

# *An Introduction to Prenatal Genetics for Healthcare Interpreters*

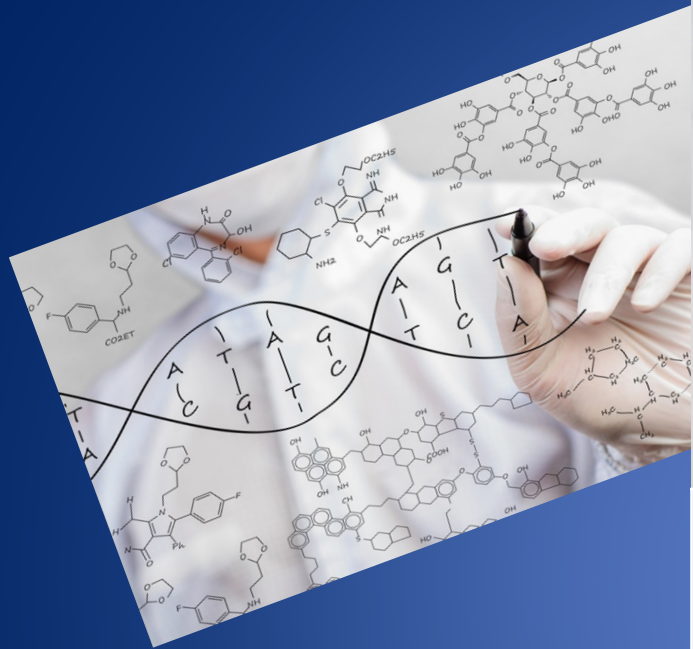
This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770-01-00 from 6/2017-5/2020 for \$800,000 per award year.

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Created 02/19, reviewed and revised 09/2024.

# Housekeeping

- Watch for a second email from me with some documents to print for next week.
- To get a certificate, you must:
  - Attend both classes: arrive on time and stay for the entire class.
  - Keep video on unless instructed to turn it off.



**UCSF**  
MAGAZINE

The First Genome  
Surgeons



Cynthia E. Roat, MPH

Lila Aiyar, MSc, CGC, CCGC

Lori Williamson Dean, MS, CGC

Alissa Bovee Terry, ScM, CGC, LCGC

Meredith Weaver, PhD, ScM, CGC

# What will we be learning today?

- Quick review of human reproduction
- Basic information about genetics
- The work of Genetic Counselors
- Genetic testing during pregnancy or related to reproductive health
- Challenges for interpreters

# What will we be doing next week?

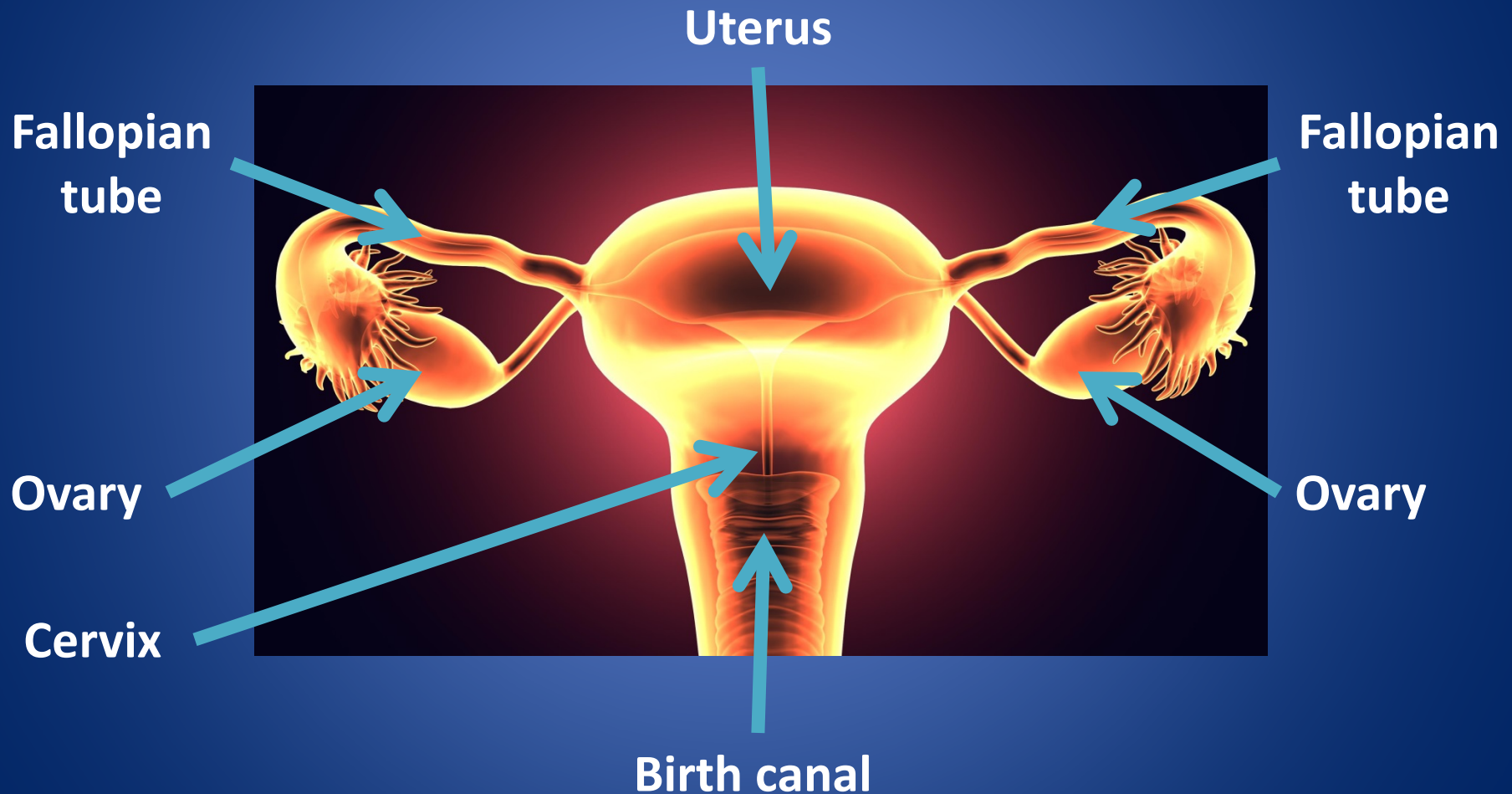
- Exercises with English vocabulary.
- Exercises in converting to another language.
- Practice interpreting exercises

# Pretest

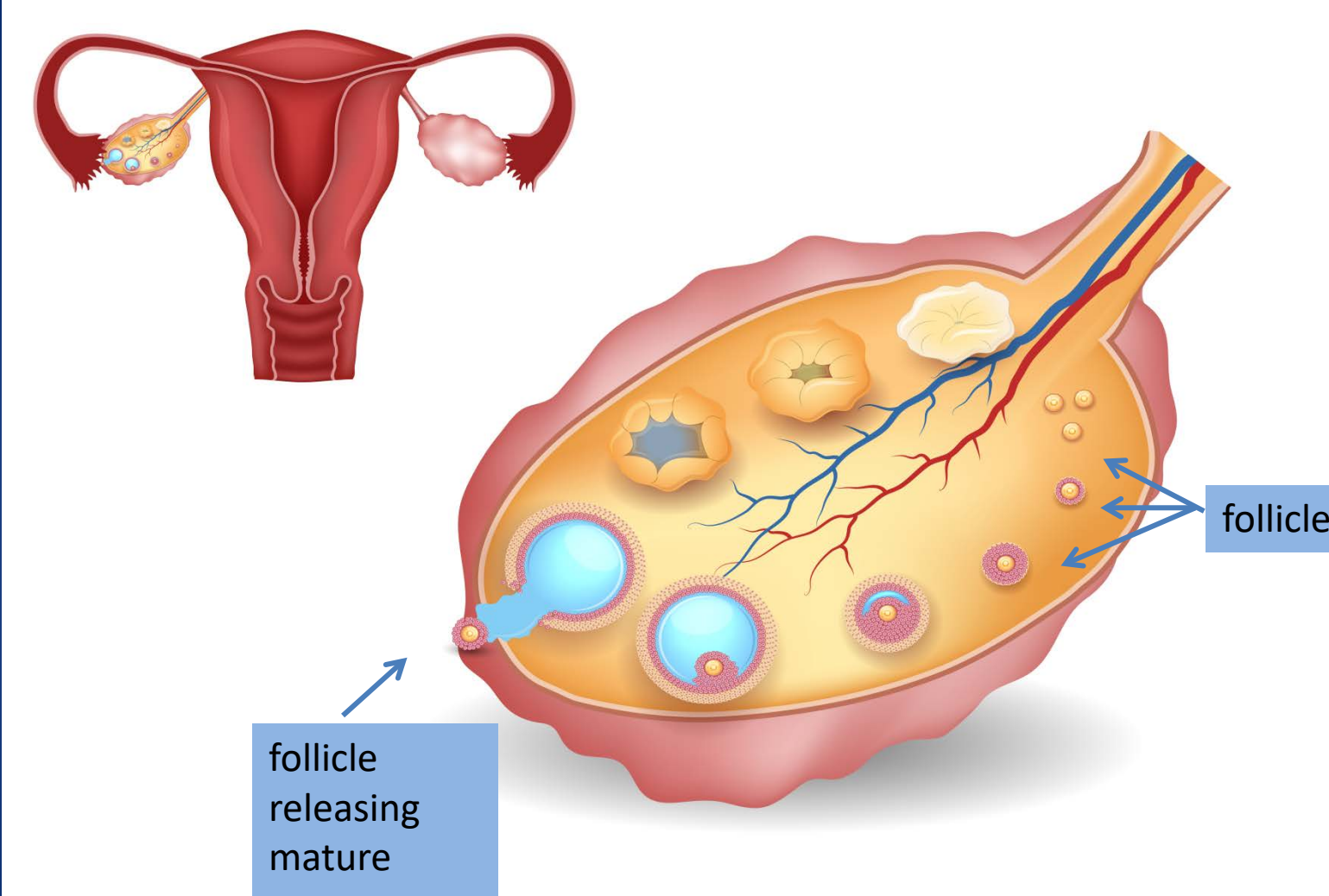


# QUICK REVIEW OF PREGNANCY

# Anatomy of the Female Reproductive System



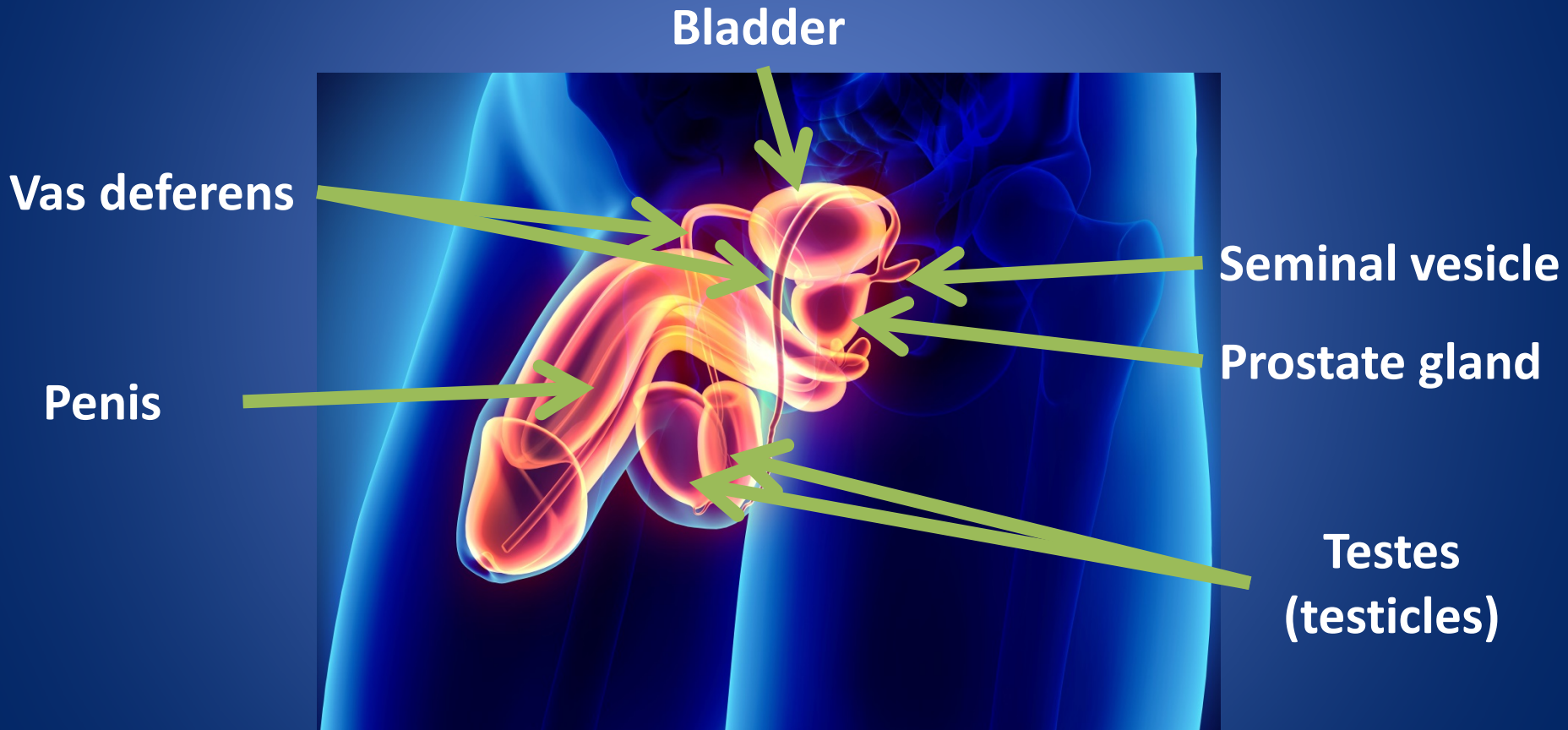




follicle  
releasing  
mature  
ovum

follicles

# Anatomy of the Male Reproductive System



# Fetal development



**Human embryo,  
4 weeks**



**Human fetus,  
16 weeks**

**Human fetus,  
40 weeks**





# Gene-what?





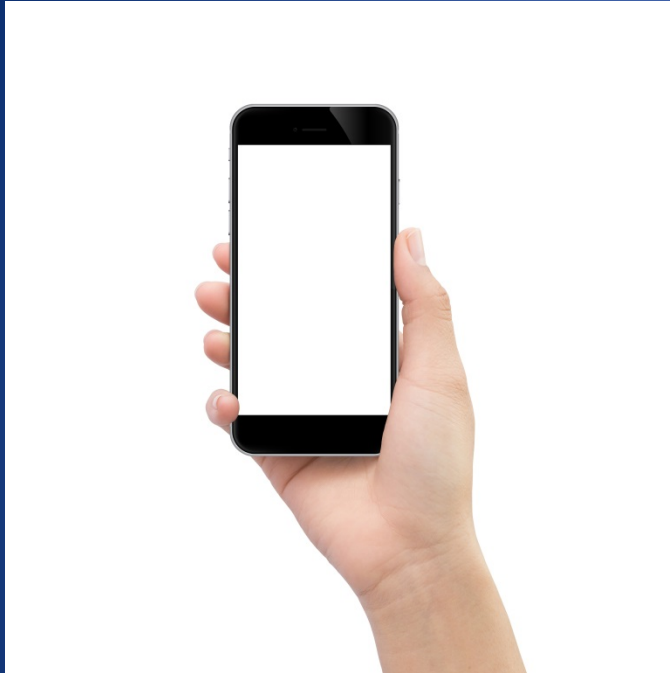
# WHAT IS GENETICS?

# What is “genetics”?

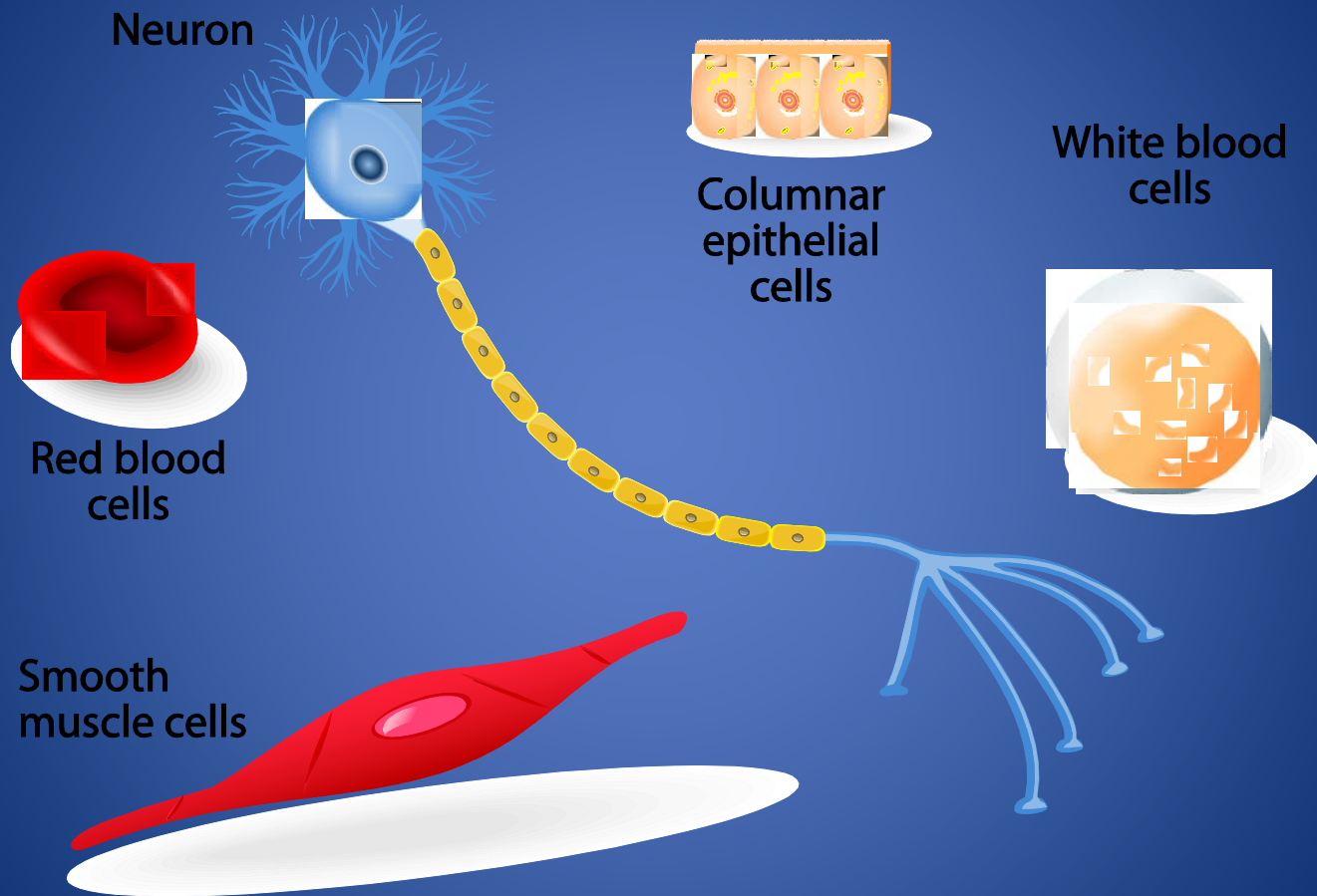
Genetics is the field of science and medicine that studies the biologic basis of heredity (how traits are passed from one generation to another) and how the instructions for life are used by all living organisms.



# Cells

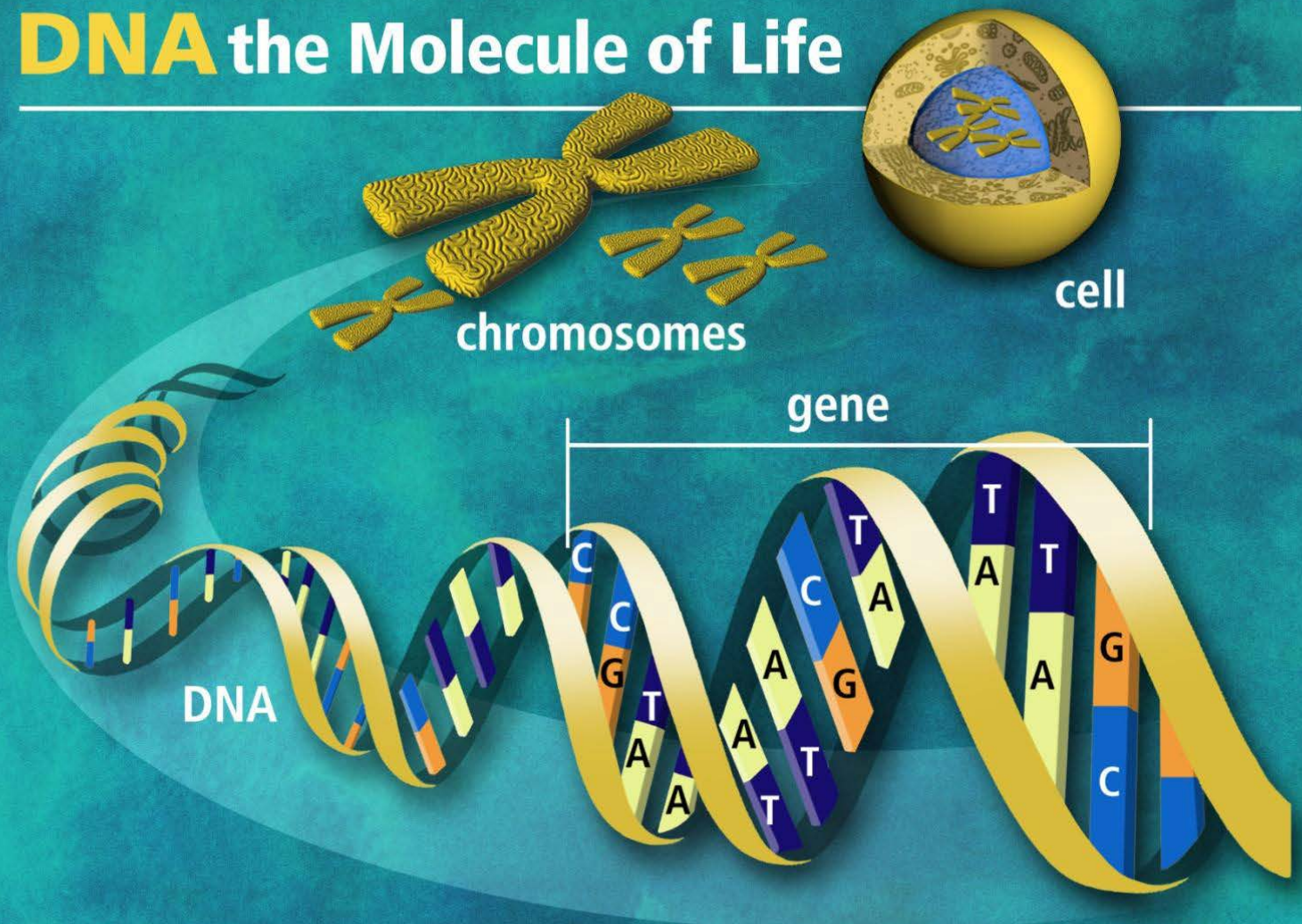


# HUMAN CELLS

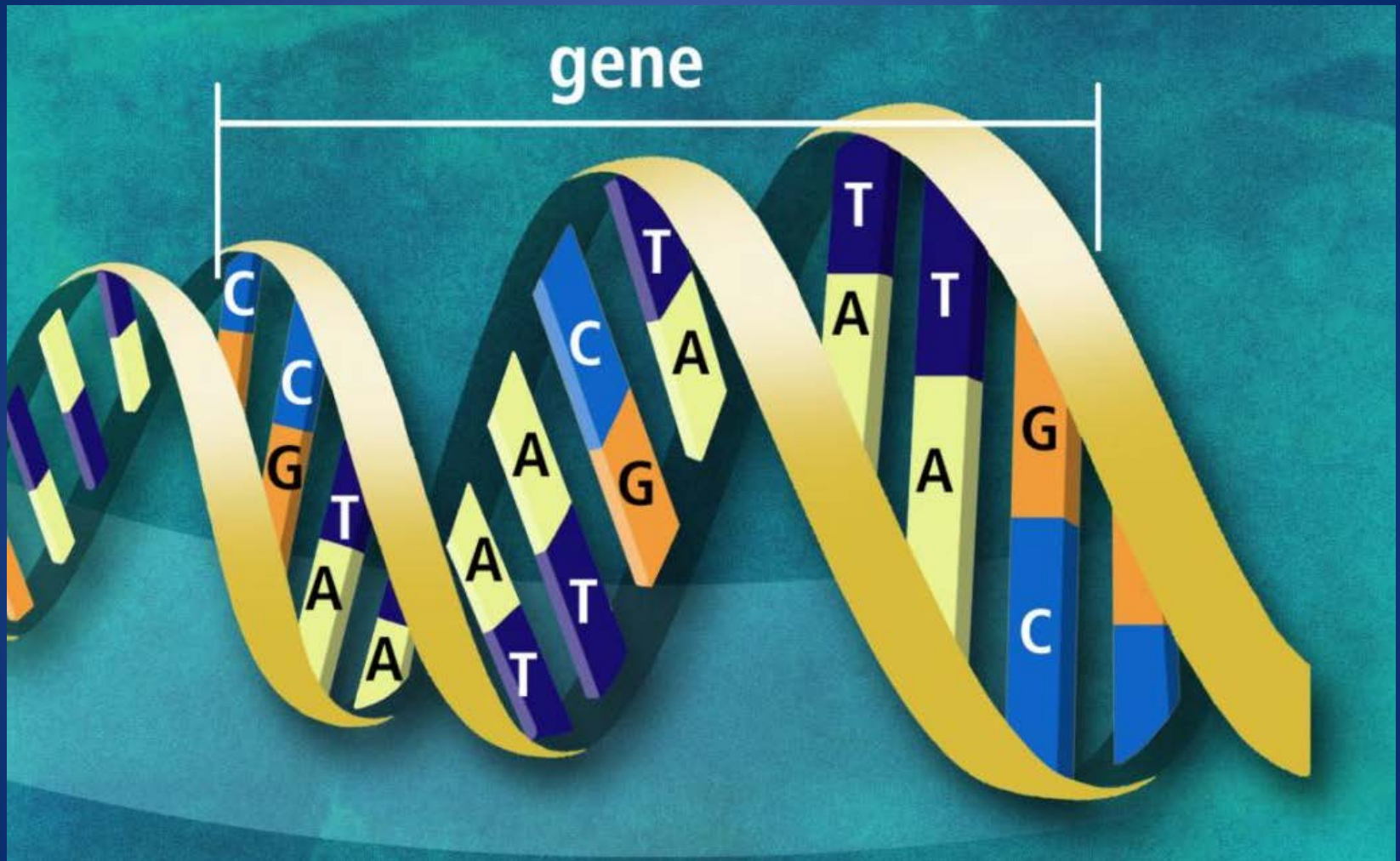




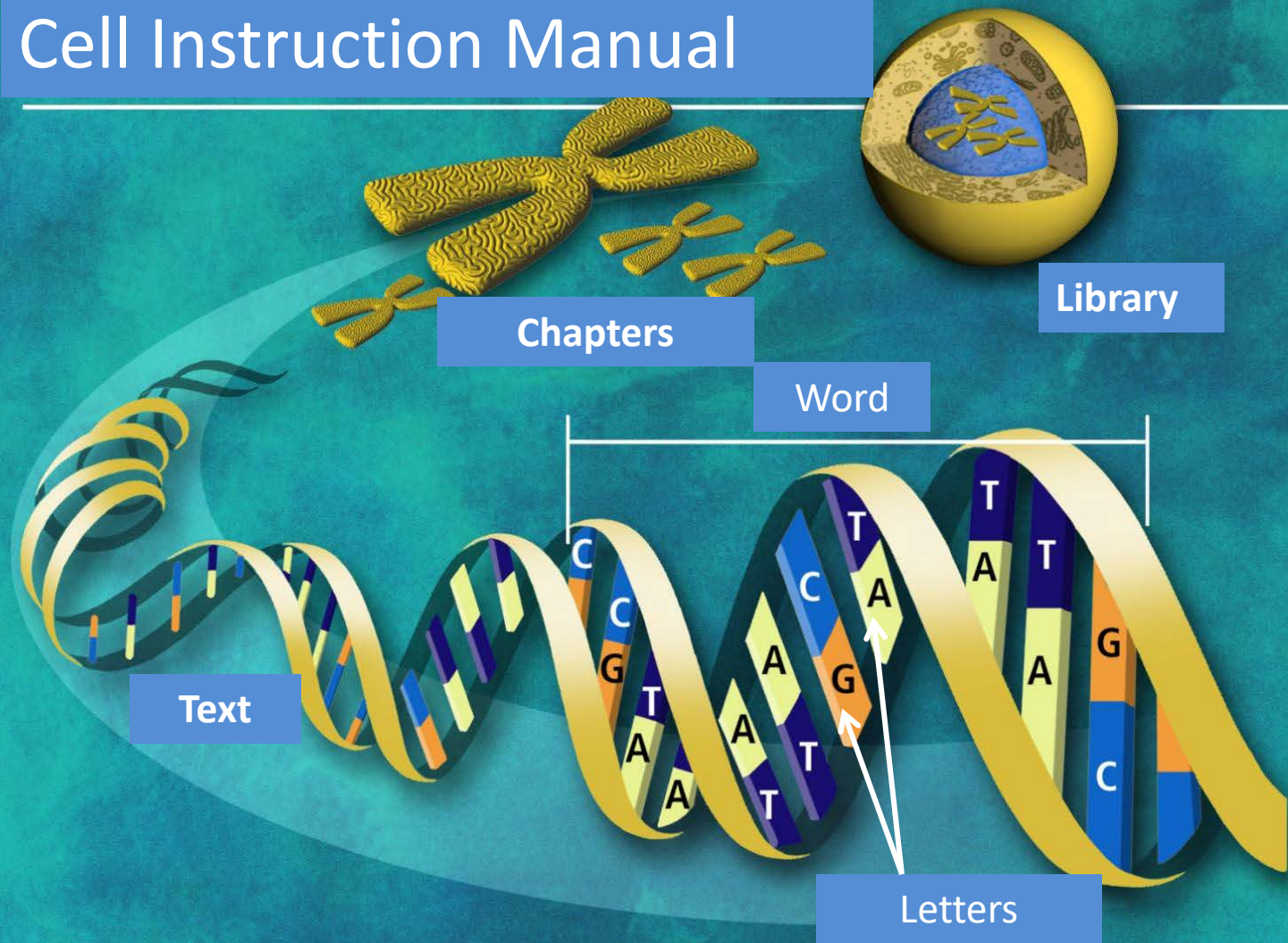
# DNA the Molecule of Life



Y-GG 00-0481

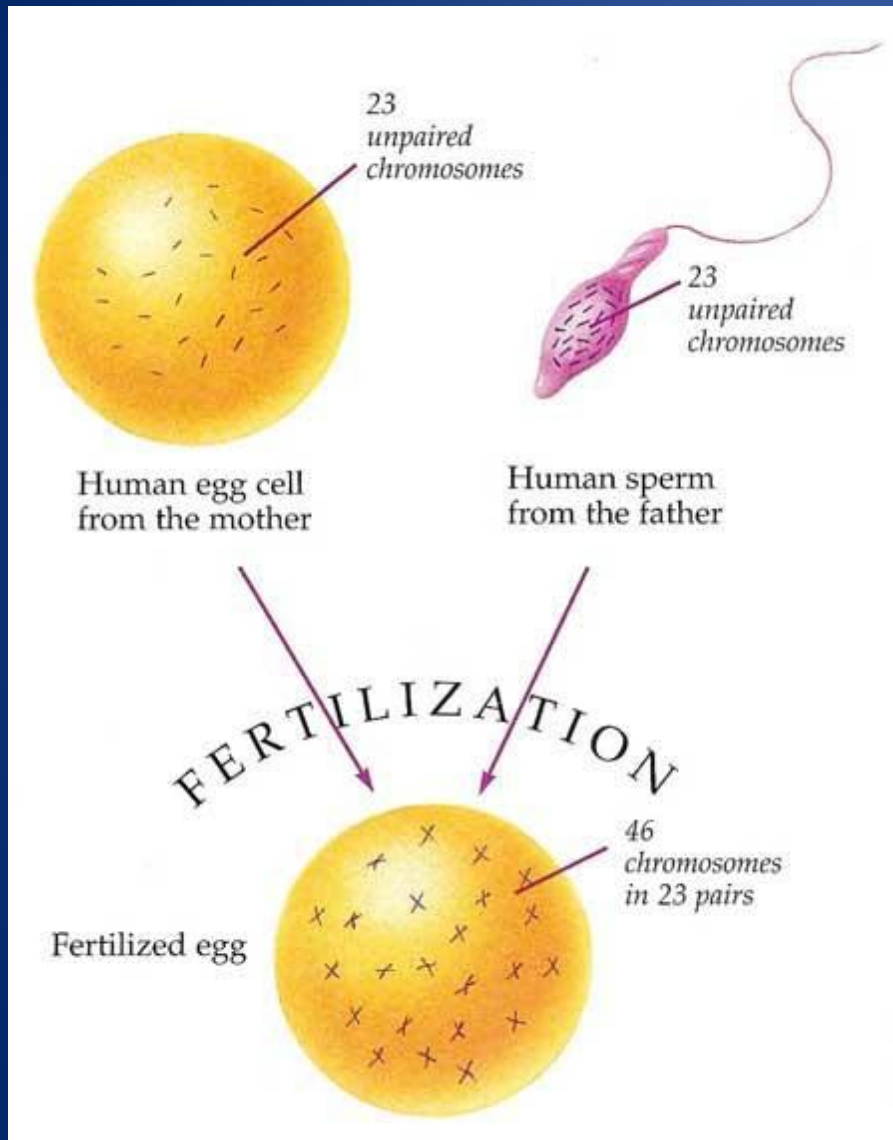


# Cell Instruction Manual



Y-GG 00-0481

# How is genetic information passed on?



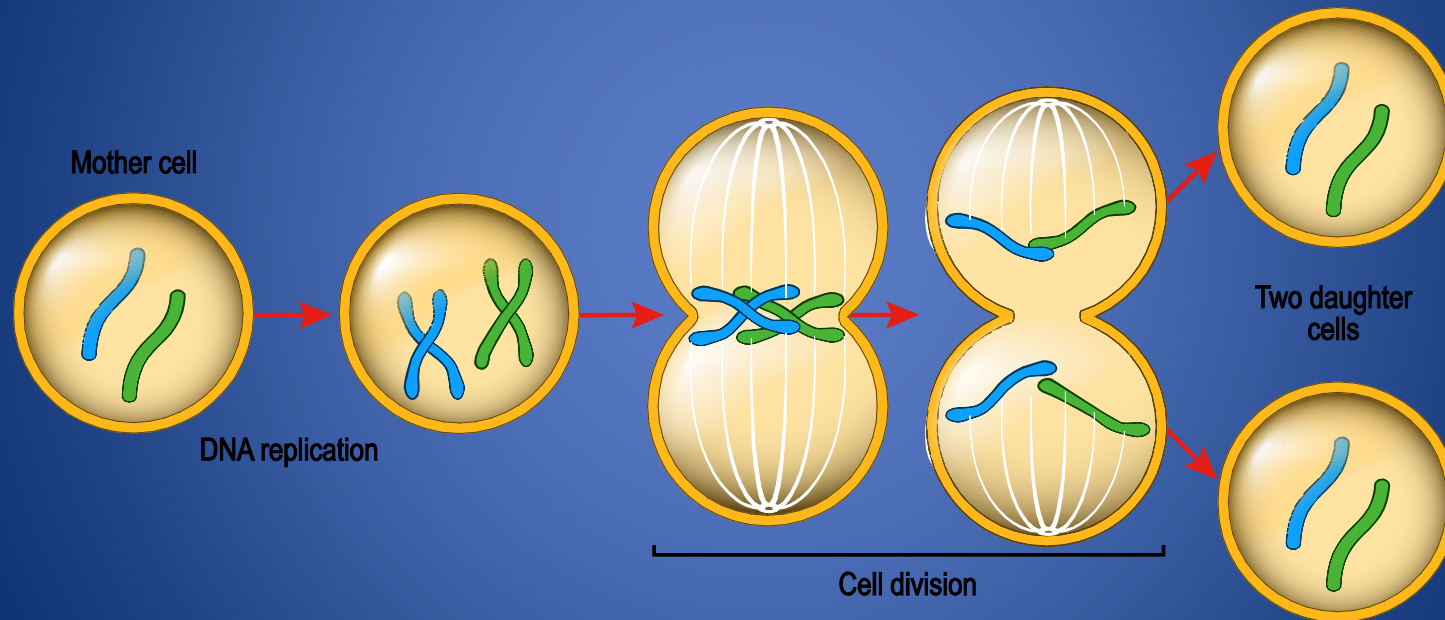
Humans have **23 pairs** of chromosomes in every cell.

The egg and sperm are special; they have **only one** of each chromosome.

When an egg and sperm come together, they typically grow into a child who has **two of each** chromosome: one from Mom, and one from Dad.

# Cell replication after that

## MITOSIS



# Changes in cell replication



# Gene variants

Original Sentence

**TIME TO DREAM**

Single Letter Change

**T<sup>I</sup><sub>A</sub>ME TO DREAM** → **TAME TO DREAM**

Reverse Order

**I<sup>M</sup><sub>T</sub>E TO DREAM** → **EMIT TO DREAM**

Deletion

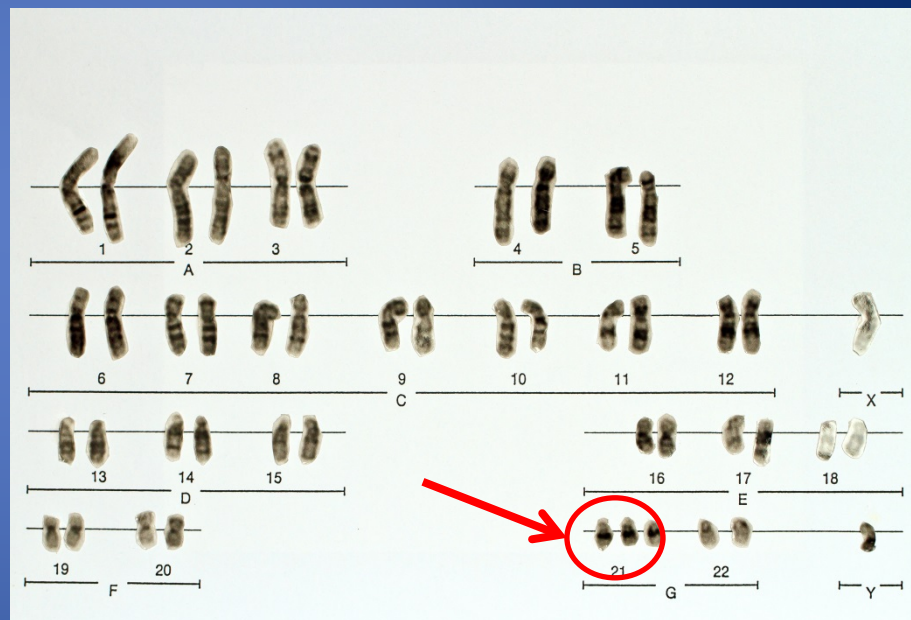
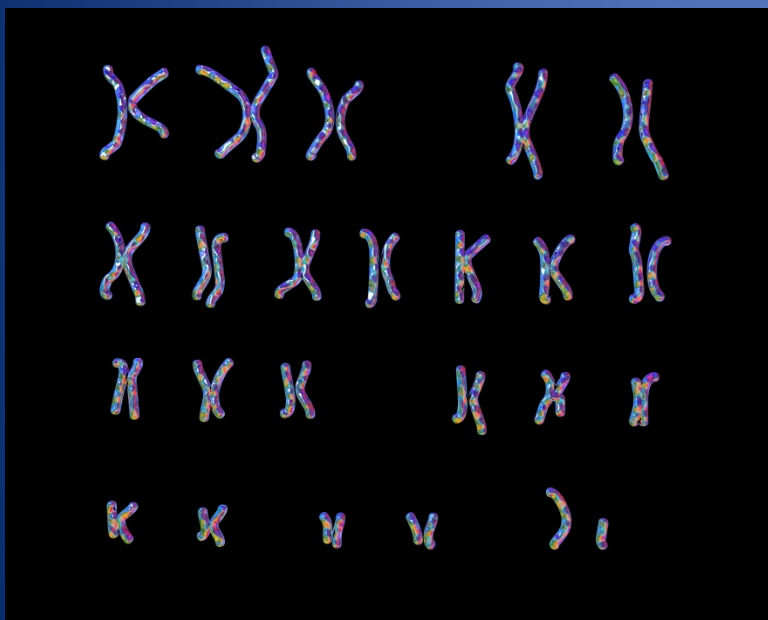
**T<sup>IM</sup>E TO DREAM** → **TETO DR EAM**

Insertion

**TI<sub>I</sub>ME TO DREAM** → **TIIM ET ODREAM**

<http://www.koshland-science-museum.org/exhibitdna/inh03.jsp>

# Chromosome abnormalities





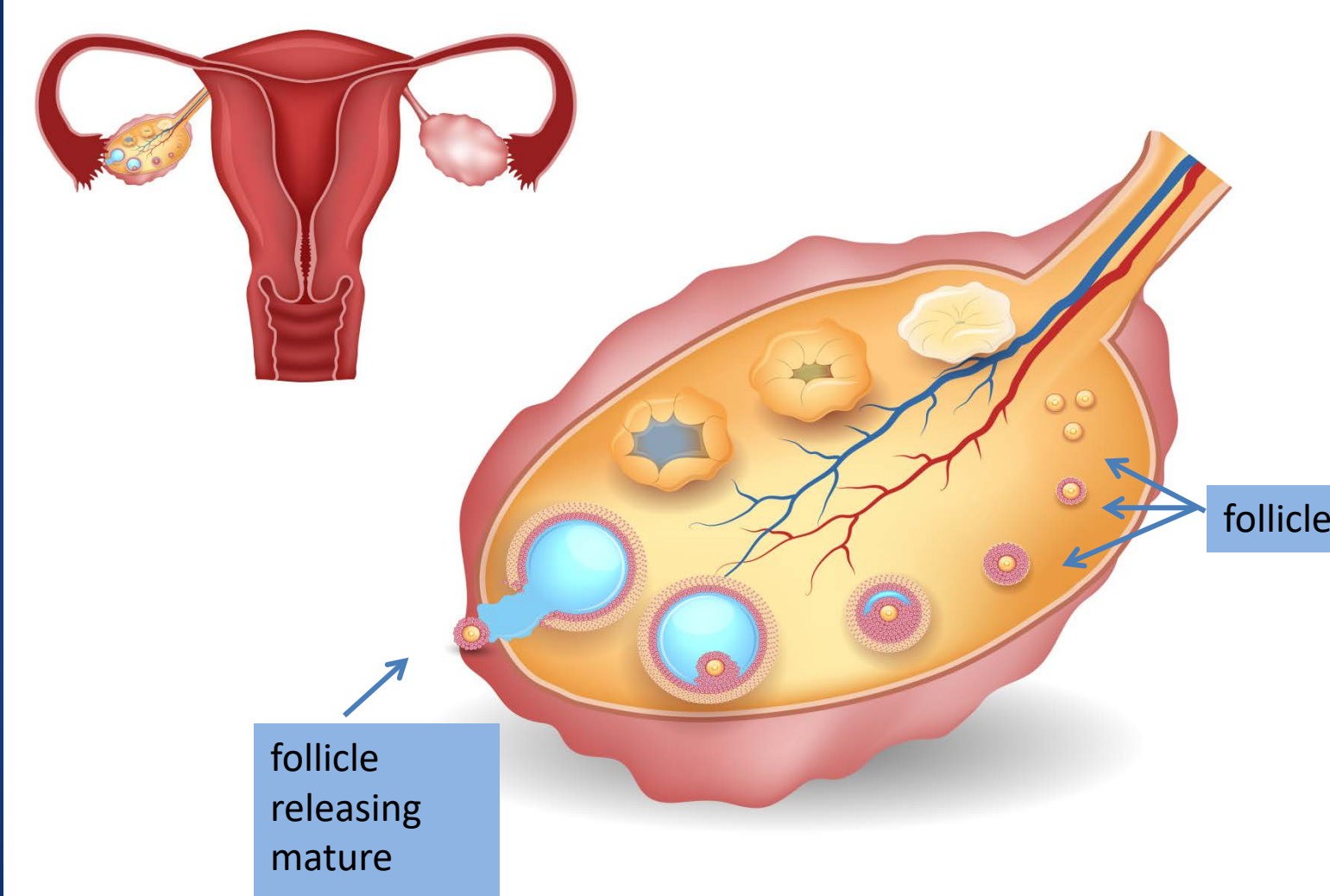


# Types of changes

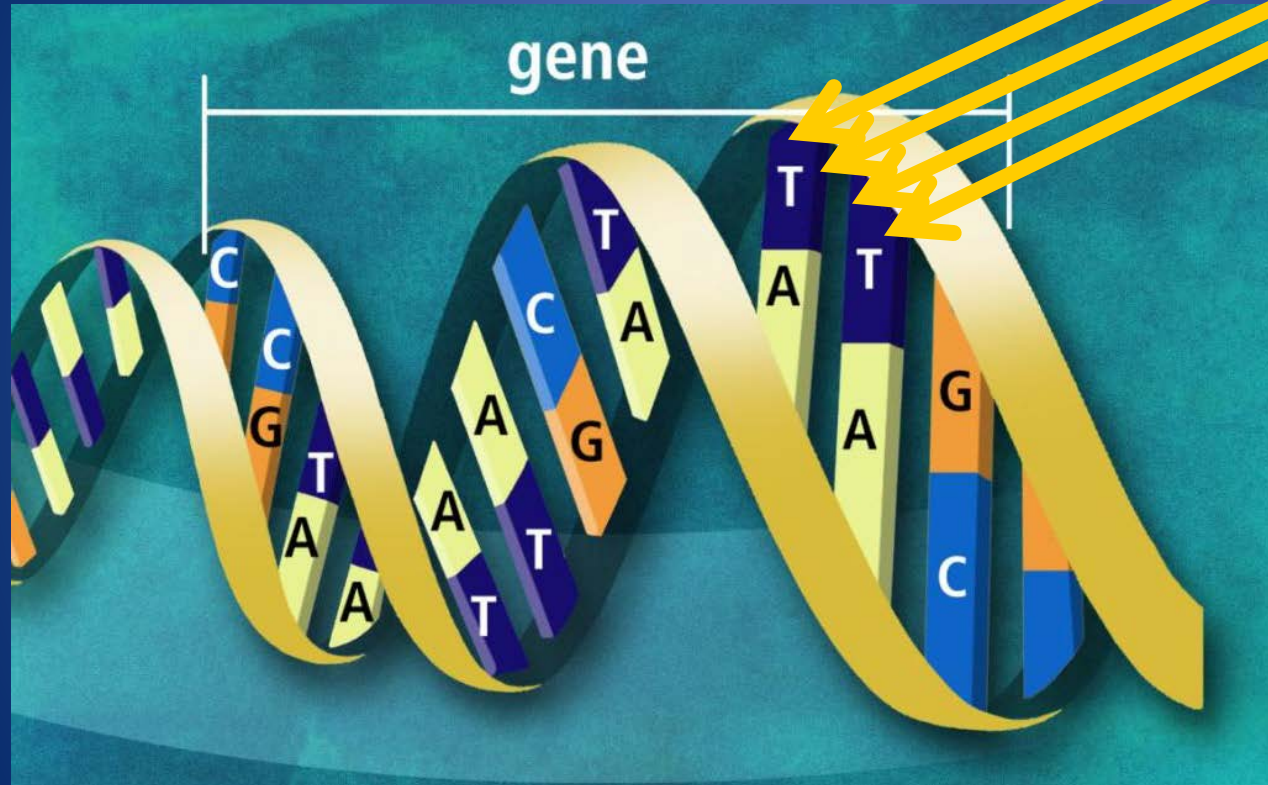
1. Inherited genetic changes  
*(“Your grandpa was just the same.”)*
2. De novo genetic changes  
*(“Now, where did that come from?”)*
3. Somatic genetic changes  
*(“I told you to stay out of the sun!”)*



## Inherited genetic change



# De novo genetic change



## Somatic or acquired genetic change

# So what?

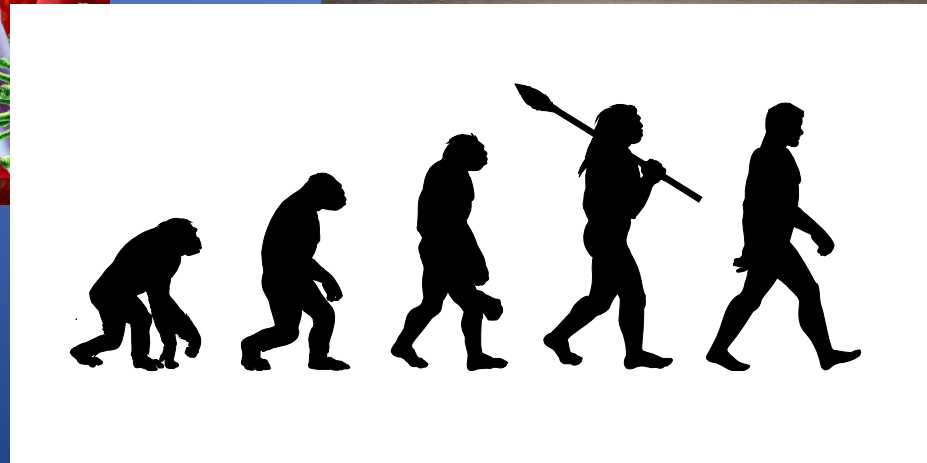
Some changes are  
**BENIGN**  
(they cause no harm).



But some are  
**CLINICALLY SIGNIFICANT**  
or “deleterious”  
or “pathogenic”  
(they cause a problem)



# Can variants or mutations be a good thing?





POLL!





# GENETIC TESTING DURING PREGNANCY





Age

35



# Certain ethnic groups

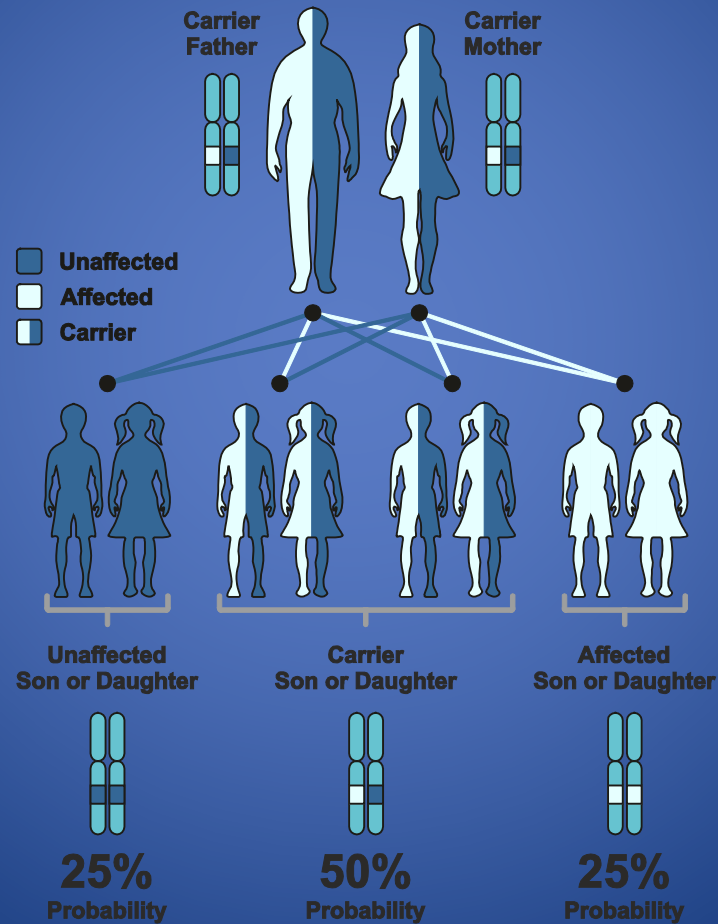


# Reproductive history



# Known carriers

## Autosomal Recessive



# Family history



# Consanguinity



First cousins  
marrying

# Ultrasound findings



# What if there is increased risk?







# Pop Quiz



List 6 reasons  
that a couple may be offered  
prenatal genetic testing.



# THE WORK OF GENETIC COUNSELORS

## PART 1



# What do prenatal genetic counselors do?

- Provide information about the risks of chromosome abnormalities, birth defects or other genetic conditions to individuals, couples or families.
- Help facilitate the decision-making process, allowing patients to make informed choices about prenatal screening and testing options.
- Help coordinate testing and communicate results to the patient.
- Provide families with resources, educational materials, and short-term psychosocial support.

*- National Society of Genetic Counselors*



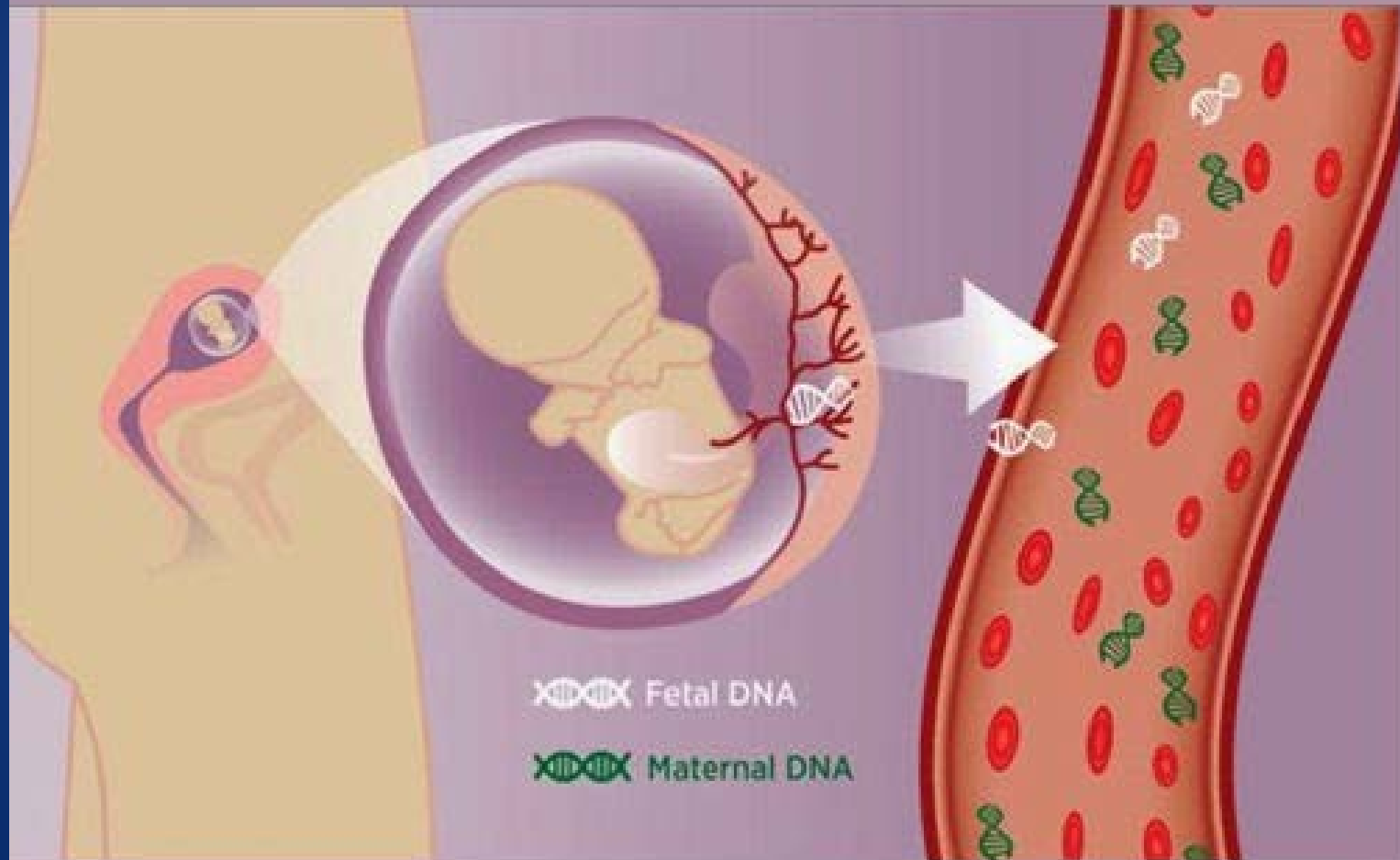
# What do GCs do in a prenatal genetic counseling session?

- Identify goals
- Take a health history
- Take a family history
- Describe genetic tests
- Help the couple decide if they want genetic testing



# SCREENING TESTS

# Non-Invasive Prenatal Testing



# First trimester screen

10<sup>th</sup> - 13<sup>th</sup> week



# Maternal serum screening (AKA quad screen or tetra screen) 15<sup>th</sup> - 20<sup>th</sup> week



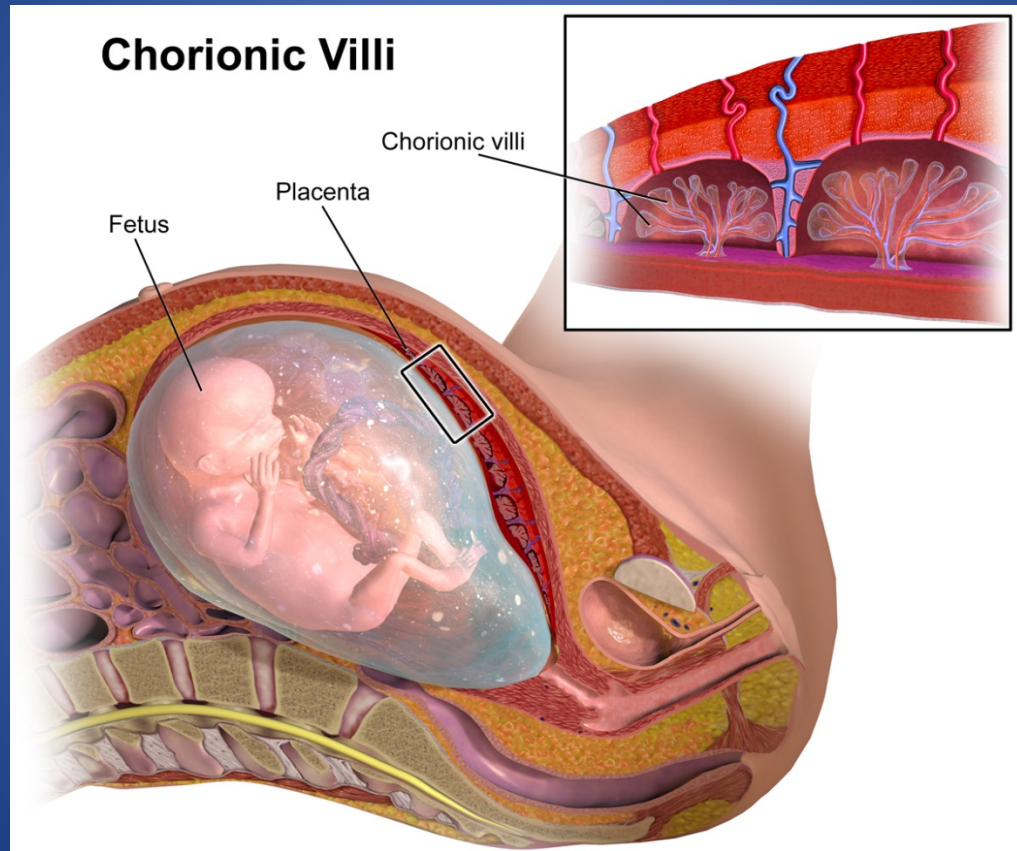




# DIAGNOSTIC TESTS

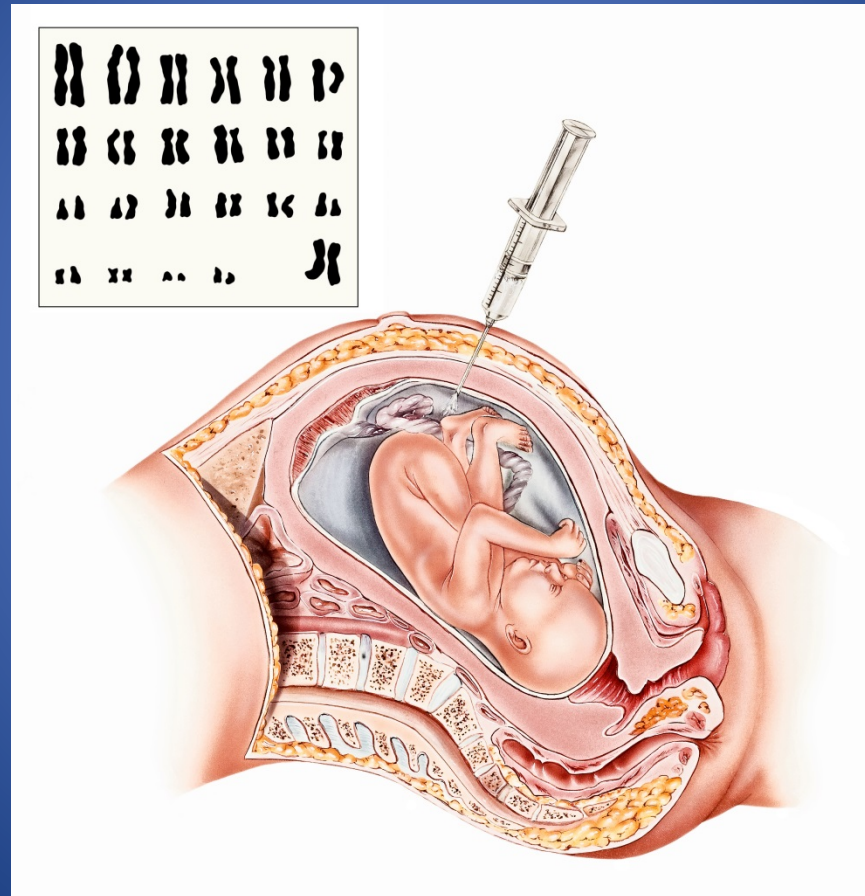
# Chorionic villus sampling

10<sup>th</sup> – 13<sup>th</sup> week



# Amniocentesis

15<sup>th</sup> - 20<sup>th</sup> week





POLL!





# WHAT ARE TESTS LOOKING FOR?

# Chromosomal changes

Karyotype

Insertions  
or  
deletions?

Uniparental disomy?

Too many of any other?

Not enough of one  
chromosome?

Marker chromosomes?

**MOSAICISM?**

Translocations?

# Trisomy 21 (Down syndrome)



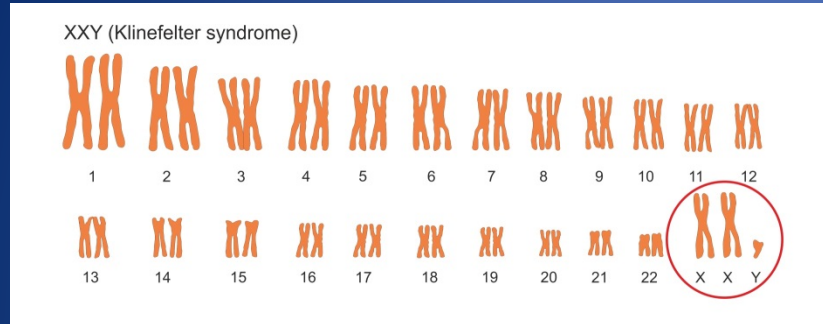
# Trisomy 18 (Edward syndrome)



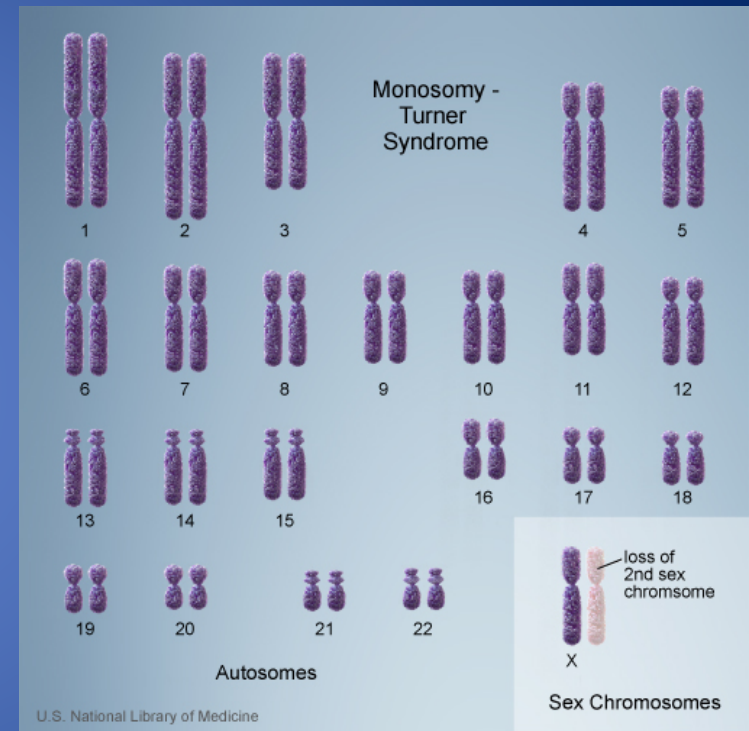
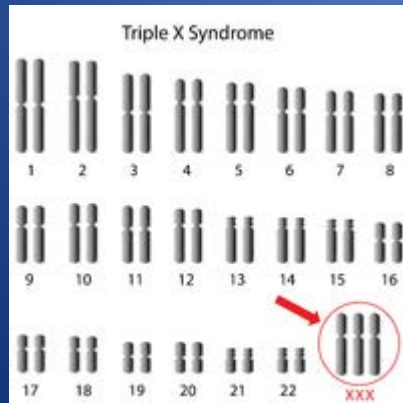


# Sex Chromosome Differences

## Klinefelter syndrome (47, XXY)



## Triple X syndrome (47, XXX)

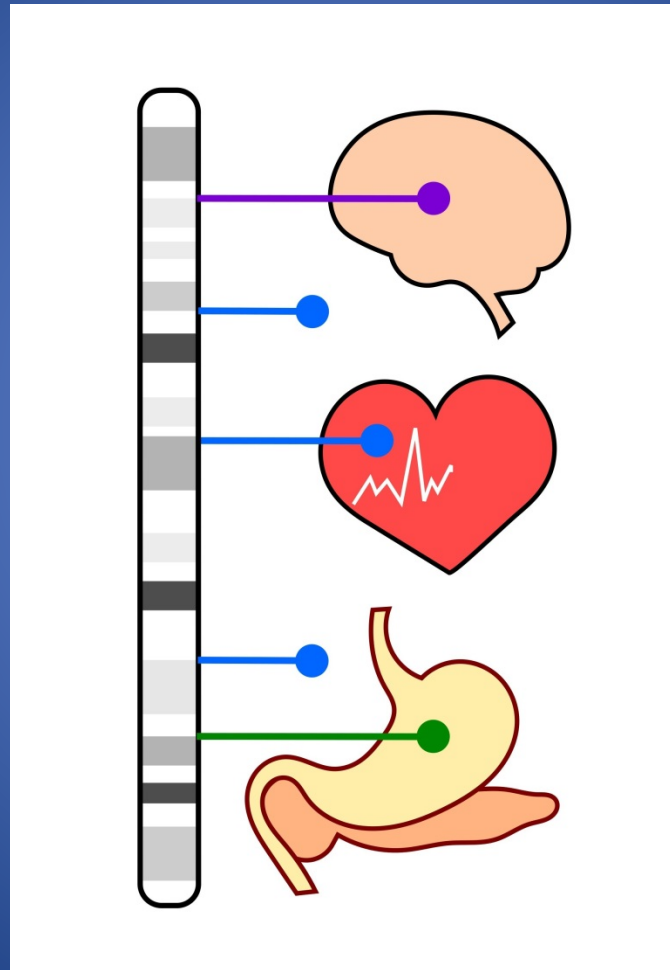


## Turner syndrome

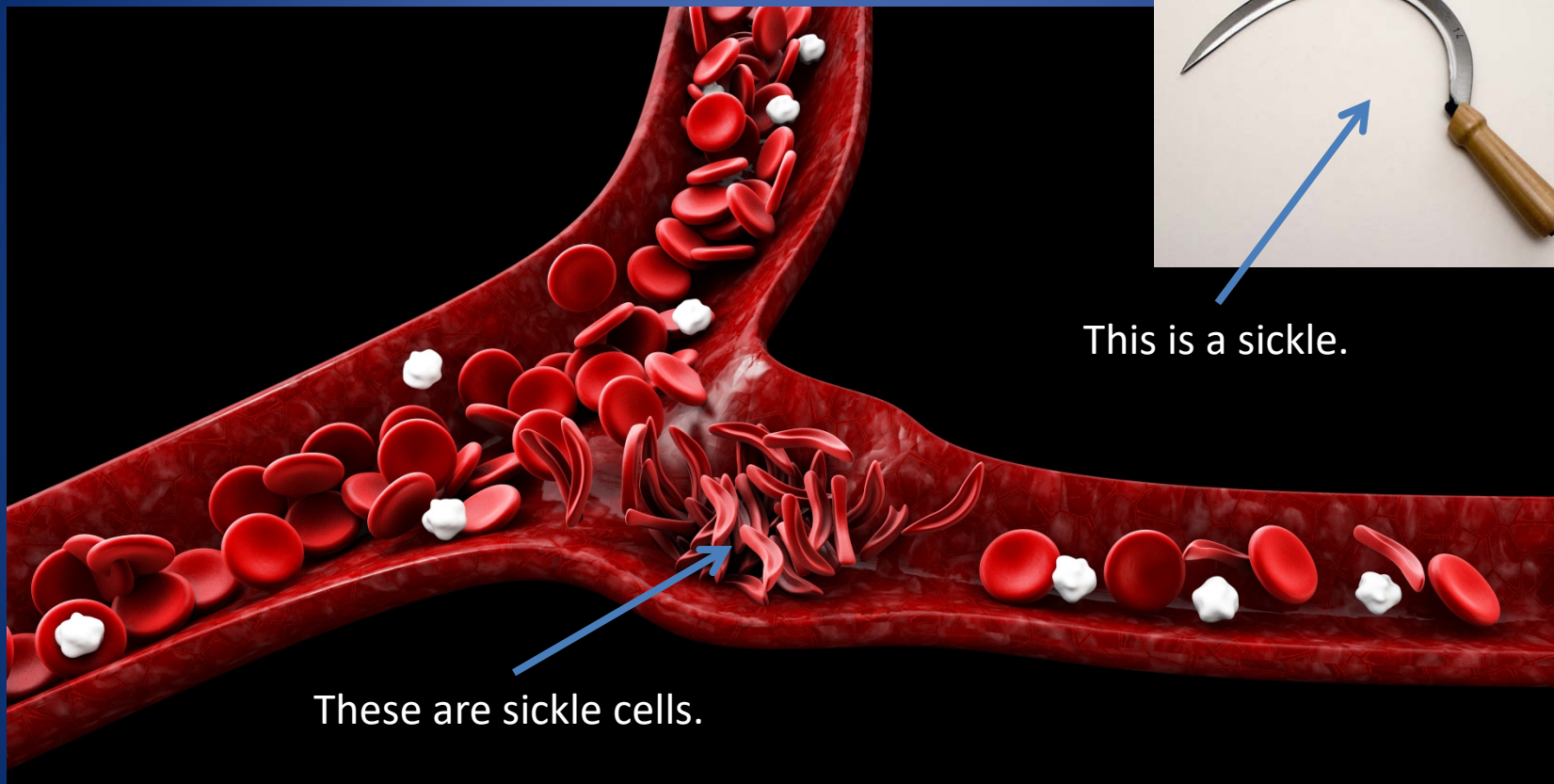
# Gene changes



# Gene changes



# Sickle cell disease



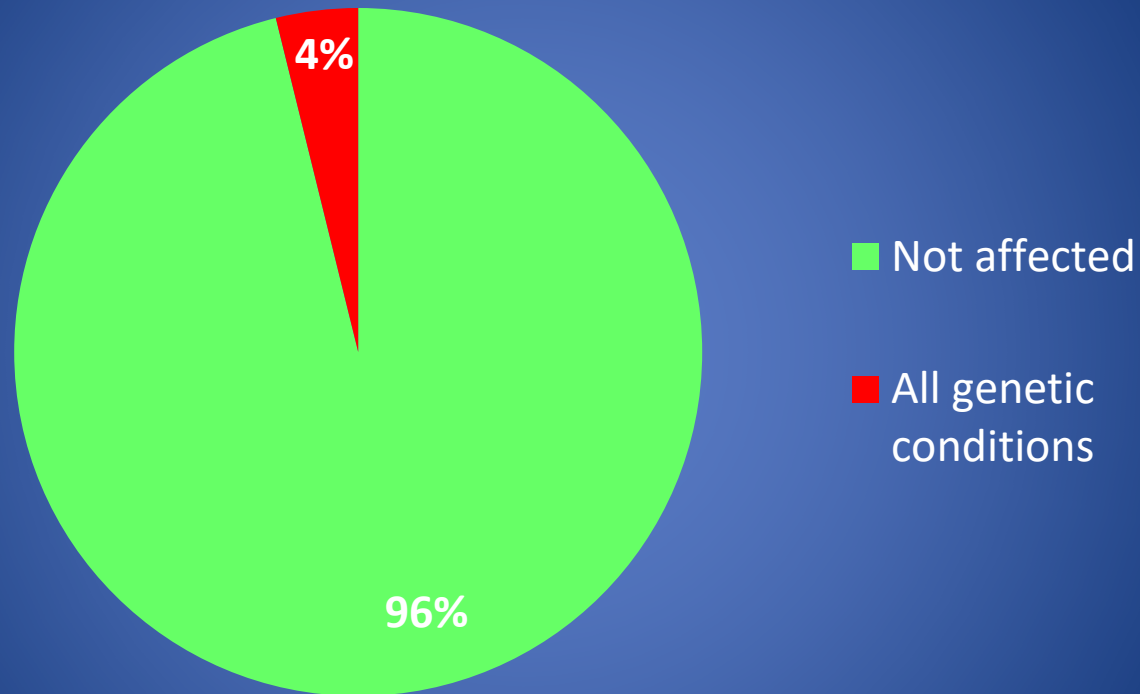
This is a sickle.

These are sickle cells.

# Cystic Fibrosis



# How common are genetic changes?





# Pop Quiz



- List three genetic conditions resulting from chromosomal abnormalities.
- List two genetic conditions resulting from gene variants.



# THE WORK OF GENETIC COUNSELORS

## PART 2



# After testing



# Understanding test results

- **Negative** – no genetic change known to cause disease is found in the genetic material tested.
- **Positive** – a genetic change is found which is known to cause disease: aka pathogenic mutation.
- **Variant of Uncertain Significance (VUS)** – a genetic change was found, but scientists don't know yet whether it causes harm or is benign.

# Screening vs. diagnostic tests

## Screening tests

- Look at hormones, chemicals, or a very small sample of genetic material.
- Results are usually reported as a level of risk. Over a certain level, the result is said to be “screen positive.”
- This only means that the fetus is at higher risk, not that the genetic condition is present for sure.

## Diagnostic tests

- Look at a significant sample of genetic material.
- Results are reported as “positive,” “negative,” “VUS” or “polymorphism.”
- The result is usually definitive.

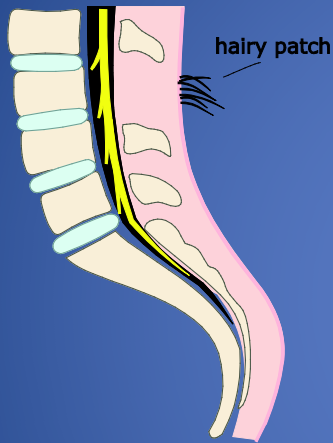
# What to do?



# Fetal surgery

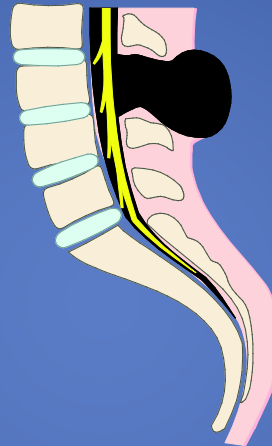
## Spina bifida occulta

(opened posterior vertebral body)



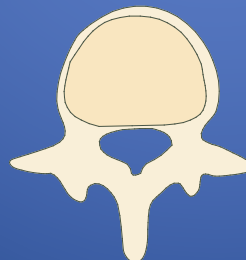
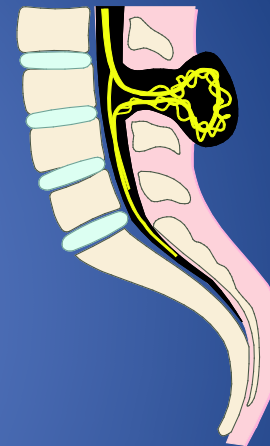
## Meningocele

(protrusion of the meninges)

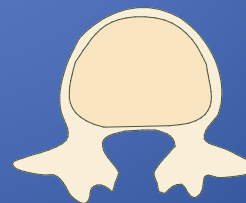


## Myelomeningocele

(protrusion and opened spinal cord)



normal vertebra



not completely closed vertebra

# Termination of Pregnancy



AS OF FEBRUARY 2024

[STATE LAWS AND POLICIES](#)

## An Overview of Abortion Laws

### Background

This resource is being updated and will be available again soon, in the meantime, please reach out to [policyinfo@guttmacher.org](mailto:policyinfo@guttmacher.org) with any inquiries.

To explore the latest abortion policies by state, visit our [interactive map](#).

Visit [our state legislation tracker](#) for policy activity on all sexual and reproductive health topics.

<https://www.guttmacher.org/state-policy/explore/overview-abortion-laws>

# Continuing the pregnancy and preparing for a child with special needs

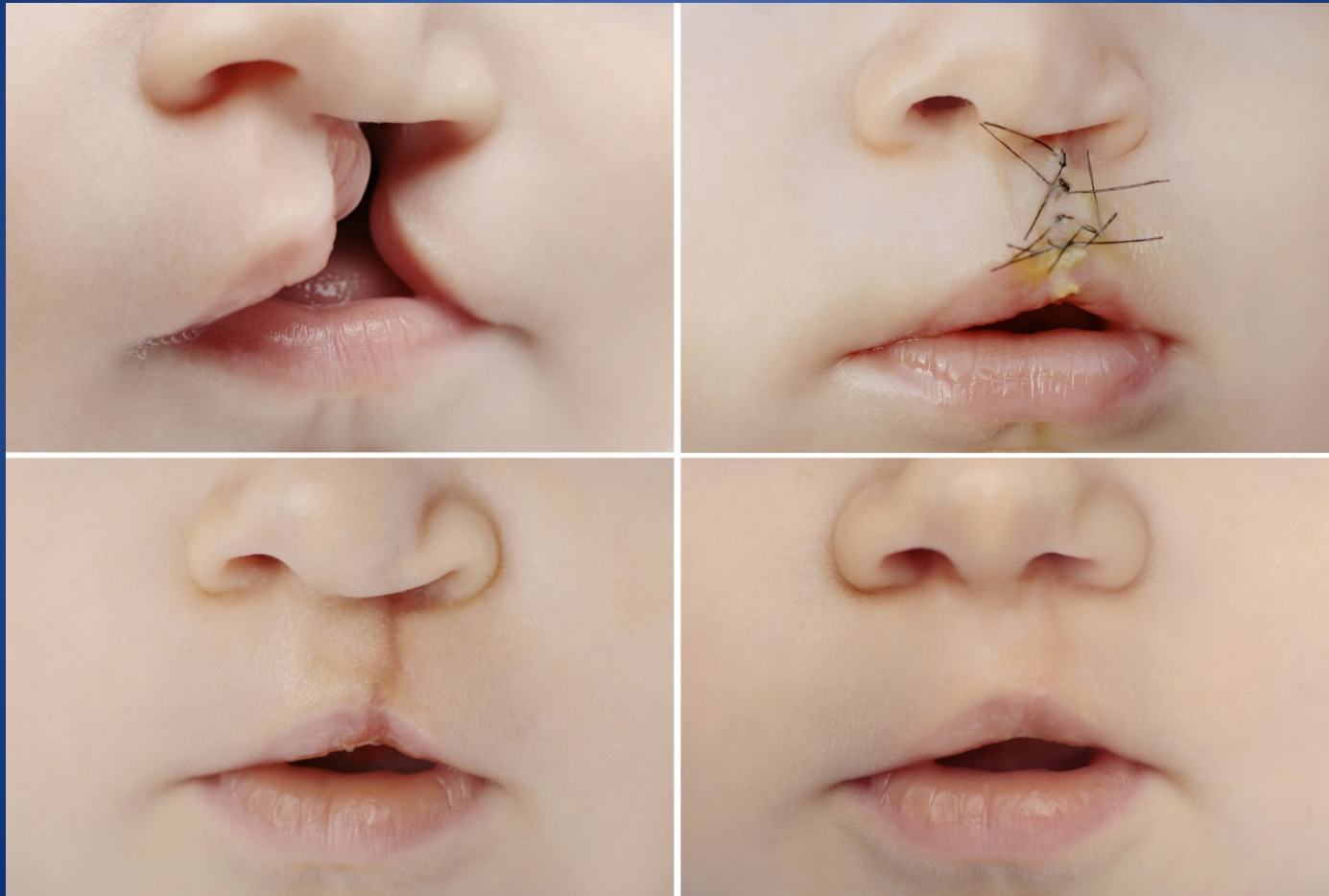


# Adoption of the child





# Postnatal treatment and management





# “What if I want to have more children?”

- Accept the risk of raising a child – or another child – with the same condition.
- Adoption
- Sperm/egg donation or embryo adoption
- In vitro fertilization (IVF) with pre-implantation genetic diagnosis.
- Prenatal testing and diagnosis in future pregnancies.

# Listen and support





# Pop Quiz



- What does it mean to have a negative result on a screening test?
- What does it mean to have a positive result?
- What does it mean to have a negative result on a diagnostic test?
- What does it mean to have a positive result?
- What is a “variant of uncertain significance?”
- List five potential options for couples with a fetus that has a genetic condition. (Some options will not apply to all conditions.)



# CHALLENGES FOR INTERPRETERS

# Family history



*You did NOT  
really  
just ask  
me that!!!!*



# Reproductive history



“Can you just read this form to the patient?”

**NO!**

## CALL Model for Deciding When to Sight Translate

Don't sight translate  
a document that is:

**C**omplex  
**A**dvanced  
**L**egal, or  
**L**ong

How to say no  
politely.

1. Recognize the validity of the requestor's goal.
2. Offer options.
3. Give reasons.

Bancroft, et al. *The Community Interpreter International*, 2015



# Termination of pregnancy





# Summary

1. Genetics is the study of how parents pass traits to their children through DNA, and how DNA controls how we grow and develop.
2. DNA is “packed” into chromosomes – normally, we have 46 chromosomes, divided into 23 pairs.
3. On the chromosomes, various sections called “genes” code for particular traits or functions.
4. If we have other than 23 pairs of chromosomes, or if the order of the base pairs in the genes is nonstandard, we get a chromosomal abnormality or a gene variant, which may lead to health problems.
5. Because these problems are more common in certain groups, we offer pregnant women in those groups the opportunity for testing.
6. Testing can be screening or diagnostic.
7. When tests come back positive, there are hard decisions to be made.

# For more information, go to

- *Genetics Home Reference*, from the National Institutes of Health:  
<https://ghr.nlm.nih.gov/>
- *Learning Genetics*, from Columbia University Medical Center:  
<http://www.learninggenetics.org/index.html>
- *Talking Glossary of Genetic Terms*, National Human Genome Research Institute:  
<https://www.genome.gov/glossary/index.cfm>  
Spanish version: <http://www.genome.gov/GlossaryS/>
- *Prenatal Genetic Testing Videos*, Washington State Department of Health, in partnership with Genetic Support Foundation:  
<https://geneticsupportfoundation.org/archive/genetics-and-you/pregnancy-and-genetics/prenatal-genetic-testing-videos>
- National Society of Genetic Counselors, to find a genetic counselor close to you:  
<http://www.nsgc.org/>
- Genetics Support Foundation, 7 videos in English and Spanish:  
<https://www.geneticsupport.org/>
- Lexigene, English-French-Spanish lexicon: <https://www.lexigene.com/en/>,