



An Introduction to Pediatric Genetics for Healthcare Interpreters

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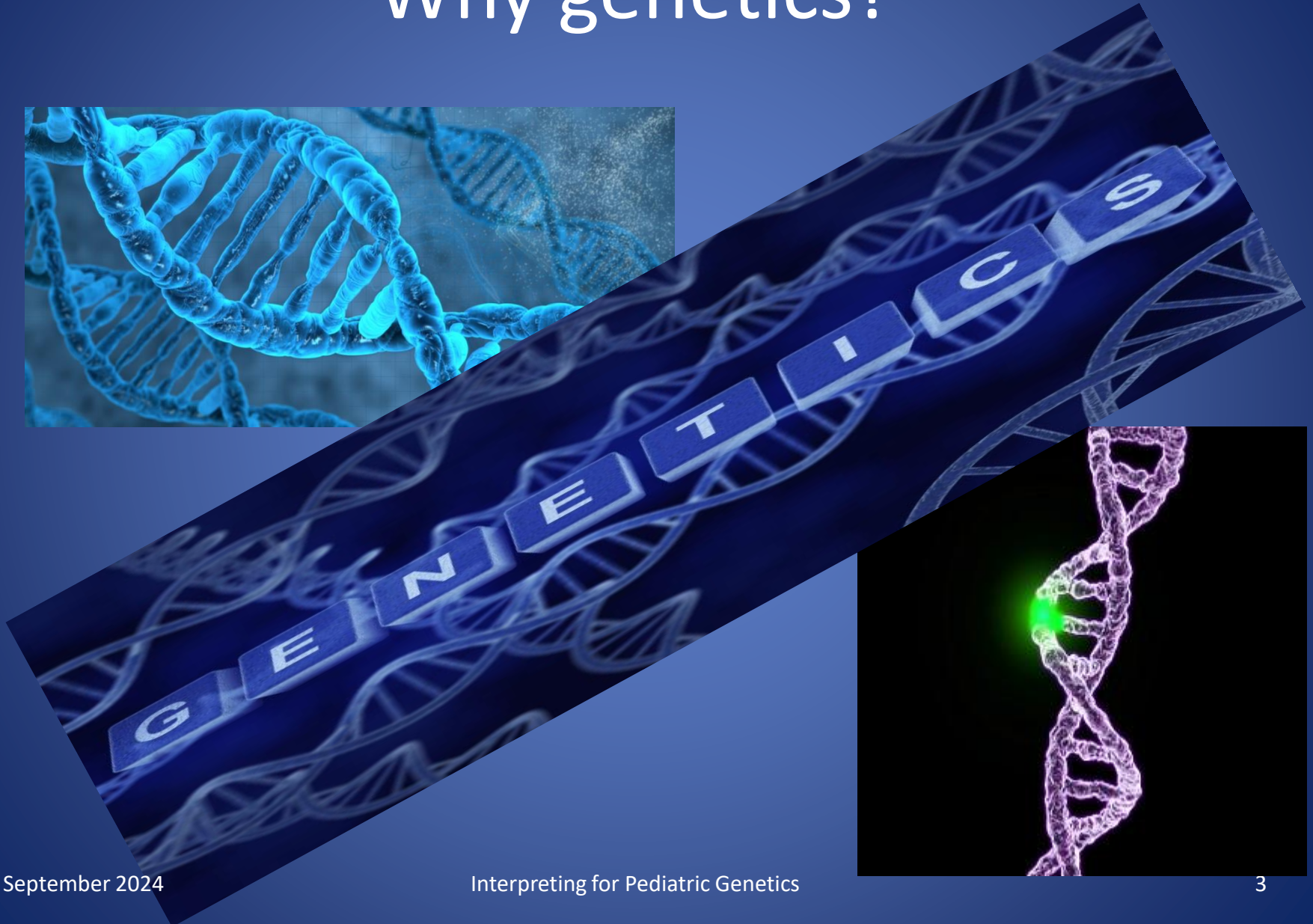
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Developed 03-2019, revised 08-2024

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.
 - Submit both the pre- and the post-test.

Why genetics?



What will we be learning today?

- Basic information about genetics
- The pediatric genetics team
- The pediatric genetics visit
- Genetic testing
- Challenges for interpreters

What will we be doing next week?

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

So, what do you know already?

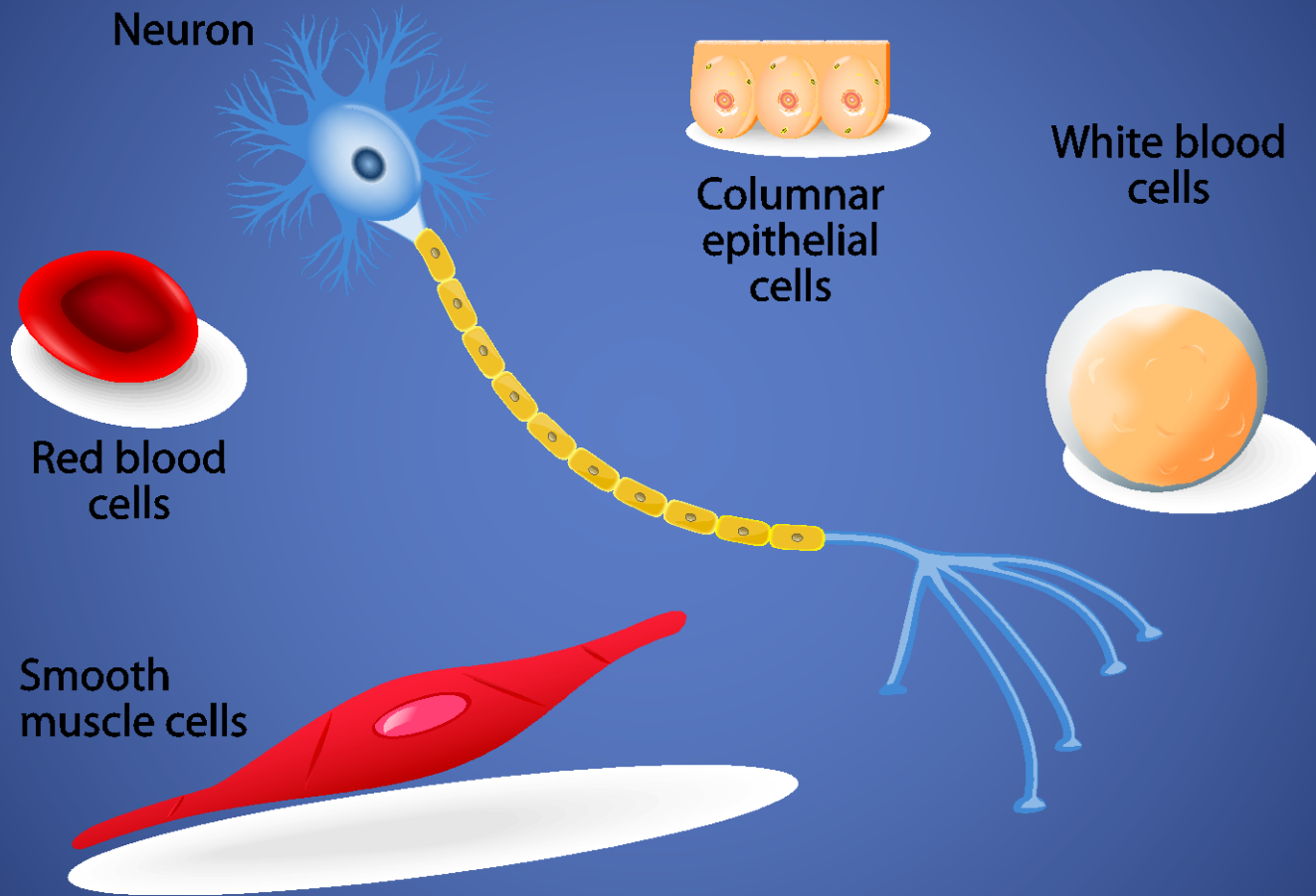
BASIC CONCEPTS IN GENETICS

What is “genetics”?

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



HUMAN CELLS

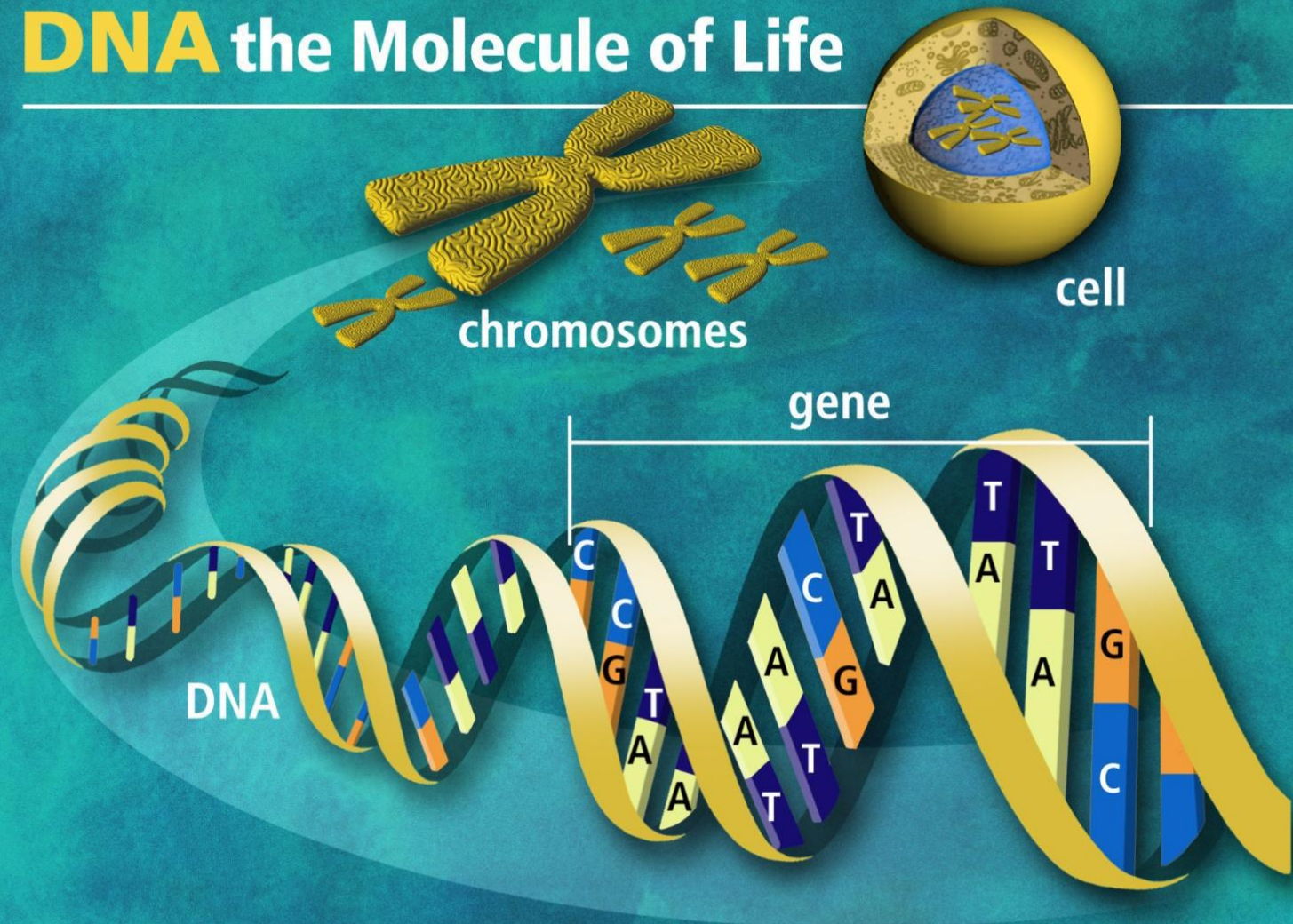


Oh Wise One!
I know I am a cell.
But what is my
True Purpose in
Life?

You must seek deep
inside yourself. Into your
nucleus, into your very
DNA!
Only then will you know
your True Purpose.

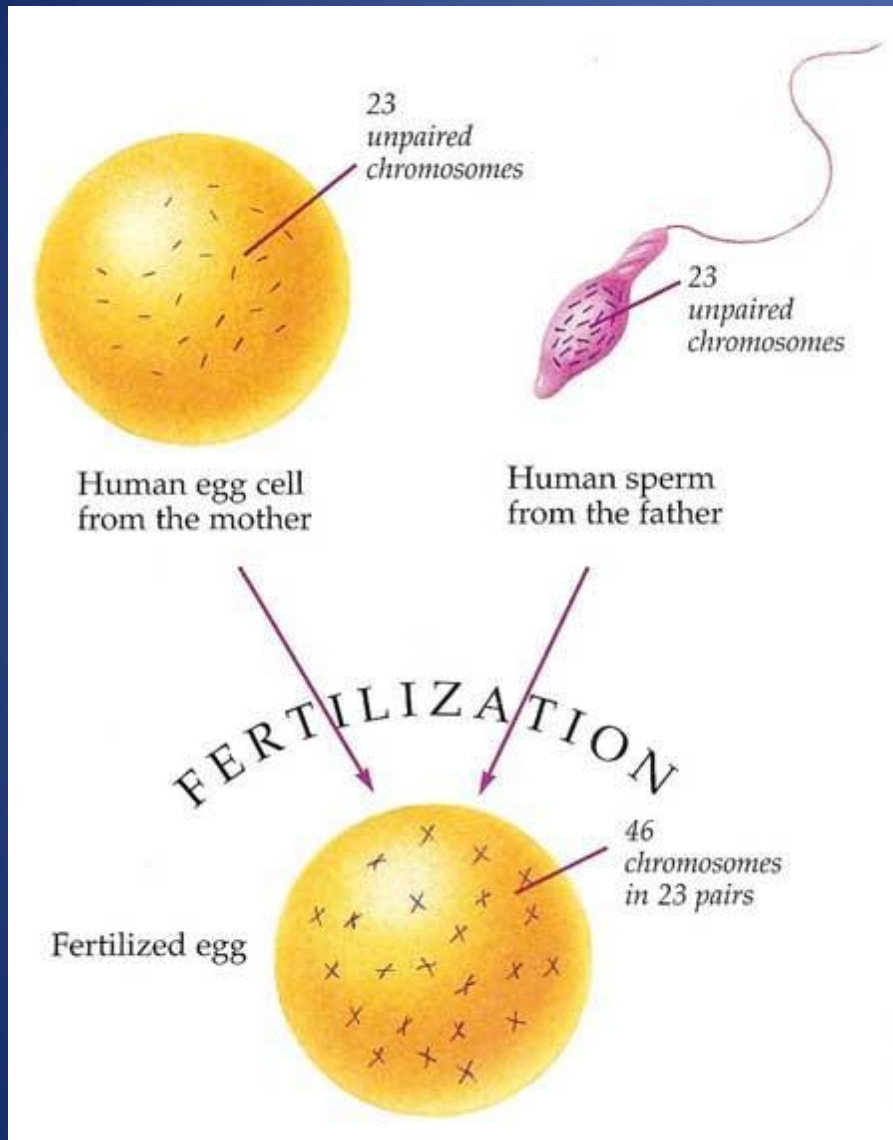


DNA the Molecule of Life



Y-GG 00-0481

How is genetic information passed on?



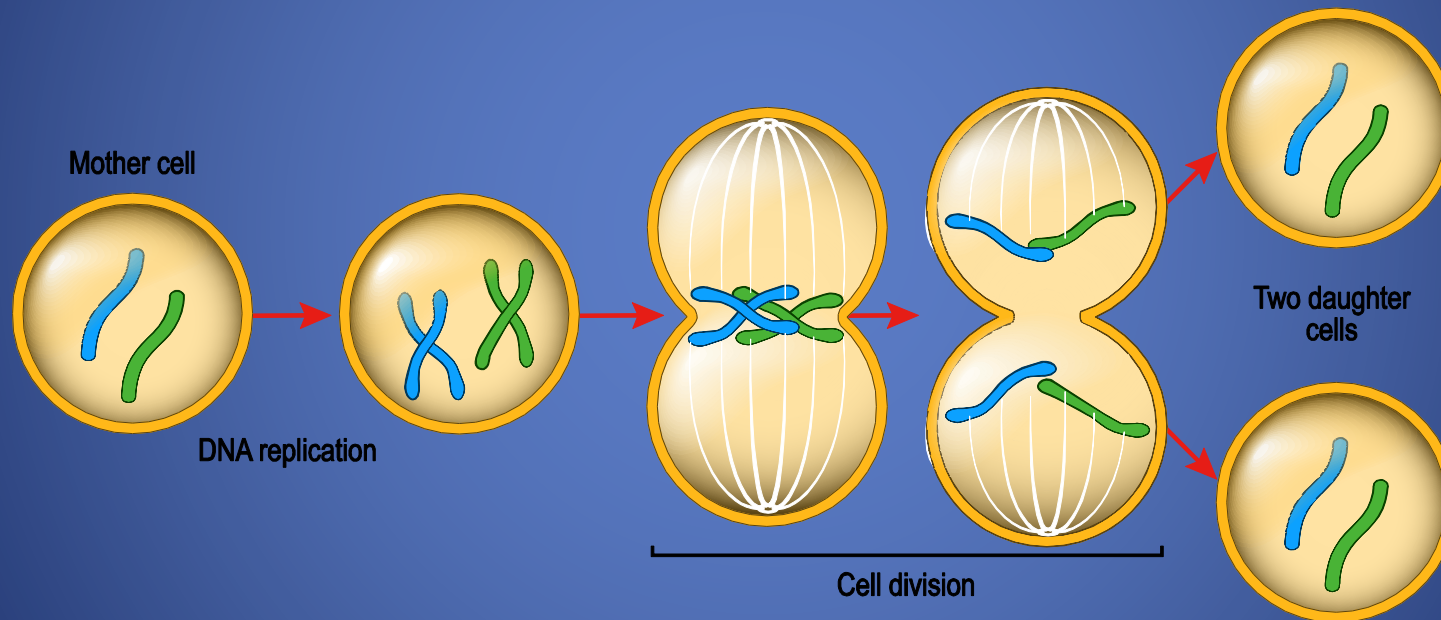
Humans have **23 pairs** of chromosomes in every cell.

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

MOVE TO THE LEFT.

Gene Change

MO^V_LE TO THE LEFT. → MOLE TO THE LEFT.

Gene Reversal

MOVE TO THE **LEFT**. → MOVE TO THE ELFT.

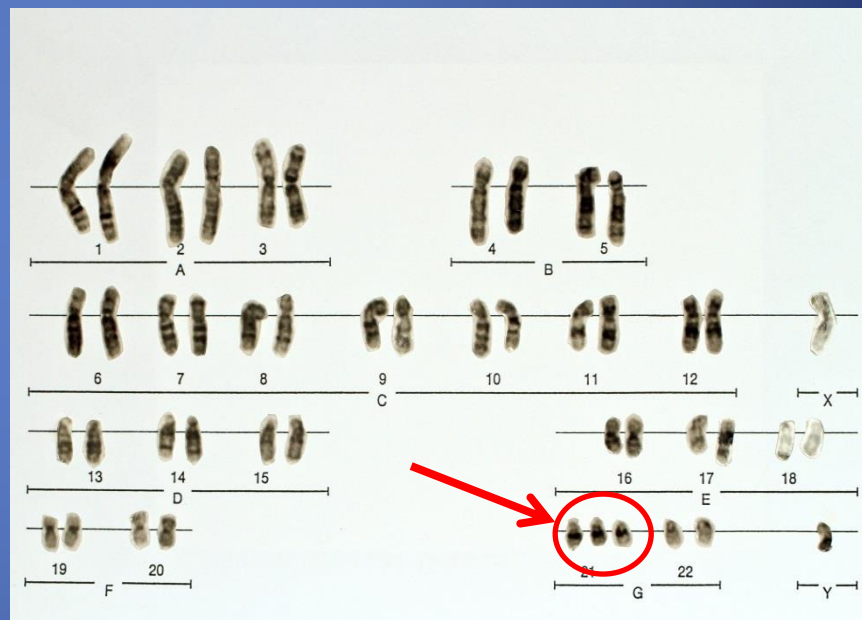
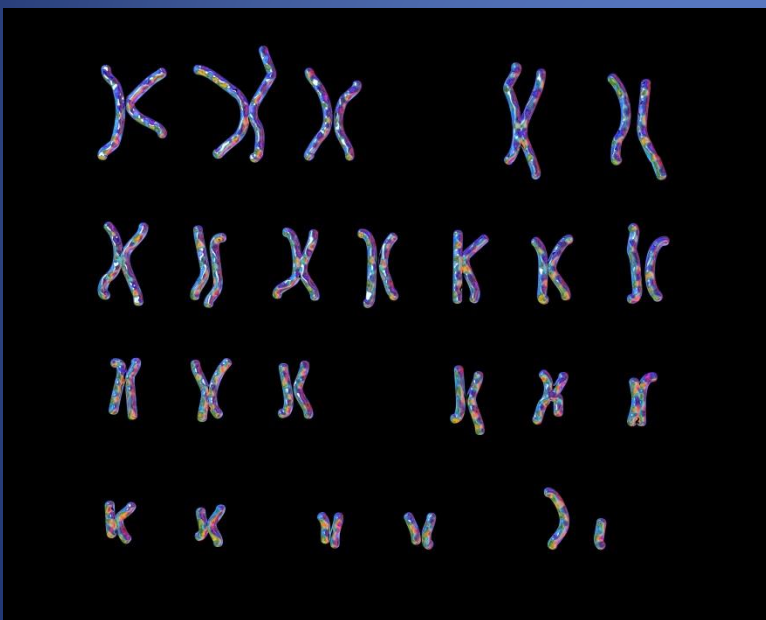
Gene Insertion

MOV_IE TO THE LEFT. → MOVIE TO THE LEFT.

Gene Deletion

MOVE TO THE **LE**FT. → MOVE TO THEFT.

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!”)

So what?

Some changes are
BENIGN
(they cause no harm).



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





POLL

PEDIATRIC GENETICS

Prenatal

GI

Cardiology

Cancer



Cancer genetics



Prenatal genetics



Pediatric genetics



The Team



Patients followed long term



Reason for referrals

*I wonder if it
could have a
genetic
component . . .*



Symptoms leading to referral

- Developmental delay
- Autism Spectrum Disorder
- Dysmorphology
- Failure to thrive
- Family member or parent has a genetic condition



Pop Quiz



- What are three major areas of genetic specialty?
- Who would you expect to see on a genetics consult team?
- Why would a primary care provider or specialist refer a patient for a genetic consult?

THE PEDIATRIC GENETIC VISIT

First visit

History of medical care

- Why was the child referred?
- When were the symptom(s) first noticed?
- What tests have already been done?
- What were the results?
- How have the symptoms progressed?
- What therapies is the child already receiving?

Family history



Physical exam

- Height
- Weight
- Head circumference
- Space between nipples
- Chest circumference
- Arm span
- Eye spacing
- Leg length
- Hand or finger length



Dysmorphology



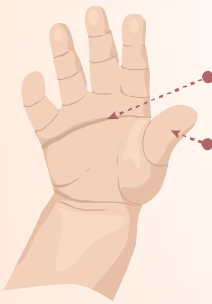
Body measurements that fall outside the normal range or that are associated with specific genetic conditions.



 **Abnormal ears**

 **Flattened nose and face**


 **Short and broad hands**



• **Simian crease**

• **Short fifth finger**

 **Intestinal blockage**

• **Small head at birth** 

• **White spots on the colored part of the eye** 

• **Congenital heart disease** 

• **Widely separated 1st and 2nd toes**

• **Increased skin creases**

• **Big toes widely spaced** 

Symptoms of Down Syndrome



Developmental history



1. At what age did your child first roll over?
2. At what age did your child take his first step?
3. When did he start saying words?
4. When did he start saying phrases?

Actual age
vs.
Adjusted age

Developmental assessment

- Communication
- Social interaction
- Motor abilities like grasping, standing and walking,
- Sensory abilities like hearing and sight
- Adaptive skills
- Unusual behaviors
- Atypical sleep patterns
- School performance (for older children)

What's next?

1.

We think we know what it is.

2.

We don't think this problem is caused by a genetic change

3.

We think there's a genetic link, but we don't know what genes are involved.



Refer to specialists.

Offer genetic testing.



POLL

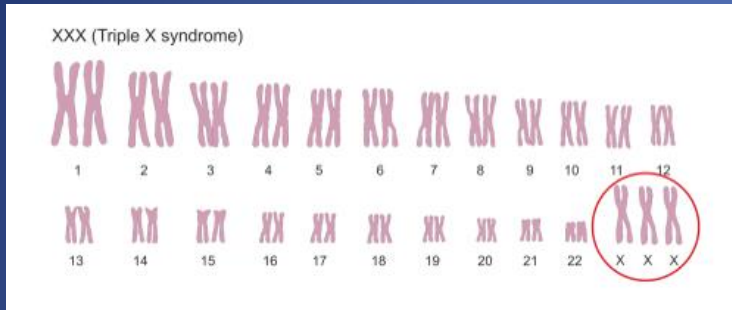
GENETIC TESTING

What is Genetic Testing?

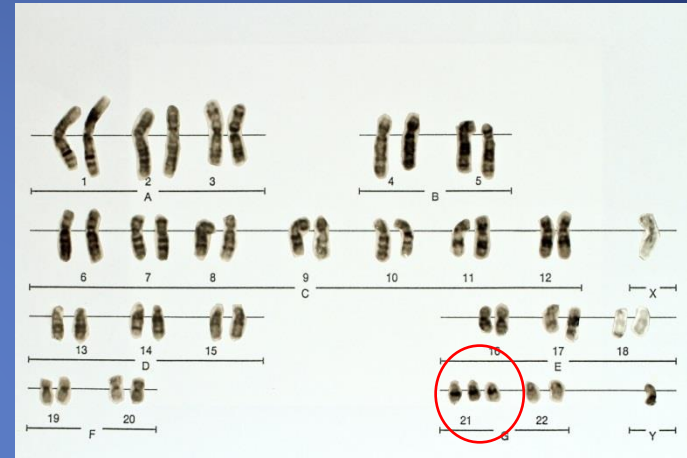
From a blood or saliva sample, the laboratory looks at a patient's actual genetic structure.



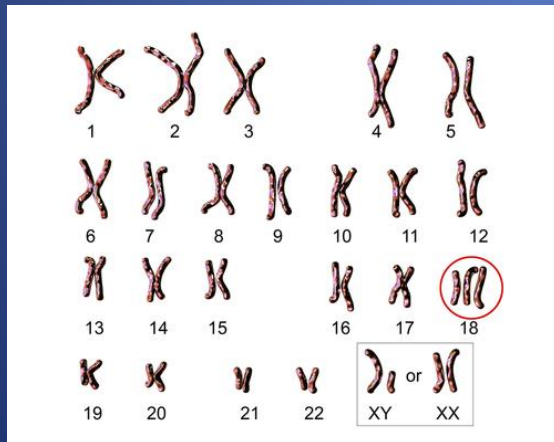
Chromosome testing



Triple X syndrome

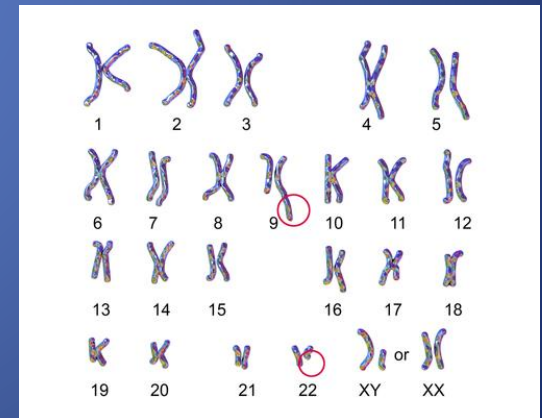


Down syndrome



Trisomy 18
(Edward syndrome)

Defective 9 and 22
chromosomes with
translocational defect



Gene testing

This test . . .

. . . examines this.

Single site analysis



One part of one gene

Individual gene testing



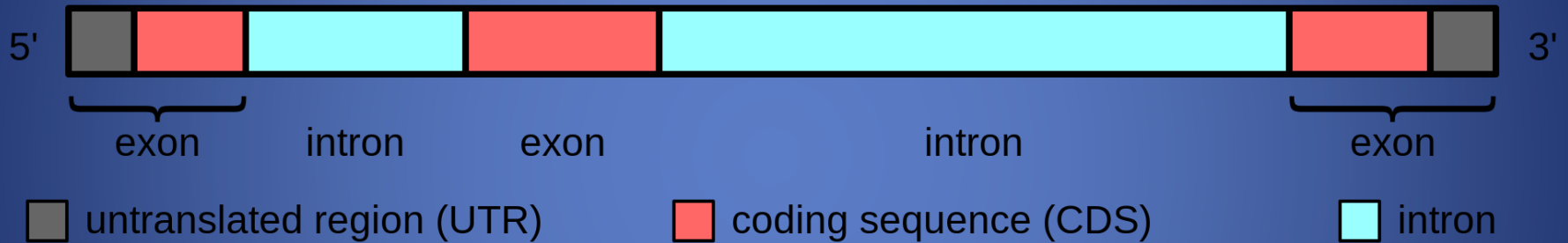
One gene in its entirety

Gene panel testing



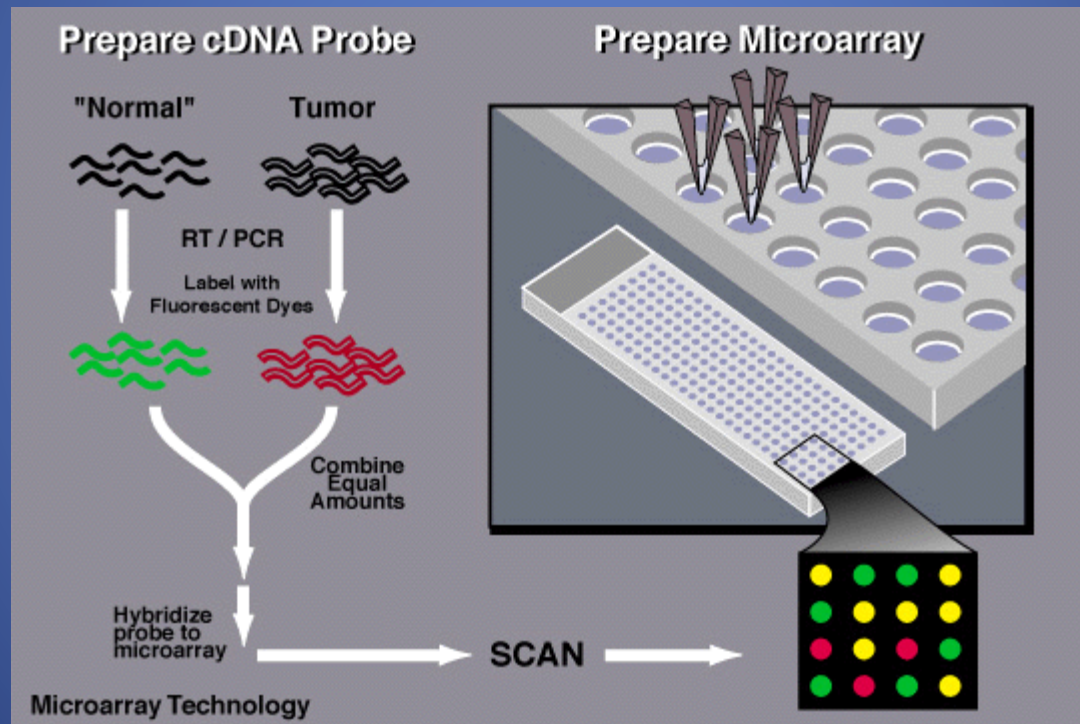
Multiple specific genes

Exome sequencing



[Daycd](#), at the English Wikipedia Project

Chromosomal microarray analysis (CMA)



National Human Genome Research Institute

Genome sequencing



Secondary findings

We were
looking at THIS
gene . .

And then,
over HERE
we found . . .



Biochemical testing





Pop Quiz



- What is sampled in order to do a genetic test?
- What is chromosome testing?
- What is looked at in single site analysis?
- What is looked at in individual gene testing?
- What is looked at in gene panel testing?
- What is looked at in exome testing?
- What is looked at in genome testing?

UNDERSTANDING RESULTS

Sharing findings



Possible results

Negative – no variant was found that would cause illness

Positive – the suspected variant was found, and this probably explains the symptoms;

Variant of Uncertain Significance (VUS) – a variant was found, but geneticists don't know yet what it means.

Limitations



“We didn’t find anything, but it may be there anyway.”

“I don’t know, I’ve never seen this finding before!”



What can the team offer?

- ✓ Make a plan that will best support the child.
- ✓ Provide psychosocial support.
- ✓ Refer to a patient advocacy group.
- ✓ Who else needs testing?



Two models of on-going care

1. Consultation model

The patient's principle medical home is with a primary care provider. The genetics team consults.

2. Continuing care model

The genetics team regularly follows patient.

What else?





Pop Quiz



- What does it mean when a genetic test comes back positive?
- What does it mean when a genetic test comes back negative?
- What is a variant of uncertain significance?
- What are some limitations of genetic testing?
- List four issues that genetic teams will want to cover with patients after discussing a positive result to a test?

CHALLENGES FOR INTERPRETERS

Who is “family”?



Sister or step-sister?

And just who IS “Auntie June?”

Birth son or adopted son?

Difficult conversations



Managing flow



Large groups



Can you see the child?



Conclusion

Pediatric Genetics

Conclusion

Pediatric **Genetics**

Conclusion

Pediatric Genetics



Pediatric Genetics

September 2024

Interpreting for Pediatric Genetics

For more information, go to

Genetics Home Reference, <https://ghr.nlm.nih.gov/>.

Talking Glossary of Genetic Terms, National Human Genome Research Institute, <https://www.genome.gov/glossary/index.cfm>

Glosario Hablado de Términos Genéticos, National Human Genome Research Institute, <http://www.genome.gov/GlossaryS/>

Learning Genetics from Columbia University Medical Center, <http://www.learninggenetics.org/index.html>

National Society of Genetic Counselors, <http://www.nsgc.org/>

Genetics Support Foundation, <https://www.geneticsupport.org/>. 7 videos in English and Spanish

Lexigene, <https://www.lexigene.com/en/>. English-French-Spanish lexicon

Eurogentest, <http://www.eurogentest.org>. Family friendly info in multiple languages