(G) Genetic cause;** when known: gene/s and mutation/s are specified using MIM symbols and mutation nomenclature^. Color code, red is for mutations that either affect or probably affect function of the protein; orange is for VUS; and green is for rare variants that may not affect function but are potentially useful for segregation studies. **(OC)** = Obligate carrier **(ONC)** = Obligated non-carrier **(DN)** De novo **(Neg)** = Genetic test negative for the known familial mutation **(N/A)** = Genetic test not available **(N)** = Genetic defect not identified **(I)** = Incomplete genotyping from segregation studies or identification of VUS **(0)** No genetic test*, any reason (no blood sample, no informed consent, etc.) **Genetic** amyloidosis (i.e. $E_{G.A-TTR}$ or $E_{G.A-Apo1}$ or $E_{G.A-Apo?}$ etc) **Hemochromatosis** (i.e. $E_{G.H-Hfe}$ or $E_{G.H-Hfe2}$ etc). **Non-genetic etiologies:** **(M)** Myocarditis **(INFL)** Inflammatory **(V)** Viral infection (eventually add the virus when identified in affected heart); **(AI)** Autoimmune/immune-mediated; suspected ($E_{I.A-suspected}$) or ($E_{I.A-proven}$); **(A)** Amyloidosis (add type of amyloidosis: $E_{I.A}$ $E_{I.A-}$ - $E_{I.A-Apo}$) **(I)** Infectious myocarditis, non viral $E_{I.A-Infectious}$ (add the non-viral infectious agent); **(T)** Toxicity (add toxic cause/drug); **(Eo)** Hypereosinophilic heart disease. **(O)** Other

MOGES Nosology summarizes the clinical and genetic work-up in cardiomyopathies



$M_0\ O_0\ G_0\ E_0\ S$

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Endorsed by the World Heart Federation

Eloisa Arbustini, MD,* Navneet Narula, MD,† G. William Dec, MD,‡
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William A. Zoghbi, MD,‖‖ Rob
Valentin Fuster, MD, PhD,** Jaq

Pavia and Cotignola, Italy; New Y
San Diego, California; Rochester, M
Ottawa, Canada; Houston, Texas;

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J Am Coll Cardiol. 2014 Jul 22;64(3):304-18. doi: 10.1016/j.jacc.2014.05.027.

The MOGE(S) classification of cardiomyopathy for clinicians.

Arbustini E[1], Narula N[2], Tavazzi L[3], Serio A[1], Grasso M[1], Favalli V[1], Bellazzi R[4], Tajik AJ[5], Bonow RO[6], Fuster V[7], Narula J[8].

Author information

Abstract

Most cardiomyopathies are familial diseases. Cascade family screening identifies asymptomatic patients and family members with early traits of disease. The inheritance is autosomal dominant in a majority of cases, and recessive, X-linked, or matrilinear in the remaining. For the last 50 years, cardiomyopathy classifications have been based on the morphofunctional phenotypes, allowing cardiologists to conveniently group them in broad descriptive categories. However, the phenotype may not always conform to the genetic characteristics, may not allow risk stratification, and may not provide pre-clinical diagnoses in the family members. Because genetic testing is now increasingly becoming a part of clinical work-up, and based on the genetic heterogeneity, numerous new names are being coined for the description of cardiomyopathies associated with mutations in different genes; a comprehensive nosology is needed that could inform the clinical phenotype and involvement of organs other than the heart, as well as the genotype and the mode of inheritance. The recently proposed MOGE(S) nosology system embodies all of these characteristics, and describes the morphofunctional phenotype (M), organ(s) involvement (O), genetic inheritance pattern (G), etiological annotation (E) including genetic defect or underlying disease/substrate, and the functional status (S) of the disease using both the American College of Cardiology/American Heart Association stage and New York Heart Association functional class. The proposed nomenclature is supported by a web-assisted application and assists in the description of cardiomyopathy in symptomatic or asymptomatic patients and family members in the context of genetic testing. It is expected that such a nomenclature would help group cardiomyopathies on their etiological basis, describe complex genetics, and create collaborative registries.

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This study was supported by Grants European Union INHERITANCE project n°241924 and Italian Ministry of Health "Diagnosis and Treatment of Hypertrophic Cardiomyopathies" (n°RF-PSM-2008-1145809)

Eloisa Arbustini*, Navneet Narula†, G. William Dec‡, K. Srinath Reddy§, Barry Greenberg‖, Sudhir Kushwaha¶, Thomas Marwick#, Sean Pinney**, Riccardo Bellazzi††, Valentina Favalli*, Christopher Kramer‡‡, Robert Roberts§§, William A. Zoghbi‖‖, Robert Bonow¶¶, Luigi Tavazzi##, Valentin Fuster**, Jagat Narula**

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PERCHE' NASCE MOGE(S)?

RAGIONI ROBUSTE

- ↑ Conoscenze cliniche, genetico-molecolari, imaging, biomarcatori, prognostiche
- Le nosologie descrittive del passato basate sulla "patologia" si stanno integrando con sistemi nosologici che non abbandonano le "radici" ma le arricchiscono di nuove informazioni
- I "BIG DATA" DI OGGI → incontrano vecchi sistemi di raccolta dati, vecchi metodi di analisi statistica
- Servono in cardiologia, come in ogni altra disciplina medica, sistemi diagnostici capaci di descrivere non solo il fenotipo ma anche di integrare tutte le nuove informazioni chiave clinico-molecolari, oggi necessarie per una nuova gestione dei work-up diagnostici e terapeutici
- Ogni nuovo sistema dovrà essere pienamente informativo, essenziale, idoneo a descrivere tutti i dati potenzialmente utili nel work-up diagnostico e terapeutico delle malattia

E’ IL MOMENTO DI “ANNOTARE” UTILIZZANDO LINGUAGGI IDENTICI E SISTEMI SEMPLICI

Cosa toglie o aggiunge MOGE(S)?

Diagnosi di dimissione

- *Cardiomiopatia Dilatativa "idiopatica"* in classe NYHA II.

Diagnosi di dimissione

Cardiomiopatia dilatativa

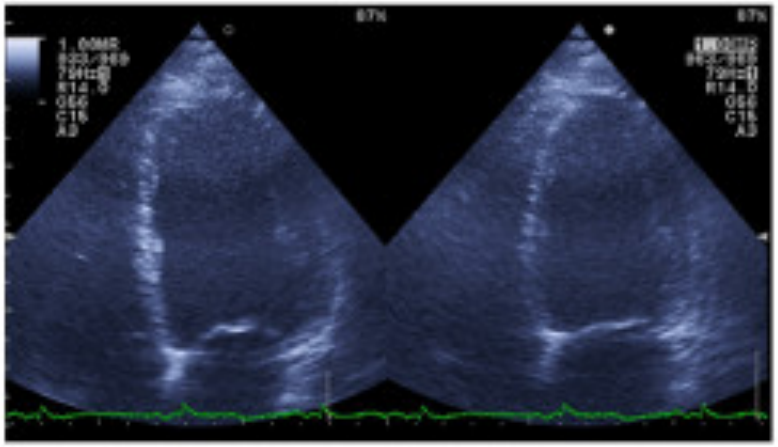
- M_D
  - (AVB) (>sCPK)
- $O_{H,M}$
- G_{AD}
- E_{LMNA}
  - P.(Arg190Trp)
- S_{C-II}

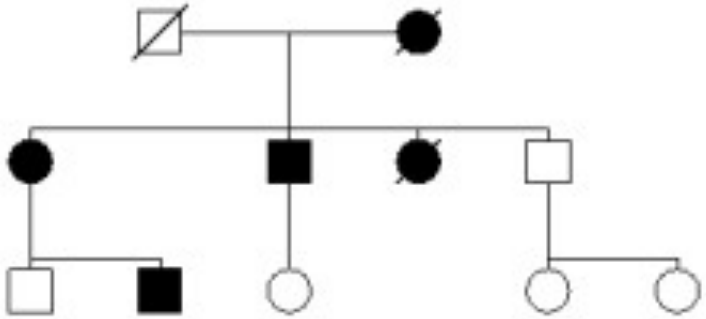
MOGE(S)

$M_H O_H G_{AD} E_{G\text{-}MYH7[p.(Arg403Glu)]} S_{B\text{-}II}$ reads as follows:

Morphofunctional phenotype (**M**): ***Hypertrophic (H)*** cardiomyopathy;
organ (**O**) involvement: ***heart (H)***;
genetic/familial (**G**) with ***autosomal dominant (AD)*** transmission;
etiology (**E**): ***genetic (G)*** and caused by the ***p.Arg403Glu*** mutation of the ***MYH7*** gene,
ACC-AHA stage (**S**) ***B, NYHA I.***

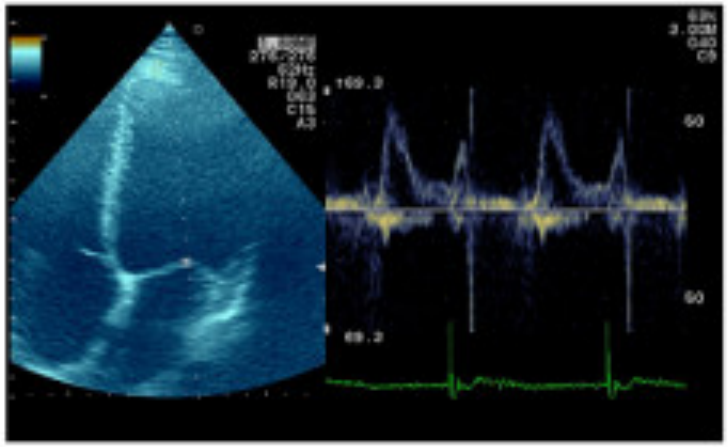
Dilated cardiolaminopathy

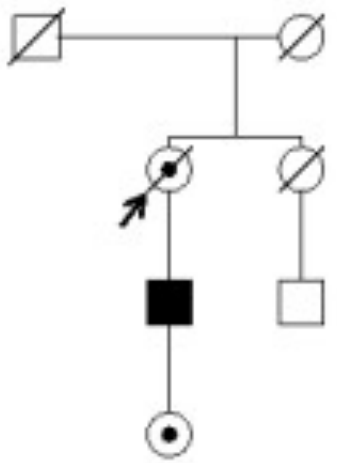




$M_{D\text{-}AVB}O_{H}G_{AD}E_{G\text{-}\textit{LMNA}\ [p.Arg190Trp]}S_{(C\text{-}II\text{-}RD3)}$

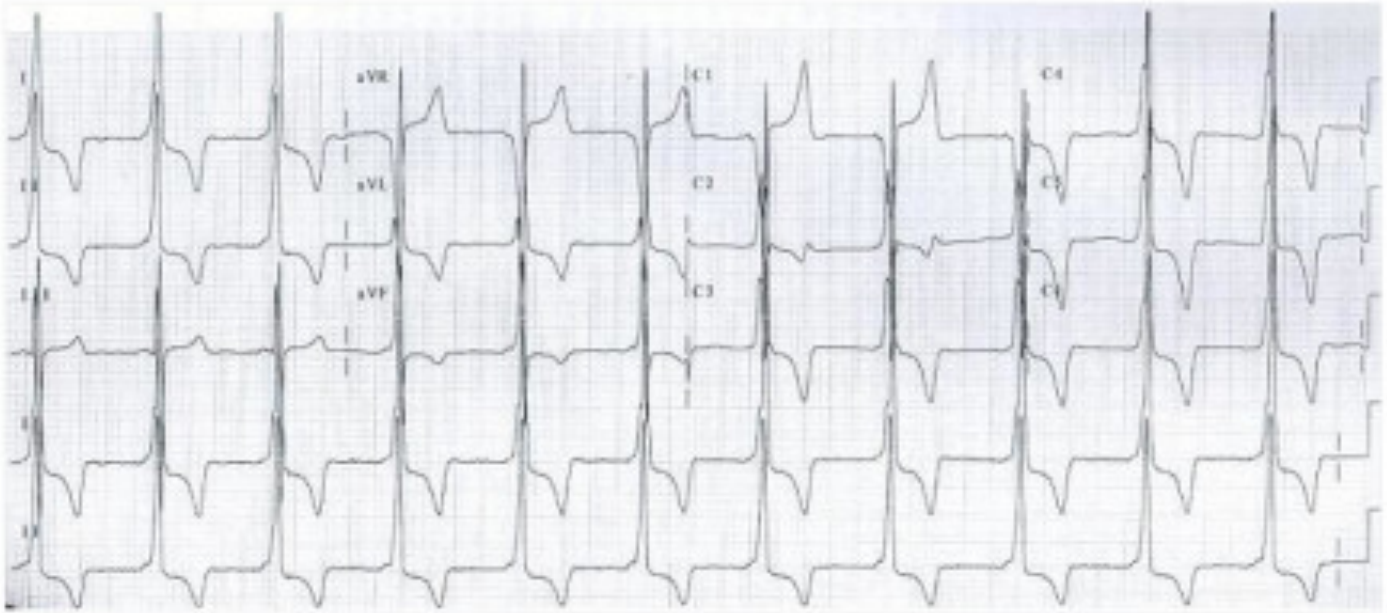
Dilated emerinopathy

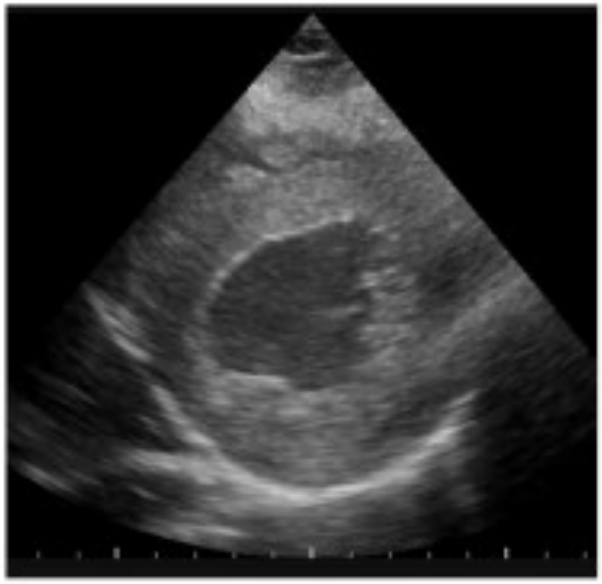


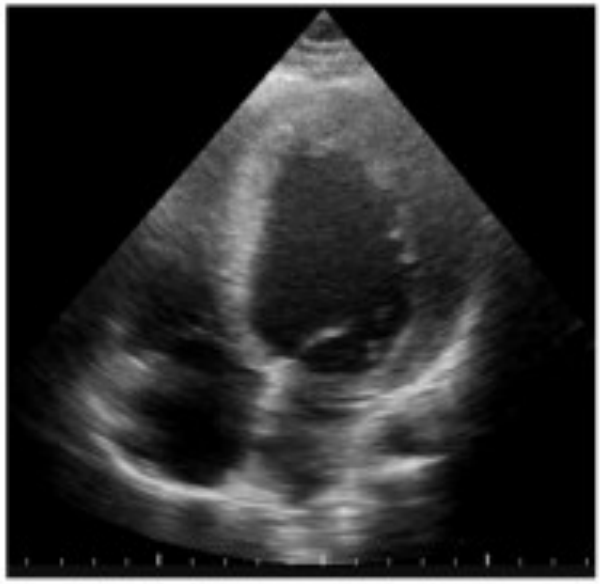


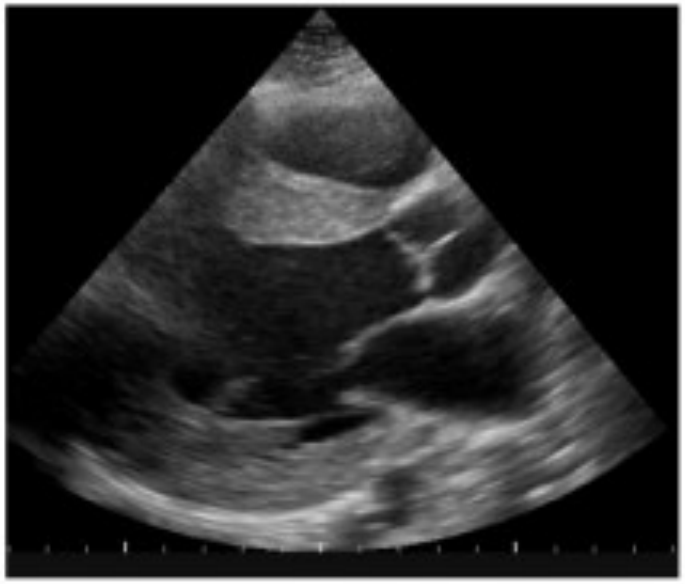
$M_{D\text{-}AVB}O_{H}G_{X\text{-}LR}E_{G\text{-}\textit{EMD}\ [p.Leu15Phe]}S_{(B\text{-}II\text{-}RD3)}$

Mitochondrial Cardiomyopathy

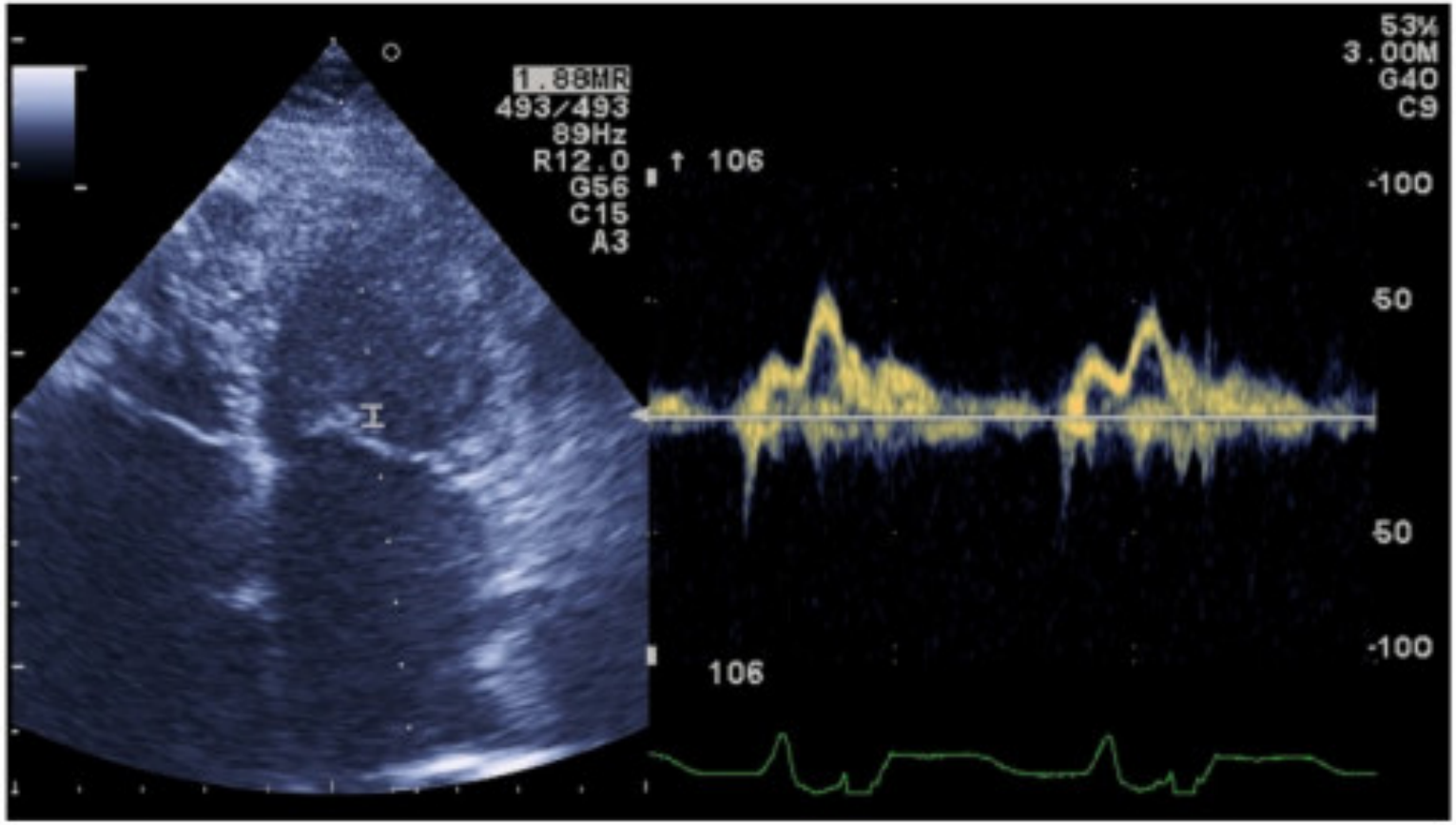








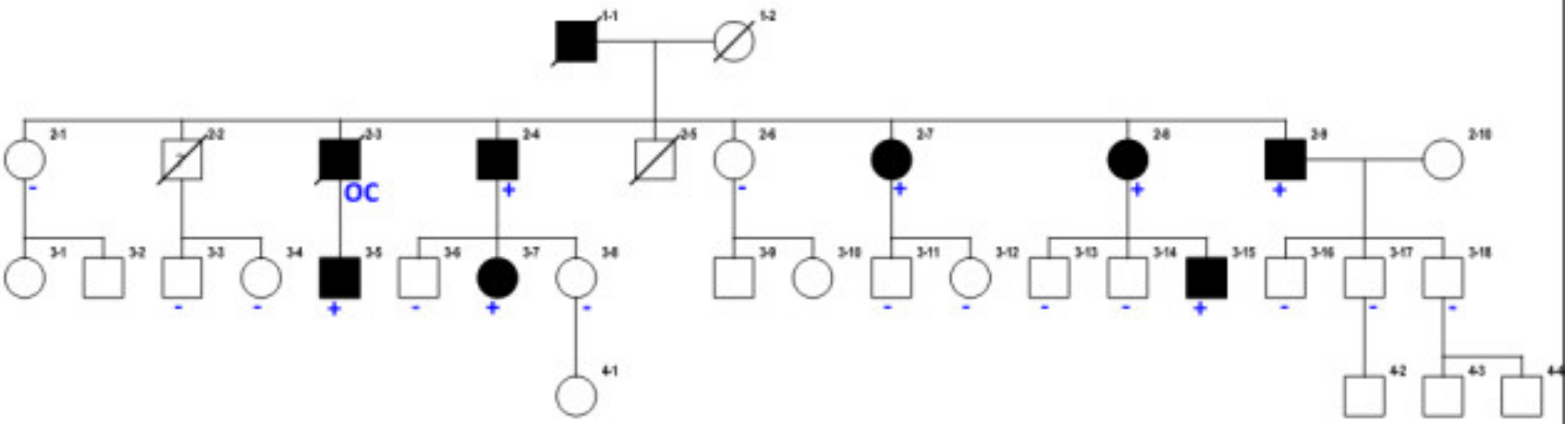
$M_{H+D\ (WPW)}\ O_{H+M+N+E+A}\ G_{M}\ E_{G\text{-}MTDNA[A3243G]}$



$$M_{H+R}\ O_{H}\ G_{DN}\ E_{G\text{-}MYL6[p.Gly162Arg]}\ S_{D\text{-}IV}$$

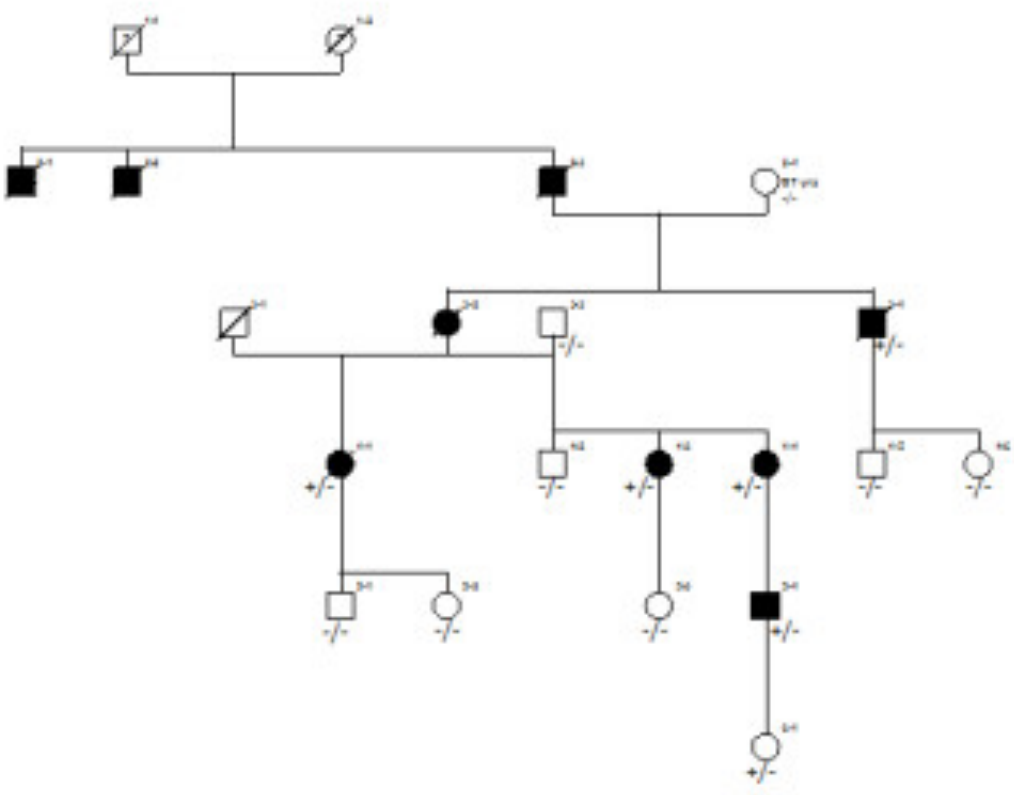
Centre for Inherited Cardiovascular Diseases
IRCCS Fondazione Policlinico San Matteo – Pavia – Italy

DILATED CARDIOMYOPATHY



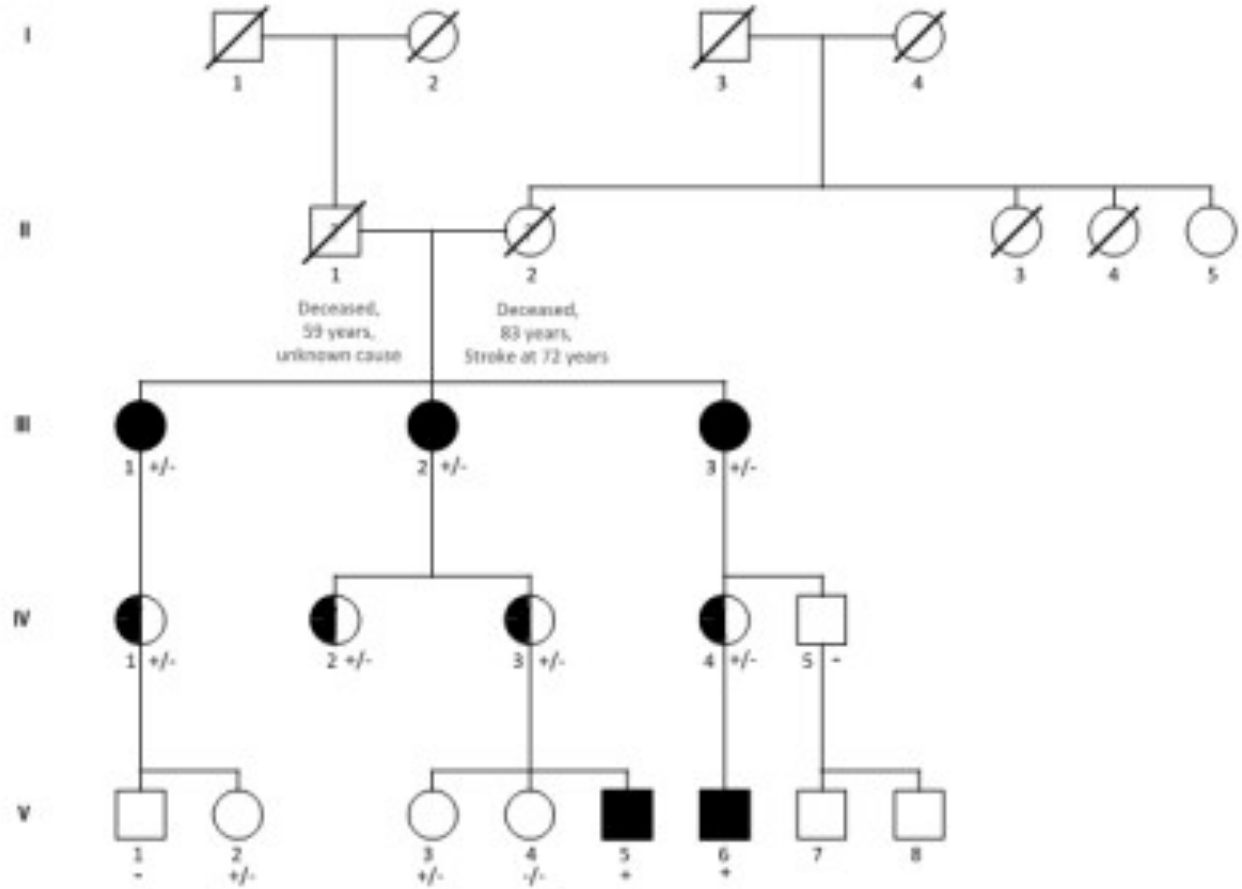
Family member	MOGE(S)
2:3	$M_{D(AVB)\ (>sCPK)}$ O_{H+M} G_{AD} $E_{G\text{-}LMNA\text{-}OC}$ [p.Glu68_Val69del] $S_{C\text{-}III}$
2:4	$M_{D(>sCPK)}$ O_{H+M} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}I}$
2:7	$M_{D(AVB)\ (>sCPK)}$ O_{H+M} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}IIb}$
2:8	$M_{D(AVB)\ (>sCPK)}$ O_{H+M} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}I}$
3:5	$M_{D(AVB)}$ O_{H} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}I}$
3:7	$M_{D(AVB)\ (>sCPK)}$ O_{H+M} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}I}$
3:15	$M_{D(AVB)}$ O_{H} G_{AD} $E_{G\text{-}LMNA}$[p.Glu68_Val69del] $S_{C\text{-}I}$

HYPERTROPHIC CARDIOMYOPATHY



Family member	MOGE(S)
II:1	$M_H O_H G_{AD} E_{G-0} S_{C-I}$
II:2	$M_H O_H G_{AD} E_{G-0} S_{C-III}$
II:3	$M_{H+D} O_H G_{AD} E_{G\text{-}MYBPC3\text{-}OC}$ [p.IVS4-2A>C] S_{C-IV}
II:4	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
III:2	$M_H O_H G_{AD} E_{G\text{-}MYBPC3\text{-}OC}$ [p.IVS4-2A>C] S_{C-III}
III:3	$M_0 O_0 G_0 E_{G-Neg-} S_{A-I}$
III:4	$M_H O_H G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{C-II}
IV:1	$M_H O_H G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{C-I}
IV:2	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
IV:3	$M_{H(AVB)} O_H G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{C-II}
IV:4	$M_{H(AVB)} O_H G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{C-IIb}
IV:5	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
IV:6	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
V:1	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
V:2	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
V:3	$M_0 O_0 G_0 E_{G-Neg} S_{A-I}$
V:4	$M_H O_H G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{C-I}
VI:1	$M_0 O_0 G_{AD} E_{G\text{-}MYBPC3}$ [p.IVS4-2A>C] S_{A-I}

Anderson-Fabry Disease



III:1	$M_{H[N\text{-}OBS]}\ O_{H,C}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{C\text{-}I}$
III:2	$M_{H[N\text{-}OBS]}\ O_{H,E,C}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{C\text{-}I}$
III:3	$M_{H[N\text{-}OBS]}\ O_{H,N[periph]}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{C\text{-}I}$
IV:1	$M_{H[E]}\ O_{H}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
IV:2	$M_{H[E]}\ O_{H}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
IV:3	$M_{H[E]}\ O_{H}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
IV:4	$M_{H[E]}\ O_{H}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
IV:5	$M_{0}\ O_{0}\ G_{XL}\ E_{G\text{-}GLA[neg]}\ S_{A\text{-}I}$
V:1	$M_{0}\ O_{0}\ G_{XL}\ E_{G\text{-}GLA[neg]}\ S_{A\text{-}I}$
V:2	$M_{0}\ O_{0}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
V:3	$M_{0}\ O_{0}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
V:4	$M_{0}\ O_{0}\ G_{XL}\ E_{G\text{-}GLA[neg]}\ S_{A\text{-}I}$
V:5	$M_{H}\ O_{H,OE}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$
V:6	$M_{H[E]}\ O_{H,OE}\ G_{XL}\ E_{G\text{-}GLA[p.Asn215Ser]}\ S_{B\text{-}I}$

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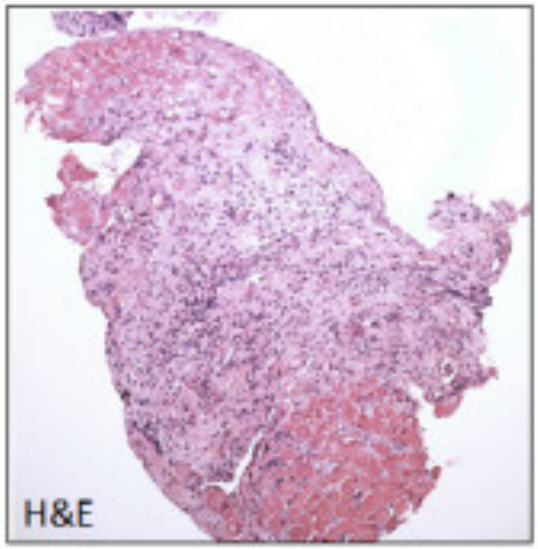
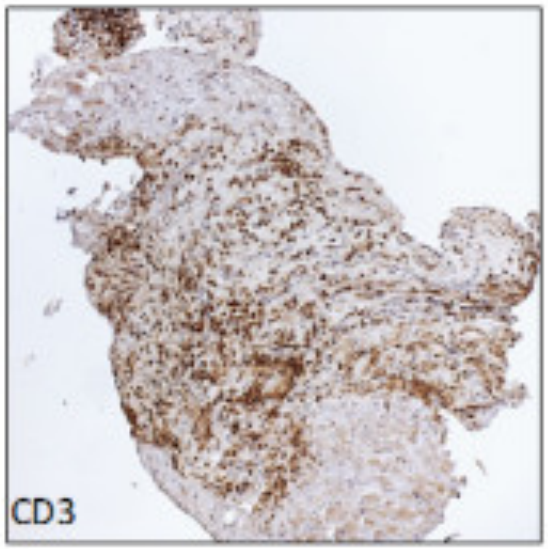
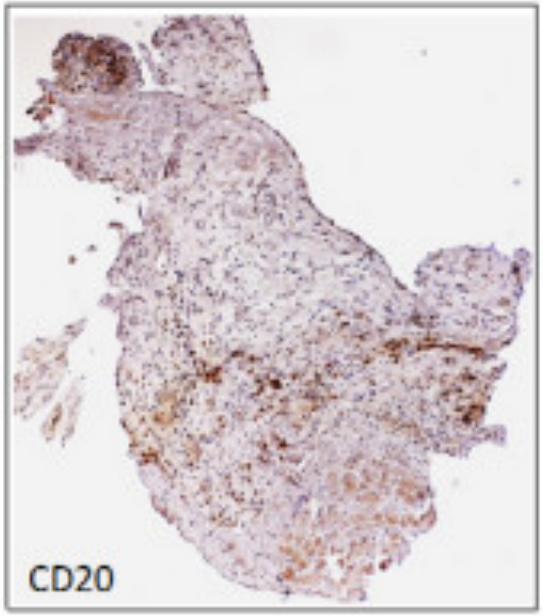
SOLO CARDIOMIOPATIE SU BASE GENETICA?

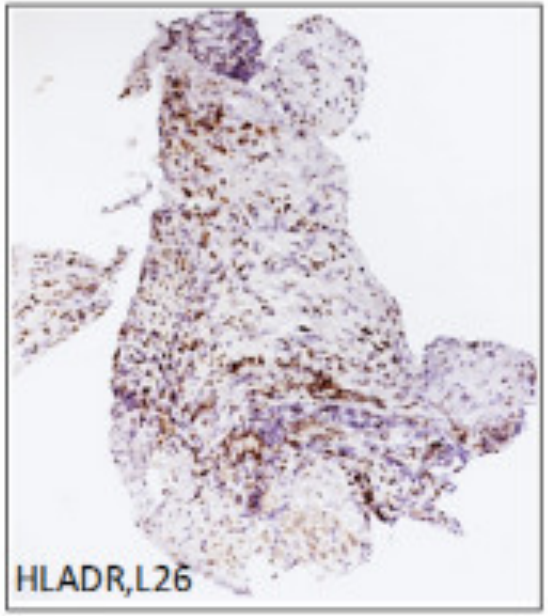
$M_{(Acute\ onset\ HF)(>sCPK)}\ O_{H+M}\ G_S\ E_{M\text{-}V[CVB3]}\ S_{D\text{-}IV}$

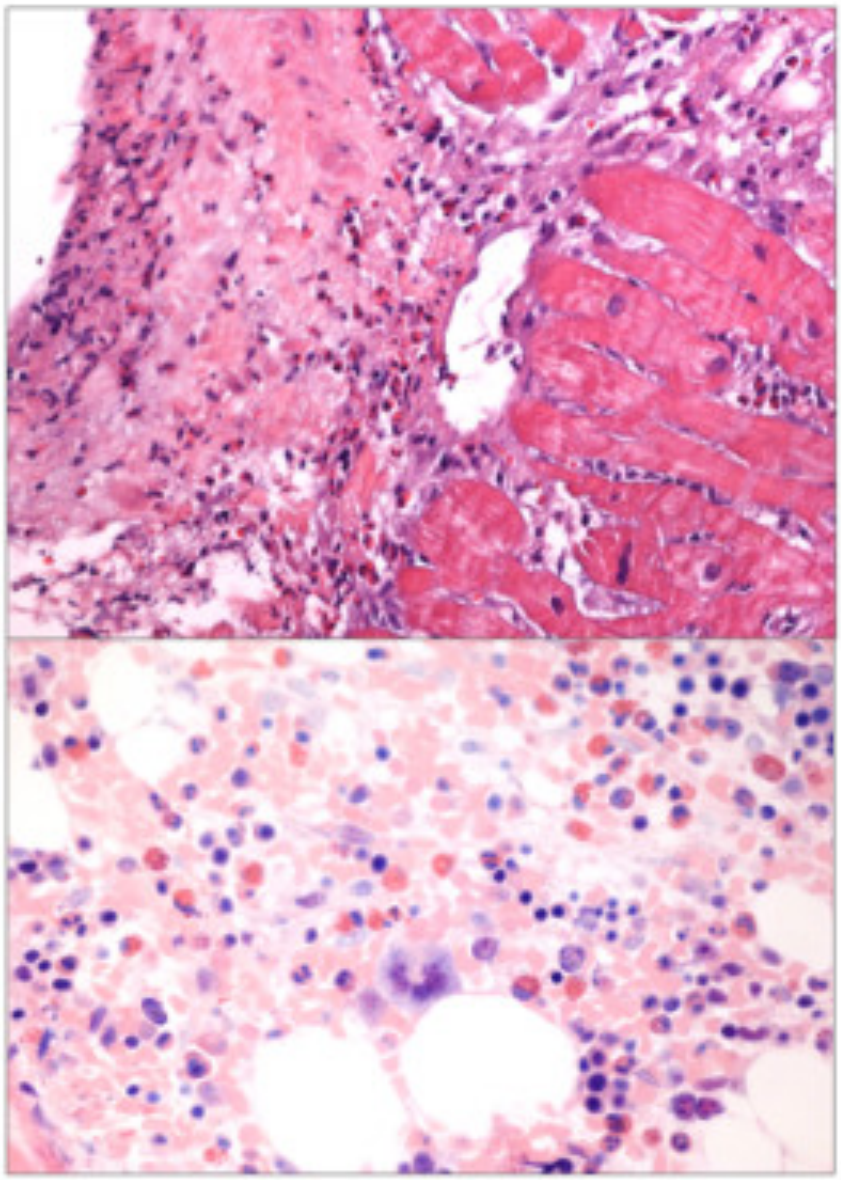
$M_R\ OH_{H+Lu}\ G_S\ E_{M\text{-}Eo}\ S_{C\text{-}II}$











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- **Viral (V),** with the virus (e.g., Coxsackie B3 virus [CB3], human cytomegalovirus [HCMV], Epstein-Barr virus [EBV]) using the Taxonomy system **as coded by the International Committee on Taxonomy of Viruses (http://www.ictvonline.org/index.asp)** [i.e. ($E_{V\text{-}HCMV}$), ($E_{V\text{-}CB3}$) or ($E_{V\text{-}EBV}$)].

- **Infectious, non-viral (E_I)** with an added description of the type of infection based on the Human Infectious Disease Taxonomy;

- **Myocarditis (E_M)** when the myocarditis is the proven cause of the myocardial disease, with specific cause such as viral ($E_{M\text{-}V\text{-}CB3}$),sarcoidosis ($E_{M\text{-}Sarcoid}$) or giant cell myocarditis ($E_{M\text{-}Giant\ cell}$);

- **Autoimmune etiology,** either suspected or proven ($E_{AI\text{-}S}$) or ($E_{AI\text{-}P}$).

- **Non-heritable amyloidosis** with kappa ($E_{A\text{-}K}$), lambda ($E_{A\text{-}L}$), or serum amyloid A protein ($E_{A\text{-}SAA}$) characterization.

- **Toxic cardiomyopathies,** such as pheochromocytoma-related ($E_{T\text{-}Pheo}$), or drug-induced ($E_{T\text{-}Chloroquine}$) cardiomyopathy. When the former is in the context of a syndrome (such as VHL, or MEN2A/2B or NF1), the name of the syndrome could be added (i.e. $E_{T\text{-}Pheo\text{-}VHL}$).

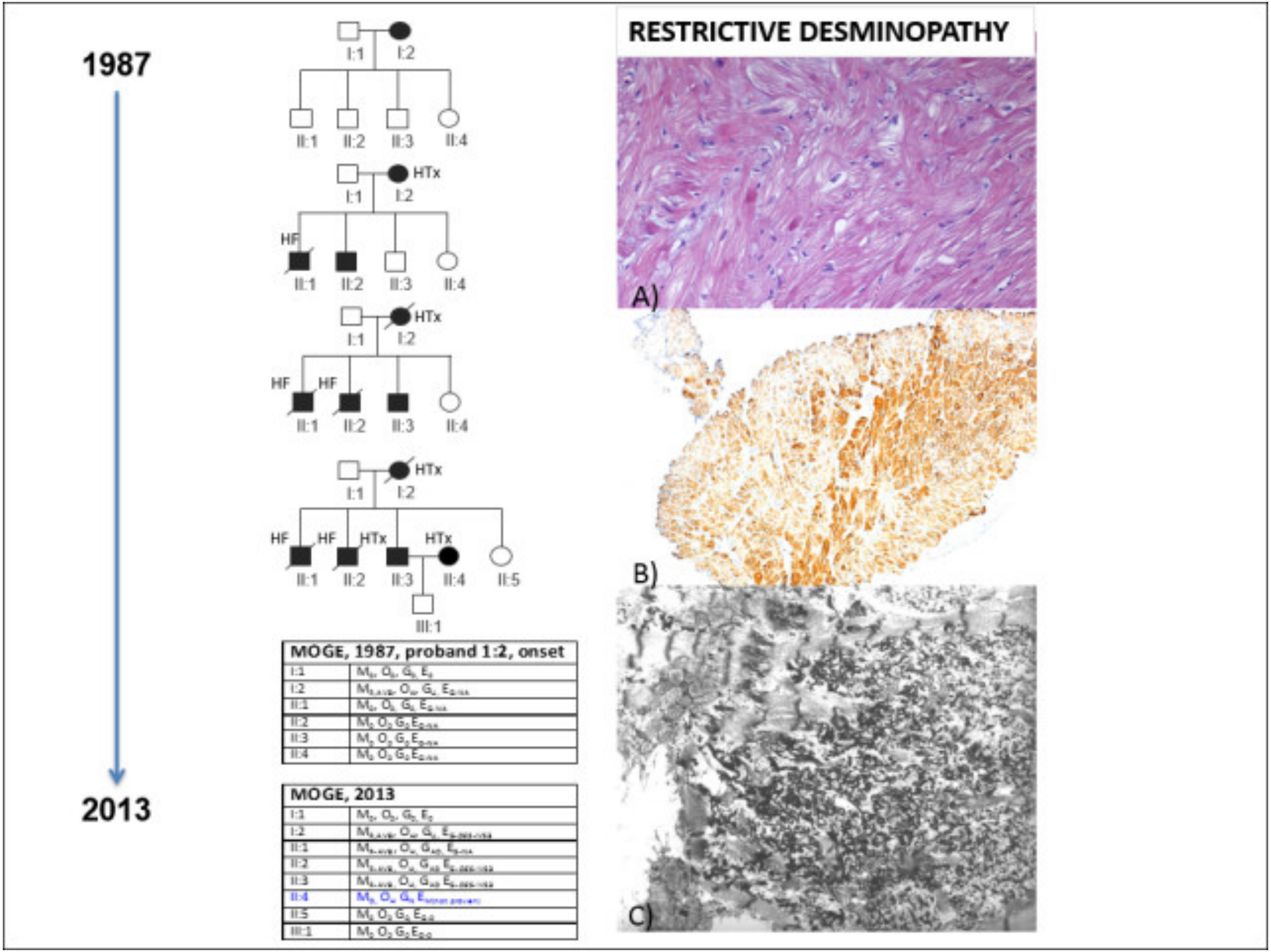
- **Eosinophilic "Loeffler" endomyocarditis** may be described, according to the cause, as either being idiopathic or part of a myeloproliferative disorder associated with the somatic chromosomal rearrangement of PDGFRa or PDGFRb genes that generate a fusion gene encoding constitutively active PDGFR tyrosine kinases.

Other applications

MOGE(S) can also be tailored to define geographically specific forms of myocardial involvement, including

- **T**oxic [such as ingestion of paraphenylene diamine) ($\mathbf{M_D\ O_H\ G_{N\text{-}S}\ E_{T\text{-}[paraphenylene\ diamine]}}$)],
- **O**ther (nutritional/deficiencies such as Keshan Disease in China ($\mathbf{M_D\ O_H\ G_{N\text{-}S}\ E_{O\text{-}[Selenium\ deficiency]}}$) causes.
- Transient causes of DCM can also be described using MOGE(S) in double-step compilation, such as Sheenan Syndrome ($\mathbf{M_D\ O_H\ G_N\ E_O}$)→ ($\mathbf{M_{D(Sheenan\ S)}\ O_{H+Pituitary,}\ G_S\ E_{O\text{-}[Post\text{-}partum\ panhypopituitarism]}}$).

FOLLOW-UP→ PRECISE AND COMPACT DATA



MOGE, 1987, proband 1:2, onset	
I:1	M_0, O_0, G_0, E_0
I:2	$M_{R (AVB)}$, O_H, G_U, E_{G-NA}
II:1	M_0, O_0, G_U, E_{G-NA}
II:2	M_0 O_0 G_0 E_{G-NA}
II:3	M_0 O_0 G_0 E_{G-NA}
II:4	M_0 O_0 G_0 E_{G-NA}

MOGE, 2013	
I:1	M_0, O_0, G_0, E_0
I:2	$M_{R (AVB)}$, O_H, G_U, $E_{G-DES-NA}$
II:1	$M_{R (AVB)}$, O_H, G_{AD}, E_{G-NA}
II:2	$M_{R (AVB)}$, O_H, G_{AD}, $E_{G-DES-NA}$
II:3	$M_{R (AVB)}$, O_H, G_{AD}, $E_{G-DES-NA}$
II:4	M_0, O_H, G_N, E[illegible]
II:5	M_0 O_0 G_0 E_{G-0}
III:1	M_0 O_0 G_0 E_{G-0}

MOGE(S) CAN DESCRIBE COMPLEX GENETICS

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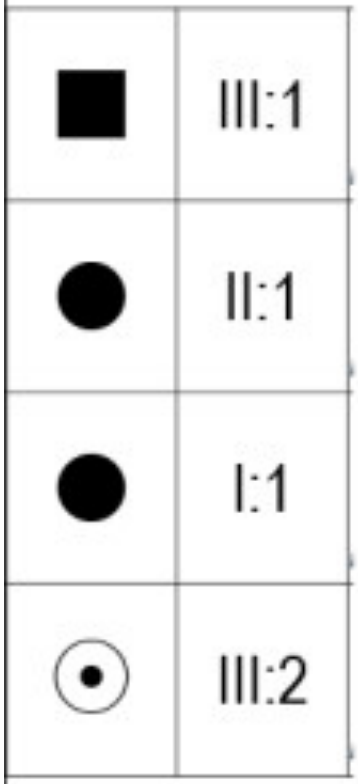
MOGE(S)
DYS Defects→ why I:1 and II:1 are affected?



MOGE(S)
DYS Defects→ why I:1 and II:1 are affected?

■	III:1	$M_{D(AVB)}\ O_{H}\ G_{XLR\text{-}AD}\ E_{G\text{-}DMD[Del\ exons\ 45]\ +\ LMNA\ [p.Met200Val]}\ S_{C\text{-}III}$
●	II:1	$M_{D(AVB)}\ O_{H}\ G_{XLR\text{-}AD}\ E_{G\text{-}DMD[Del\ exons\ 45)\ ]\ +\ LMNA\ [p.Met200Val]}\ S_{C\text{-}II}$
●	I:1	$M_{D(AVB)}\ O_{H}\ G_{XLR\text{-}AD}\ E_{G\text{-}DMD[Del\ exons\ 45)\ ]\ +\ LMNA\ [p.Met200Val]}\ S_{C\text{-}II}$
⊙	III:2	$M_{0(PR=192msec)}\ O_{0}\ G_{XLR\text{-}AD}\ E_{G\text{-}DMD[Del\ exon\ 45\ ]\ +\ LMNA\ [p.Met200VAl\ ]}\ S_{A\text{-}I}$

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NATURE REVIEWS CARDIOLOGY | NEWS AND VIEWS

Cardiomyopathies: MOGE(S): a standardized classification of cardiomyopathies?

Bongani M. Mayosi

Nature Reviews Cardiology **11**, 134–135 (2014) | doi:10.1038/nrcardio.2013.219

Published online 14 January 2014

MOGE(S) is an admirable achievement on the path towards a globally accepted nomenclature for cardiomyopathy. The efforts to reach a consensus on the definition and classification of cardiomyopathy have been spearheaded by single-country or single-continent societies, such as the AHA and ESC. Future development of the MOGE(S) classification should include participation from experts from all regions of the world, including Africa, where cardiomyopathies are endemic.[10] The MOGE(S) nosology provides a sound basis on which a global consensus on the definition and classification of cardiomyopathy can be achieved, under the auspices of the World Heart Federation and its global affiliates.

Nature Reviews Cardiology published online XX Month 2014; doi:10.1038/nrcardio.2013.219-c1

MOGE(S) nosology in low-to-middle-income countries

Eloisa Arbustini, Navneet Narula, G. William Dec, K. Srinath Reddy, Barry Greenberg, Sudhir Kushwaha, Thomas Marwick, Sean Pinney, Riccardo Bellazzi, Valentina Favalli, Christopher Kramer, Robert Roberts, William A. Zoghbi, Robert Bonow, Luigi Tavazzi, Valentin Fuster and Jagat Narula

Box 1 | MOGE(S) classifications of tropical EMF

$M_{R\ (EMF\ LV)}\ O_{H+\uparrow Eo}\ G_N\ E_{I\text{-}S\text{-}Schistosomiasis}\ S_{C\text{-}IV}$

M Restrictive cardiomyopathy (R) associated with EMF predominantly involving the left ventricle (LV)
O Heart involvement (H), and ↑ Eo
G Absence of familial aggregation (N)
E Infectious aetiology (I), possibly schistosomiasis, which is suspected (S) as the cause of the EMF
S AHA stage C (structural heart disease with previous or current symptoms of heart failure), in NYHA functional class IV

$M_{R\ (EMF\ RV+LV)}\ O_{H+L+\uparrow Eo}\ G_N\ E_{I\text{-}U}\ S_{C\text{-}III}$

M Restrictive cardiomyopathy (R) associated with EMF involving both the right ventricle (RV) and the left ventricle (LV)
O Heart (H) and liver (L) involvement, and ↑ Eo

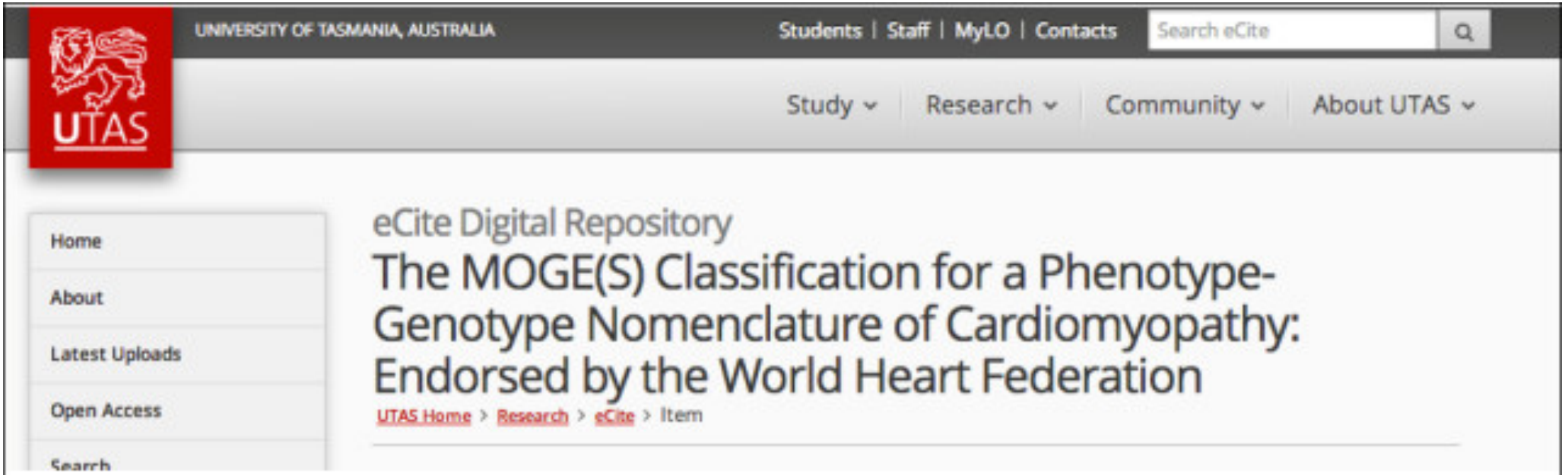
O Heart (H) and liver (L) involvement, and ↑ Eo
G Absence of familial aggregation (N)
E Infectious aetiology (I), possibly helminthiasis, which is suspected (S) as the cause of the EMF
S AHA stage C, in NYHA functional class IV

$M_{R\ (EMF\ LV)}\ O_{H+\uparrow Eo}\ G_N\ E_{Eo\text{-}HES\text{-}PDGFRA/PDGFRB}\ S_{C\text{-}II}$*

M Restrictive cardiomyopathy (R) associated with EMF predominantly involving the left ventricle (LV)
O Heart (H) involvement, and ↑ Eo
G Absence of familial aggregation (N)
E Eosinophilic heart disease (Eo) and hypereosinophilic syndrome (HES), in which a *PDGFRA/PDGFRB* fusion gene has been identified
S AHA stage C, in NYHA functional class II

*This example is provided to describe the EMF variety associated with hypereosinophilic syndrome and mutations in oncogenic tyrosine kinase receptors. Abbreviations: EMF, endomyocardial fibrosis; ↑ Eo, hypereosinophilia.

The MOGE(S) Classification for a Phenotype-Genotype Nomenclature of Cardiomyopathy: Endorsed by the World Heart Federation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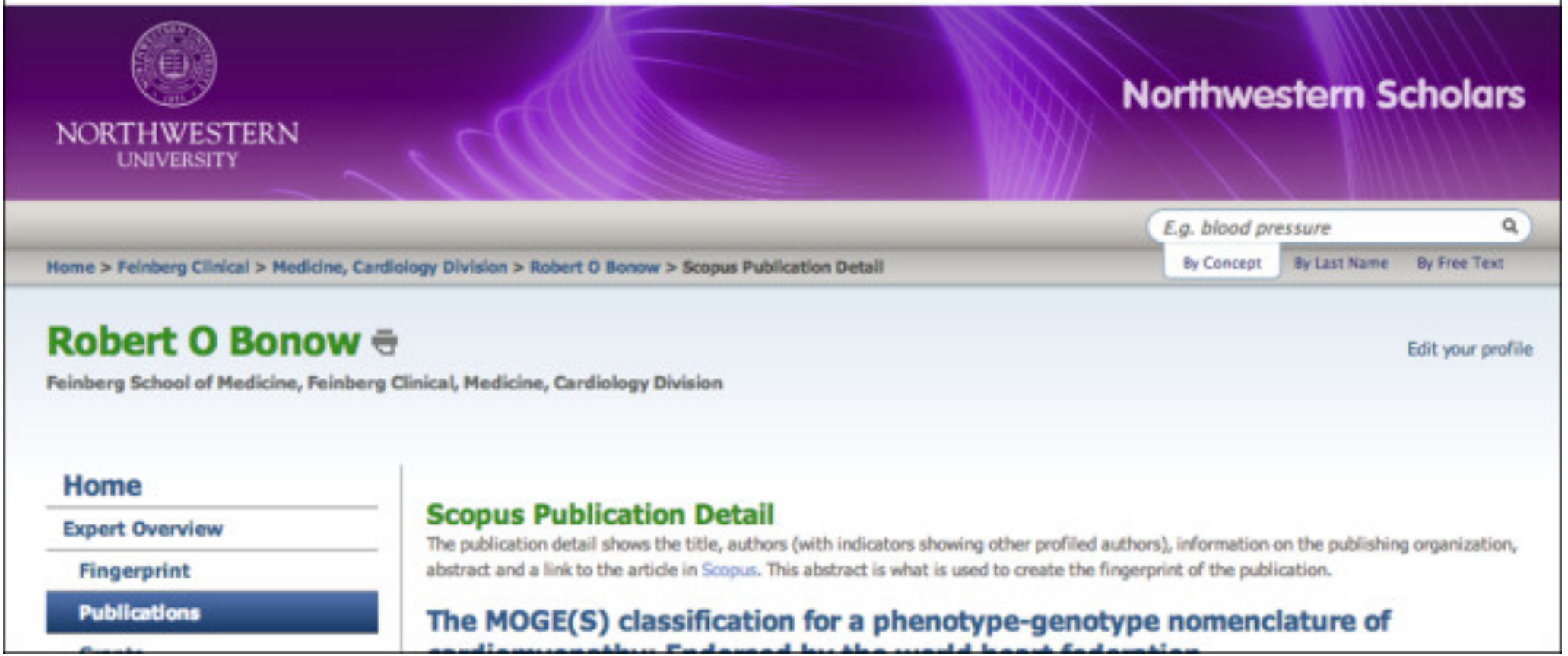
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The MOGE(S) classification for a phenotype-genotype nomenclature of

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The MOGE(S) Classification for a Phenotype–Genotype Nomenclature of Cardiomyopathy: Endorsed by the World Heart Federation

01 Dec 2013

Presented by : Jose Maria López-Ayala, Jesus Martin and Juan Gimeno, Inherited Cardiac Unit, University Hospital Virgen Arixaca

Authors: Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J.

The authors of the paper, endorsed by the World Heart Federation, propose a new classification for Cardiomyopathies based on the current knowledge on phenotype-genotype correlation. A 5 letter scheme called MOGE(S), similar to the TNM classification in oncology, is described in an attempt for standardization. Current classifications (AHA and ESC) have their limitations to appropriately address a constantly changing scenario with increasing sophistication.

The MOGE(S) classification similarly as the TNM is dynamic and patients are coded differently as they develop the disease or after additional information arises from the examinations. A web application for MOGE(S) nomenclature is available at **http://moges.biomeris.com**

MNT Cardiovascular
@mnt_cardiovasc

Segui

Newly proposed MOGE(S) classification system for cardiomyopathy disorders mnt.to/4jkJ #cardiovascular

Risposta Retweet Preferito ••• Altro

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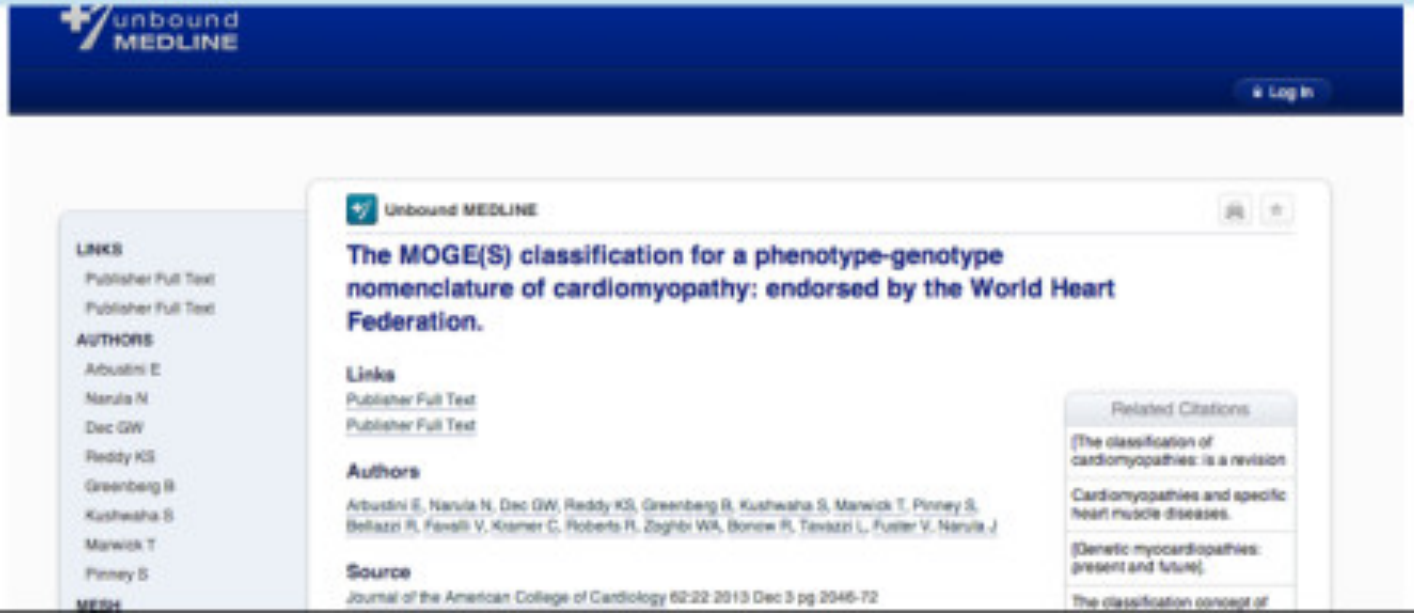

The Desminopathy Reporter

Making sense of missense, nonsense, and other vexsome gene mutations

CardioBuzz: Annotating With MOGES

In the last 10 years, what we know about the genetics of cardiomyopathies has evolved exponentially. Of the 60 or so disease genes confirmed to date (or putative candidate genes), the gene for desmin (*DES*), as well as other familiar suspects—namely, genes associated with myofibrillar myopathy (MFM), *previously known as desmin-related myopathy (DRM)*—figure prominently. Now a new "nosology" (a word most people are unlikely ever to encounter) has been proposed—**a new classification system for heart disease that is endorsed by the World Heart Federation and represents a global Expert Consensus Statement**.

Key Excerpts from MOGES

It's called **MOGES**, an acronym we'll explore in a

CardioBuzz: Annotating With MOGES

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Key Excerpts from MOGES

It's called **MOGES**, an acronym we'll explore in a

Here's how MOGES would convey this information:

$$M_{E(R)[AVB]}O_{H+M}G_{AD}E_{G\text{-}Des[p.Gly84Ser]}S_{A\text{-}I}$$

Now let's unpack this, element by element.

First off, representing **(M)**, the "morphofunctional" phenotype, entries are made for "*early*" (**E**) restrictive cardiomyopathy (**R**) with **AVB**; next, **(O)** for "organ involvement" indicates both the heart (**H**) and skeletal muscle (**M**); the genetic basis **(G)** is autosomal dominant (**AD**) transmission; etiology **(E)** is made explicit with details of the genetic (**G**) defect caused by the ***p.Gly84Ser*** mutation in the gene for desmin (***DES***); and lastly, **(S)** for "functional status" is invoked using ACC/AHA stage (**A**) and NYHA class (**I**).

How's that for powerful and compact annotation!

In this Expert Consensus Statement, the narrative that explains the development of MOGES is replete with numerous, helpful examples of how to use it. Additionally, the authors also offer an easy and convenient, Web-assisted application for on demand use by busy clinicians. **Online help with applying MOGES to complete descriptive classification entries is available at http://moges.biomeris.com.**

Denoix PF. Enquete permanent dans les centres anticancereaux. Bull Inst Nat Hyg 1946;1:70–5.

Edition 1 published 1977 and went into effect 1978

Edition 7 published 2009 and went into effect 2010

THERE WAS A TIME WHEN…
TNM WAS SIMPLE

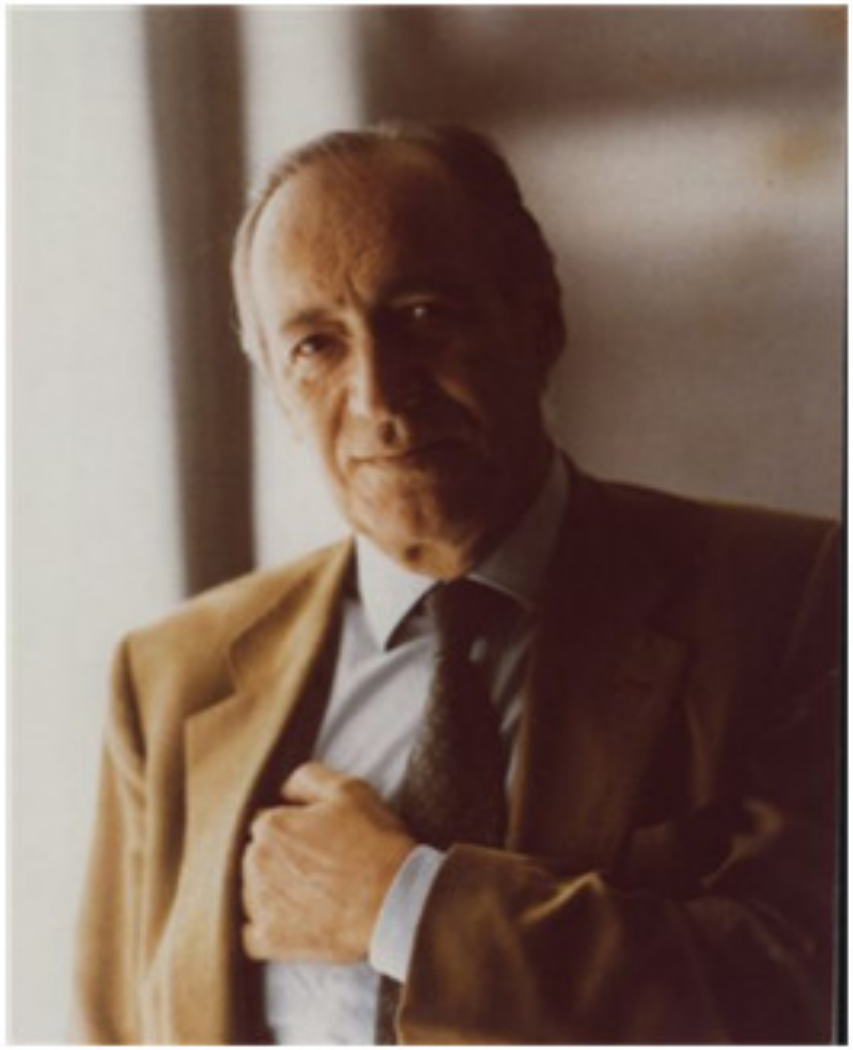
ypT4(m) N0 (i+) M1b G3 LVI + R2.

y denotes that the patient has received neoadjuvant therapy prior to resection,
p presents pathological stage after resection,
T4 offers the extent of tumor which in this case has multiple residual tumor nodules (m) in different lobes of ipsilateral lung,
N denotes the nodal status [N0(i+)] isolated tumor cells only in a lymph node that are considered node negative or N0,
and M represents metastases where M1b means distant metastases (in contrast to M1a, which is thoracic metastases such as contralateral lung, pleural nodules or malignant pleural, or pericardial effusion).
G in this staging is histological grade (1 . Well differentiated; 2 . moderate; 3 . poorly differentiated),
LVI+ represents lymphovascular invasion (LVI, absent)
R is residual disease after treatment (R0 . no residual disease; R1 . microscopic residual disease; R2 . Grossly identified residual disease).
Howsoever complex it may sound, oncologists are expected to use standard TNMstaging. TNM nosology is constantly expanding, is very flexible, but ensures completeness.

Simply looking at [ypT4(m) N0(i+)M1b G3 LVI+ R2] gives physicians all the information about the patient in question. However, in the common practice, this patient is considered to have lung cancer.

Take home message

- MOGE(S)→
  - introduce un sistema descrittivo che aggiunge informazioni sul fenotipo cardiaco ed extracardiaco, sulla famiglia e sulle basi eziologiche, genetico-molecolari della malattia
  - Segue il work-up diagnostico delle cardiomiopatie (e di qualsiasi altra malattia)
  - Flessibile, modificabile, espandibile (app, 3rd edition)
  - Compatta dati essenziali scritti con uno stesso linguaggio internazionale
  - Non modifica e non conclude: annota!



LUI AVREBBE CAPITO!

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