QUESTION 16
A doctor makes a diagnosis of hypertrophic cardiomyopathy in a 34-year-old woman. She responds well to medication and returns for regular follow-up appointments. Which one of the following is the most appropriate action to extend care to the woman's family?
A. Advise the woman of the genetic basis of hypertrophic cardiomyopathy.
B. Advise the woman that her immediate relatives should seek medical advice regarding their risk of hypertrophic cardiomyopathy.
C. Contact the relatives regarding the risk of them having hypertrophic cardiomyopathy.
D. Contact the relatives' doctors and recommend that the relatives be assessed regarding the possibility of hypertrophic cardiomyopathy.
E. Confirm the familial basis of the diagnosis of hypertrophic cardiomyopathy by DNA testing.

The genetics of the disease has to be understood before answering the question. Although I have to say I struggled between the decision of Option A or B.

Genetics of disease
- autosomal dominant disease
- 10 causative genes identified (missense type)
- function of genes:
  encoding proteins of cardiac sarcomere components of the thick or thin filaments with contractile, structural or regulatory functions

Varied phenotype
1Wide variation in pattern of L ventricular hypertrophy within a single family pedigree
- environmental influences and modifier genes
- Hypertrophy could represent an adaptive response to malfunctioning contractile ability of cardiac myocytes
- 4 main types:
  1) Hypertrophic
  2) Restrictive
  3) Arrhythmogenic Right Ventricular
  4) Intracellular pathophysiological mechanisms of different mutations poor understood

Routine genotyping
is currently not recommended
- large no. of possible mutations
- expensive and complex lab techniques

Family screening
- advised that all 1st degree relatives are screened for the condition
- this is recommended as HCOM may be present without any Sx
- evaluation involves physical examination, ECG and ECHO

1 Hypertrophic cardiomyopathy: state of the art review, with focus on the management of outflow obstruction, Internal Medicine Journal 2003: 521-529
2 http://www.cardiomyopathy.org
3 http://www.cardiomyopathy.org