

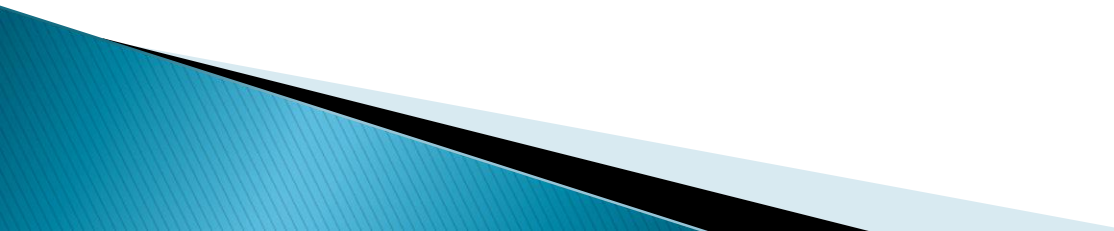
# Medical Genetics, Law and Ethics

Genetic Conditions and the Family

# The patient is embedded in a family...

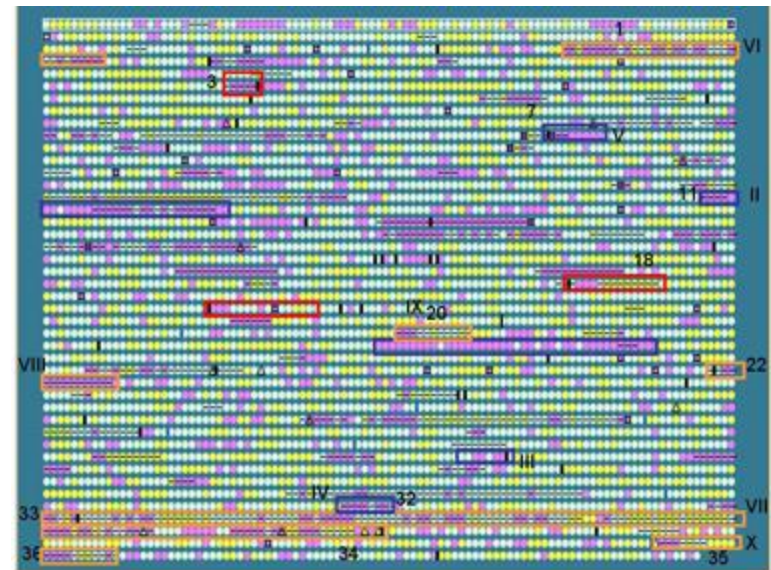
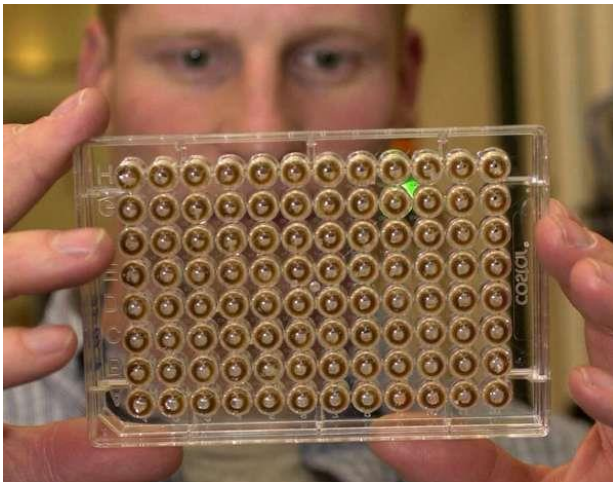


# Case scenario #1

- ▶ 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.
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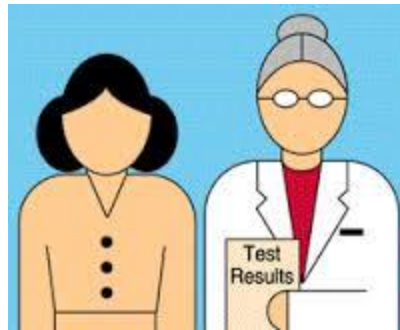
# 14 gene panel

- ▶ A combination of genes for hereditary breast/ovarian, colon cancer and polyposes syndromes



# Case scenario #1 (cont.)

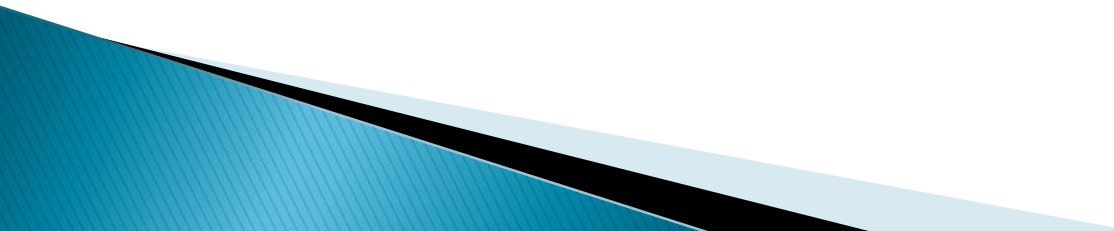
- ▶ Clinics are needing to prepare for both possible expected and unexpected results. Should all results be divulged? Should results be tailored to only the specific indication bringing the family in?



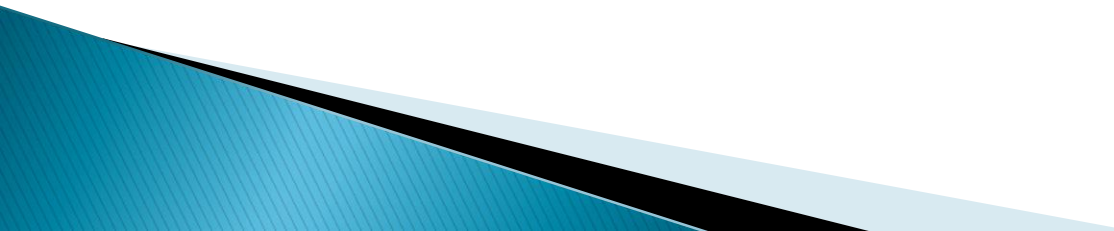
# Is there guidance?

- ▶ **ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing (March 2013)**
- ▶ **Clinical labs to seek and report mutations for a list of genes (57)**
  - Preventative measures available
  - Pathogenic mutations might be asymptomatic
- ▶ **Children, adults but not prenatal**

# issues

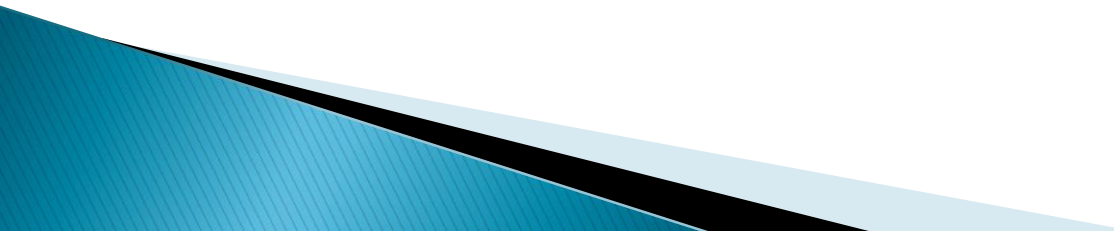
- ▶ Autonomy – could a patient choose to have directed testing only?
  - ▶ What about parents receiving results of adult onset conditions for their children?
  - ▶ Pre-test counselling will be crucial
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# Case scenario #2

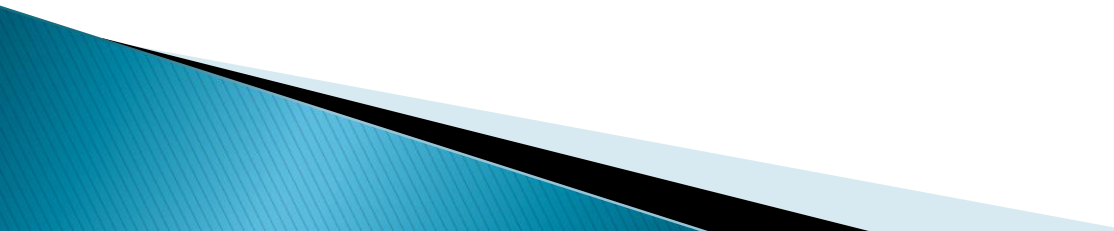
- ▶ Prenatal diagnosis is not an acceptable option for all couples at risk to have offspring with serious genetic disorders. For these couples, pre-implantation genetic diagnosis may be preferable. Early embryos, comprised of a small number of cells, can safely have a cell or supporting cells removed for genetic testing. Those embryos free of the family mutation can then be implanted.
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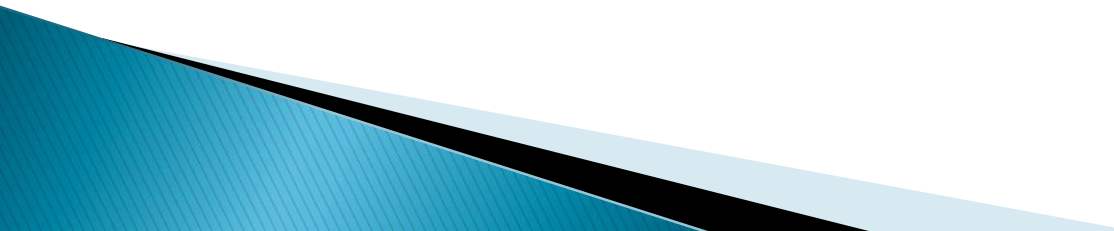
# Case scenario #2 (cont)

- ▶ Should this type of testing be available for adult onset disorders (for example, Huntington Disease) or be restricted to serious childhood onset disorders?
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# What might be the issues?

- ▶ Woman's right to reproductive decisions vs right to life of unborn child
  - ▶ Rights of disabled against the promotion of public health
  - ▶ How serious is the disorder?
    - Offit et al – age of onset, penetrance of disorder, severity and risks, availability of interventions or cures
  - ▶ Number of affected in the family
  - ▶ Emotional impact of potential mother; suffering of future child
  - ▶ Moral difference between future young child and future adult?
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# Other issues?

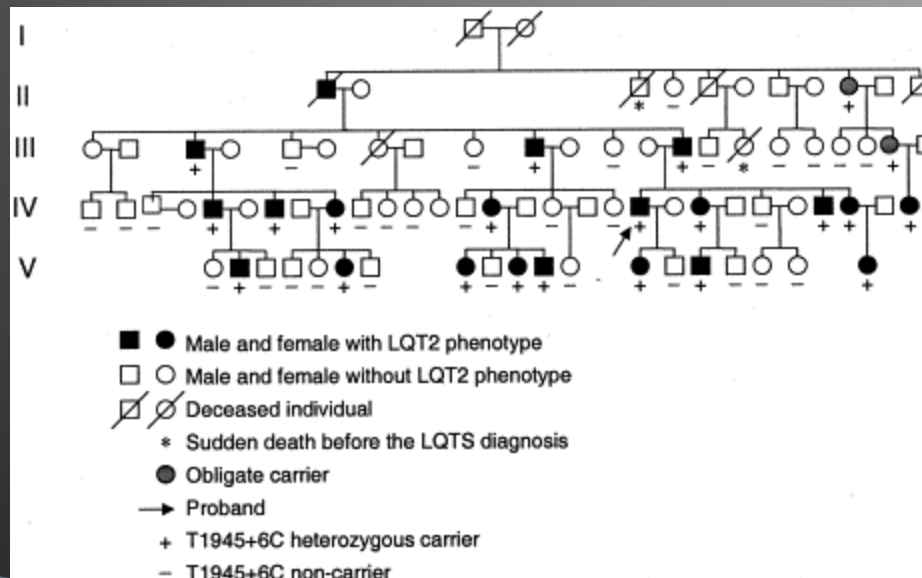
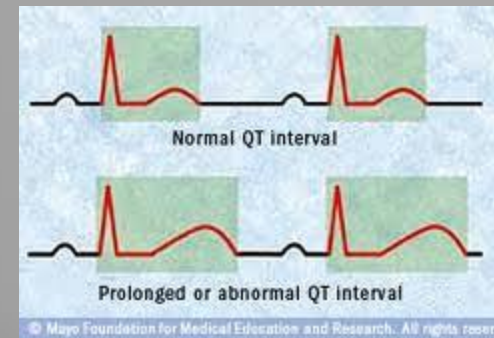
- ▶ If choose to continue an affected pregnancy – the diagnosis of an adult disorder is made in a child
  - ▶ Costs \$2500 to 7000 per cycle
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# Case scenario #3

- ▶ A young man drops on the soccer field, is found to be in cardiac arrest, and is safely resuscitated. 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?

# What is long QT?

- ▶ Fainting
- ▶ Seizures
- ▶ Sudden death



# approaches

- ▶ What principles are in conflict?
  - ▶ Do you have all the facts?
  - ▶ Is there direction or are there developing standards?
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