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Transforming Science Learning

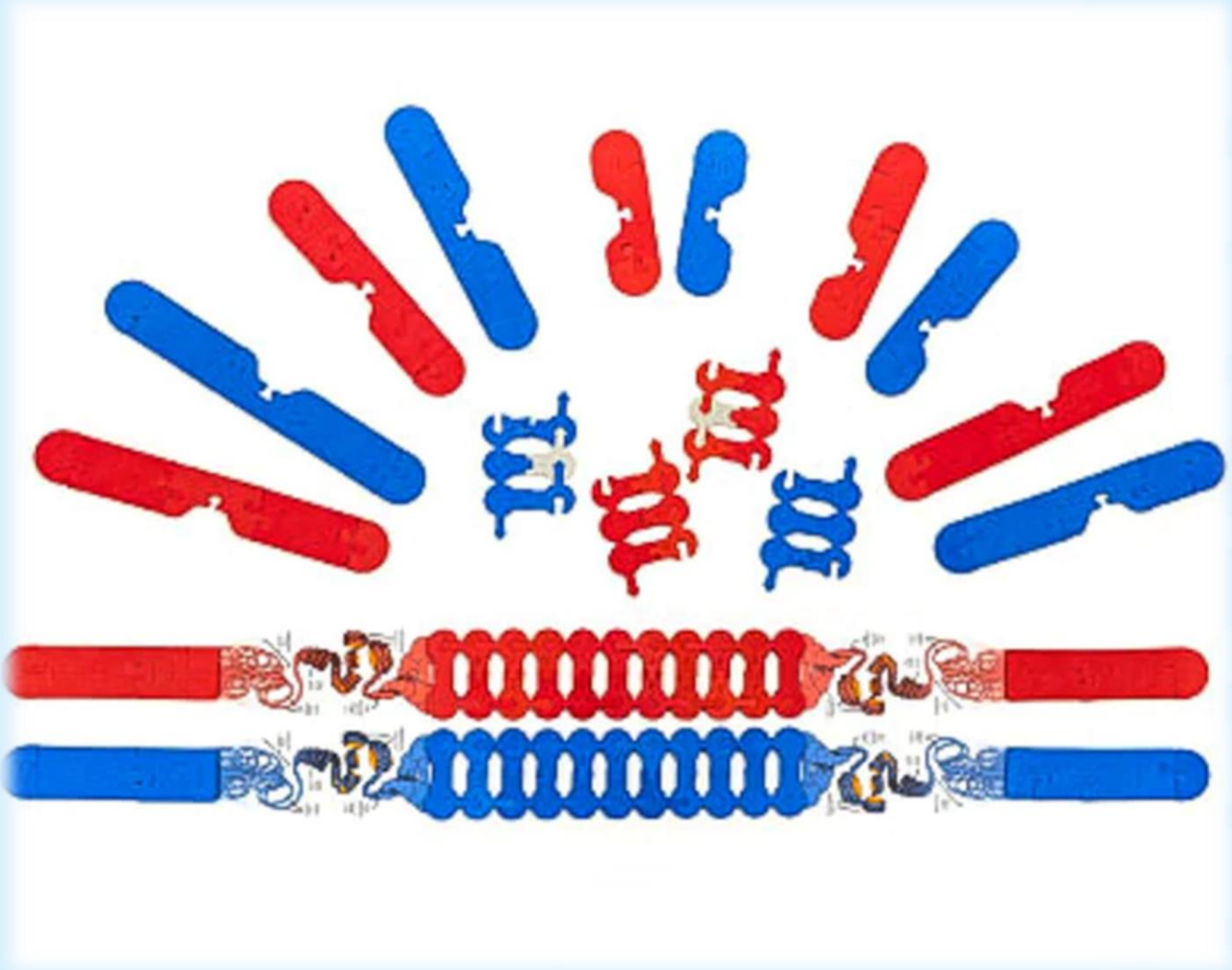
Georgia Science Teachers Association

Genes Unraveled: Modeling Inheritance Mysteries

Ruth Hutson, MSSE

3d molecular designs

Trans*forming* Science Learning



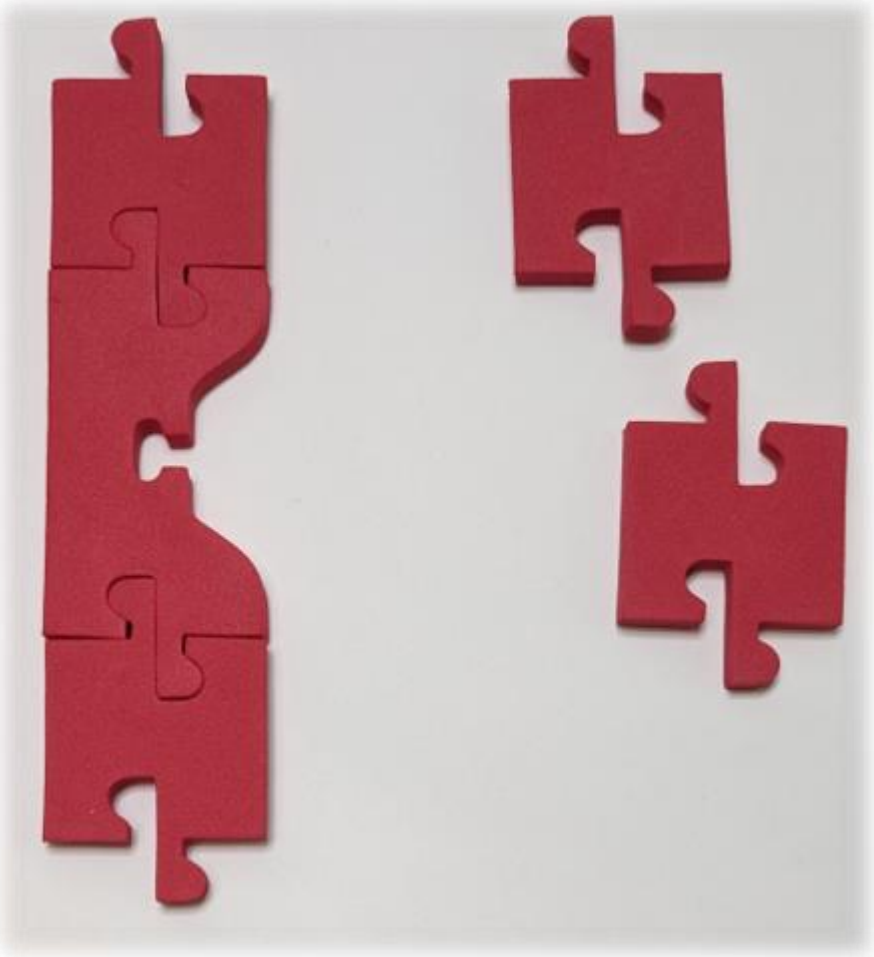
Workshop Outcomes

- Construct Punnett squares with gene sequences as alleles to connect inheritance of traits to chromosomes at the molecular level.
- Model several types of inheritance using the Chromosome Connection Kit©.
- Make and defend a claim based on evidence that genetic variations result from a variety of factors.

Construct a chromosome

- **Start with one centromere**
- Add 3 to 4 chromosome connectors to each side of the centromere
- Add a cap piece to each end of the chromatid.

Construct a chromosome



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Create a sister chromatid

- Create a duplicate copy of the chromosome to make a sister chromatid.

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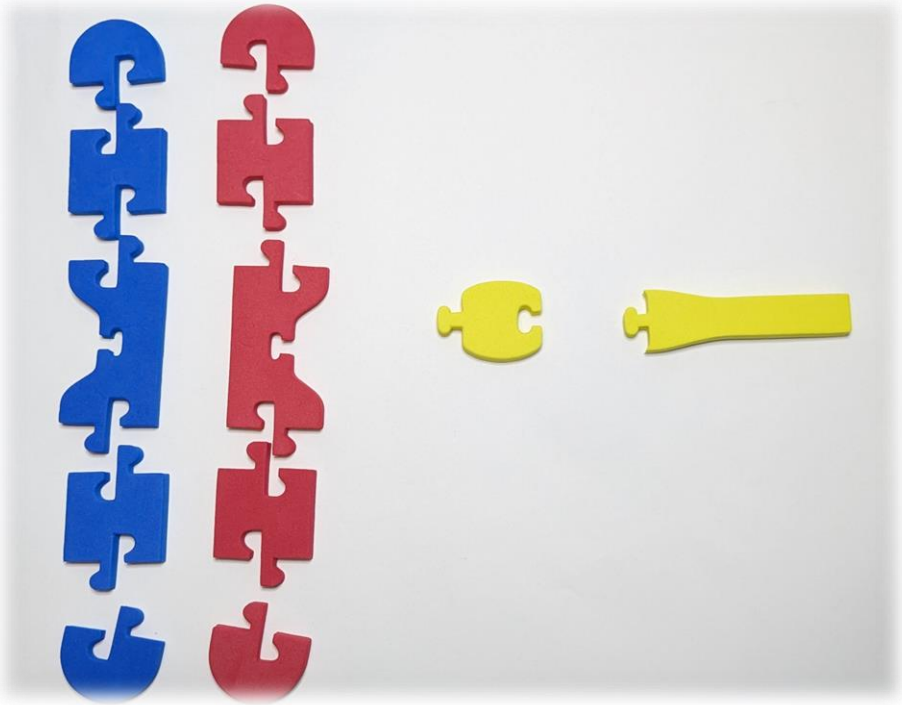
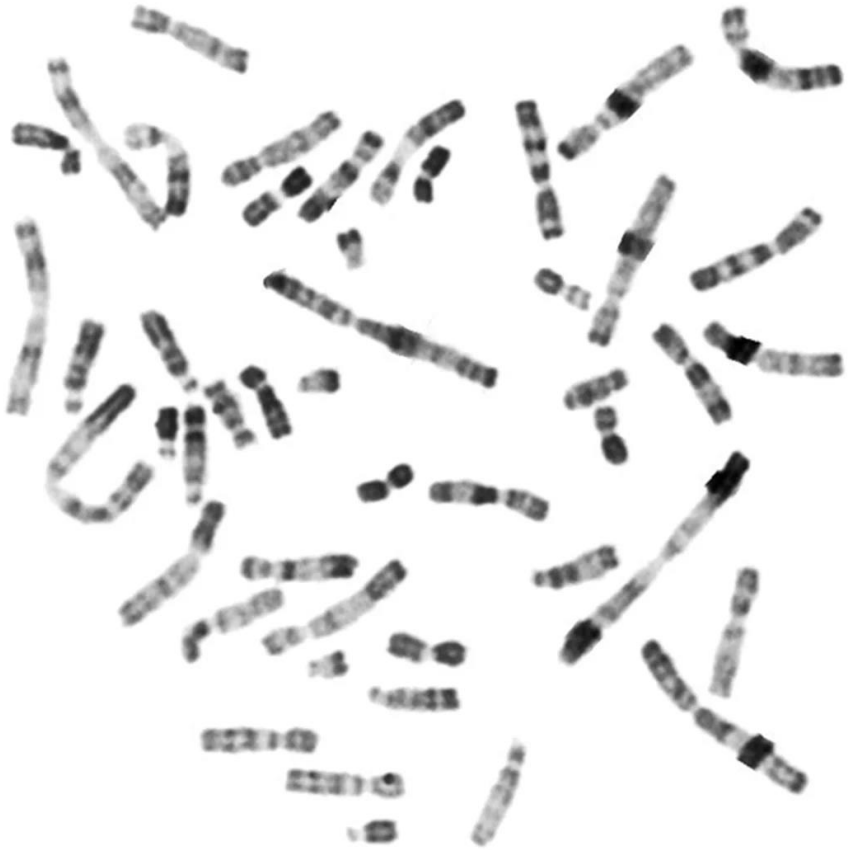
Suggested Chromosome Assembly

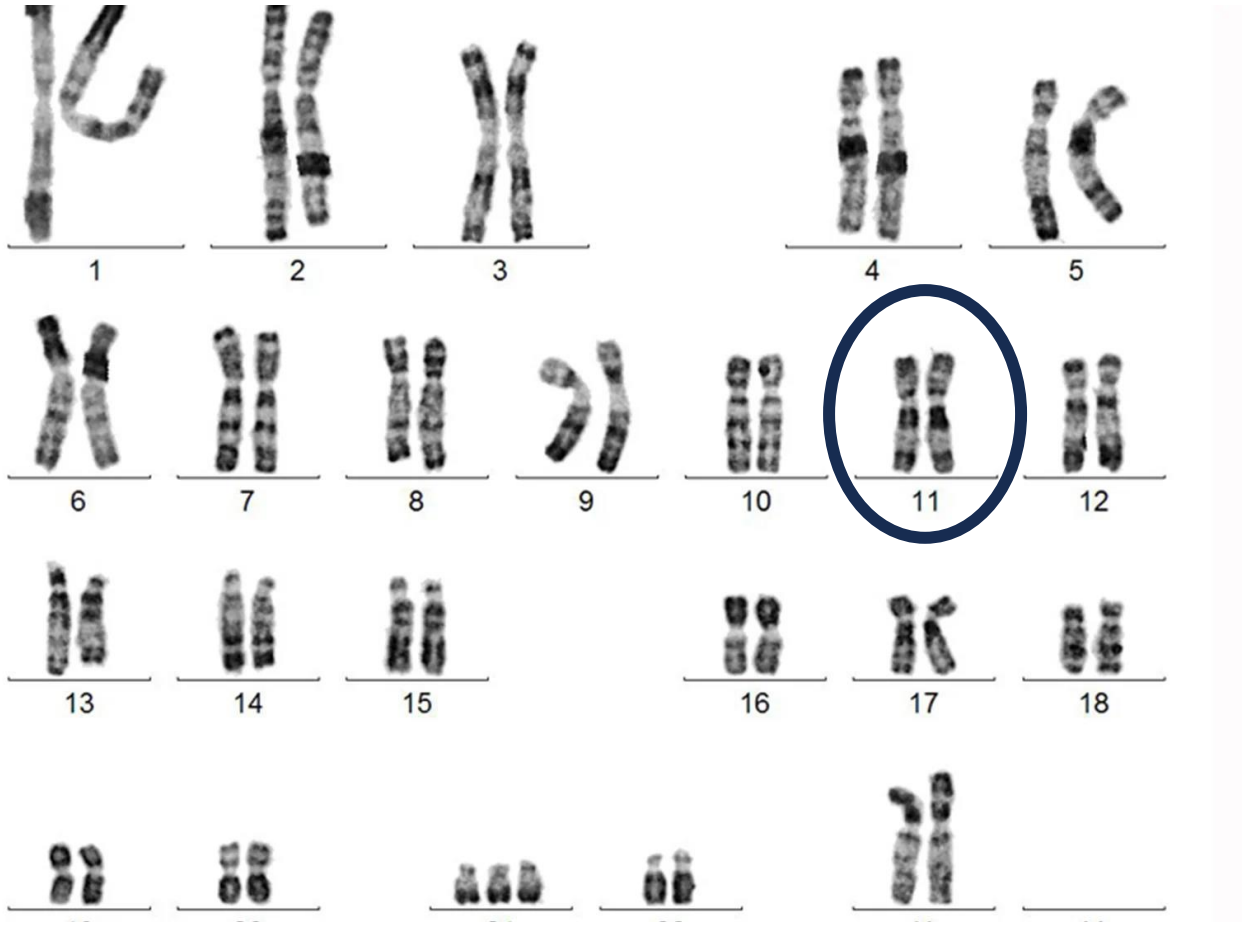


Chromosome Size	Part	Egg donating parent chromosome (Red Foam)	Sperm donating parent chromosome (Blue Foam)
Short chromosome (Acrocentric)	p arm linkers	0	0
	q arm linkers	1	1
Medium Chromosome (Metacentric)	p arm linkers	1	1
	q arm linkers	1	1
Long Chromosome (Submetacentric)	p arm linkers	1	1
	q arm linkers	2	2

Why do I look different than my siblings?

Let’s look at chromosome 11



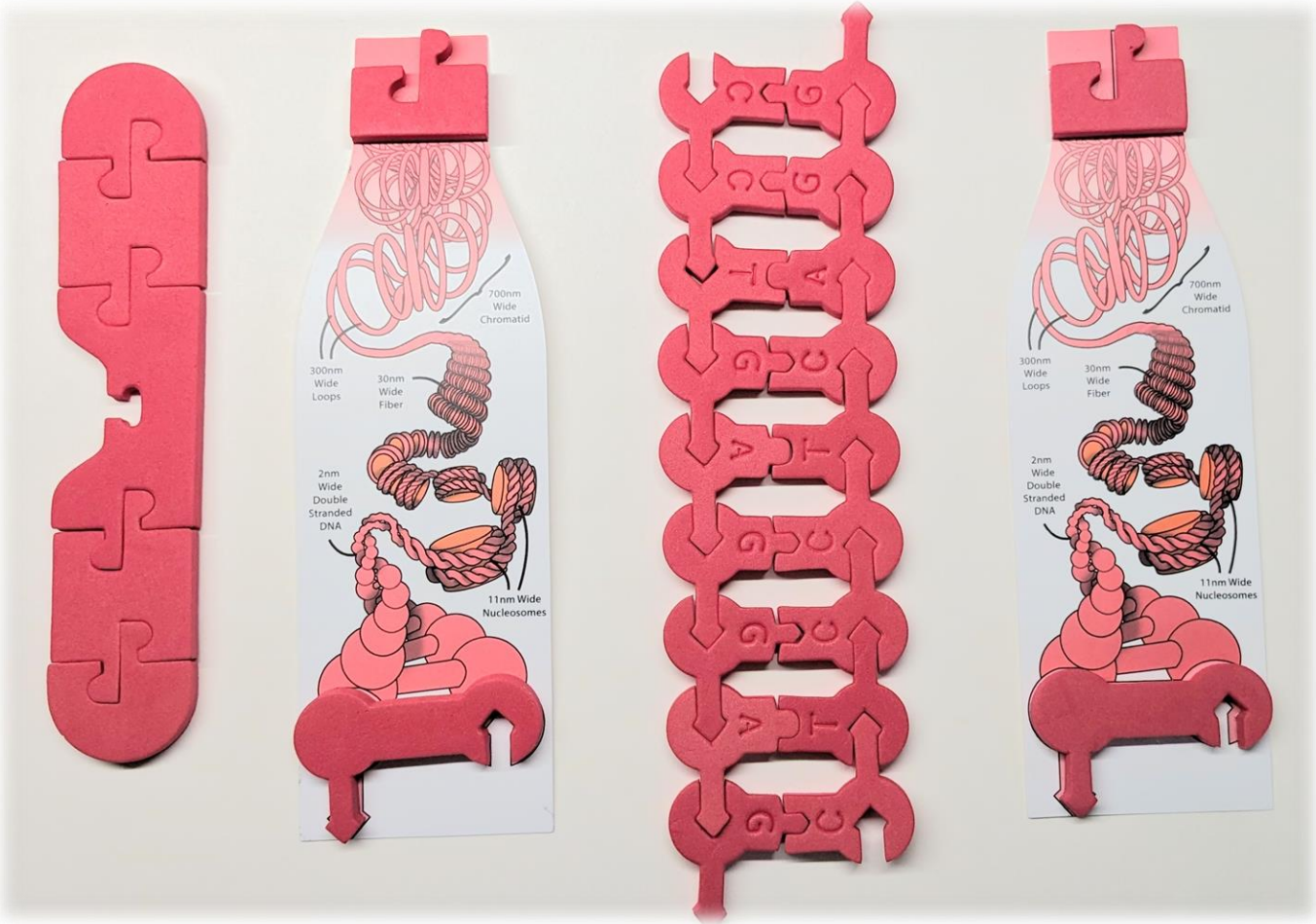


Create a normal beta-globin DNA gene sequence

3′ G G A C T C C T C 5′

5′ C C T G A G G A G 3′

Use insert pieces to assemble the maternal chromosome



Use insert pieces to assemble the maternal chromosome

Human B-globin is located on the p-arm of chromosome 11.

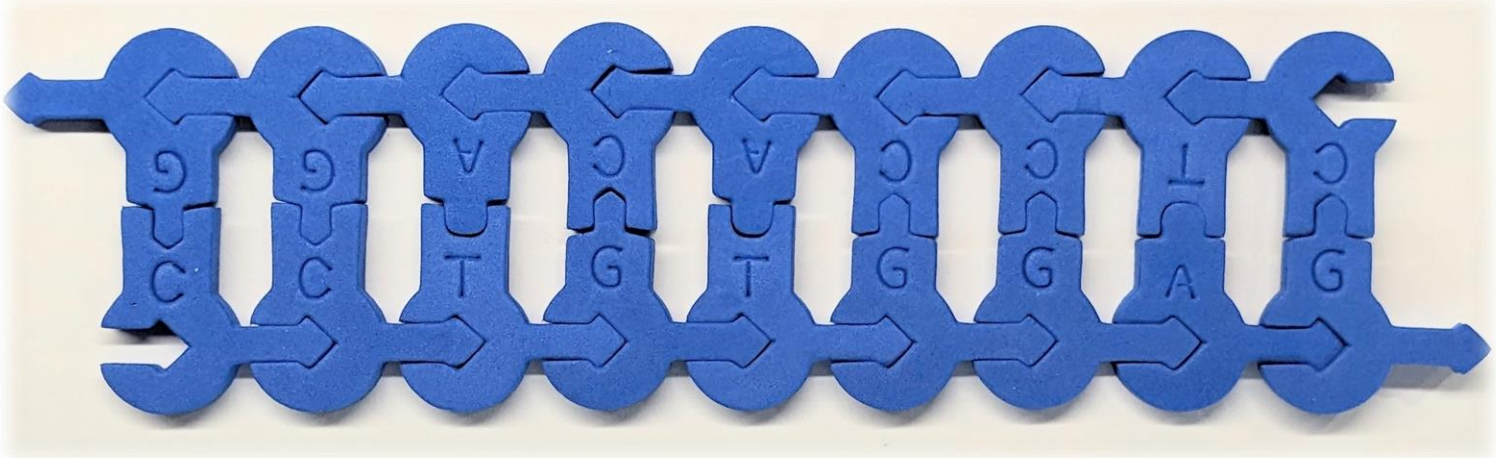
The human B-globin locus is a 60-kb segment on the short arm and contains five functional genes and two pseudogenes. We will look at a small part of this locus.

It is responsible for the creation of the beta parts of the oxygen transport protein, hemoglobin.

Create a sickle cell beta-globin DNA gene sequence

3′ G G A C A C C T C 5′

5′ C C T G T G G A G 3′



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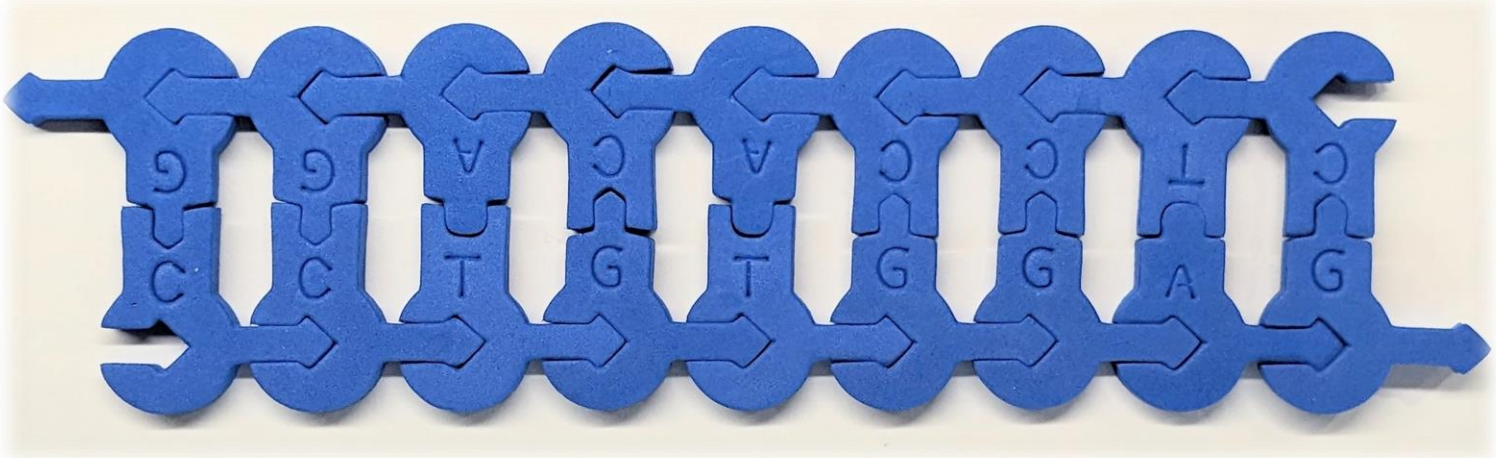
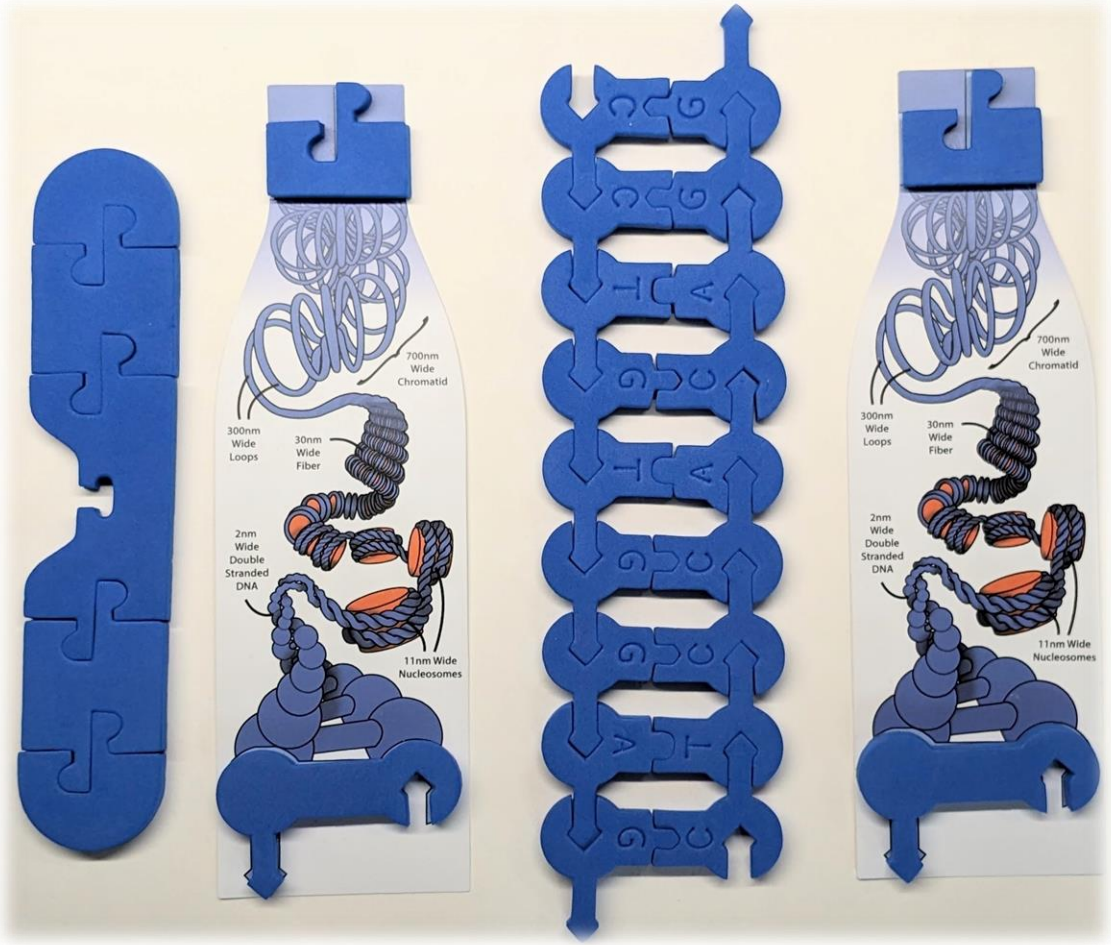
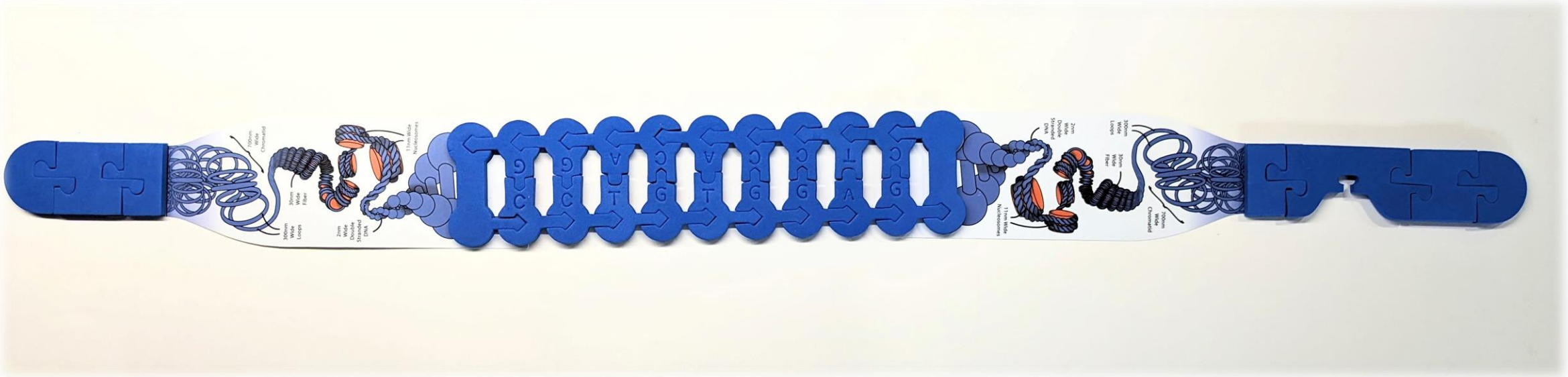


Use insert pieces to assemble the paternal chromosome

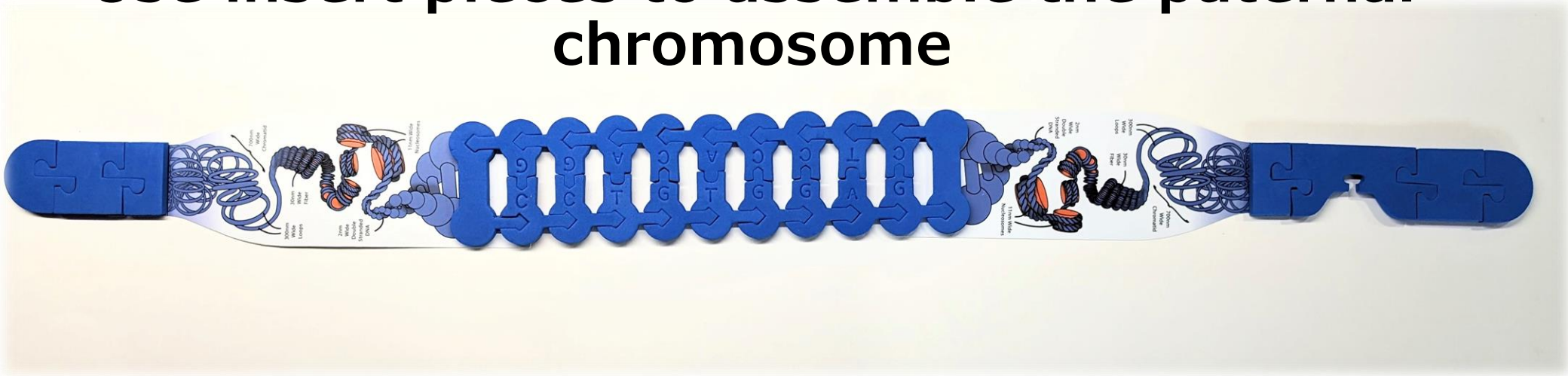


Use insert pieces to assemble the paternal chromosome



What is the difference between the two sequences?

Use insert pieces to assemble the paternal chromosome



What is the difference between the two sequences?

G – C	G – C
A – T	T – A
G – C	G – C

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The Genetic Codon Chart©

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Amino Acid Properties

- Translation Start Codon
- Translation Stop Codon
- Hydrophobic / Non-polar
- Hydrophilic / Polar
- Negative Charge
- Positive Charge
- Cysteine

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How might that affect the resulting protein?

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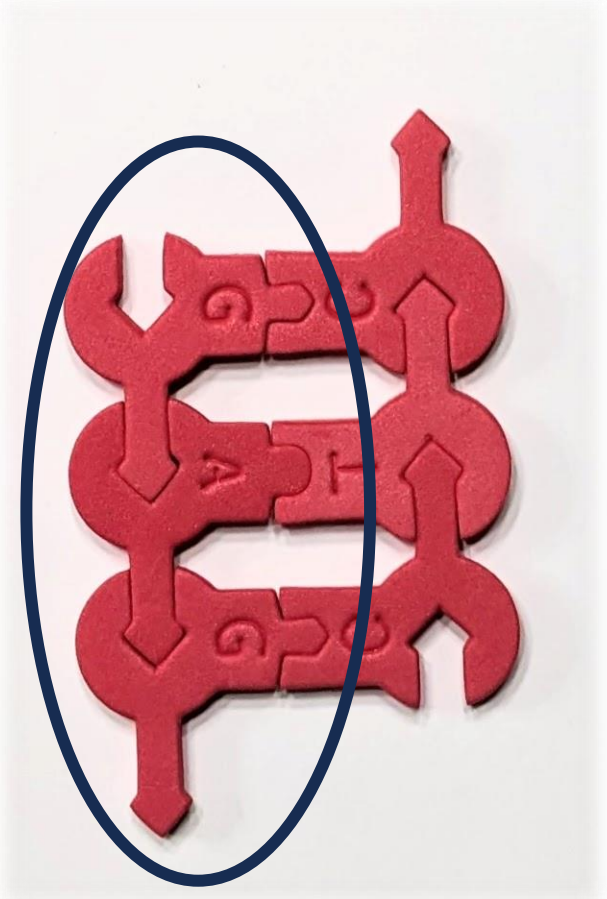

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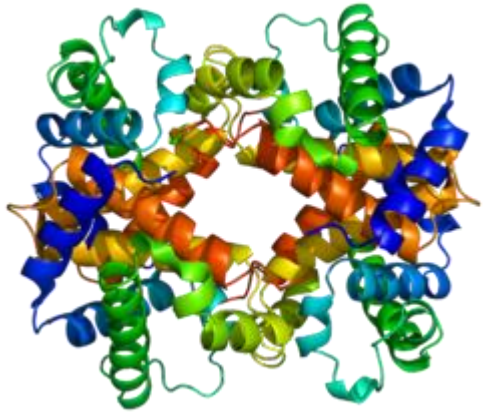
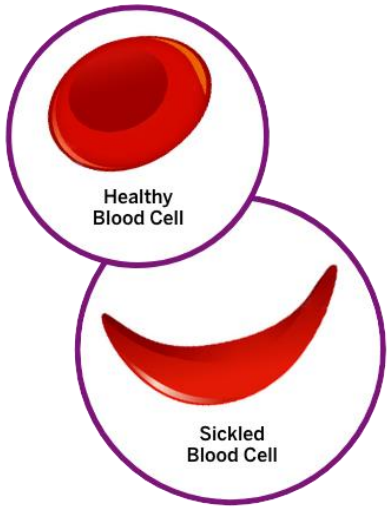
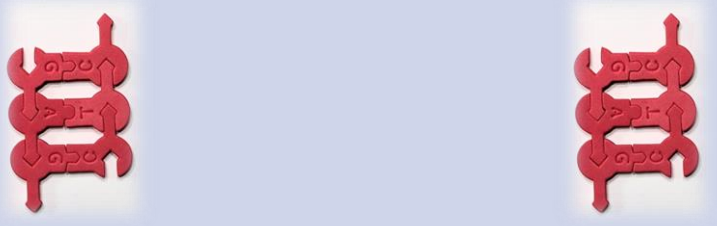
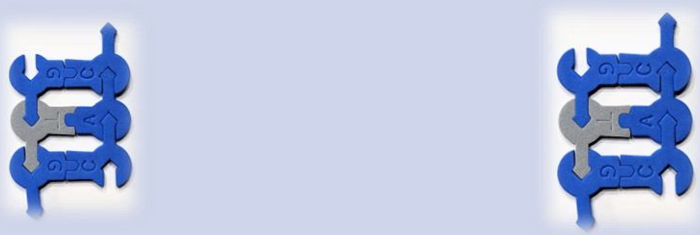


Beta-globin



Understanding Codominance

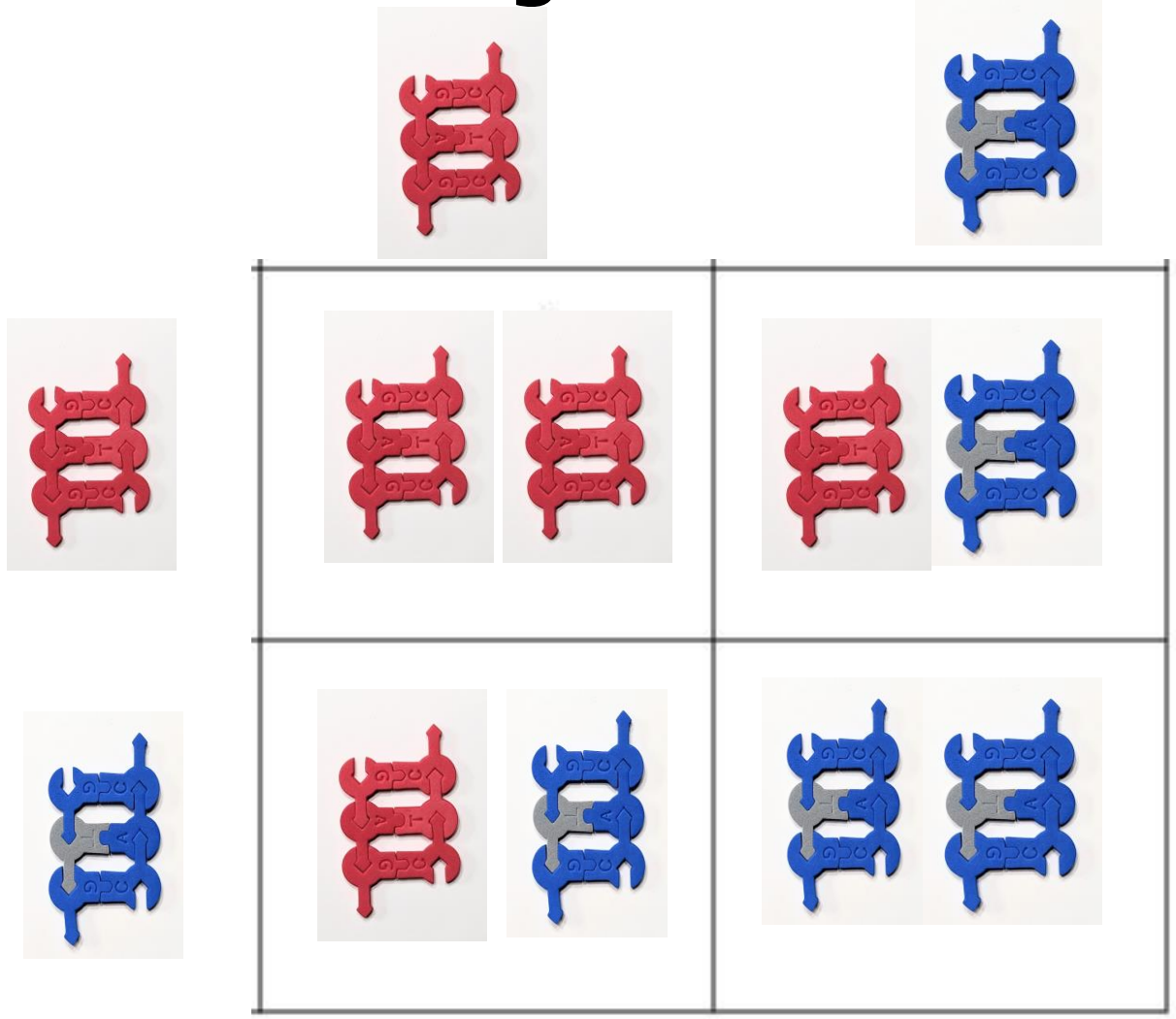
Sickle Cell Anemia Possible Genotypes and Phenotypes	DNA sequence
SS Homozygous Typical beta-globin	
Ss Heterozygous Carrier Typical beta-globin and Atypical beta-globin	
ss Homozygous Atypical beta-globin	

A visit to the genetic counselor

Two individuals with sickle
cell trait want to know their chances of having a child with sick cell anemia. Complete the Punnett square using DNA sequences to fill in each of the squares.

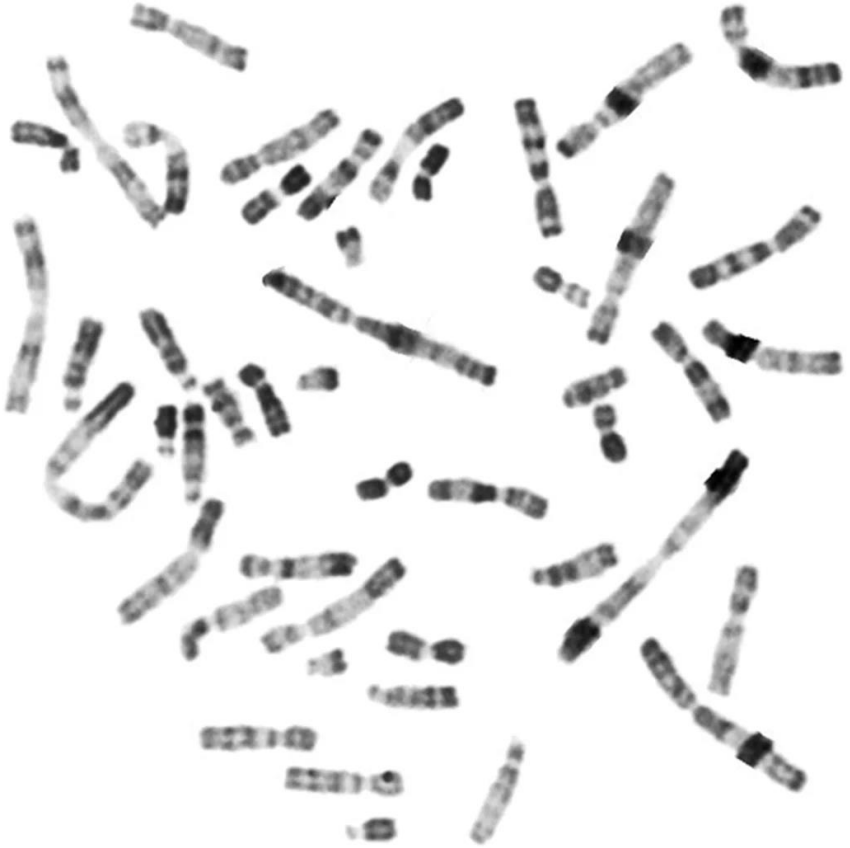
How would you advise them?

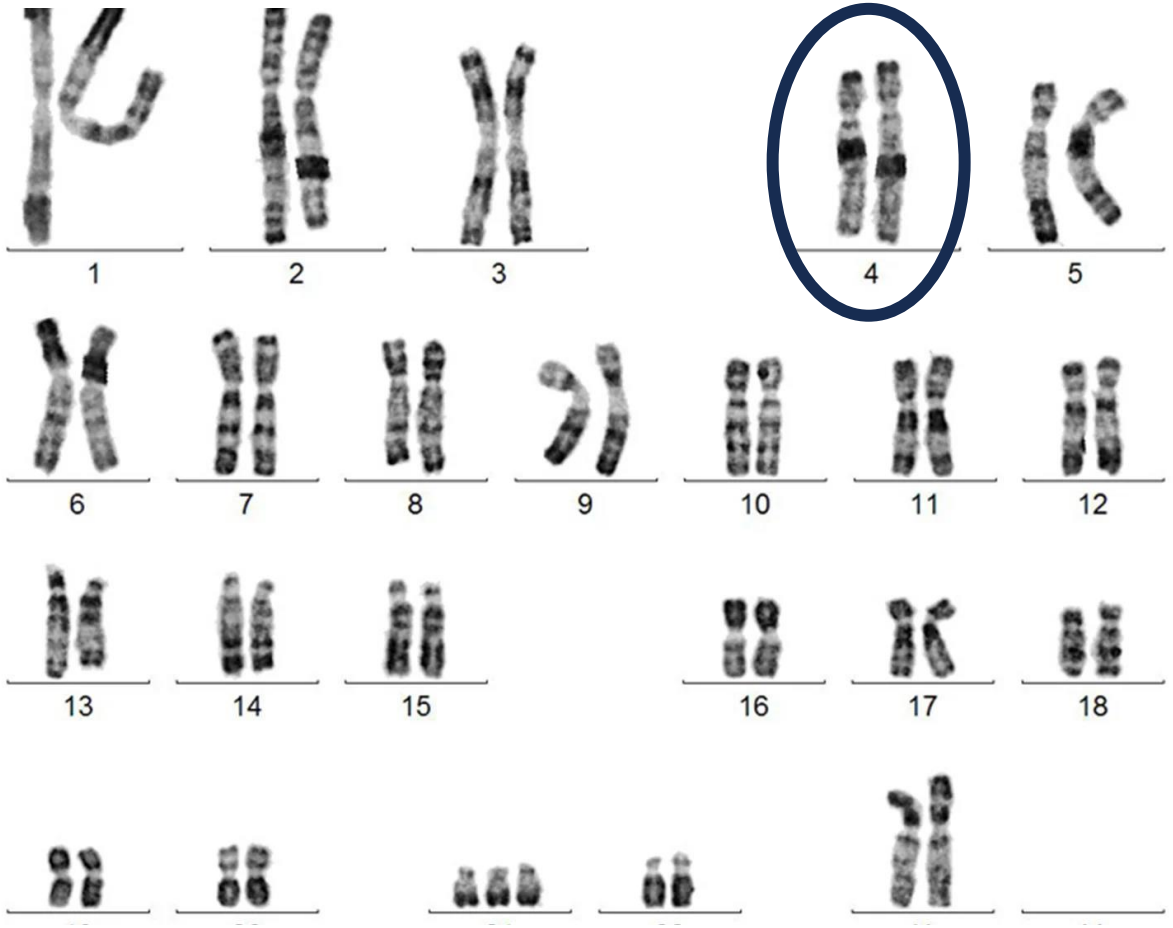
A visit to the genetic counselor



What are the implications?

Moving to chromosome 4





Huntington disease is caused by multiple repeats of CAG at the 5′ end of the HTT gene

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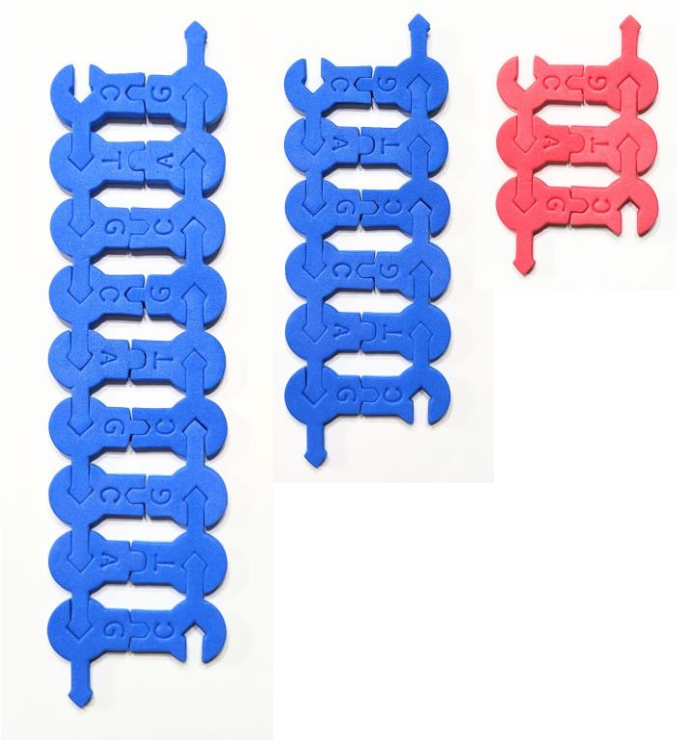
How might that affect the resulting protein?



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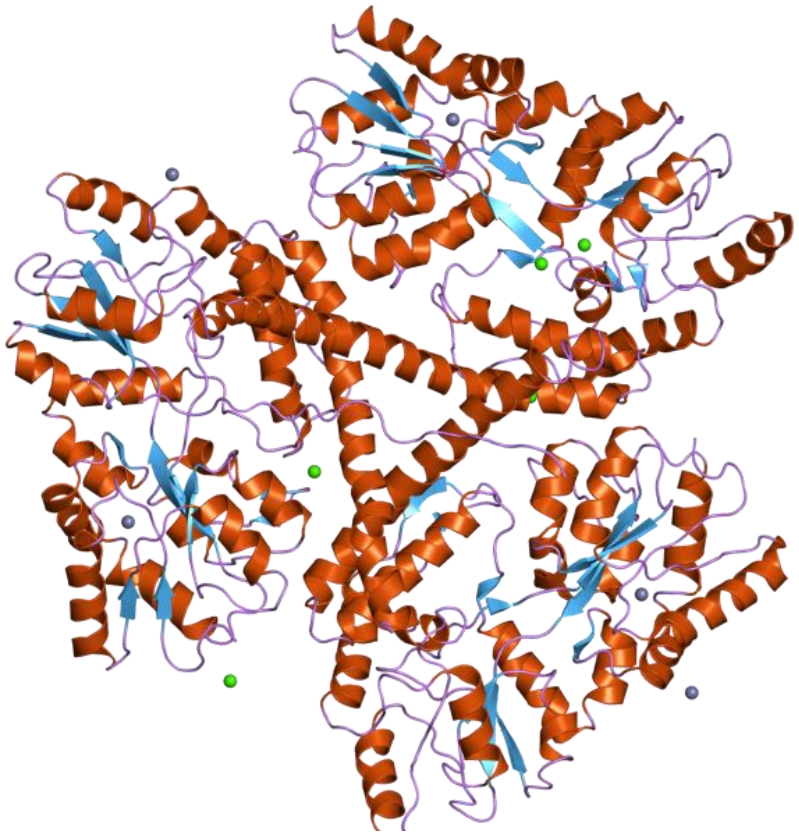
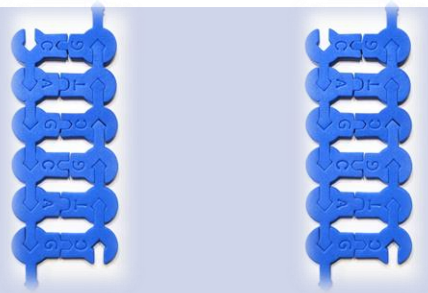
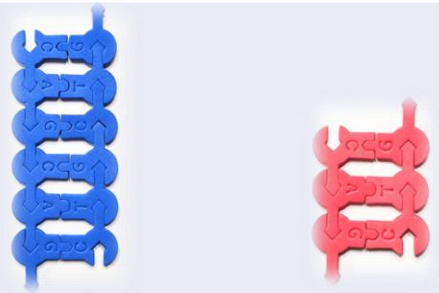


Huntingtin

Understanding Complete Dominance

Huntington disease Possible Genotypes and Phenotypes	DNA sequences
HH Atypical huntingtin protein Huntington disease	
Hh Atypical huntingtin protein Huntington disease	
hh Normal huntingtin protein	

A visit to the genetic counselor

One individual with severe Huntington disease and a healthy individual want to know their chances of having a child with Huntington disease. Complete the Punnett square using DNA sequences to fill in each of the squares.

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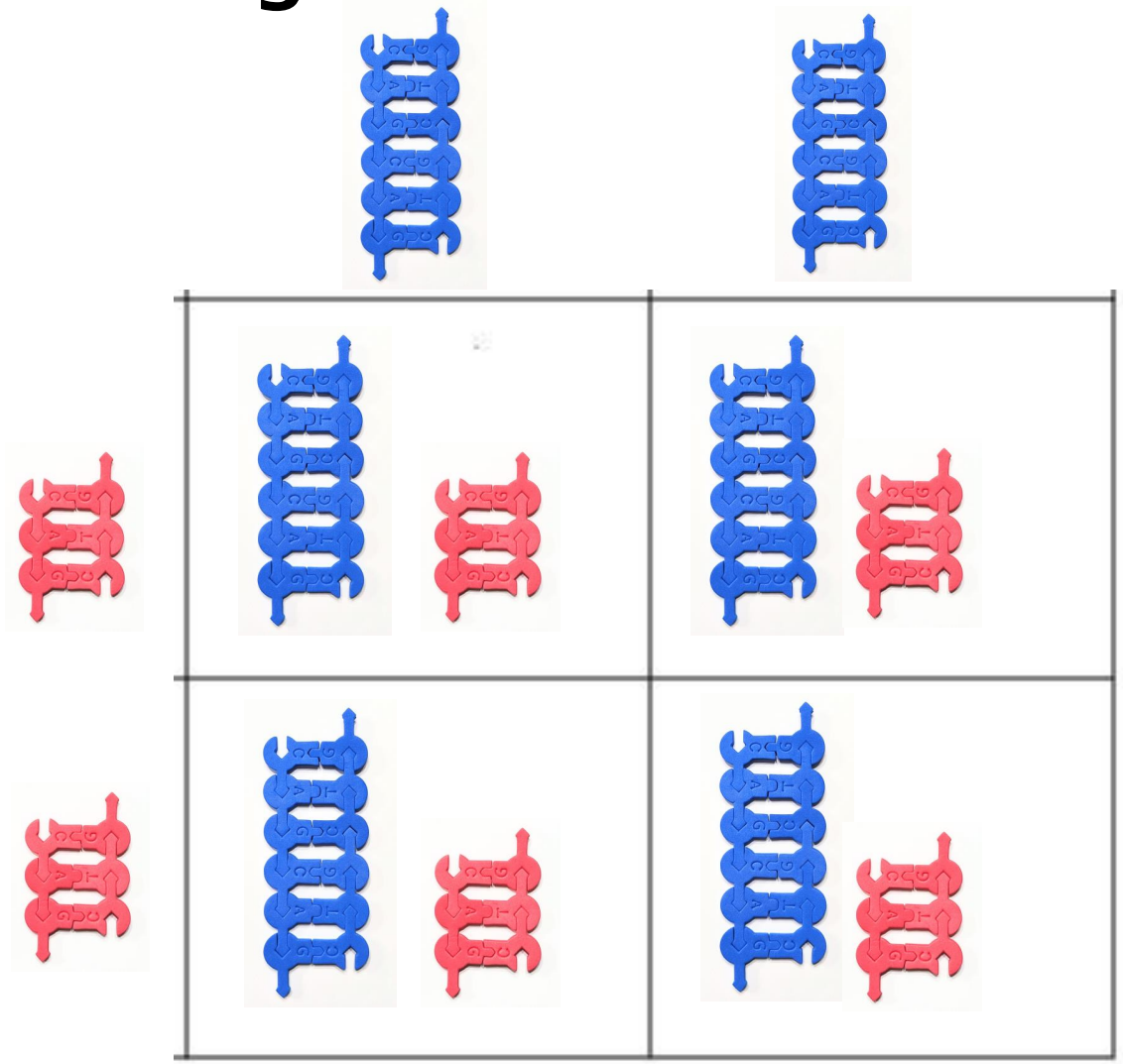
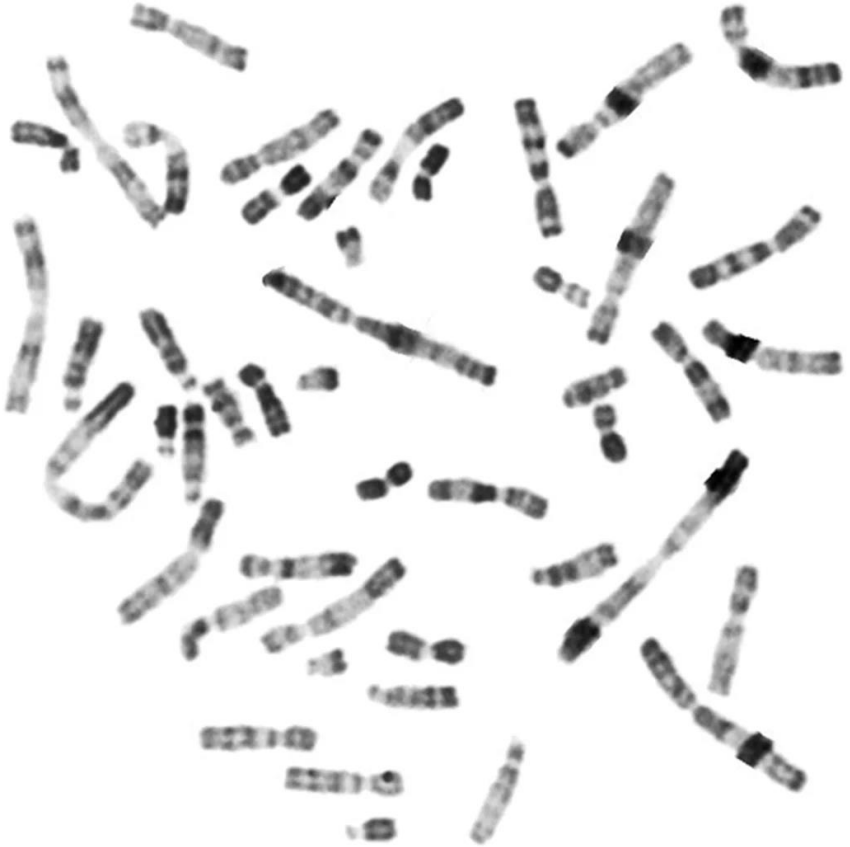
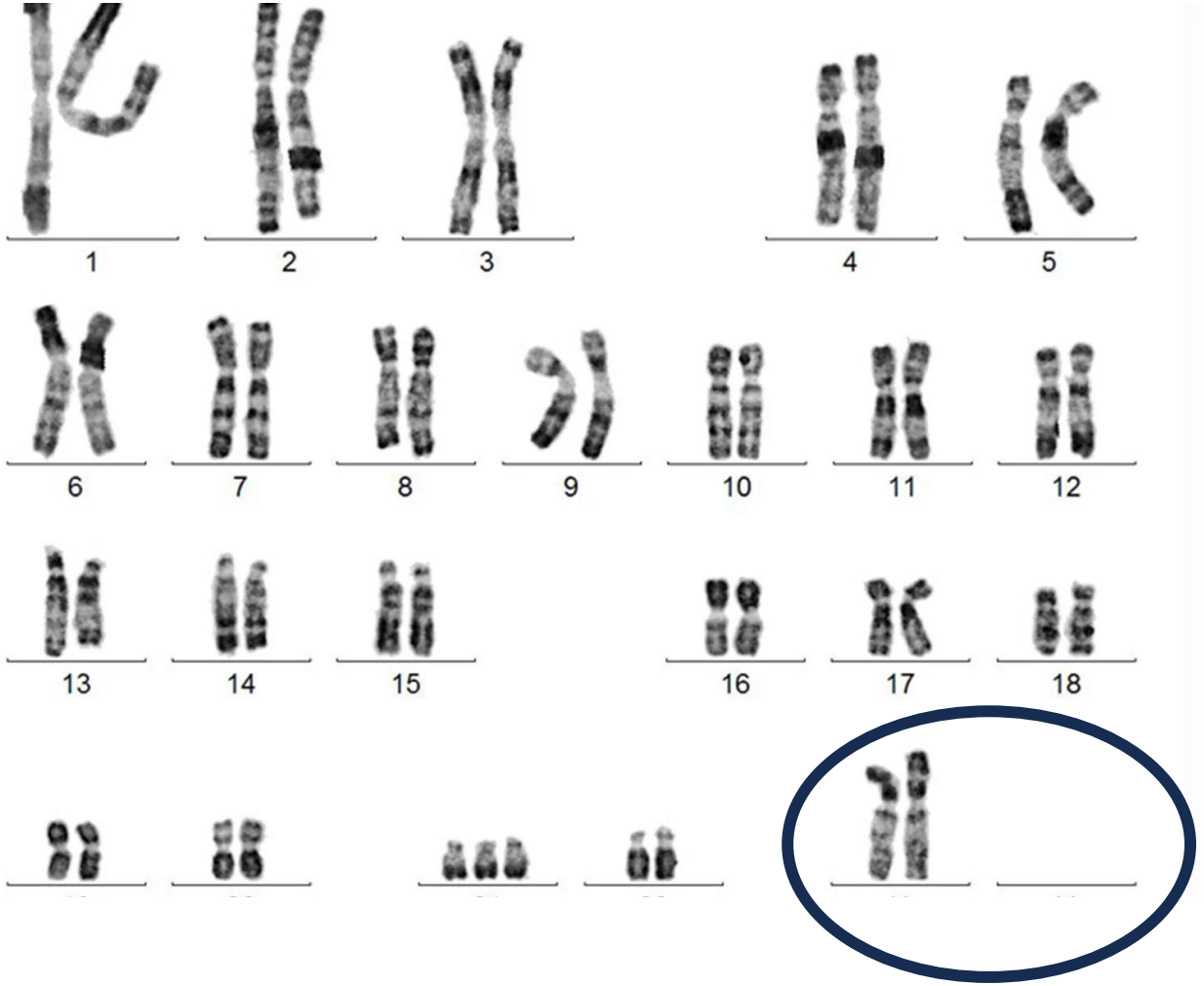


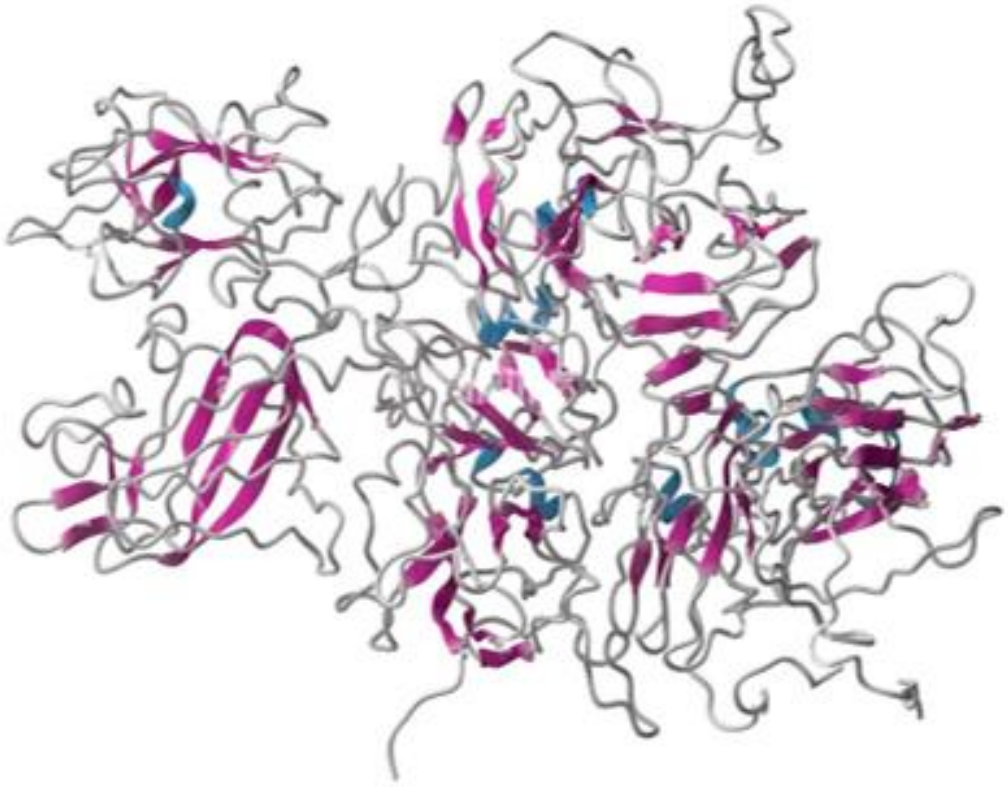
What are the implications?

Let’s look at the sex chromosomes



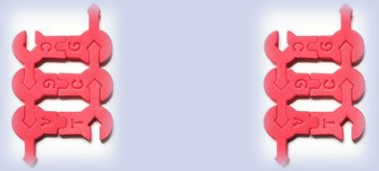
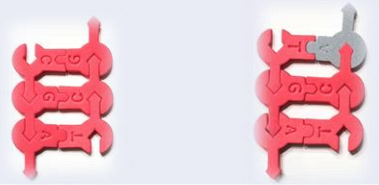


Hemophilia A is caused by a mutation in the F8 gene on the X sex chromosome.



Clotting Factor VIII

Understanding Sex-linked Inheritance

Hemophilia Possible Genotypes and Phenotypes	DNA sequences
$X^H X^H$ Non-hemophilic biological female	
$X^H X^h$ Biological female carrier for hemophilia	
$X^h X^h$ Biological female with hemophilia	
X^HY Non-hemophilic biological male	
X^hY Biological male with hemophilia	

A visit to the genetic counselor

A carrier female and non-hemophiliac male want to know if they have a chance of having a child with hemophilia. Complete the Punnett square using DNA sequences to fill in each of the squares.

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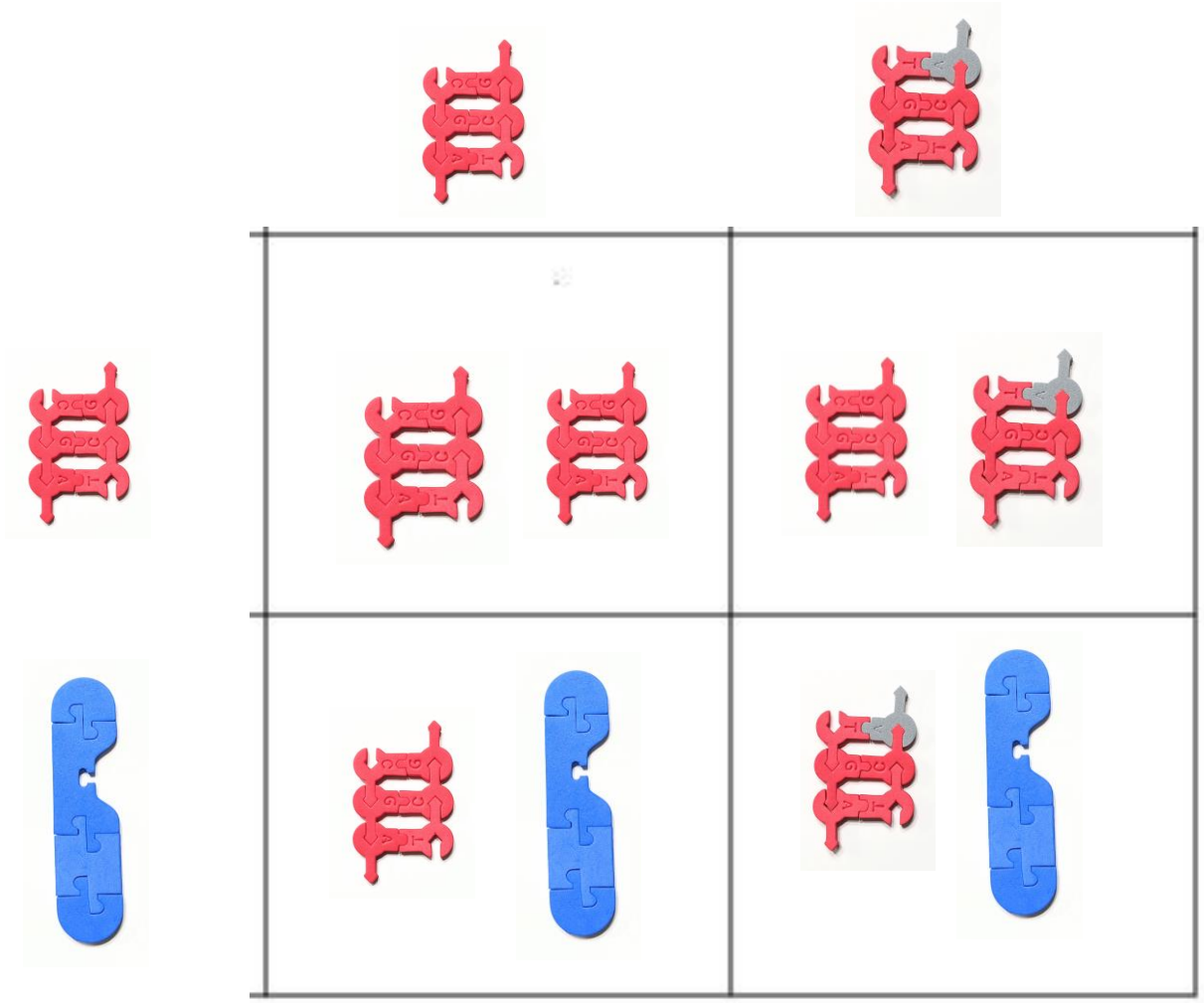
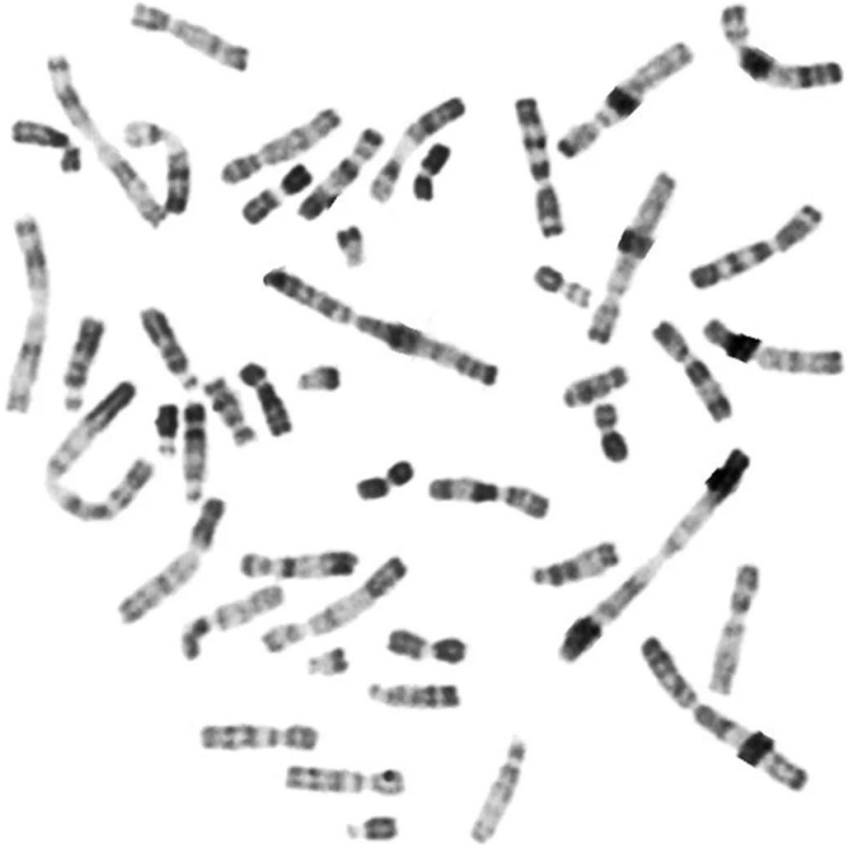
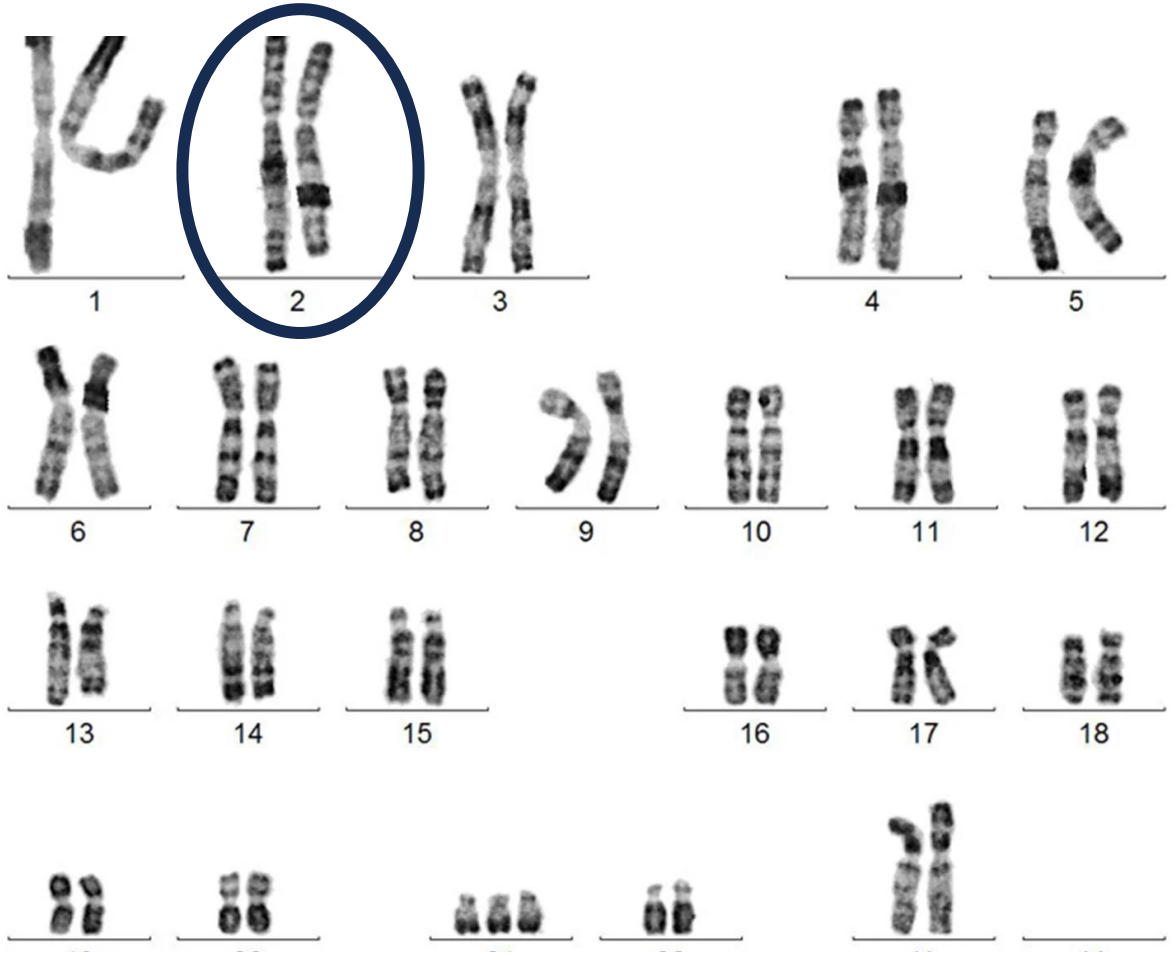


Our last stop is chromosome 2.





The gene for myostatin has a variant that causes significantly increased muscle mass and strength.

The Genetic Codon Chart©

	U	C	A	G	
U	UUU → Phe F	UCU → Ser S	UAU → Tyr Y	UGU → Cys C	U
	UUC → Phe F	UCC → Ser S	UAC → Tyr Y	UGC → Cys C	C
	UUA → Leu L	UCA → Ser S	UAA → Stop	UGA → Stop	A
	UUG → Leu L	UCG → Ser S	UAG → Stop	UGG → Trp W	G
C	CUU → Leu L	CCU → Pro P	CAU → His H	CGU → Arg R	U
	CUC → Leu L	CCC → Pro P	CAC → His H	CGC → Arg R	C
	CUA → Leu L	CCA → Pro P	CAA → Gln Q	CGA → Arg R	A
	CUG → Leu L	CCG → Pro P	CAG → Gln Q	CGG → Arg R	G
A	AUU → Ile I	ACU → Thr T	AAU → Asn N	AGU → Ser S	U
	AUC → Ile I	ACC → Thr T	AAC → Asn N	AGC → Ser S	C
	AUA → Ile I	ACA → Thr T	AAA → Lys K	AGA → Arg R	A
	AUG → Met M	ACG → Thr T	AAG → Lys K	AGG → Arg R	G
G	GUU → Val V	GCU → Ala A	GAU → Asp D	GGU → Gly G	U
	GUC → Val V	GCC → Ala A	GAC → Asp D	GGC → Gly G	C
	GUA → Val V	GCA → Ala A	GAA → Glu E	GGA → Gly G	A
	GUG → Val V	GCG → Ala A	GAG → Glu E	GGG → Gly G	G

Amino Acid Properties

- Translation Start Codon
- Translation Stop Codon
- Hydrophobic / Non-polar
- Hydrophilic / Polar
- Negative Charge
- Positive Charge
- Cysteine

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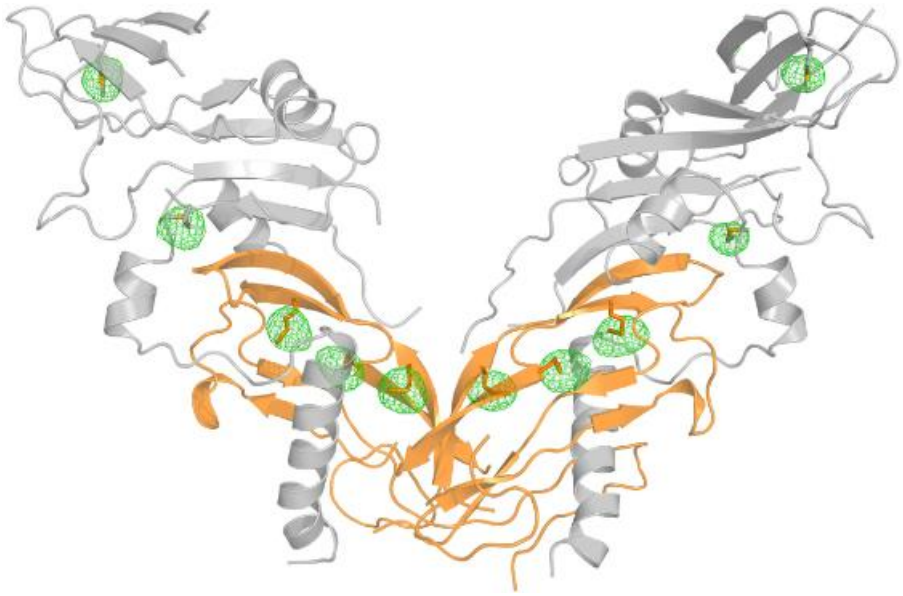

Myostatin

A guanine was replaced by an adenine. What are the implications of this mutation?

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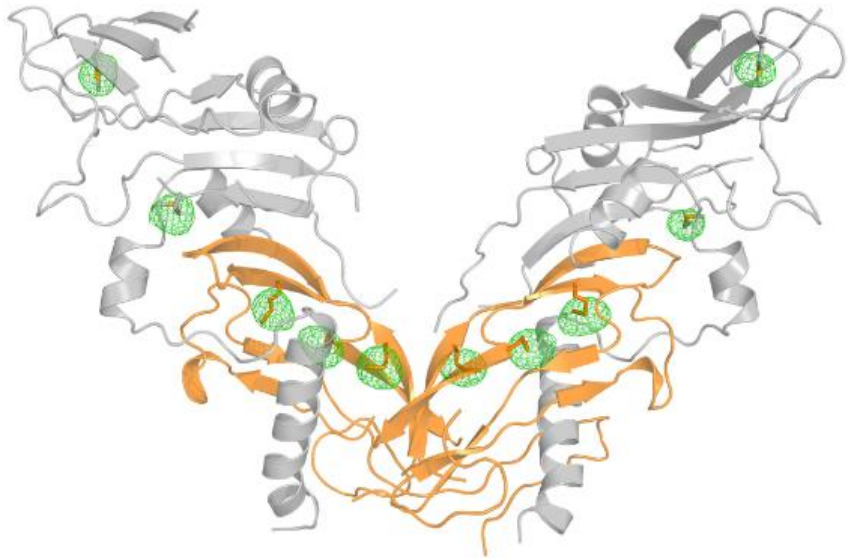
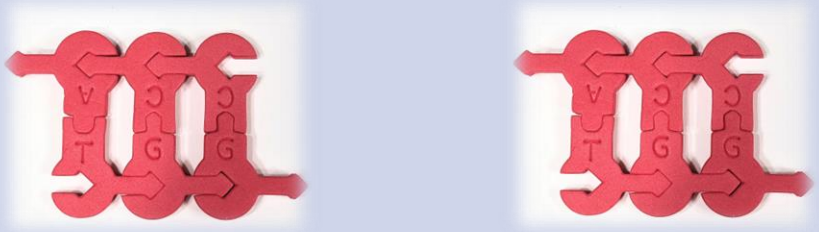
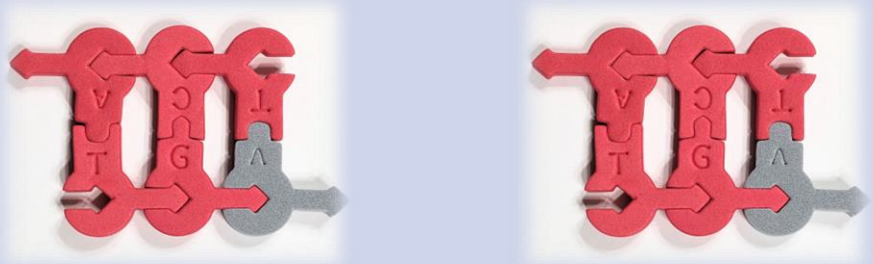


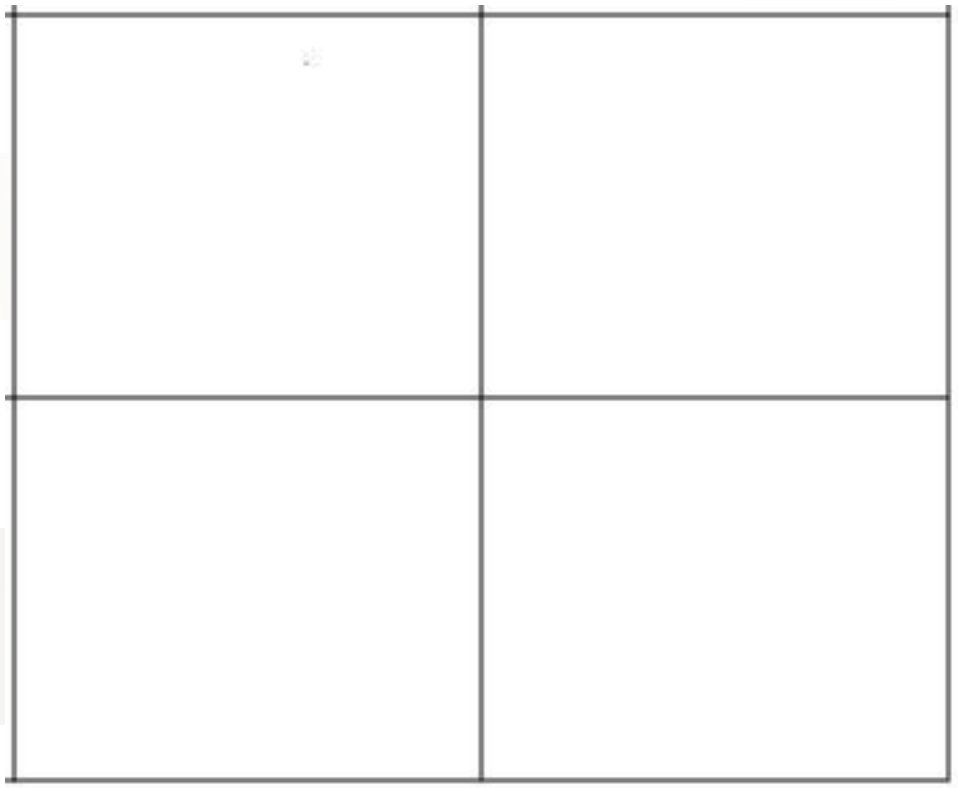
Myostatin

Understanding Incomplete Dominance

Myostatin-related muscle hypertrophy Possible Genotypes and Phenotypes	DNA Sequence
Homozygous dominant MM	
Heterozygous Mm	
Homozygous recessive mm	

A visit to the genetic counselor

Two heterozygous individuals with muscle hypertrophy syndrome want to know if they might have affected children. Complete the Punnett square using DNA sequences to fill in each of the squares.



What are the implications?

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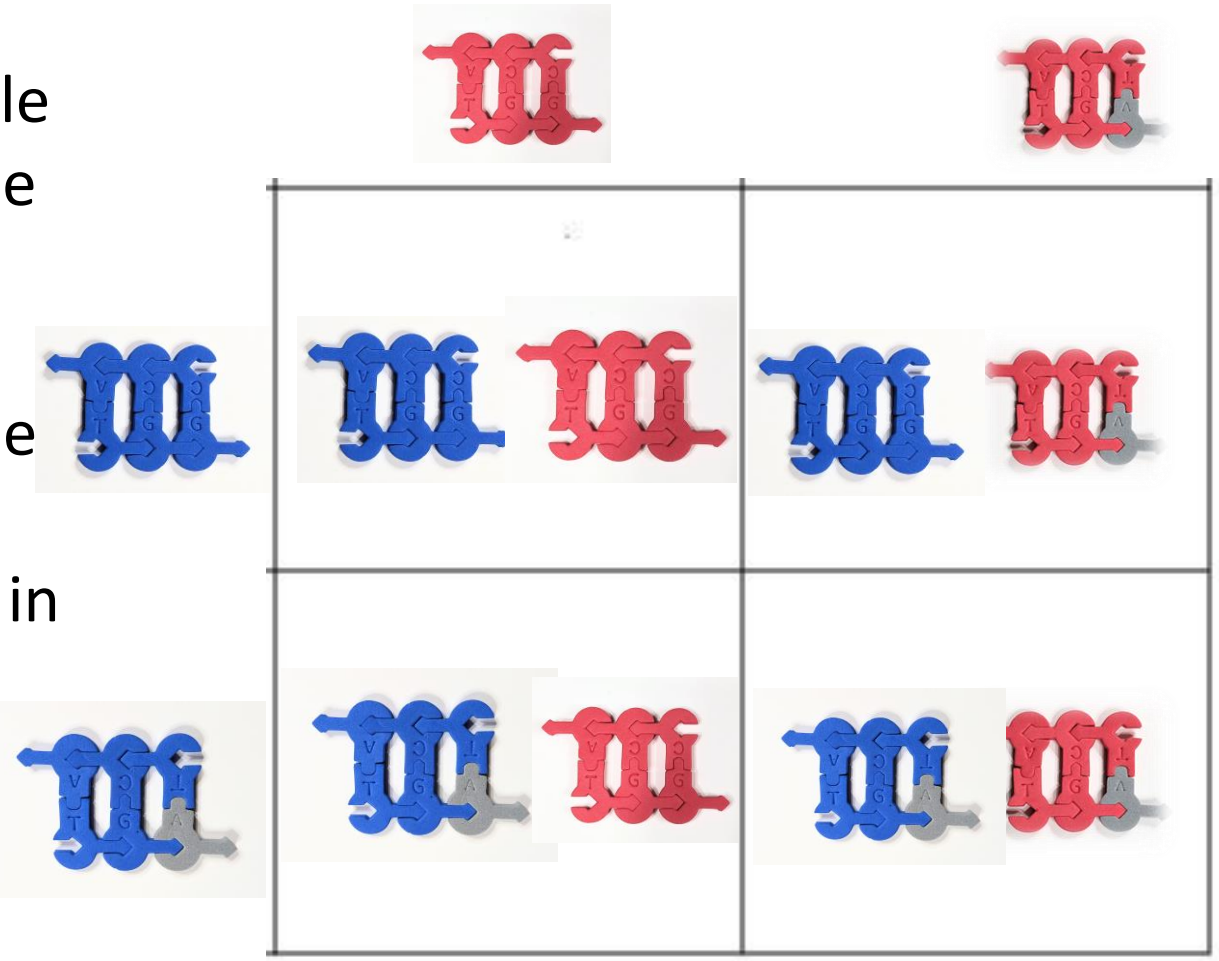

What are the implications?

Genetic databanks to help you develop other cases

- Home - OMIM - NCBI (nih.gov)

- MedlinePlus: Genetics

Additional Resources

- Understanding Sickle Cell - Sickle Cell Speaks
- Sickle hemoglobin (HbS) allele and sickle cell disease: a HuGE review - PubMed (nih.gov)
- What is HD? - Huntington's Disease Society of America (hdsa.org)
- Home - Hemophilia Federation of America
- Myostatin-related muscle hypertrophy: MedlinePlus Genetics
- Entry - *601788 - MYOSTATIN; MSTN – OMIM
- Myostatin Mutation Associated with Gross Muscle Hypertrophy in a Child | NEJM

Additional Resources

- The status of the human gene catalogue | Nature
- Human Genome Is Much More Than Just Genes | Science | AAAS
- An integrated encyclopedia of DNA elements in the human genome | Nature

Workshop NGSS Connections

SEPs	CCCs	DCIs
Asking Questions and Defining Problems	Systems and System Models	HS-LS1-4
Developing and Using Models	Cause and Effect	HS-LS3-2
	Scale, Proportion, and Quantity	Georgia Standards: SB3, SB6

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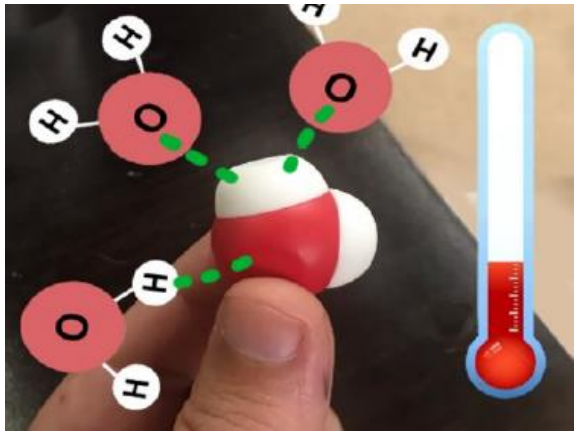

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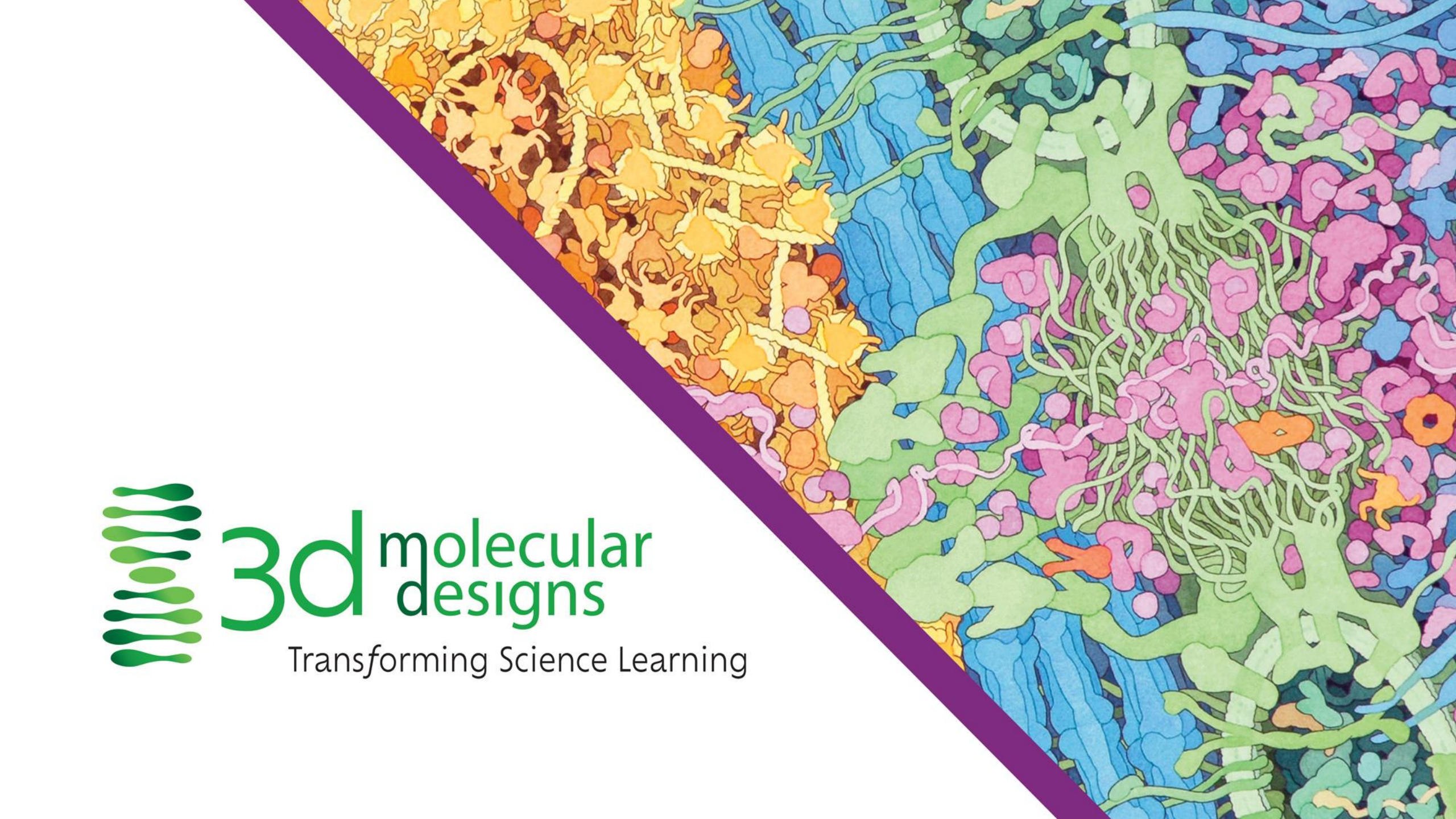

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