



Physician Instructions for Cardiac Screening in FDC Research Participants

Dear Physician:

The purpose of this letter is to inform you of our cardiovascular screening recommendations for participants in the Familial Dilated Cardiomyopathy (FDC) Research Project. Your patient is a participant in the FDC Research Project because his/her relative(s) has idiopathic dilated cardiomyopathy (IDC).

As a participant in our research study, we have asked your patient to accomplish the following:

- 1) **Blood draw:** The tubes provided by our research study should be labeled and shipped (according to the instructions provided) to the FDC Research Project in Miami, FL. This blood draw is for research purposes only. If your staff is able to do this as a courtesy, we would appreciate it very much, or the study can reimburse the patient for the phlebotomy fee. All materials are provided by the study in an enrollment kit given to your patient.
- 2) **Cardiovascular screening:** The **physical exam** should include elements of an annual physical exam, including height, weight, blood pressure and pulse. This exam should be performed by an appropriate health care provider (physician, physician's assistant or nurse practitioner). The **electrocardiogram and echocardiogram** can be done in your office or at another facility that you recommend to the patient. A brief screening echo (i.e. no doppler unless indicated), principally to assess ventricular size and function, is adequate. We will request a copy of the EKG and the echo DVD. Please submit this to the patient's insurance company, as our research study cannot reimburse for these tests. There are diagnostic codes (ICD-9 codes) based on a family history of cardiac disease that can be used when billing for this screening. These include:
 - V17.4: Family history of other cardiovascular diseases
 - V17.41: Family history of sudden cardiac death (SCD)*Excludes: family history of ischemic heart disease or myocardial infarction (V17.3)*

We have provided a list of citations for your reference on the back of this letter, including the Heart Failure Society of America's (HFSA's) recent guidelines and a brief summary of the DCM cardiovascular screening recommendations in this publication. If you have any questions, please feel free to call us at anytime at 1-877-800-3430.

Sincerely,

The FDC Research Team

Cardiovascular Division

Familial Dilated Cardiomyopathy Research Project

Mailing address:

The FDC Project Group,
Cardiovascular Division
P.O. Box 019132, C-205
Miami, FL 33101

Physical address:

Clinical Research Building
1120 NW 14th Street
Suite 1112
Miami, FL 33136

Toll Free: 877 800-3430

Fax: 305-243-2661

www.fdc.to

Ray E. Hershberger, MD

Principal Investigator
rhershberger@med.miami.edu

Ana Morales, MS, CGC

*Genetic Counselor,
Research Coordinator*
amorales4@med.miami.edu

Jill Siegfried, MS, CGC

*Genetic Counselor,
Research Coordinator*
jsiegfried@med.miami.edu

Yves Baptiste

*Clinical Research
Coordinator*
ybaptiste2@med.miami.edu

HFSA Guideline:

Hershberger RE, Lindenfeld J, Mestroni L, Seidman C, Taylor MRG, Towbin JA. Genetic Evaluation of Cardiomyopathy: A Heart Failure Society of America Practice Guideline. *Journal of Cardiac Failure* 2009;15:83-96.

HFSA recommends that all 1st degree relatives (parents, children, siblings) of a person with idiopathic dilated cardiomyopathy undergo cardiac screening, including a physical exam, electrocardiogram and echocardiogram every 3-5 years beginning in childhood. If there are any abnormal results, screening should be repeated at 1 year. This screening is recommended due to the possible familial nature of this disease and because people can have signs of DCM and reduced cardiac function, but remain asymptomatic. These guidelines also provide important evidence for insurance coverage based on a family history of IDC in at least 1 relative.

Related References:

Burkett EL, Hershberger RE. State of the Art: Clinical and genetic issues in familial dilated cardiomyopathy. *J Am Coll Cardiol* 2005; 45:969-81.

Hershberger RE, Cowan J, Morales A, Siegfried JD. Progress with genetic cardiomyopathies: Screening, counseling, and testing in dilated, hypertrophic, and arrhythmogenic right ventricular dysplasia/ cardiomyopathy. *Circ Heart Fail* 2009;2:253-261.

Parks SB, Kushner JD, Nauman D, Burgess D, Ludwigsen S, Peterson A, Li D, Jakobs P, Litt M, Porter C, Rahko P, Hershberger RE. Lamin A/C mutation analysis in a cohort of 324 unrelated patients with idiopathic or familial dilated cardiomyopathy. *Am Heart J* 2008;156:161-9.

Hershberger RE, Parks SD, Kushner JD, Li D, Ludwigsen S, Jakobs PM, Nauman D, Burgess D, Partain J, Litt M. Coding sequence mutations identified in MYH7, TNNT2, SCN5A, CSRP3, LDB3, and TCAP from 313 patients with familial or idiopathic dilated cardiomyopathy. *Clin Trans Sci* 2008; 1:21-26(6).

Hershberger RE, Cardiovascular Genetic Medicine: Evolving concepts, rationale, and implementation. *J Cardiovasc Trans Res* 2008; 1:137-43.