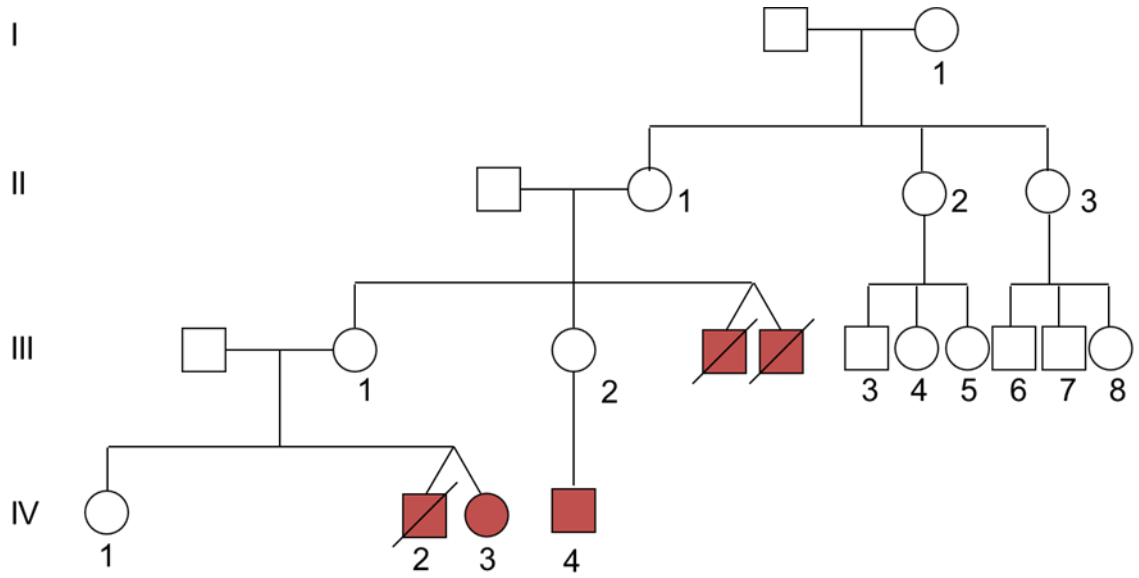
Molecular genetics example

Your cancer geneticist has referred a 35-year old patient with colorectal cancer for testing for Lynch syndrome.	
a)	Which genes are frequently found mutated in this syndrome? [4] ....
b)	What is their role? [2] ....
c)	What particular problem makes searching for mutations in one of these genes more complex? [2] ....
d)	Name two features which tumours from these patients exhibit. [4] ....
e)	Why do some sporadic tumours show the same abnormalities? [4] .... …….. ……..
f)	What other test will suggest the tumour is sporadic? [4] ..... ……. ……. ……..

Joint Cytogenetics and molecular genetics example

In the family tree below. IV-2 died in infancy from an X linked disorder.



a)	Name 4 possible mechanisms whereby a female could manifest an X linked disease normally only expressed in males. [4] ....
b)	If a sequence change is found in a potential causative gene, describe three factors which can be considered when establishing pathogenicity. [6] ....
c)	Name two dominant X linked diseases. [2] ....
d)	Given an equal mutation rate in males and females what is the proportion of cases with a de novo mutation? [2] ....

e)	If only III.1 ,her husband and IV.1 were available for testing, but the family history was known, what genetic tests could you do to reduce the risk of IV.1 having an affected child? [4] .... …….. ……...
f)	II1 is tested and shown not to carry the familial mutation in this X-linked pedigree. What is the explanation for this? [2] ..... …….