EC CARDIOLOGY Special Issue - 2021

Homozygous Familial Hypercholesterolemia, Sudden Adult Death, Sudden Infant Death, Epilepsy and Sudden Death due to Travet Syndrome. Genetic Test or Subsequent Molecular Autopsy?

Henry F Collet Camarillo[1]*, Daniel Collet Salgueiro[2] and Manuel Velasco[3]

[1]Cardiologist, Medical Director of the Collet Foundation for Cardiac Catheterization, El Avila Clinic, Altamira, Caracas, Venezuela

[2]Physician Attached to the Collet Cardiac Catheterization Foundation, El Avila Clinic, Altamira, Caracas, Venezuela

[3]Full Professor of Pharmacology,

Director of the Unit of Clinical Pharmacology. Jose Maria Vargas School of Medicine, Central University of Venezuela, Caracas, Venezuela

***Corresponding Author:** Henry F Collet Camarillo, Cardiologist, Medical Director of the Collet Foundation for Cardiac Catheterization, El Avila Clinic, Altamira, Caracas, Venezuela.

Received: March 05, 2021; **Published:** March 17, 2021

The medical and family history is very important but the genetic test helps to clarify the symptoms and justify a more aggressive treatment to avoid strokes, acute ischemic heart disease, and sudden death.

There are guides such as the Dutch lipid clinic network or u.s medped diagnostic that guide patients.

Genetic tests: 29 to 35% of individuals have criteria for familial hypercholesterolemia before performing the test depending on the clinical criteria that is accepted.

If we talk about clinical lipidology, we can guide patients who have possible familial hyperlipidemia or obvious familial hyperlipidemia. Initially these studies were very expensive thousands of dollars, currently there are different genetic teas whose cost is hundreds of dollars which would help to guide these complicated patients. Of the known pathological variants it is in LD LR or apo B. Genetic tests have had a boom today With diseases like mine heart disease, rhythm disorders, Aortic diseases, familial hyperlipidemia. Genome experts have described long QT syndromes and Text has been commercialized to rule out these abnormalities of the myocardial channels of these genes are associated with Lqts-kcnq1, kch2, scn5a, in adults there are other pediatric genes calm1, calm2, calm3trd1 and cacna1C. There are currently more than 36 laboratories in North America, Europe, Australia, New Zealand where genes are screened for long QT syndromes. Ackerman includes the calcium channel gene the Catlett one I know he discovered and he calls it irrefutable for arrhythmias. Of the most common long QT syndromes, 85% are proven, followed by polymorphic ventricular tachycardia, catecholaminergic tac, and Brugada syndrome. Approximately 300,000 to 400,000 deaths per year in the United States are due to sudden death. A molecular autopsy must be implemented and the genetic study must be performed.

Case 1: 50-year-old male patient with multiple risk factors, positive family history of coronary artery disease, history of repair of ascending aortic aneurysm, bentall operation, history of smoking, aortic root dilation, hemochromatosis, documented by genetic test. At present CT angiography with 50% lesions in the left coronary anterior descending artery, 50% stenosis, 50% stenosis in the circumflex, 50% stenosis in the right coronary artery. Ischemia induction tests such as stress test and negative myocardial viability study. Medical treatment and follow-up were indicated.

Citation: Henry F Collet Camarillo., et al. "Homozygous Familial Hypercholesterolemia, Sudden Adult Death, Sudden Infant Death, Epilepsy and Sudden Death due to Travet Syndrome. Genetic Test or Subsequent Molecular Autopsy?". EC Cardiology SI.03 (2021): 01-02.

Case 2: 82-year-old male patient with a family history of coronary artery disease, Huntington's disease, generalized atherosclerosis, homozygous familial hypercholesterolemia under treatment with Alirocumab 150 mg, PSK9-type monoclonal antibody currently on dialysis awaiting kidney graft. In both patients, a genetic study was performed, being monogenic diseases, which were very useful for patient follow-up and effective treatment. There are currently two TLR mutations, seven where patients have a greater predisposition for Covid 19. The syndrome of travet convulsion and sudden death with new treatment with fenfluramine/fintepla anorectic agent and elevation of the pulmonary pressure, which was withdrawn in 1997 by cardiac valve calcification and phentermine [1-3].

Ethical Responsibilities

The authors declare that they have no conflicts of interest when writing the manuscript.

Bibliography

1. D Best., *et al.* "Molecular pathology methods". *Molecular Pathology in Clinical Practice* (2016): 19-52.
2. NJ Boczek., *et al*. "The molecular autopsy: An indispensable step following suddem cardiac death in the Young?". *Herzschr Elektrophys* 23 (2012): 167-173.
3. C Semsarian., *et al.* "Sudden cardiac death in the Young: The molecular autopsy and a practical approach to surviving relatives". *European Heart Journal* 36 (2015): 1290-1296.

Citation: Henry F Collet Camarillo., et al. "Homozygous Familial Hypercholesterolemia, Sudden Adult Death, Sudden Infant Death, Epilepsy and Sudden Death due to Travet Syndrome. Genetic Test or Subsequent Molecular Autopsy?". EC Cardiology SI.03 (2021): 01-02.