

Screening echocardiography in inherited cardiomyopathies

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In order not to overlap with the previous excellent speakers, I will start with a dilemma...

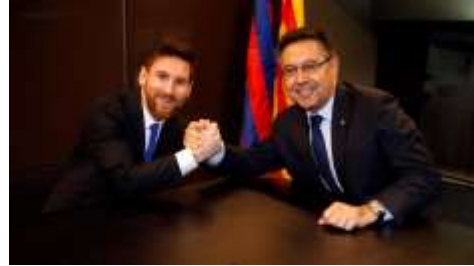
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25 yrs old professional footballer is coming for annual screening...

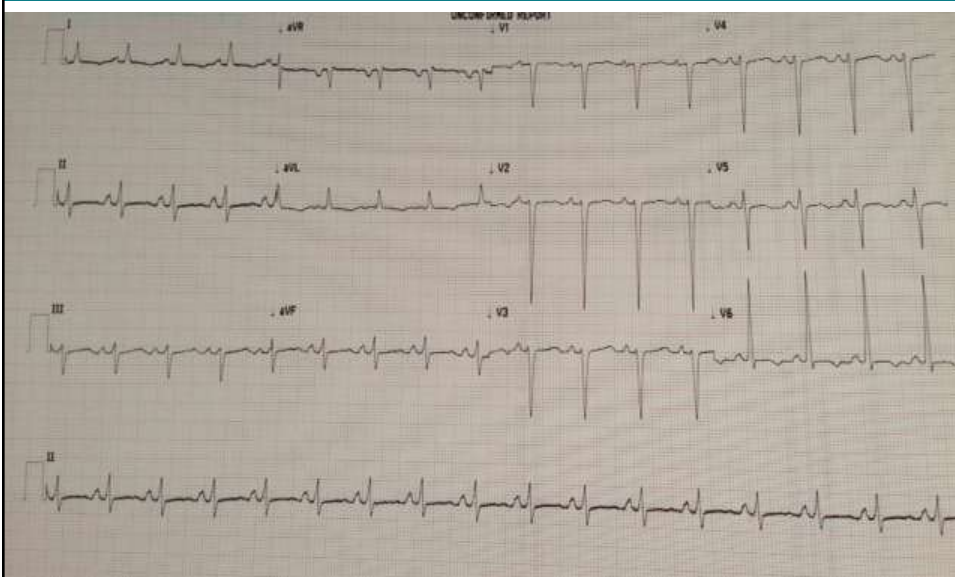


You as the cardiologist...

Vs the professional footballer ...



ECG - asymptomatic



What would be your next step?

1. I would perform stress test
2. I would do a bedside echocardiogram
3. I would let him play football
4. I wouldn't let him play football

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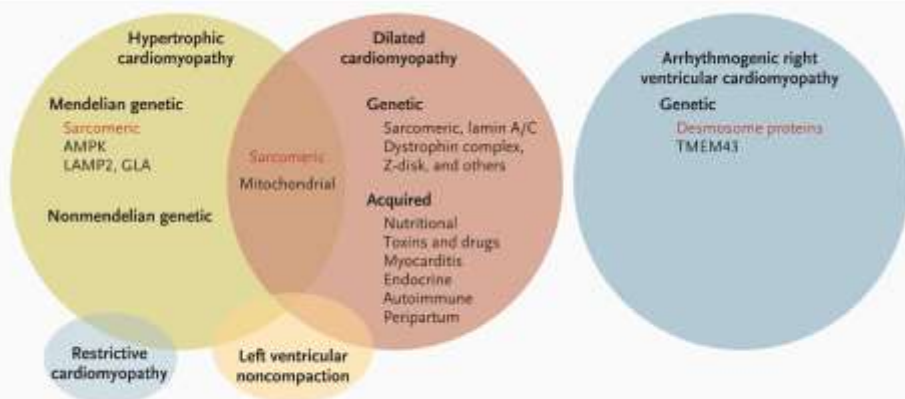
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What happened?

- The patient was diagnosed with dilated cardiomyopathy
- He was intolerant to b-blockers
- MDT decided in favour of urgent cardiac transplant

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Be aware of ...the inherited cardiomyopathies



N Engl J Med 2011; 364:1643-1656

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ORIGINAL ARTICLE

A New Diagnostic Test for Arrhythmogenic Right Ventricular Cardiomyopathy

Angeliki Agapay, Ph.D., Harishchandra Tendin, M.D., Myoung Hoang, Ph.D., Marc R. Halachmi, M.D., Ph.D., Shiva Gadhani, Ph.D., Cristina Salazar, M.D., Ph.D., Gustavo Torres, M.D., Adeline Fatsopoulos, M.D., Nikos Papanicolaou, M.D., William Mulamba, M.D., D.Sc., Hugh Collins, M.D., and Jeffrey E. Gold, M.D., Ph.D.

TEST THE HYPOTHESES ABOUT VENTRICULAR COUPLING

Figure 1. Histopathological images of trichrome-stained double-headed mice. Top left: Heart with normal and non-obstructive ARVC. Middle left: Heart with obstructive ARVC. Right: Heart with normal and non-obstructive ARVC. Bottom left: Heart with normal and non-obstructive ARVC. Bottom right: Heart with normal and non-obstructive ARVC. Scale bars are shown in the bottom right of each panel.

N Engl J Med 2011; 364:1643-1656

Hypertrophic cardiomyopathy

- Present in 1/500 individuals
- Most common cause of sudden cardiac death (> 33% of cases)
- Usually asymptomatic with abnormal ECG



Normal heart



Heart with hypertrophic cardiomyopathy (HCM)



Heart with obstructive hypertrophic cardiomyopathy (HOCM)

Screening Hx for cardiac disease

- Hx of discomfort or chest pain on exertion
- Unexplained syncope or near syncope (especially on exertion)
- Excessive SOB on exertion
- Prior heart murmur
- Increased blood pressure
- Premature death (< 50 yrs old) in 1 relative due to heart condition
- Premature (< 50 yrs old) disability in close relative due to cardiac condition
- Family Hx of HCM, DCM, long QT syndrome, Marfan syndrome, arrhythmia etc

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Other diseases mimicking HOCM...

Condition	Genetics	Inc.	Sex	Phenotype	Substrate
L1/L2/3/4/5/6/7/8/9/10/11/12/13/14/15/16/17/18/19/20/21/22/23/24/25/26/27/28/29/30/31/32/33/34/35/36/37/38/39/40/41/42/43/44/45/46/47/48/49/50/51/52/53/54/55/56/57/58/59/60/61/62/63/64/65/66/67/68/69/70/71/72/73/74/75/76/77/78/79/80/81/82/83/84/85/86/87/88/89/90/91/92/93/94/95/96/97/98/99/100	ADP-actin-1/2/3/4/5/6/7/8/9/10/11/12/13/14/15/16/17/18/19/20/21/22/23/24/25/26/27/28/29/30/31/32/33/34/35/36/37/38/39/40/41/42/43/44/45/46/47/48/49/50/51/52/53/54/55/56/57/58/59/60/61/62/63/64/65/66/67/68/69/70/71/72/73/74/75/76/77/78/79/80/81/82/83/84/85/86/87/88/89/90/91/92/93/94/95/96/97/98/99/100	100%	70% M	Cardiac hypertrophy, pre-excitation	Substrate
Tricuspid regurgitation	Tricuspiditis	7%	100% F	Low voltage, narrow QRS, ST-segment depression	Substrate
Coronary anomalies	Patent coronary artery, anomalous origin of coronary artery (ACA), anomalous origin of coronary artery (ACA), anomalous origin of coronary artery (ACA)	0.5-1%	100% F	Normal ECG, normal chest X-ray, normal echocardiogram, normal cardiac catheterization, normal coronary angiography	Substrate
Left bundle branch block	Aglycosylated A	0.5%	50% M	Normal ECG, normal chest X-ray, normal echocardiogram	Substrate
Right bundle branch block	Unilateral congenital valvular disease	1.5-2%	50% M	Normal ECG, normal chest X-ray, normal echocardiogram	Substrate
Dissecting aortic aneurysm	Marfan, Loeys-Dietrich, Ehlers-Danlos	0.5%	50% M	Normal ECG, normal chest X-ray, normal echocardiogram	Substrate
Myocarditis	Autoimmune, infectious, drug-induced	0.5%	50% M	Normal ECG, normal chest X-ray, normal echocardiogram	Substrate

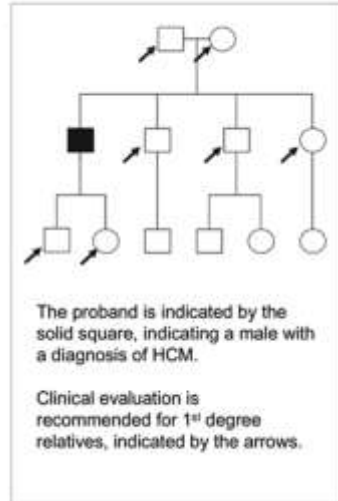


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Hypertrophic cardiomyopathy

Recommended Clinical Screening of Family Members: Physical Examination, Echocardiography, and Electrocardiogram

<12 years old	<p><u>Optional Unless:</u></p> <ul style="list-style-type: none"> Severe family history of early HCM-related death, early development of LV hypertrophy, or other adverse complications Competitive athlete in intense training Suspected symptoms
12-21 years old	Repeat evaluation every 12-18 months
>21 years old	Repeat evaluation approximately <u>every 5 years</u> , or in response to symptoms.
If genetic results available:	<p>Genotype (+) family members: serial clinical evaluation, as above</p> <p>Genotype (-) family members: reassurance; no need for further testing</p>

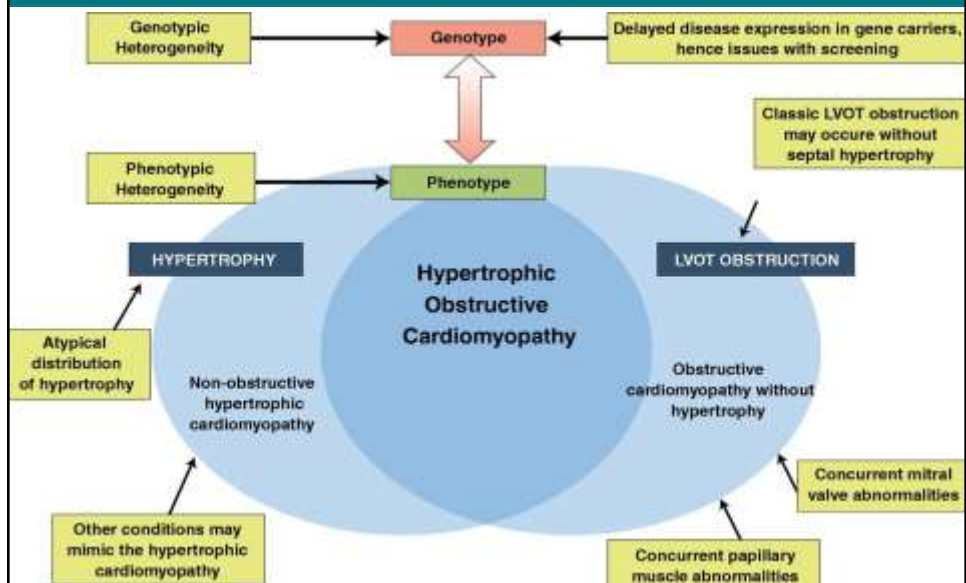


Circulation: Genomic and Precision Medicine. 2013;6:118-131

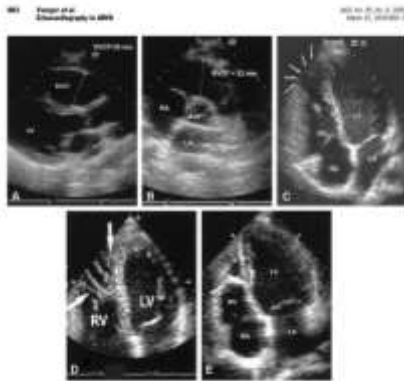
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Cardiac Magnetic Resonance in Hypertrophic Cardiomyopathy

Andrew C.Y. To, Ashwat Dhillon and Milind Y. Desai



ARVC



Echocardiographic Findings in Patients Meeting Task Force Criteria for Arrhythmogenic Right Ventricular Dysplasia
 J Am Coll Cardiol 2005;45:860-5

Approach to family screening in arrhythmogenic right ventricular dysplasia/cardiomyopathy

Annaliese S.J.M. de Riva^{1,2}, Cynthia A. James³, Judith A. Greenberg^{4,5}, Abhishek C. Sawani⁶, Kai Kammann⁶, Brittnay Murray⁶, Crystal Tidwell⁶, James F. van der Haeghe⁷, Daniel P. Judge⁸, Dennis Doores⁹, J. Peter van Tasseler¹⁰, Richard N.W. Hauer¹¹, Hugh Calkins¹², and Marlenebe Tazari^{1*}

Received 26 May 2010; accepted 22 July 2010; online publication 27 August 2010

- Male
- Age
- Symptoms
- FHx
- ECG abnormality
- Holter
- Imaging

Table 3 Characteristics of relatives with definite arrhythmogenic right ventricular dysplasia/cardiomyopathy stratified by arrhythmic outcome

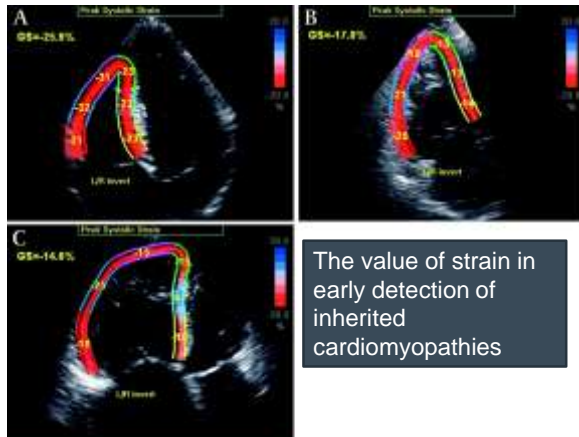
	Overall (n = 96)	No sustained ventricular arrhythmia (n = 75)	Sustained ventricular arrhythmia (n = 21)	P-Value
Male, n (%)	53 (55)	39 (52)	14 (67)	0.076
Age at presentation (year)	62.2 ± 14.4	62.7 ± 14.7	59.2 ± 14.7	0.612
Symptoms at presentation, n (%)	82 (85)	58 (77)	24 (100)	<0.001
Polysymptomatic, n (%)	55 (57)	34 (45)	21 (100)	0.010
Genetics				
None, n (%)	47 (49)	47 (63)	0 (0)	
None, n (%)	54 (57)	55 (73)	0 (0)	
None, n (%)	79 (82)	78 (103)	1 (5)	0.007
Plaque family history, n (%)	41 (43)	36 (48)	5 (24)	
Clinical evaluation				
ECG abnormal, n (%)	79 (82)	67 (89)	12 (57)	0.040
T-wave inversion V1-V3, n (%)	57 (59)	38 (51)	19 (90)	0.006
T-wave inversion V1-V3, n (%)	12 (13)	12 (16)	0 (0)	0.040
T-wave inversion V1-V6 with (T488), n (%)	0 (0)	0 (0)	0 (0)	0.145
T-wave inversion V4-V6, n (%)	67 (70)	61 (81)	6 (29)	0.000
Spindle waves, n (%)	0 (0)	0 (0)	0 (0)	0.006
Prolonged QTc, n (%)	36 (38)	34 (45)	2 (10)	0.030
Holter monitoring (≥ 500 PVCs/24 h), n (%)	68 (71)	49 (65)	19 (90)	0.017
PVC morpho-metric (Q2), n (%)	191 (199-199)	161 (212-212)	30 (143-143)	0.001
Signal-averaged ECG abnormal, n (%)	104 (108)	103 (137)	1 (5)	0.010
Imaging abnormal, n (%)	179 (187)	171 (226)	8 (38)	<0.001
Plaque identified, n (%)	36 (38)	32 (42)	4 (19)	0.003
Pituitary adenoma, n (%)	21 (22)	18 (24)	3 (14)	0.512
Diagnosis per TIC independent of family history, n (%)	45 (47)	34 (45)	11 (52)	<0.001

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Instead of take home messages , I would like to focus on two issues...



The importance of handheld echocardiography in schools and as a screening tool in areas which don't have access to tertiary healthcare



The value of strain in early detection of inherited cardiomyopathies

Thank you



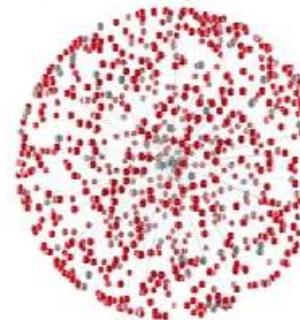
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- www.escardio.org

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