

Genetic Mutation and the Family - some case outlines

1. Mary is seen in the Hereditary Cancer Clinic not long before her death from ovarian cancer. She has three adult daughters, but attends the session with her second husband (not the father of her children). Mary elects to store DNA for future testing. Two years after her death, two of the daughters are seen and wish to have testing initiated on the mother's DNA for the breast/ovary cancer genes. Mary's second husband, the executor of her estate, refuses to give permission.

Can Mary's daughters wishes be taken into account? What should the genetic counsellor do?

2. Many laboratories are now switching from testing individual genes for mutations to preparing "gene panels" or even whole genome sequencing. This means that there may be information regarding mutations (and therefore increased lifetime cancer risks) involving many genes. Clinics are needing to prepare for both possible expected and unexpected results. Should all results be divulged? Should results be tailored to only the specific indication bringing the family in?

3. Mutation analysis in children. Li Fraumeni syndrome is a rare but significant cancer predisposition syndrome with presentations even in early childhood with leukemia, sarcoma, and malignant tumours of the adrenal glands. At this time, there are no well evaluated screening modalities for such families. In this situation, should children be tested for an identified family mutation?

4. Prenatal diagnosis is not an acceptable option for all couples at risk to have offspring with serious genetic disorders. For these couples, pre-implantation genetic diagnosis may be preferable. Early embryos, comprised of a small number of cells, can safely have a cell removed for genetic testing. Those embryos free of the family mutation can then be implanted. Should this type of testing be available for adult onset disorders (for example, Huntington Disease), or be restricted to serious childhood onset disorders?

5. A young man drops on the soccer field, is found to be in cardiac arrest and is safely resuscitated. His parents are terrified. After seeing a cardiologist, the young man is identified to have a hereditary arrhythmia syndrome, Long QT, and the causative mutation is identified. Dad has the mutation as well. Both are treated with an implantable pacemaker. The geneticist discusses the importance of dad letting his extended family know of these results. Dad refuses, as he feels this is a private matter.

In this situation, does the geneticist have a responsibility to the extended family?