

Content

- Oct 20 – Applied genetics: DNA forensics --- chapter 6, esp. pp. 146-149
- Oct 22 – Applied genetics: prenatal medicine I --- pp. 282-283
- Oct 24 – Applied genetics: prenatal medicine II
- *Lab: Forensic genetics (Exercise 11) + quiz*

Primary disciplinary learning goals

- To understand the basics of the forensic use of DNA, with a high-technology lab that allows students to use standard procedures themselves to solve a hypothetical crime. As with the Amanda Knox murder case, jurists must understand the forensic use of DNA in order to make informed decisions.
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- To understand the potential that prenatal technologies have in the diagnosis, treatment – and perhaps cure – of human diseases and disorders.
- The goal is to detect, and hopefully cure (well, at least treat), certain kinds of genetic diseases.
- Human development from fertilized egg to new-born baby can go wrong for many reasons, including environmental insults (e.g., alcohol abuse during pregnancy giving rise to fetal alcohol syndrome) or genes (perhaps inherited from one or both parents) that do not function properly.
- Ultrasonography can detect large-scale anatomical problems. But what about more subtle problems, such as disorders of biochemistry?
- Amniocentesis and chorionic villus sampling (let's call the latter CVS) can be used to detect certain genetic abnormalities, by looking at chromosomes and genes, or by looking at gene products (proteins). An extra copy of chromosome 21 might be detected. Such trisomy results in a disorder known as Down syndrome.
- Genetic counselors advise people with known genetic diseases on the odds of having children that may be affected. With that advice, affected couples can then make informed decisions about whether they wish to become biological parents.
- Amniocentesis and CVS can detect subtle problems fairly early in pregnancy, but the technique of pre-implantation genetic diagnosis (or PGD) allows detection of problems even before a very young embryo implants into its mother's womb. How is that possible?
- In PGD, cells from very young embryos produced by *in vitro* fertilization are screened for chromosomal abnormalities and defective genes. Only those

embryos whose cells reveal normal chromosomes and genes are then put into their mothers' wombs.

- Could the technique of PGD be modified to include treatment or even a cure, not just diagnosis? Could we transfer functional genes into cells that have defective ones?
- And what about other uses of PGD? For example, do you think it would be O.K. to use the technique to screen embryos for such traits as sex, eye color or (if we could do it) future intelligence?
- We fairly recently developed another tool for prenatal diagnosis. Believe it or not, DNA from a fetus can be found in a pregnant woman's blood. We can isolate that fetal DNA, make copies (by PCR) and sequence it to see if it is normal. This is a very low-risk procedure because all it involves is taking some of Mom's blood.

We can determine the health and the sex of the baby (ask yourself how). Do you feel comfortable with that?