The clinical case of the month: *What is your diagnosis?*

Answers will be given in the next news letter and in the web sit

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A young male infant, without previous medical history, developed congestive heart failure at two month of age. There was no fever, no other organ failure.

Echocardiography exhibited enlarged and hypokinetic left ventricle. Despite rapid hospitalisation and medical treatment, the evolution was rapidly unfavourable and the infant died within few days. Extensive etiological examinations were negative and the diagnosis of Idiopathic Dilated Cardiomyopathy was assessed by the cardiopediatric team. There was no history of similar cardiac disease within the family. The mother and the father were not related by their respective families.

The parents were subsequently addressed to the cardiogenetics consultation and the questions they addressed were to determine whether the disease could be of genetic origin, whether the family should underwent cardiac examination (namely the parents and another child, four years of age), and whether there was a risk for another future child.

What would have been your answer ?

Ten month later, the parents came again to the consultation. Cardiac examination, including echocardiography and ECG, was normal within the family. A heterozygous mutation was found in the cardiac troponin T gene (R141W mutation) in the deceased infant (blood sample was performed before death). The consequences of this genetic result were discussed with the parents. In the same time and consultation, the mother indicated that in fact she was pregnant again (three months).

The parents asked for the mean to prevent the transmission of the disease, including the possibility to perform prenatal diagnosis through amniocentesis, and to discuss medical abortion if the foetus would carry the mutation.

What is your answer and attitude ?

Myocardial and Pericardial Diseases
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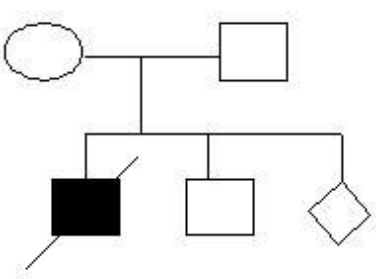
Pedigree of the family :



For more information:
www.escardio.org/CMP

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