Paciente masculino, 33 años, dolor precordial de tipo pleurítico con fiebre y tos con expectoración purulenta – 2008

Prof Dr. Paul Levine (RIP)

Querido Edgardinho: Acabo de receber este maravilhoso caso do Prof Paul Levine (RIP) e de nenhuma maneira poderia deixar de enviar aos integrantes de nosso amado foro.

Como o foro tem grande número de pessoas inteligentes e muito informadas seguramente teremos aportes de grande valia.

Abraços

Andrés R. Pérez Riera

Paciente masculino, 33 años, dolor precordial de tipo pleurítico con fiebre y tos con expectoración purulenta.

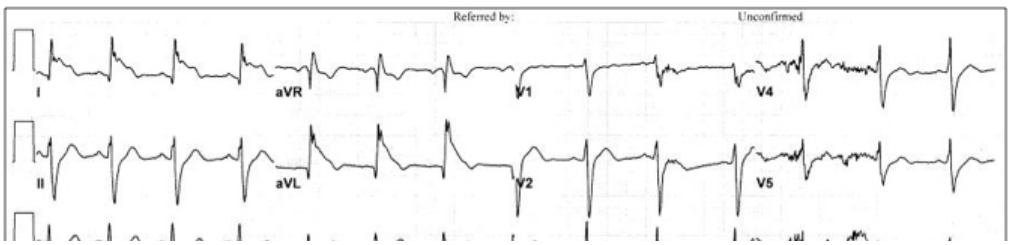
Enzimas cardíacas y ecocardiograma normal.

Sin antecedentes familiares de importancia

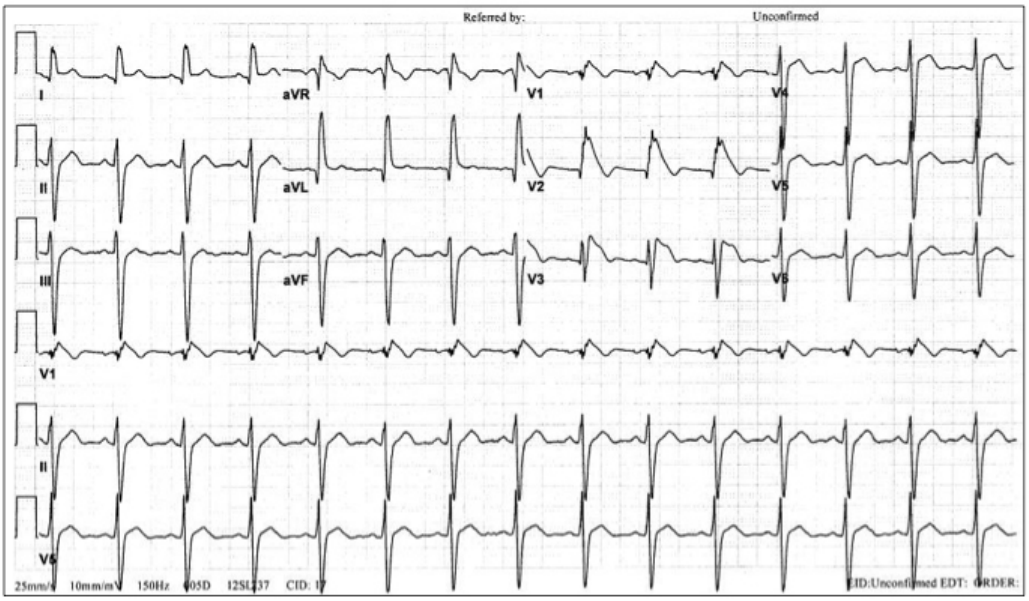
Clinical case

33 year old Caucasian male. Presented with pleuritic chest pain on the evening of February 20, 2008. He also had a low grade fever and a productive cough. Because of the chest pain, a 12 lead ECG was obtained. He was admitted to the hospital where a myocardial infarction was excluded, the cardiac exam was normal as was an echocardiogram.

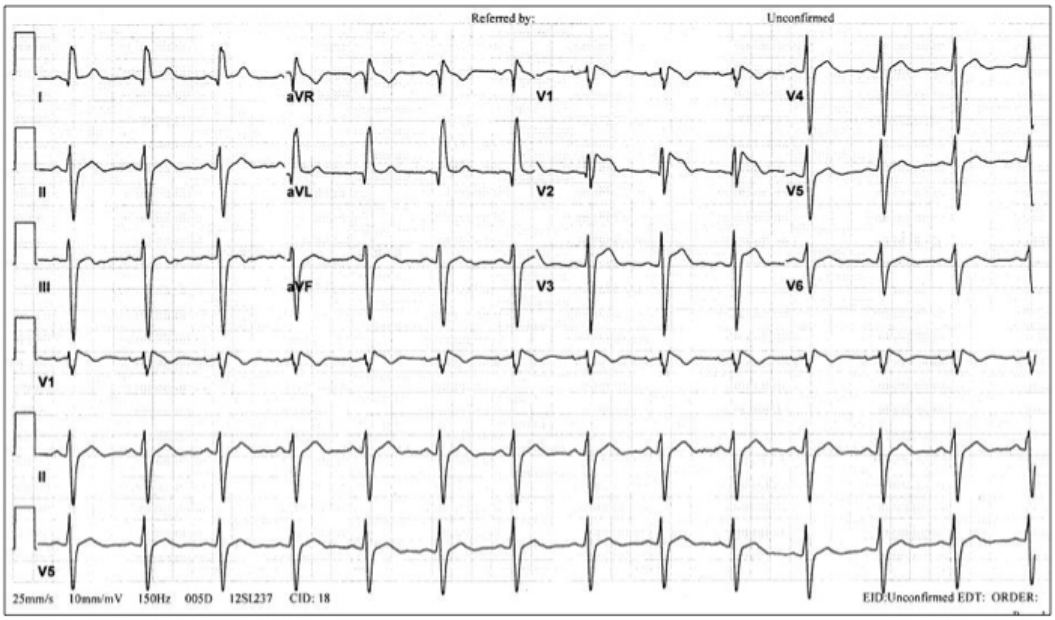
12 Lead ECG recorded at 2209 hours on Feb 20



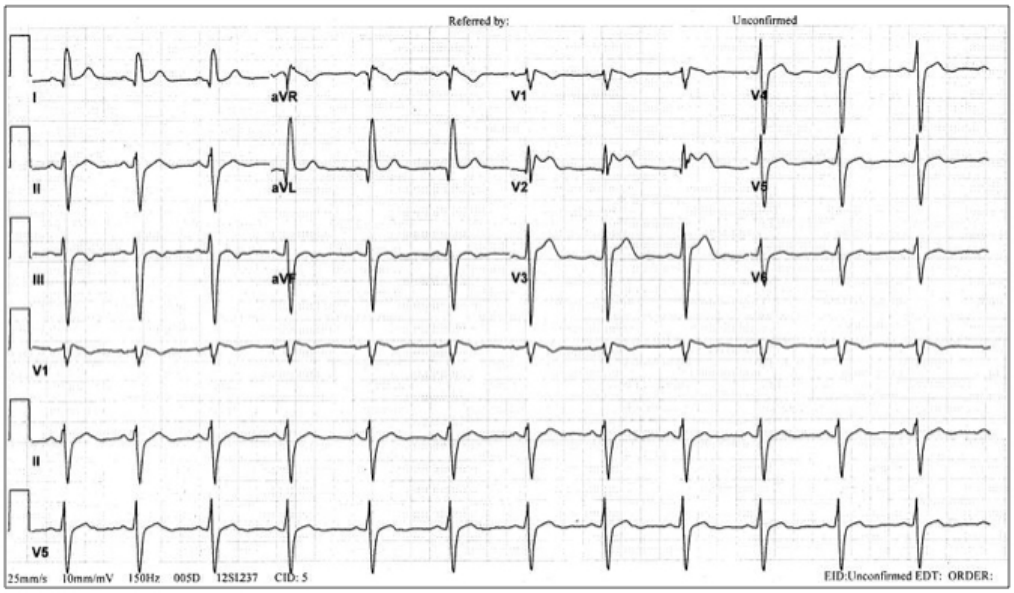
12 Lead ECG recorded at 1123 hours on Feb 20



12 Lead ECG recorded at 0056 hours on Feb 21



12 Lead ECG recorded at 0806 hours on Feb 21



OPINIONES DE COLEGAS

Queridos amigos:

El Profesor Levine (RIP) nos envía dos aclaraciones acerca de su paciente

- El ECG Nro 2 fue tomado a las 11:11 PM (23:11 hs)
- Además de lo aportado, el paciente era portador de una **distrofia miotónica**

Un abrazo

Edgardo Schapachnik

Estimado Andrés,

¡Qué hermoso e interesante ECG!! Hay que agradecerte a vos y al Dr. Levine (RIP) por compartirlo con todos nosotros. A pesar de que no siempre obtenemos estos casos aprendemos de Uds para cuando (ojalá) podamos verlo en nuestra práctica.

a) Si uno hiciera una secuencia de análisis de los ECG sin conocer los posteriores deberíamos hacer con el primer ECG 2 o 3 diagnósticos diferenciales:

1. Paciente jóven con dolor pleurítico, tos y fiebre deberíamos pensar primero en pericarditis, lo extraño es el supradesnivel ST tan localizado (DI-aVL), pero podría ocurrir.

2. Vasoespasmo coronario, que cedería con nitroglicerina

3. IAM como fue descartado.

El dato de la distrofia muscular puede explicar el hemibloqueo anterior izquierdo, ya que esta patología cursa con trastornos de conducción intraventricular.

Hace poco tiempo en nuestro servicio hubo un paciente con distrofia muscular de Steinert. Hice una revisión y aprendí que puede existir esta alteración.

b) El segundo ECG a mi juicio es una patente de Brugada tipo IA. La pregunta que me surge, ¿cómo se explica el supradesnivel del ST lateral sin imagen en espejo en V1-V3; después resuelve y aparece el supradesnivel del ST V1-V3?

Posiblemente el proceso infeccioso puso de manifiesto el Brugada.

c) Finalmente evoluciona a un Brugada tipo II.

d) De acuerdo a esto uno debería pensar que tiene dos malformaciones genéticas, **la distrofia miotónica y el Brugada asintomático.**

Creo que es interesante analizar ¿qué se hace con este paciente? Desde mi punto de vista, por el momento, seguimiento y ver cómo evoluciona la distrofia y el Brugada. ¿Cuál de los dos tiene mayor riesgo?

Un abrazo y gracias nuevamente por este caso.

Oscar Pellizzón.

Brilhante raciocínio Oscarsinho!. Não deixaste nada para ninguém!

Como dizem os yankis: *You are a fast learner.*

Quero fazer um pequeno comentário de tua brilhante análise.

A subclassificação do padrão Brugada tipo 1 em 1A (*ST segment elevation coved to the top*) e 1B (*straight oblique rectilinear descendent*) é uma coisa minha todavia não publicada na literatura indexada.

Digo isto porque muitos dos integrantes do foro não fizeram nosso curso de ECG/VCG e quedarão perguntando-se: ¿qué significa isto?

Andrés R. Pérez Riera

Claro, como vos lo enviaste, me parecíó oportuno señalarlo ya que es una observación interesante y de un electrocardiografista brillante. Yo lo aprendí del curso.

Sería interesante que después me explicaras lo que yo no sé. Gran abrazo.

Oscar Pellizzón

Edgardinho está é minha análise.****

Andres T. Pérez Riera

Dr Levine Case report

1) 2) 3)

I think

1) 2)

3)

ECG 080220 2209: LAFB + ST segment elevation confined in high lateral leads DI and aVL

ECG 080220 2314: LAFB + ST segment elevation in right precordial leads V1- V2 type 1B Brugada pattern

ECG 080221 0056: LAFB+ ST segment elevation in right precordial leads V1- V2 type 1A Brugada pattern, Positive aVR sign (prominent final R wave)

that the diagnosis possibilities are:

¿Prinzmetal angina or vasospastic angina? ¿Acquired form of Brugada syndrome? (2a)
¿Pericarditis?
(2b) Muscular dystrophy?

¿Atypical Brugada syndrome?

1) Prinzmetal angina
DIFFERENTIAL DIAGNOSIS WITH VARIANT ANGINA OR PRIZMETAL ANGINA

In the following table the main differential features between both entities are explained:

DIFFERENTIAL CHARACTERISTICS BETWEEN BRUGADA SYNDROME AND VASOSPASTIC ANGINA

Prinzmetal angina

	BRUGADA SYNDROME	VASOSPASTIC ANGINA
Precordial pain:	No.	Yes.
Tendency to VT/VF:	High.	High.
Structural heart disease:	Absent.	Could exist.
Response to nitrates and nitroglycerine:	Null.	Improves or suppresses clinical/electrocardiographic manifestations.
Permanence of ST segment elevation:	Persistent (or fluctuating) and without pain.	Brief, transitory and accompanied by pain.
Cause:	Genetic alteration of Na+ channel.	Possible alteration in production nitrous oxide in vascular wall.
Presence of image in mirror or reciprocal in ECG:	Could be present.	Present.
Topography of ST elevation:	Right precordial leads of V1 to V3; it could rarely be observed in inferior, lateral wall and **triggered or**	Variable. It could alternate between precordial leads and inferior ones. It could be triggered by hyperventilation.

	increased by antiarrhythmic	

	agents of the IC (Nakamura W, Segawa K, Ito H, et al. Class IC antiarrhytmic drugs: flecainide and pilsicainide, produce ST segment elevation simulating inferior myocardial ischemia. J Cardiovasc Electrophysiol 1998; 9: 855-85.) and IA classes .	
Dromotropic disorders:	AV block of the first degree by extension of H- V in 50% of casesand in carriers of the mutation.	It could happen in a transitory way until a high degree of AV block during the episode; it is associated with a higher risk of arrhythmia and SCD.
Persistent T wave inversion:	Negative T wave in precordial leads from V1 to V3, characteristic of type 1.	Inverted and deep T waves from V1 to V4 associated to anterior hypokinesia, suggesting myocardial “stunning” that indicates critical lesion of the anterior descending artery: “LAD-T wave pattern”.
Presence of transitory Q wave:	No.	It could happen.
Effort test:	It could normalized the variation during effort.	Variable response.
Myocardial scintigraphy with thallium 201:	Normal.	Transitory transmural hypo -uptake.
Response to test of maleate of ergonovine in doses of 0.05 to 0.40 mg (stimulant of alpha adrenergic and seratoninergic receptor)	There could be a mild diffuse reduction of caliber without spasm when doses are equal to or less than 0.40 mg are used.	Intense coronary spasm accompanied by pain and ST elevation. Possible cardiac block, asystole and VT.
Response to hyperventilation:	It does not modify.	Severe spasm and reproduction of clinical electrocardiographic manifestations.
Response to	It could worse the ST elevation	Severe spasm and

intracoronary acetylcholine, each dose given in a time above one minute,	with paradoxical dilation of coronary vessels.	reproduction of clinical electrocardiographic manifestations.

in doses of 10, 25, 50 and 100μg doses separated by five minute intervals:		
Response to magnesium sulfate:	Not mentioned.	Suppresses attacks induced by hyperventilation and exercise.
Treatment:	Automatic implantable cardioverter defibrillator in association with, a drug that contributes to diminish the number of shocks: quinidine Isoproterenol indicated in elctric storm associated with general anesthesia and cardiopulmonary “bypass” or eventually quinidine	Calcium antagonists, such as nifedipine, diltiazem, verapamil, and felodipine associated to nitrates. Benefit with prazosin is mentioned.

1) Acquired form of Brugada syndrome? (2a) Pericarditis
(2b) Muscular dystrophy?

Pericarditis:

Differential diagnosis in chest pain can be broad, and the ECG is often helpful in differentiating acute pericarditis from other causes. Some distinguishing features seen on the ECG in patients with pericarditis include:

1. 1) Concave ST segments: In this case is localized
2. 2) Diffuse ST-segment elevation
3. 3) Absence of reciprocal changes
4. 4) Markers of acute inflammation, such as C-reactive protein, erythrocyte sedimentation rate, and leukocyte count, are commonly elevated
5. 5) Markers of myocardial injury, such as creatine kinase-MB and cardiac troponin I, also may be elevated in patients with acute pericarditis
6. 6) Although troponin levels are often used in the diagnosis of acute coronary syndromes, several reports have documented elevations of troponin I in up to 71% of patients with acute pericarditis. It is hypothesized that the mechanism involves inflammation of the subepicardial myocytes. In one study the median peak cardiac

troponin I concentration was 21.4 ng/mL, which is within the range commonly seen in acute coronary syndromes. In a study by Bonnefoy and colleagues an increase in cardiac troponin I level was seen only in patients with ST-segment elevation and was more likely to occur in younger patients and those with a recent infection.

7) **Chest radiography**: Chest radiography usually has a limited role in the evaluation of acute pericarditis. In the presence of a large pericardial effusion (at least 250 mL), a radiograph may show an enlarged cardiac silhouette.

8) **Echocardiography**: Most authorities recommend obtaining an echocardiogram in patients in whom acute pericarditis is suspected. Although the echocardiogram does not help distinguish between causes of acute pericarditis, it is sensitive for detecting a pericardial effusion. When there is clinical suspicion of acute pericarditis, the presence of an effusion supports the diagnosis; however, this finding is not specific, because 8% to 15% of healthy, asymptomatic people have effusions.

9) **Pericardiocentesis**: The role of pericardiocentesis in the evaluation of acute pericarditis has been controversial. Many investigators have evaluated its role as both a diagnostic and a therapeutic procedure. In one prospective evaluation of patients with acute pericarditis, the yield of diagnostic pericardiocentesis (in the presence of persistent illness or suspicion of purulent pericarditis) was 6%, whereas the diagnostic yield of therapeutic pericardiocentesis (for treatment of tamponade) was 29%. In another study,investigators reviewed their 6-year experience with patients who had a large pericardial effusion without hemodynamic compromise. Pericardial drainage in these patients had a diagnostic yield of 7% and had a limited role in the evolution of the effusion. On the basis of these studies, routine diagnostic pericardiocentesis is not recommended, but the procedure is useful for treatment of cardiac tamponade and in cases of suspected purulent pericarditis.

Hermida et al report the case of a man, treated with mesalazine for Crohn's disease who developed drug-induced pericarditis. The ECG showed a coved ST-segment elevation in the right precordial leads V1-V3. The ECG normalized in a few days after mesalazine withdrawal and the follow-up was uneventful. The ECG remained normal. Two ajmaline tests were both negative and ruled out the diagnosis of BS. This observation illustrates that a coved ST-segment elevation from V1-V3. should not be, systematically, regarded as a marker of a specific syndrome, but may also reflect a common electrical manifestation of abnormalities in the right ventricle or pericardium. **Hermida JS, Six I, Jarry G.Drug-induced pericarditis mimicking Brugada syndrome. Europace. 2007; 9: 66-68.**

OZeke et al. from Turkey reported two cases of acute pericarditis presented with interesting ECGs resembling Brugada-like or early repolarization patterns. **Ozeke O, Selcuk MT, Topaloglu S, Maden O, Aras D.Brugada-like early repolarisation pattern associated with acute pericarditis.** Emerg Med J. 2006;23(12):e64.

A 26-year-old man was admitted to the hospital because of acute pericarditis. The patient had a saddle-back type ST-segment elevation shortly after the onset of acute pericarditis. Interestingly, it converted into a type 1 Brugada pattern, subsequently regressed gradually

as acute inflammation improved. After 3 months, right ventricular rapid pacing induced VF, and intravenous sodium channel blocker induced a Type 1 Brugada pattern. The current case implies that a Brugada-type ST-segment elevation, which is thought to be false in acute pericarditis, may be true in some patients with asymptomatic Brugada syndrome. **Kurisu S**, **Inoue I**, **Kawagoe T**, **Ishihara M**, **Shimatani Y**, **Mitsuba N**, **Hata T**, **Nakama Y**, **Kijima Y**, **Kisaka T**.Acute pericarditis unmasks ST-segment elevation in asymptomatic Brugada syndrome.

(2b) Muscular dystrophy: This patient has a muscular dystrophy.

Steinert's disease is a heredo-familial autosomal dominant entity, nearly exclusive of the male sex, caused by an error located either at chromosome 19 (DM1) or at chromosome 3 (DM2 or proximal myotonic myopathy). Being a disease that can cause cardiomyopathy, it presents several similarities with Brugada Syndrome. The latter is an entity without structural heart disease. We believe that this is the first case in Brazil of Steinert's myotonic disease with Electrocardiogram presenting characteristics typical of Brugada syndrome: complete right bundle branch block, J point and ST segment elevation in V1-V2, first degree AV block, normal QTc, and high resolution ECG with abnormal late potentials to the end of filtered QRS.

Clinical case presentation

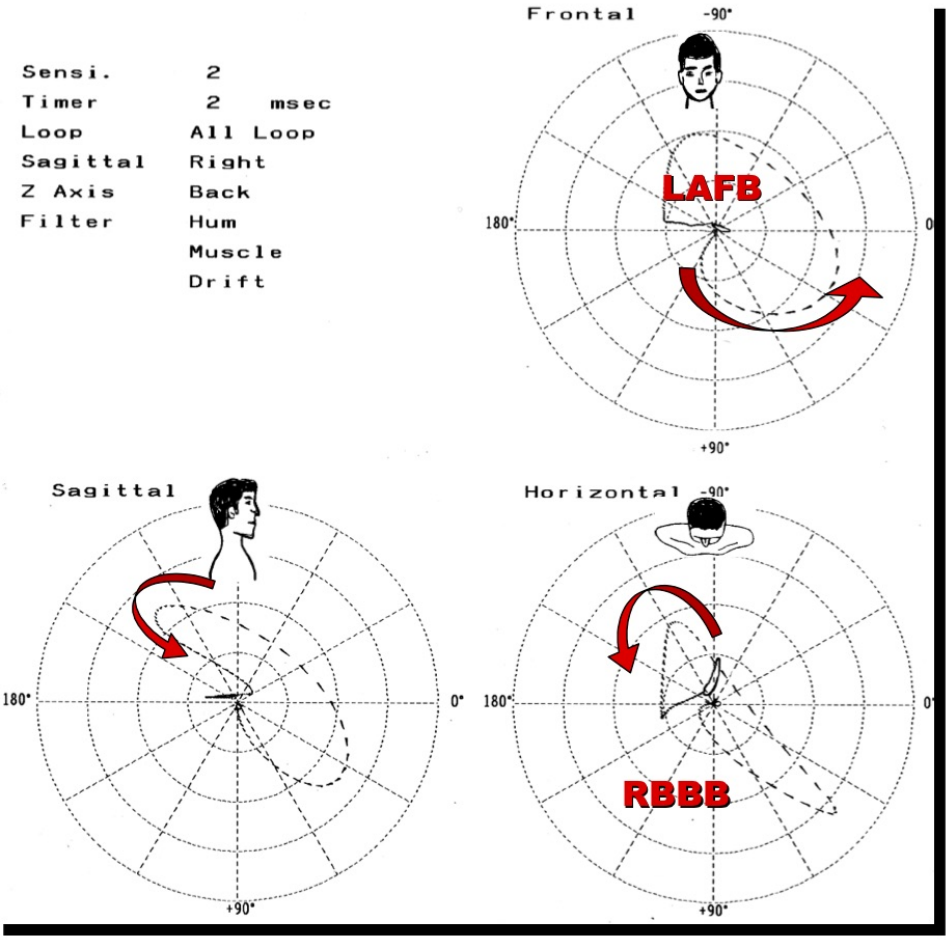
Male patient, 52 years old, from São Paulo (capital city), yellow race (he descends from a Japanese family). He came to our office for the first time on 01/14/2003, on a wheelchair for a cardiologic evaluation prior to a cataract extraction surgery (crystalline removal).

He said he was a carrier of a "family disease that affects muscles" that manifested in him 20 years ago, with progressive characteristics, which confined him to a wheelchair a year ago.
He has known for 5 years now that he suffers from non-insulin dependent diabetes mellitus, under control with a diet.

He found out he is hypertensive 3 years ago, regularly using ACE inhibitor.
For three years he has been noticing growing problems in his sight. He was diagnosed cataracts secondary to his basal disease, and prescribed cataract extraction surgery.

Family background: several brothers affected. He is the third son among 12 siblings. Four of them are affected besides himself. All were studied through genetic mapping. One of the affected siblings died last year due to respiratory failure. A sister affected in a lesser degree still suffers some problems, and from the other 2 with a positive gene, one is asymptomatic and one presents mild symptoms.

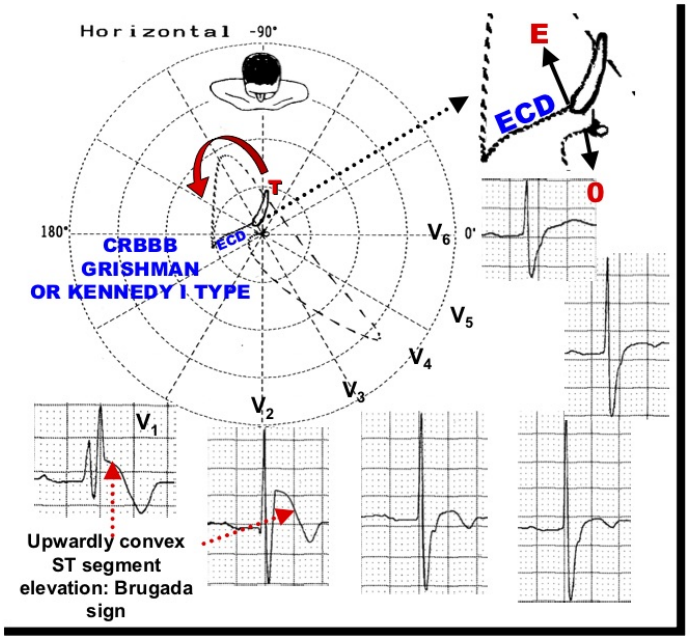
VETORCARDIOGRAM
NAME: **N.M.** DATE: **01/14/2003** AGE: **52 YEARS OLD.** SEX: **MALE** WEIGHT: **82Kg.**
HEIGHT: **1.70m.** RACE: **YELLOW.**
MEDICATION IN USE: **METFORMIN 850mg 2x, ENALAPRIL 10mg 2x.**



Physical examination: abasic patient; notorious proximal atrophy in the deltoid area, face with a mildly dropped eyelids and sleepy expression; the bilateral crystalline opacification

is clearly noticeable even at a distance, extreme difficulty to move upper limbs and impossibility in lower limbs, nasal voice, and suppression of deep reflexes. BP 150/90, about the rest nothing worth mentioning. **Figures 1,2,3 and 4 shows ECG and VCG.**

ECG/VCG HORIZONTAL PLANE



THE END OF THE QRS LOOP (E POINT) DOES NOT COINCIDE WITH THE 0 POINT, JUST AS IT HAPPENS IN BRUGADA DISEASE VCG.

Echocardiogram: signs of decrease in ventricular compliance and discrete concentric myocardial hypertrophy with 12mm septum and posterior wall. Normal-sized LA and discrete posterior basal hypokinetics.
Signal average ECG: presence of late potentials of abnormal ones at the end of the filtered QRS.
Holter: frequent atrial arrhythmias. Frequent polymorphic ventricular extra-systoles. Two episodes of NSVT.

DISCUSSION

Steinert's disease is a neuromuscular disease, heredo-familial, with autosomal dominant transmission, and caused by a genetic mutation located either in the long arm of chromosome 19q13.3 (Myotonic Dystrophy type 1 or MD1)**Harley HG, Brook JD, Rundle SA, et al. Expansion of an unstable DNA region and phenotypic variation in myotonic dystrophy. Nature 1992;355:545-546.)(Buxton J, Shelbourne P, Davies J, et al. Detection of an unstable fragment of DNA specific to individuals with myotonic dystrophy. Nature 1992;355:547-548.)(Mahadevan M, Tsilfidis C, Sabourin L, et al. Myotonic dystrophy mutation: an unstable CTG repeat in the 3' untranslated region of the gene. Science 1992;255:1253-1255.)(Fu YH, Pizzuti A, Fenwick RG Jr, et al. An unstable triplet repeat in a gene related to myotonic muscular dystrophy. Science 1992;255:1256-1258**.) or in chromosome 3 (MD type 2)[**Ricker K, Koch MC, Lehmann-Horn F, et al. Proximal myotonic myopathy: a new dominant disorder with myotonia, muscle weakness, and cataracts. Neurology 1994;44:1448-1452.)(Abbruzzese C, Krahe R, Liguori M, et al. Myotonic dystrophy phenotype without expansion of (CTG)n repeat: an entity distinct from proximal myotonic myopathy (PROMM)? J Neurol 1996;243:715-**

721.) (Rowland LP. Thornton-Griggs-Moxley disease: myotonic dystrophy type 2. Ann Neurol 1994; 36:803-804.)

Clinically, it is characterized by association of difficulties in skeletal muscular relaxation after contraction (myotonic phenomenon) and selective muscular atrophy that causes progressive muscular weakness and myotonic spasms secondary to abnormalities in intracellular calcium content. The entity is multisystemic because it widely attacks the organism; thus, it can cause: cataracts, heart involvement, predominantly involvement of the His system below the His bundle, responsible for different types and degrees of blocks, tendencies to atrial and ventricular arrhythmias, hypogonadism due to testicular atrophy, resistance to insulin with hyperinsulinemia and appearance of diabetes mellitus, dysthyroidism, possible mind deterioration by involvement of the frontal and temporal areas, including some degree of oligophrenics and neuropsychiatric conditions, excess of sleep, involvement of the enteric smooth muscle with dysphagia, sphincter alterations, cholecystitis neurogenic bladder, and anicteric cholestasis, and a skin condition, specially pilomatricoma.

Incidence: this is the most common muscular dystrophy in adults, being the second most frequent muscular dystrophy after Duchenne-Erb's malignant infant myopathy. It affects 1 individual each 8,000, and it is estimated that there are 30,000 carriers in USA and North America.

Age: it begins in average, between 10 and 30 years of age, with a higher incidence between 20 and 25 years. The so-called phenomenon of anticipation has been observed, which consists of earlier appearance and more severe behavior from generation to generation. The evolution in general, is slowly progressive with a wide range of degrees of involvement and advancement of evolution, generally being slow. **Sex:** it affects both sexes in the same proportion.

Race: high incidence between French Canadian and very low in African-American. The MD2 variant is observed with a higher frequency in German descendants.

Clinical and genetic types Three forms are described:

1. 1) Congenital form of myotonic dystrophy (CMyD): it affects chromosome 19q13.3.
2. 2) Classical myotonic dystrophy type 1 (MD1): it affects chromosome 19q13.3.
3. 3) Myotonic dystrophy type MD2 or PROMM (Proximal Myotonic Myopathy): it affects chromosome 3.

1) Congenital form of myotonic dystrophy (CMyD)

This is the most severe form, with a more accelerated progression than the initial forms in adulthood. It is present from birth. It affects children who are sons of mothers carriers of the disease. When the father suffers from this illness, there is no risk for the children. The condition present in the mother is obligatory. Infants appear with extreme muscular

weakness, including face, hypotonia, club feet, sweat, difficulty to suck and to breath, and suppression of deep reflexes.

If they survive after a while of being born, they present problems in language and motor development, and mental deterioration.
The condition occurs in chromosome 19q13.3, and a widening greater than 45 is observed in trinucleotids repeats units.

2) Classical myotonic dystrophy type 1 (MD1)

This is the most common form, which affects the long arm of chromosome 19q13.3, immediately adjacent to the home domain of the SIX5 gene. This is translated into an unstable mutation that affects the production the muscular protein, called protein kinase gene or DMPK (**Ueda H, Ohno S, Kobasyashi T. Myotonic dystrophy protein kinase. Prog Histochem Cytochem 2000; 35: 187-251.)**
The genetic mutation causes a triple repetition or amplification of an unstable trinucleotid, i.e. cytosine-thymine-guanine in chromosome 19. Surprisingly, the mutation occurs in an area of the gene that cannot be translated without affecting the part of the DMPK protein that is important in contractile function.

There are several DMPK protein isoforms in the cardiac and skeletal muscles, nervous system myotendinous tissues, and lymphocytes **(Depardon F, Cisneros B, Alonso-Vilatela E, Montanez C. Myotonic dystrophy protein kinase (DMPK) gene expression in lymphocytes of patients with myotonic dystrophy. Arch Med Res 2001; 32: 123-128).**

The functional role of DMPK still isn't very clear, however it is known that it is fundamental in Ca2+ homeostasis and in signal transduction.
Could the symptoms be caused by a reduction in the amount of DMPK protein?
Could it be the expansion of the DNA segment that affects adjacent genes?

Or could it be that the expansion of the RNA segment intermediary between the protein and the DNA is the responsible one?
The three mechanisms seem to take place in MD1.

3) Myotonic dystrophy type MD2 or PROMM (Proximal Myotonic Myopathy)

1. 1) It is due to the involvement of the long arm of chromosome 3 that affects an "intron" of the gene called ZNF9 with the numbers OMIN 602668, 60109 and 116955;
2. 2) Introns are non-movable parts of the gene that are removed completely before protein production. Researchers think that the ZNF9 protein is affected by the mutation. It is conjectured that the abnormality lies in an excess of RNA, which seems to be decisive for the appearance of manifestations of the disease without affecting the protein.
3. 3) Predominance of proximal muscular involvement of the limbs, however it can affect muscles distally;

4. 4) Evolution less severe than the classical form;
5. 5) It is observed more in German descendants.

MD mechanism

The mechanism of muscular dysfunction is located in slow muscular fibers called type 1, which present atrophy and dysfunction of the endoplasmic-reticulum system (ER). There would be a defect in the Calsequestrin-binding protein, the same one affected in catecholaminergic ventricular tachycardia.

Studies of ultra-structure and biochemicals have proved that the ER affects Ca2+ uptaking **(Mussini I, DiMauro S, Angelini C: Early ultrastructural and biochemical changes in muscle of myotonic dystrophy. J Neurol Sci 1970; 10: 585-604) (Salvatori S, Biral D, Furlan S, Marin O: Evidence for localization of the myotonic dystrophy protein kinase to the terminal cisternae of the sarcoplasmic reticulum. J Muscle Res Cell Motil 1997; 18: 429-440)**, which is found in a lower amount. In the ER terminal cisterna of the type 1 fiber, there is MPK (myotonin-protein-kinase) in a low concentration. Moreover, there is a decrease in expression of a poly-expanded mRNA with negative effects on contractile function **(Wang JZ, Pegoraro E, Menegazzo E, et. al.: Myotonic dystrophy: evidence for a possible dominant negative RNA mutation. Hum Molec Genet 1995; 4: 599-606)**.

CTG repeat in MD reduces the expression of a DNA site called DMAHP gene **(Klesert TR, Otten AD, Bird TD, Tapscott SJ: Trinucleotide repeat expansion at the myotonic dystrophy locus reduces expression of DMAHP. Nature Genet 1997; 16: 402-406)**.

Some endocrine alterations observed (diabetes, hypogonadism **(Mastrogiacomo I, Bonanni G, Menegazzo E, et. al.: Clinical and hormonal aspects of male hypogonadism in myotonic dystrophy. Ital J Neurol Sci 1996; 17: 59-66), dysthyroidism), as well as cardiac dromotropic disorders and late potentials observed in high resolution ECG, are related to the expansion of CTG (Melacini P, Villanova C, Menegazzo E, et. al.: Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. J Am Coll Cardiol 1995; 25: 239-245); while others can have a different source. Nowadays, MD is considered as a channel disease.**

Involvement in MD

In this entity there is a high percentage of patients with cardiac involvement, and severity in these problems is directly related to the number of genetic repeats (CTG triplets: cytosine-thymine-guanine); this being significant in prognosis, which will be the more severe the higher the number of repeats **(Gennarelli M, Novelli G, Andreassi C, et al: Prediction of myotonic dystrophy severity based on the number of the intragenic [CTG]n trinucleotide. Am J Med Genet 1996; 65: 342-347).**

It is more frequent to observe a progressive disease in the intraventricular conduction system of a variable severity correlated with the number of CTG triplets.

Electrocardiogram

Rhythm: supraventricular bigeminy has been reported in MD2 **(von zur M¨uhlen F; Klass C; Kreuzer H ; et al; :Cardiac involvement in proximal myotonic myopathy. Heart 1998; 79:619-21.).** Just as AF and flutter (8% of cases), atrial arrhythmias are more frequent than ventricular ones.

HR: tendency to sinus bradycardia.

P wave: low voltage is mentioned.

PR: it is prolonged in a 40% of instances. Values ≥ 240msec are a good predictor of cardiac events, such as syncope by complete block. Many patients need a permanent pacemaker implantation due to the progression of intraventricular dromotropic alterations. Anesthetics can increase the risk of AV conduction defects and other arrhythmias. PR extension is caused by the increase of the HV interval of the electrogram.

SÂQRS: an extreme deviation to the left has been described, of the electric axis of QRS in the frontal plane **(Evans W. The Heart in myotonia atrophica. Br Heart J. 1944; 6:41-47)**.

Blocks: complete RBBB, is frequent, as well as complete LBBB, bifascicular and trifascicular block, and possible complete block with junctional escape.

Q waves have been described, associated to myocardial infarction.

ST segment and T wave alterations **(Church SC. The Heart in myotonia atrophica. Arch Intern Med 1967; 119:176-181)** are common; ST elevation in right precordial leads having been described as similar to Brugada syndrome.

SMVT by interfascicular reentry is mentioned, which requires radio-frequency catheter ablation.

Sudden cardiac death: terminal events are observed in a 30% of the cases by complete AV block or VT **(Hiromasa S, Ikeda T, Kubota K, et al. Ventricular tachycardia and sudden death in myotonic dystrophy. Am Heart J 1988; 115: 914- 915)**. The events can be detected by 24h Holter **(Fragola PV, Ruscitti GC, Autore C, et al. Ambulatory ECG monitoring in myotonic dystrophy (Steinert's Disease). A study of 22 patients. Cardiol 1987; 74: 362-368) (Forsberg H, Olofsson BO, Andersson S, et al. 24-hour electrocardiographic study in myotonic dystrophy. Cardiol 1988; 75: 241-249).**

Signal average ECG (Milner MR, Hawley RJ, Jachim M, et al. Ventricular late potentials in myotonic dystrophy. Ann Intern Med 1991;115 (no. 8):607-613) (Nalos PC, Gang ES, Mandle WJ, et al. The signal-average electrocardiogram as a screening test for inducibility of sustained ventricular tachycardia in high risk patients: a prospective study. J Am Coll Cardiol 1987;9:539-548): it shows a significant incidence of late potentials, and it constitutes a "good screening" that indicates with certainty sustained VT.

Echocardiogram: the following are described **(Badano L, Autore C, Fragola PV, et al. Left ventricular myocardial function in myotonic dystrophy. Am J Cardiol 1993; 71: 987-991) (Bu'Lock FA ; Sood M ; De Giovanni JV ; Green SH Left ventricular diastolic function in congenital myotonic dystrophy. Arch Dis Child, 1999;80:267-270):**

1. 1) Mitral valve prolapse;
2. 2) Diastolic dysfunction;

3. 3) Prolongation of isovolumetric relaxation time;
4. 4) Prolongation in early filling duration;
5. 5) Increase in velocity of E peak.

Electrophysiologic Study: it must not be prescribed as a routine. The most frequent one is the prolongation of the HV interval. Values above 65msec point out the probability of pacemaker implantation, which will depend on the presence of symptoms such as dizziness, syncope or fatigue. Autopsy reveals fibrosis in conduction system **(Lazarus A, Varin J, Ounnoughene Z, et al. Relationships among electrophysiological findings and clinical status, heart function, and extent of DNA mutation in myotonic dystrophy. Circulation, 1999;99:1041-1046).**

RELATIONSHIP BETWEEN STEINERT'S MYOTONIA AND BRUGADA SYNDROME

We observe peculiar and many coincidences between both entities:

1. 1) Both have autosomal dominant transmission;
2. 2) Both are manifested predominantly initially in the productive time of life (between 20 and 30 years old)
3. 3) Both present a higher incidence of atrial arrhythmias;
4. 4) Both present a higher incidence of ventricular arrhythmias;
5. 5) Both may present an extreme deviation of the electric axis of QRS in the frontal plane;
6. 6) Both present a frequent prolongation of the PR interval;
7. 7) Both have involvement of the intraventricular His system with prolongation of the HV interval in the electrogram;
8. 8) Both present frequent complete RBBB in ECG;
9. 9) Our case shows that both can present upwardly convex ST segment elevation from V1 to V2;
10. 10) Both present high prevalence of late potentials in signal averaged ECG;
11. 11) Both may present E point in QRS loop in VCG, which does not coincide with 0 point;
12. 12) Both are considered channel diseases;
13. 13) Both can affect chromosome 3;
14. 14) Both present tendency to sudden cardiac death;
15. 15) Both present enhanced risk of arrhythmias with anesthetics.

All the best.
Andrés Ricardo Pérez Riera