GENETIC TESTING
Serena and Larry have been trying to have a baby for several years. However, every time they have tried she had a miscarriage.

Who might Serena and Larry’s doctor recommend they go see?
Which woman has a greater chance of having a child with Down syndrome?

Selena Gomez, Age 26

Jennifer Aniston, Age 50

Sorry Jen :/
REASONS SOME MAY SEEK GENETIC COUNSELING

- Advanced maternal/paternal age
- Mother has been exposed to certain chemicals, like mutagens or teratogens
- Mother has a history of certain health issues like alcoholism, diabetes, or seizures
- Couple is having trouble getting pregnant (infertility issues)
Karen is pregnant and went to see her doctor for a regularly scheduled ultrasound. Here, they discovered that the baby has an **omphalocele**.

An omphalocele is when the baby’s intestine is on the outside of its body.

What is an **omphalocele**?

What may the doctor recommend to Karen as her next steps?

Karen should get genetic testing done on the fetus to determine why the baby suffers from this issue.
Jess is looking to conceive, however she has Cystic Fibrosis. She is unsure if her husband, Jim, is a carrier.

What is a carrier?

What might a genetic counselor recommend to this patient?

Results come back from the carrier screening panel and show that Jim is a carrier for the cystic fibrosis gene mutation. What is the chance that their child will have the condition?

50% (and the other 50% represents the chance their child will be a carrier)

A carrier does not have a condition but can pass it on to their children because they carry an allele.

Jim should undergo carrier screening. It will tell him if he carries a disease-causing allele.
REASONS SOME MAY SEEK GENETIC COUNSELING CONT.

- Abnormal finding on ultrasound
- Recurrent miscarriages
- Family history of genetic conditions
- Parents are of a background prone to a certain condition
- Parents are carriers for a condition

Those of Ashkenazi Jew descent are at a greater risk for diseases like Tay-Sachs
Emma is interested in learning whether or not her child is at risk for Edwards syndrome. She is 10 weeks pregnant.

What does it mean if someone is at risk for a condition?

What is another term for Edwards syndrome?

What genetic screening method would Emma be able to seek out?

Noninvasive Prenatal Screening

Risk means someone may have a condition but it is not a definitive answer

Trisomy 18
A simple blood test that takes cell-free DNA (cfDNA)

*cfDNA is DNA that is not inside a cell that is floating around inside the mother’s bloodstream*

Can be performed once there is a high enough *fetal fraction* (enough baby DNA in the mother’s bloodstream) *Occurs around 10 weeks of pregnancy*

Can screen for *aneuploidies* (extra or missing chromosomes) *Trisomies 13, 18 and 21* *Sex chromosome aneuploidy (X’s and Y’s)*

Is a *screening* method *Will not definitively say if the baby has the condition; provides a *risk factor*
OTHER GENETIC TESTING OPTIONS

- Newborn screening
- Diagnostic testing
- Carrier testing
- Prenatal testing
- Preimplantation genetic diagnosis (performed on embryos made by assisted reproductive technology (ART))
GENETIC TESTING
SAMPLES

- Blood
- Amniotic fluid
- Chorionic villi
- Tissue
- Buccal (cheek) cells
- And more…
Henry is a 65 year old man that has recently been experiencing jerking movements, unsteady gait, and slower cognitive processing.

What kind of genetic testing may be performed?
What kind of sample may be used to perform the test?

Sequencing results come back. Henry has an abnormally high number of CAG repeats in the huntingtin gene. What might be the diagnosis?

Huntington disease

Blood
GENETIC TESTING CAN BE DONE FOR A VARIETY OF REASONS

- PRENATAL
- NEWBORN
- CHILDHOOD-ONSET
- ADULT
- CANCER
- NEUROLOGICAL
- CARDIOVASCULAR
- AND MORE!
WHAT LABORATORY TESTING IS DONE TO LOOK FOR ANOMALIES

- Next-generation sequencing
- Sanger sequencing
- PCR
- Southern blots
- Microarrays
- Fluorescence in situ hybridization
- Karyotyping
KARYOTYPING IS PERFORMED WHEN A CHROMOSOME ABNORMALITY IS EXPECTED

...AND CAN BE PERFORMED ON ANYTHING WITH CHROMOSOMES

...THIS IS ACTUALLY WHAT HELPED US LEARN HOW CLOSELY RELATED WE ARE TO CHIMPANZEES

LET’S SEE IF YOU CAN TELL WHO IS WHO
Step 1: Count how many chromosomes you have

Step 2: Arrange by size

Step 3: Band for band analysis
IMAGE CREDITS

- Slide 3: https://people.com/tag/selena-gomez/
- Slide 5: https://www.google.com/search?q=omphalocele&source=lnms&tbm=isch&sa=X&ved=0ahUKEwiOy4G6qrDjAhVUCM0KHXSUBBQQAUIECgB&biw=1536&bih=747#imgrc=_AvmiHVmyhuk_M:
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- Slide 14: https://www.genengnews.com/magazine/286/next-generation-sequencing-challenges/
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- Slide 10: https://ghr.nlm.nih.gov/primer/testing/uses
- Slide 11: https://www.acog.org/Patients/FAQs/Prenatal-Genetic-Screening-Tests?IsMobileSet=false
- Remainder of info gathered from DGS 3222 course