NSDAR Educational Resources Committee

"DNA and Genealogy"

Lesson Plan

Contributor: Marcia M. Yates **Grade Level:** High School or Adult Education

1. **Statement of the objective and lesson outcomes**:
   Objective: To provide a basic understanding of human genetics and how currently-available commercial genetics tests can be used in genealogy. Outcome: Understand stated objective.

2. **Materials, resources, and technology to be used by teacher/students**:
   Presentation is in PowerPoint format, and is saved with Notes pages that provide the script for each slide. Requires a projector and either a Mac or PC. Download the PowerPoint here.

3. **Introduction of the topic**:
   This presentation, suitable for use for high school/advanced middle school or adult education, gives a basic understanding of genetics and how genetic evidence can be used in establishing family connections.

4. **Procedure for instruction**:
   The presentation itself contains everything needed -- slides, a script for each slide, and references for attendees interested in learning more. Slides may be freely distributed.

5. **Lesson closure**:
   Invite attendees to learn more by going on to the commercial DNA test websites (Ancestry.com, FamilyTreeDNA, 23andMe, etc.) and by reading the references supplied at the end of the presentation.

6. **Assessment of student understanding**:
   A short, interactive quiz on genetics basics is in the first part of the presentation. Quizzes and tests could be easily developed from the presentation material and references given.

DNA for Genealogy

NSDAR Educational Resources Committee

Marcia M. Yates, M.S.

August 15, 2018

Presentation Author's Bio

- Consultant at Philips Innovation Lab in Cambridge, MA
- Previously Chief Informatics Officer (CIO) at Claritas Genomics, Cambridge, MA
- Formerly held executive positions at PatientsLikeMe, Good Start Genetics, the Whitehead Institute, the Broad Institute of MIT & Harvard, Thomson Financial, Fidelity Investments, others
- 30+ years of experience in high tech, the past 15 years exclusively in biotechnology – learned genetics "on the job"
- Teaches part-time at Boston University
  - Graduate courses in computer science

DNA and Genealogy

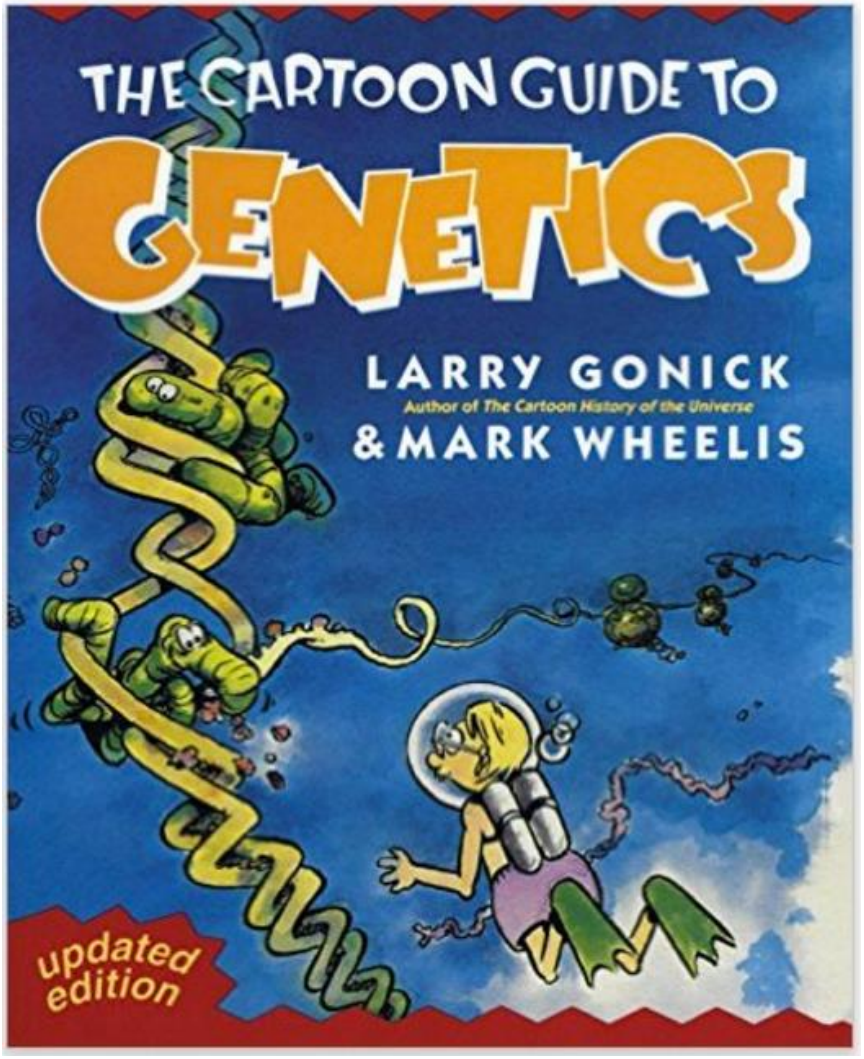
"The body has a long memory indeed. Written in the quirky tongue of DNA and wound into the nucleus of nearly every human cell are biological mementos of the family who came before us. And science is finding ways to dig them out, rummaging through our DNA as if it were a trunk in the attic."

-- Carolyn Abraham (journalist)

Motivation for Presentation

- As of March 2015, the DAR is now accepting genetic testing (Y-chromosome) as supplemental proof of ancestry
- Many people, not just DARs, are interested in their ancestry, hoping to prove/disprove their family lore on country-of-origin and other family myths and legends
- Hope to give some starter background information on the topic – genetics is a large and complex topic, and this presentation only scratches the surface

Highly Recommended!



- This book does a wonderful job explaining the basics
- Very memorable
- Recently revised!
- One example…

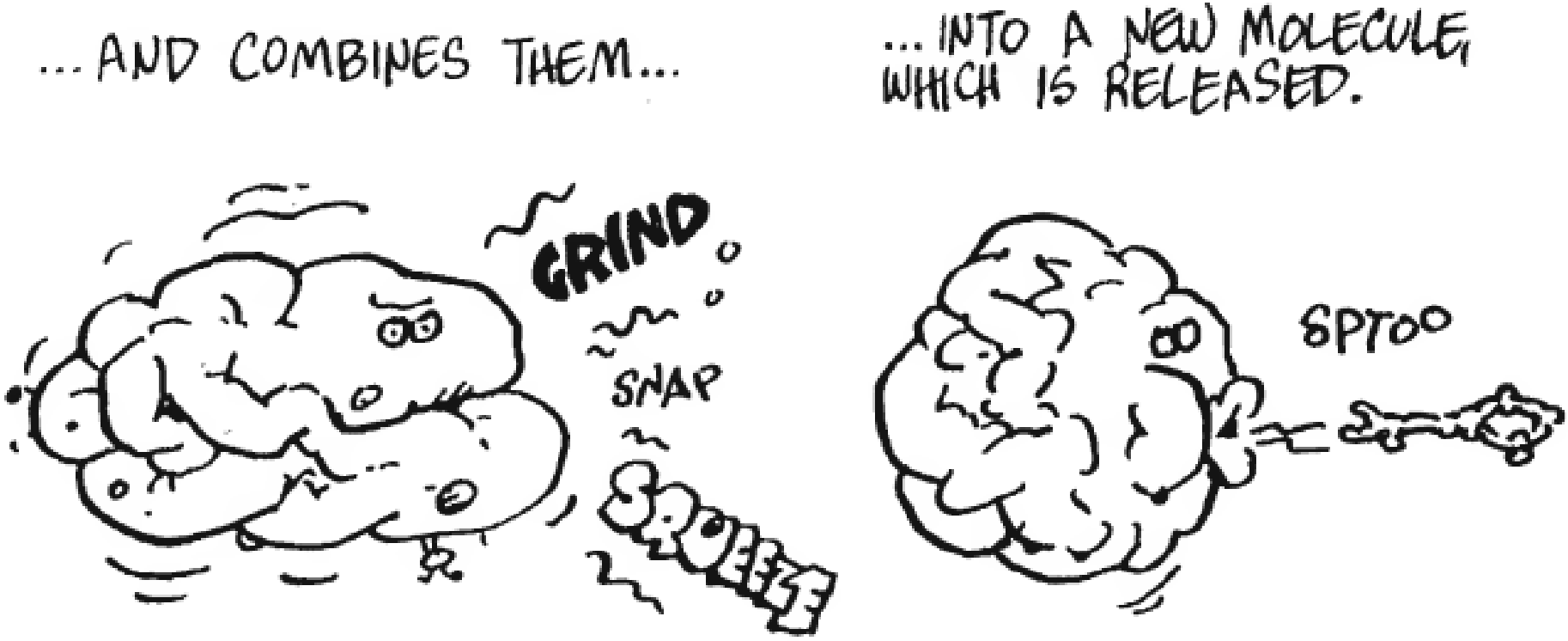
Enzymes – Part 1

ENZYMES ARE PROTEINS WHICH TAKE APART OR PUT TOGETHER OTHER MOLECULES. EACH ENZYME IS RESPONSIBLE FOR JUST ONE SPECIFIC REACTION.

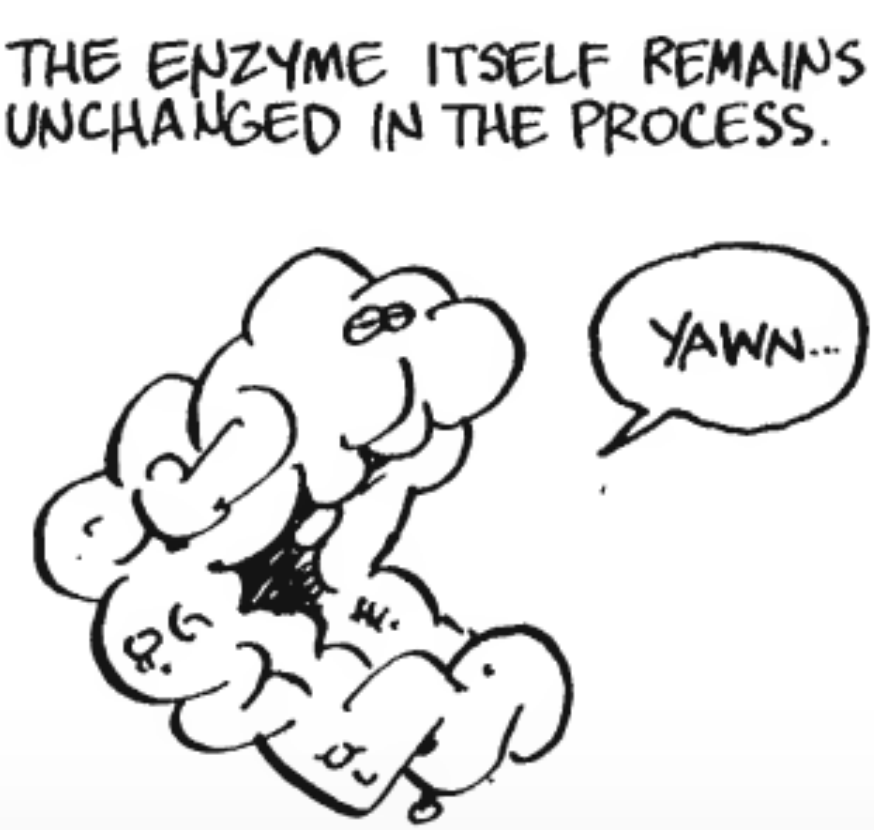
A TYPICAL ENZYME LIES IN WAIT FOR THE RIGHT MOLECULES TO COME AROUND.

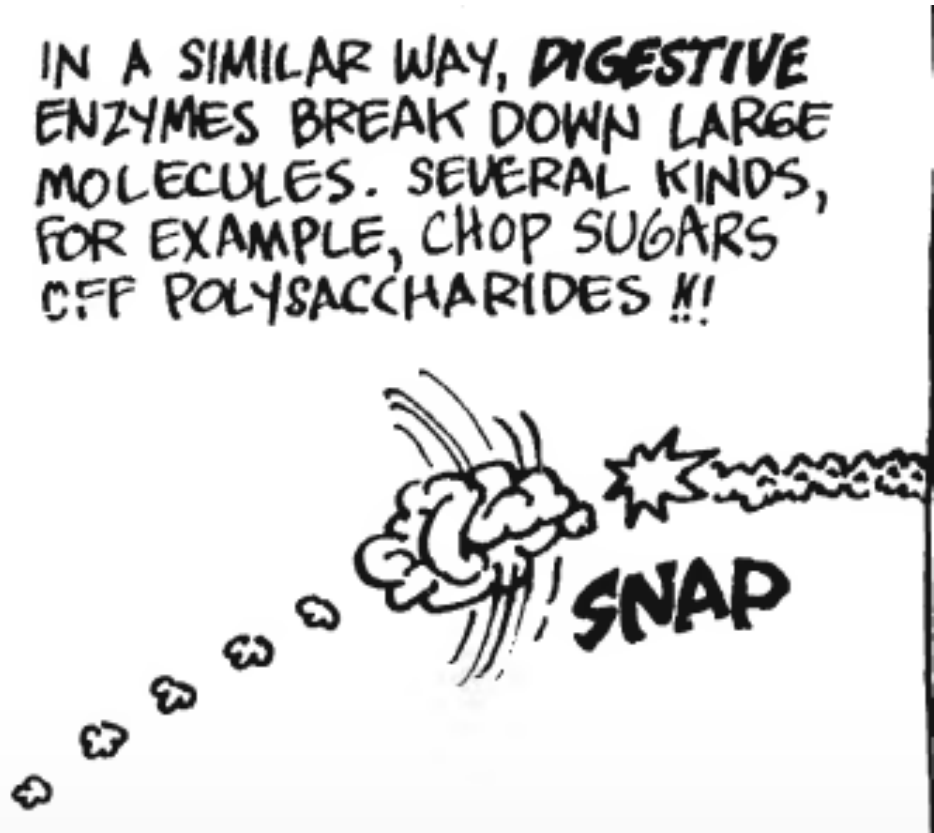
THE ENZYME BINDS TO THE SMALL MOLECULES...

Enzymes – Part 2



Enzymes – Part 3





Topics

- Basic genetics primer
- Genetic variation and inheritance
- Using genetic tests in genealogical research

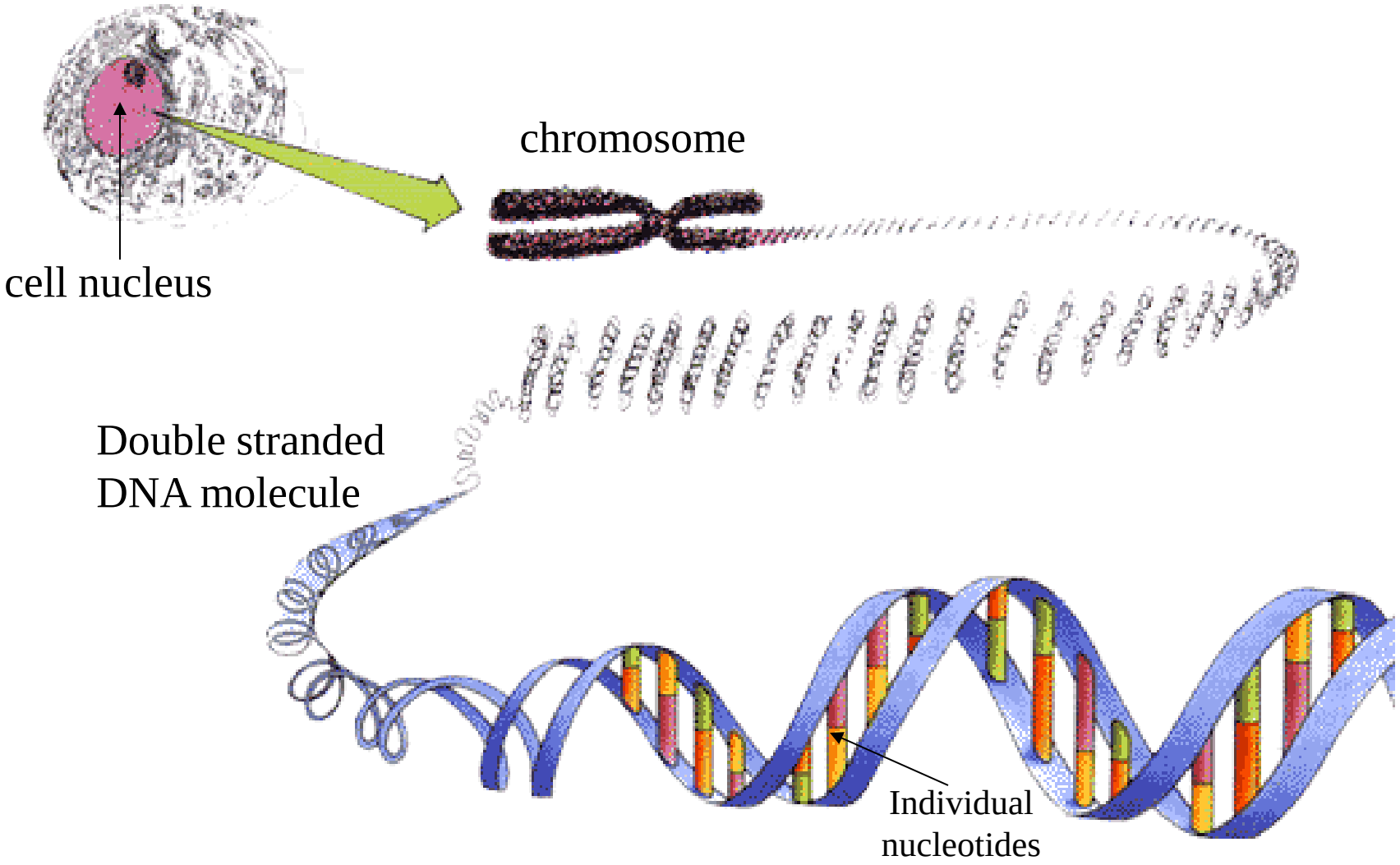
It’s hard to understand how the tests work without the basics…

BASIC GENETICS PRIMER

The Players…in our drama

- In the nucleus of most of our cells is our **DNA**
- Our DNA consists of 46 chromosomes, 23 from mom and 23 from dad; our **genome** is that complete set of DNA
- **Genes** are located on the chromosomes; there are around 25,000 genes in the human genome
- Genes provide the blueprint to create **proteins**
- **Proteins** do the bulk of the work in the cell

DNA in the Cell



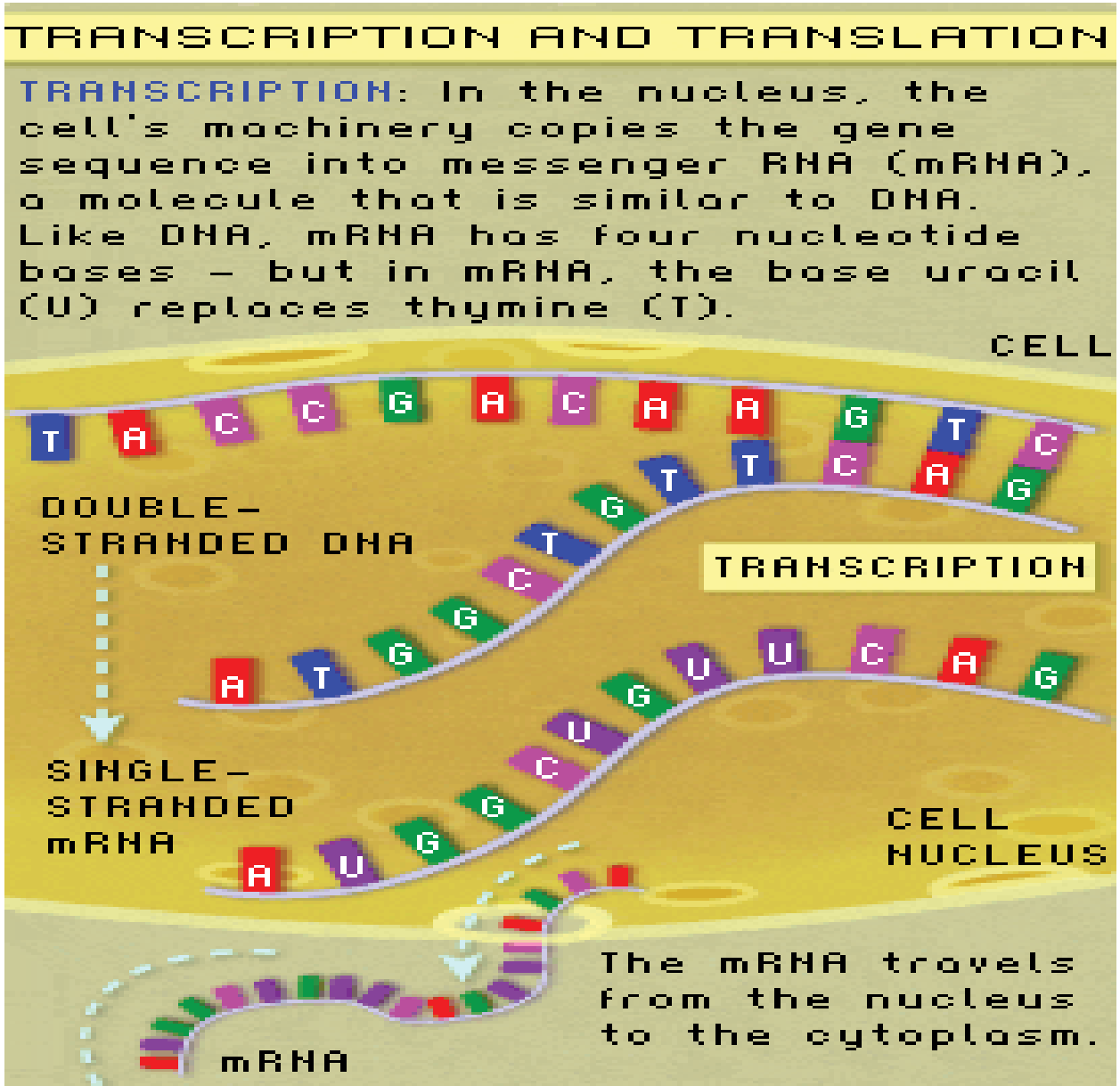
How are proteins manufactured?

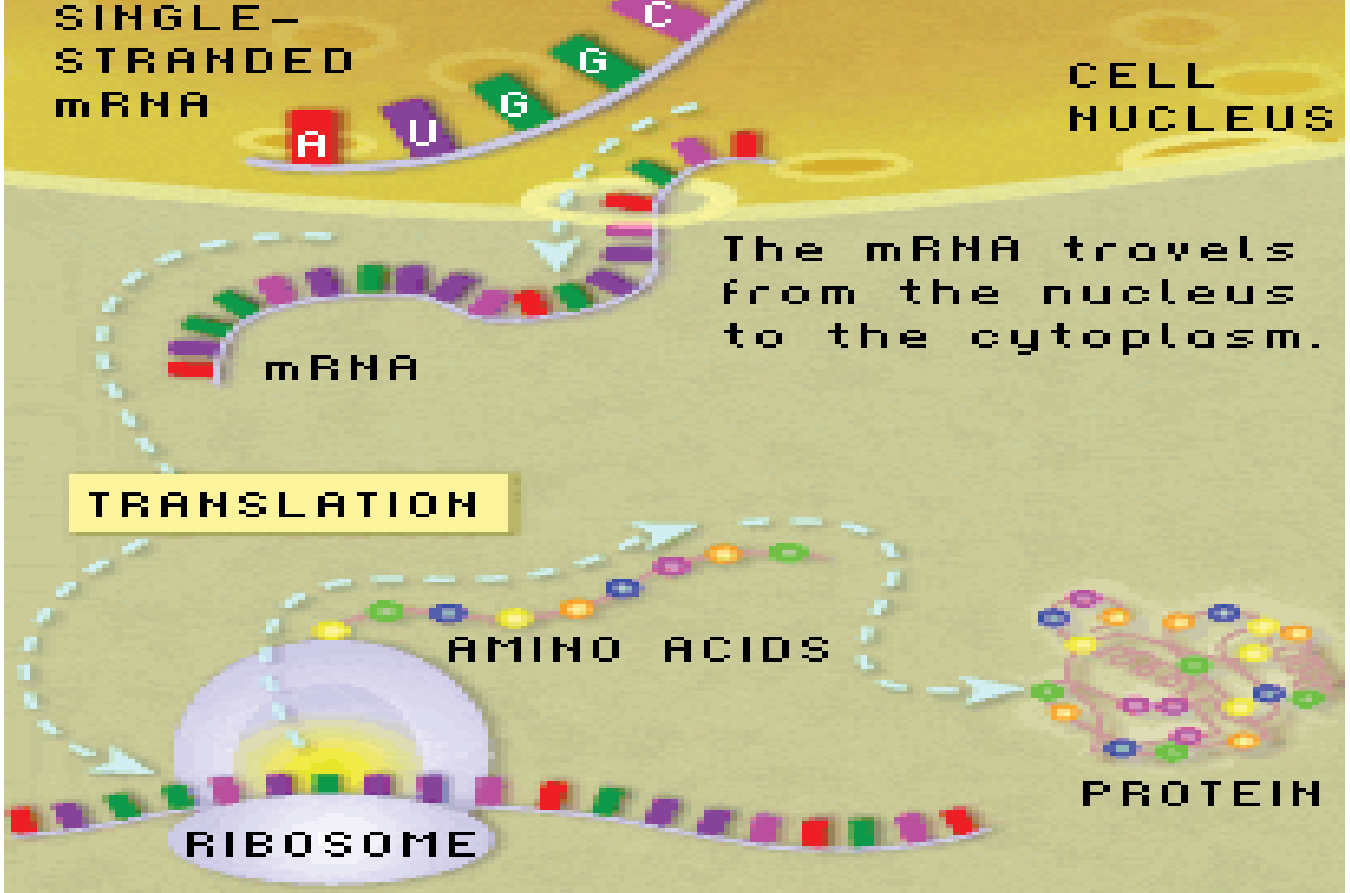
- The DNA in our genes provides the blueprint
- DNA consists of four small molecules called nucleotides, represented by the letters A, C, G, and T
- They form the “rungs” of the DNA helix molecule
- A’s always go with T’s; G’s always go with C’s…

Getting to the Proteins…

- Transcription: The DNA blueprint is used to create **messenger RNA (mRNA)**, which then travels from the nucleus to the cytoplasm of the cell
- Translation: In the cytoplasm, the ribosome uses the mRNA sequence to manufacture the correct **amino acids** from 3-letter sequences called **codons**
- The **protein** is built up from amino acids

DNA and RNA


TRANSCRIPTION AND TRANSLATION
TRANSCRIPTION: In the nucleus, the cell's machinery copies the gene sequence into messenger RNA (mRNA), a molecule that is similar to DNA. Like DNA, mRNA has four nucleotide bases – but in mRNA, the base uracil (U) replaces thymine (T).
CELL
DOUBLE-STRANDED DNA
TRANSCRIPTION
SINGLE-STRANDED mRNA
CELL NUCLEUS
The mRNA travels from the nucleus to the cytoplasm.
mRNA



TRANSLATION: The protein-making machinery, called the ribosome, reads the mRNA sequence and translates it into the amino acid sequence of the protein. The ribosome starts at the sequence AUG, then reads three nucleotides at a time. Each three-nucleotide codon specifies a particular amino acid. The "stop" codons (UAA, UAG and UGA) tell the ribosome that the protein is complete.

DNA and RNA

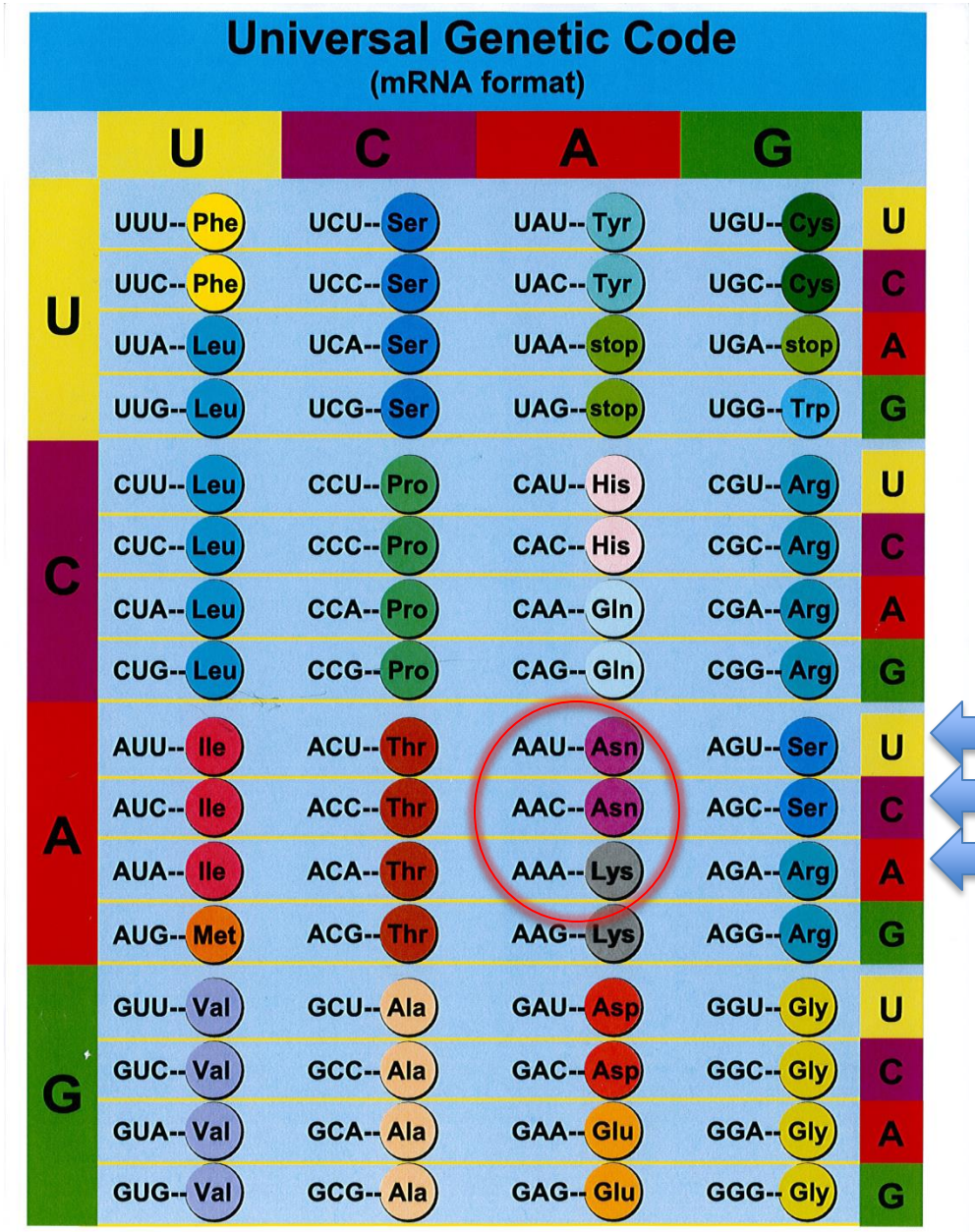
The ribosome is a cell organelle

GENETIC VARIATION AND INHERITANCE

What makes us different from each other?

- Although our human genomes are ~99.9% the same, the .1% differences amount to millions of opportunities for differences!
- A one-nucleotide difference in a coding region is called a **SNP** (single nucleotide polymorphism)
- Most SNPs are benign, *but some can cause disease* – it depends on the protein produced
- Note there are many other types of mutations (deletions, insertions, tandem repeats, other accidents of translation, etc. that might cause disease – or not)

Why are some SNPs important?



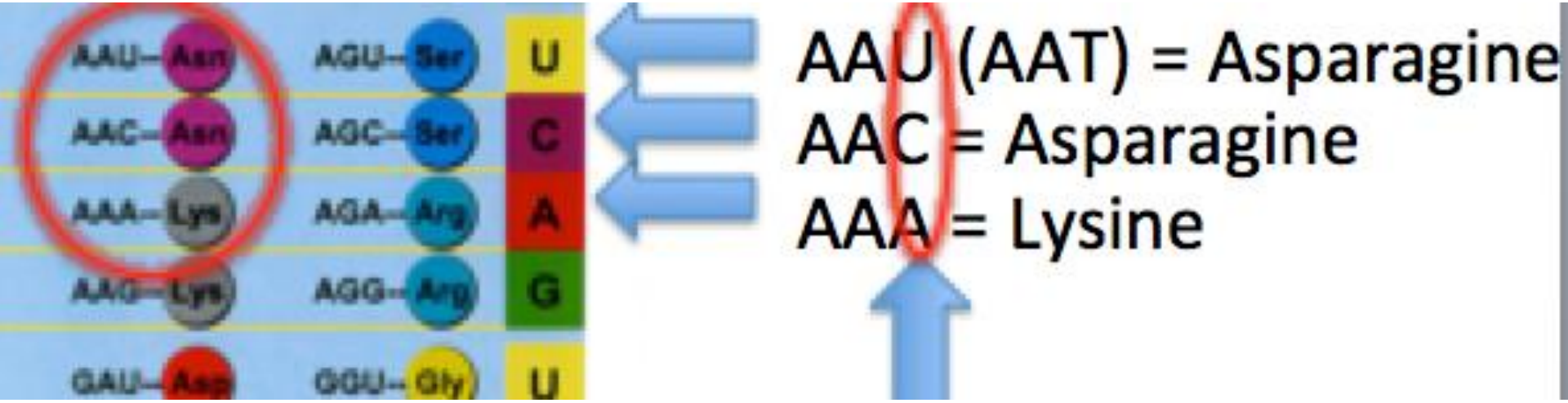
Remember, U (uracil) replaces T (thymine) in the transcription process.

Each three letter codon translates to an amino acid. Proteins are built from amino acids.

There is redundancy – some three letter combinations are aliases for the same amino acid.

AAU (AAT) = Asparagine
AAC = Asparagine
AAA = Lysine

It may or may not matter…



- In this example, two codons, AAT and AAC are *synonyms*; they both code for the amino acid **Asparagine**
- The AAA codon codes for **Lysine**, a *different amino acid*

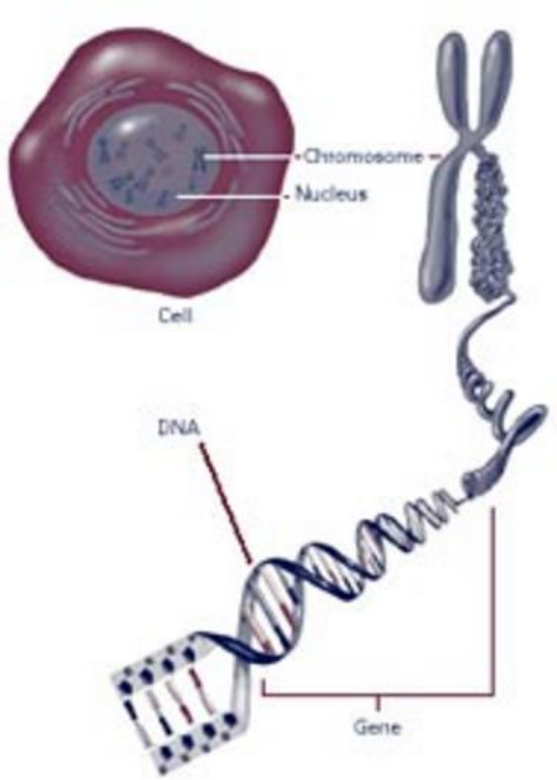
Example – What can go wrong?

AAU (AAT) = Asparagine – Person 1
AAC = Asparagine – Person 2
AAA = Lysine – Person 3

- Let's say AAT and AAC contribute to a normal protein (Persons 1 and 2)
  - Therefore, no negative effect of either allele/variation
- AAA codes for a low-functioning or non-functioning protein (Person 3)
  - Could **lead** to disease, such as sickle-cell anemia
  - Could cause **susceptibility** to disease, e.g., lung cancer

An analogy…

- Chromosomes are like cookbooks
- Genes are the individual recipes
- *Genetic variation* is like substitution of different letters or words in the recipe



Chromosomes are like cookbooks.
And genes are the recipes.

Brief Look at Genetic Variation

- Until recently, most scientists focused on small changes or *variations* within a gene. For example, imagine this set of instructions:
  - *Get a glass. Fill the glass with milk. Add 2 tbsp of chocolate powder. Stir.*
- Here are a couple of single letter differences that completely change the meaning of this recipe:
  - *Get a glass. Fill the glass with **s**ilk. Add 2 tbsp of chocolate powder. Stir.*
  - Get a glass. Fill the glass with milk. Add 2 tsp of chocolate powder. Stir.
  - Now instead of a glass of chocolate milk, you get a glass of chocolate silk. Or in the second case, a very weak glass of chocolate milk.
- This is how DNA works too. A change in a letter can cause a change in how a gene gets used. Or whether it gets used at all.

From: http://genetics.thetech.org/original_news/news38

Copy Number Variation Example

- *Get a glass. Fill the glass with milk. Add 2 tbsp of chocolate powder. Add 2 tbsp of chocolate powder. Stir.*
  - Now you're going to get a stronger chocolate milk. Or imagine the whole recipe just repeats. Now you get twice as much chocolate milk.
- Until a few years ago, scientists thought that most of our differences came from small changes in our DNA. The idea was that every 1000 letters or so, you and I have a different letter. These 6 million differences made me distinct from you.
- But over the past few years, scientists have started to notice changes where big chunks of DNA are repeated. Or missing.
- As they looked harder, more and more of these changes became apparent. As of November 2006, more than 600 of these CNVs had been identified that covered 104 million DNA letters (called bases). That's 4% of our DNA!

From: http://genetics.thetech.org/original_news/news38

Quiz!

1. How many chromosomes come from mom?
2. How many chromosomes come from dad?
3. If a person has two X chromosomes, it that person male or female?
4. What is a SNP?
5. What are the letters that represent the four nucleotides in DNA?
6. What does a codon encode for?

The “short subjects” version

HOW INHERITANCE WORKS

It starts simple…

22 autosomal chromosomes +
X chromosome + mtDNA
From Mom

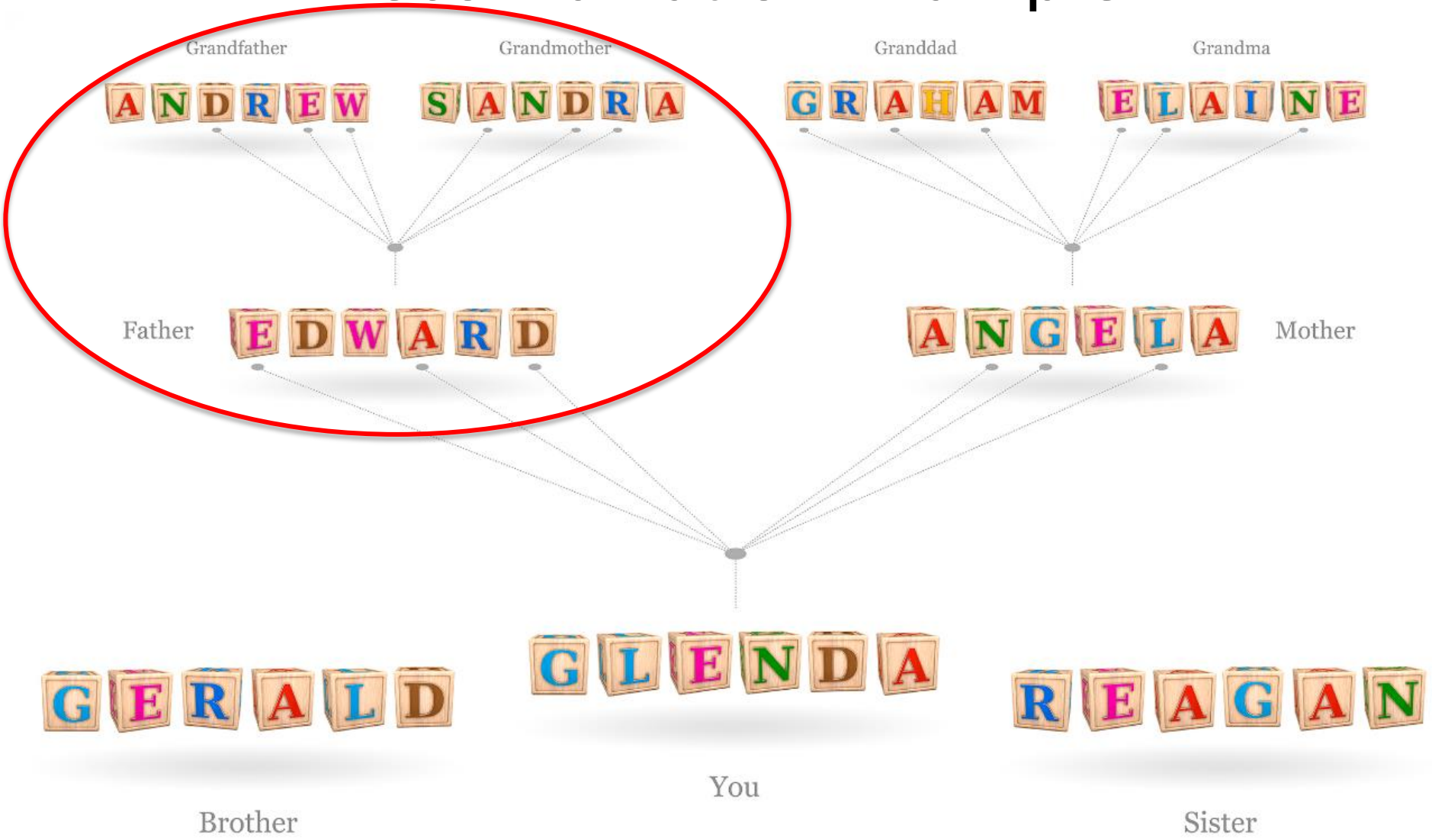
22 autosomal chromosomes +
X chromosome
From Dad

Female child with
44 autosomal chromosomes + mtDNA from mom + X from mom + X from dad =
46 chromosomes + maternal mitochondria DNA (mtDNA)

But it's complicated!

- When the chromosomes combine at meiosis (cell division) to create a zygote, they can exchange genes -- **so pieces of chromosomes from each parent make up the genome of the new offspring**
- The child has approximately 50% of the genetic makeup of each parent, but it's not exact
- These chromosome fracture and assembly events are called **recombination**
  - There are an average of 33 recombination events per transmitted genome!
- Because the recombination changes with each pregnancy, siblings are only about 50% the same
  - Of course, identical twins are the exception

Recombination Example



Recombination

- **Roughly 50% of each parent's chromosomal "chunks" are present in the child**
- **There are an average of 33 to 50 recombination events per transmitted genome**
- **It is not exact; and other *de novo* mutations also enter in**
- **10 generations back would be 1,024 opportunities times the number of recombination events possible, wow!**

Which brings us to…

Therefore….

- **Recombination** is the reason why siblings in a family, if not identical twins, have different *phenotypes*, or appearances
- However:
  - The Y chromosome is very stable and is passed down from father to son
  - Mitochondrial DNA is passed down from the mother
- The X chromosome does try to recombine in women (as they have two X chromosomes)
  - This gets complicated in dealing with men…more to come!

DNA TESTING FOR GENEALOGICAL RESEARCH

There are Four Types of Testing

- Autosomal DNA
- Mitochondrial DNA
- X-Chromosome
- Y-Chromosome

Note that…

- Mitochondrial DNA and Y-Chromosome are considered “lineage markers” because they **do not recombine** and are passed down unchanged (except for mutations)
- Because they do not recombine, they’re not useful for forensics that require direct match
- You cannot separate out direct relatives
  - Y-Chromosome profile can be any male in the family…

It’s all about the matching…

- In resources like Family Tree DNA, there are tools that can help you compare chunks of DNA from any relatives that may have been tested
- Y-Chromosome and mitochondria are relatively stable, while *autosomal* DNA is subject to much change over generations due to recombination
- Let’s quickly look at each of the four types of tests and the information we can learn from them

But…wait…What about the mito?

- Autosomal DNA and the Y and X chromosomes are the “23andMe” chromosomes
- But, what are mitochondria, and why isn’t there a separate mito chromosome?
  - Well, there actually **is** a separate chromosome, it’s just quite different!

Mitochondria

- Classified as an organelle in our cells, called mtDNA or ChrM by geneticists
- Thought to have evolved from early bacteria that were incorporated into our cells
- A fertilized zygote will have ~2,000 mitochondria, nearly all from the mother
  - Thus the *matrilineal inheritance* pattern
- **Mitochondria have their own genome, which can be independently sequenced**
- They are the energy powerhouse of the cell

Mito (cont'd)

- Mutations in mitochondria can result in mild to severe disease, often affecting nervous system and muscle
  - Disease can be severe, as those systems require the most energy
- Inheritance pattern can be seen with:
  - Mothers passing on disease to both sons and daughters
  - The daughters (not the sons) passing disease onto grandchildren
  - Note that some mitochondrial disease is actually X-linked recessive inheritance, including color-blindness and the most common muscular dystrophy
- The mitochondrial or "ChrM" can be traced back through the generations

Genealogical Case Study with mtDNA

- From 1976 to 1983, Argentina's military dictators "disappeared" dissidents and opponents
- Many people were abducted and killed, their children were adopted by childless couples with ties to the police and military
- In 1977, a group of the grandmothers set out to find over 400 missing children; around 120 have been located so far

Case Study (cont'd)

- Tracking down these children required painstaking work with adoption and birth records
- The grandmothers lobbied in the 1980s for a national genetic database to store blood samples of grandparents and other relatives
- A geneticist found a novel way to use mtDNA to identify the children

THE GENEALOGICAL GENETICS TESTS

For each test type, we will ask…

- Who has it?
- Passed on from?
- Where located?
- How many are passed down?
- From whom inherited?
- Who can be tested for it?
- What is reported in the most common “consumer” tests?

Autosomal DNA

- Who has it? Everyone
- Passed on from? Each parent in approx 50%
- Where located? In nucleus of cells
- How many are passed down? One copy of each of the 22 pairs of chromosomes in each cell
- From whom inherited? ½ from mom, ½ from dad, approximately
- Who can be tested for it? Everyone
- What is reported in the most common "consumer" tests? *Length* of matching segments shared between people

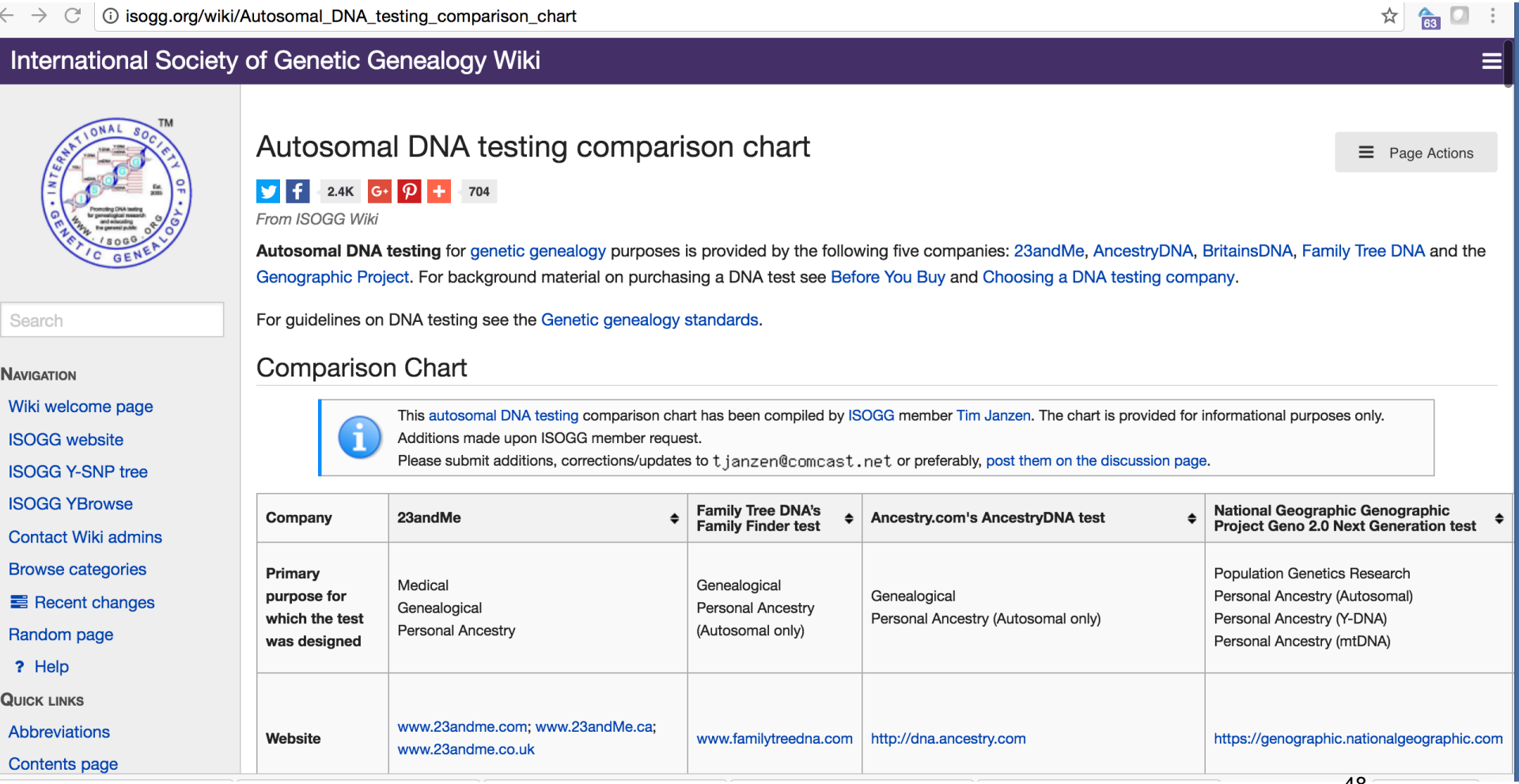
Tests

- These tests can help locate relatives (e.g., cousins) of either sex across your family tree
- Autosomal DNA tests report back matching by length and/or a probability (likely match)
- Similar to forensic cases, where there's a "99.2%" chance *this* one is the perpetrator
- Lengths of DNA that are always 'travel together' are called *haplotypes*; they are used in population genetics, too

Commercial Testing

- There are several companies that do autosomal DNA testing for genetic genealogy:
  - 23andMe
  - AncestryDNA
  - MyHeritage (replaced BritansDNA in the ISOGG chart)
  - Family Tree DNA
  - Genographic Project (National Geographic)
- The ISOGG website is an excellent resource for one-stop-shopping comparison http://isogg.org/wiki/Autosomal_DNA_testing_comparison_chart

ISOGG Wikiweb – Tim Jensen's Comparison Chart



isogg.org/wiki/Autosomal_DNA_testing_comparison_chart

International Society of Genetic Genealogy Wiki

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Autosomal DNA testing comparison chart

Page Actions

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From ISOGG Wiki

Autosomal DNA testing for genetic genealogy purposes is provided by the following five companies: 23andMe, AncestryDNA, BritainsDNA, Family Tree DNA and the Genographic Project. For background material on purchasing a DNA test see Before You Buy and Choosing a DNA testing company.

For guidelines on DNA testing see the Genetic genealogy standards.

Comparison Chart

This autosomal DNA testing comparison chart has been compiled by ISOGG member Tim Janzen. The chart is provided for informational purposes only.
Additions made upon ISOGG member request.
Please submit additions, corrections/updates to tjanzen@comcast.net or preferably, post them on the discussion page.

Company	23andMe	Family Tree DNA's Family Finder test	Ancestry.com's AncestryDNA test	National Geographic Genographic Project Geno 2.0 Next Generation test
Primary purpose for which the test was designed	Medical Genealogical Personal Ancestry	Genealogical Personal Ancestry (Autosomal only)	Genealogical Personal Ancestry (Autosomal only)	Population Genetics Research Personal Ancestry (Autosomal) Personal Ancestry (Y-DNA) Personal Ancestry (mtDNA)
Website	www.23andme.com; www.23andMe.ca; www.23andme.co.uk	www.familytreedna.com	http://dna.ancestry.com	https://genographic.nationalgeographic.com

Determining a Match

- Each company sets its own thresholds for what they determine is a match
- A match is asserted by concluding that two individuals likely inherited their ½ identical matching segments from a recent common ancestor (compare # of SNPs per length of DNA)
- Phasing helps improve accuracy and eliminate false positive matches
  - *Phasing* assigns the alleles (variations) to maternal or paternal chromosomes
  - Ancestry.com does phasing at this time; Family Tree DNA may add the feature in the future

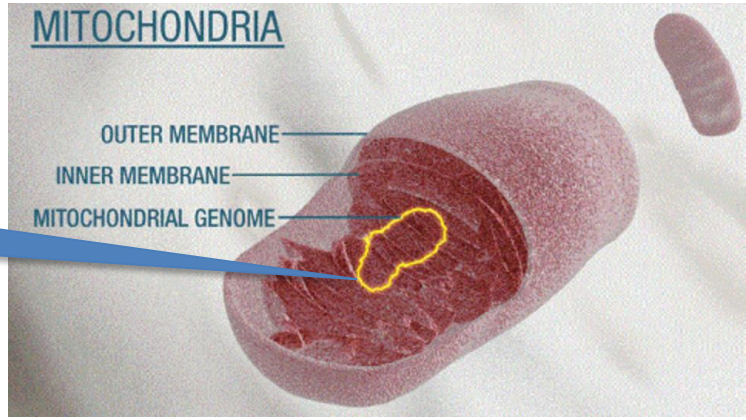
- In the ISOGG comparison (just a few points):
  - 23andMe has the largest dataset
  - Info on lengths and SNPs in matching segments and matching criteria are given by 23andMe and Family Tree DNA, not given by ancestryDNA
  - 23andMe has the best biogeographical ancestry analysis, Family Tree DNA is runner-up
  - All provide some way to contact others you match
- **However, there isn't a common database!!!**
  - You need to be tested by each company to have maximum possibility for a match
  - There are tools you can use to import all your results so you can look at them in one place
  - Some companies may offer import of others' results…

Mitochondrial DNA

- Who has it? Everyone
- Passed on from? Mother, to children of either sex
- Where located? In cells, *outside* of nucleus
- How many are passed down? Multiple copies in each cell
- From whom inherited? The mother to all children
- Who can be tested for it? Everyone
- What is reported in the most common "consumer" tests? The actual bases (C,G,A,T) found; there are only about 16,000 locations, each is important; two regions are analyzed, plus additional SNPs

What is the Mitochondrial Test?

Mitochondria have their own genome

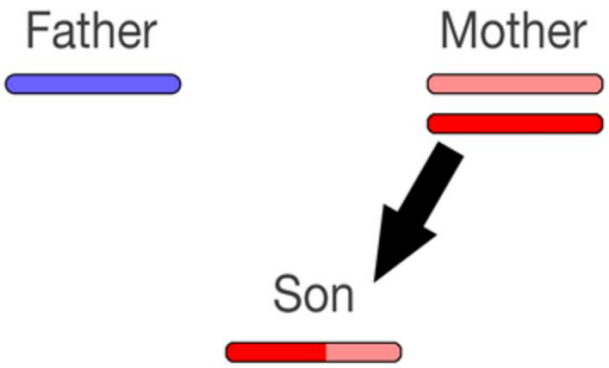


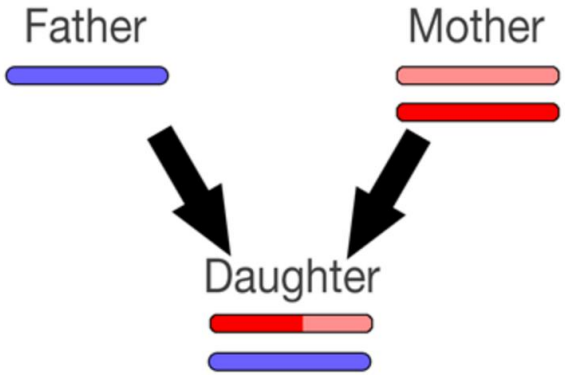
- The mitochondria are passed down relatively unchanged through the maternal line
- The small, separate genome is tested at two regions, and sometimes additional SNPs are included
- Assignment is made to a pool of relatives through these results – called a “matrilineal haplogroup”

X-Chromosome

- Who has it? Men have one, women have two
- Passed on from? Mother to each child; father to daughters only
- Where located? In nucleus of cells
- How many are passed down? One for men, two for women
- From whom inherited? Men get 1 chromosome from mother only; women get 1 chromosome from each parent
- Who can be tested for it? Everyone – but men have only a maternal contribution
- What is reported in the most common "consumer" tests? *Length* of matching segments shared between people, sometimes including actual SNPs

Men have a maternal contribution? What's that mean?



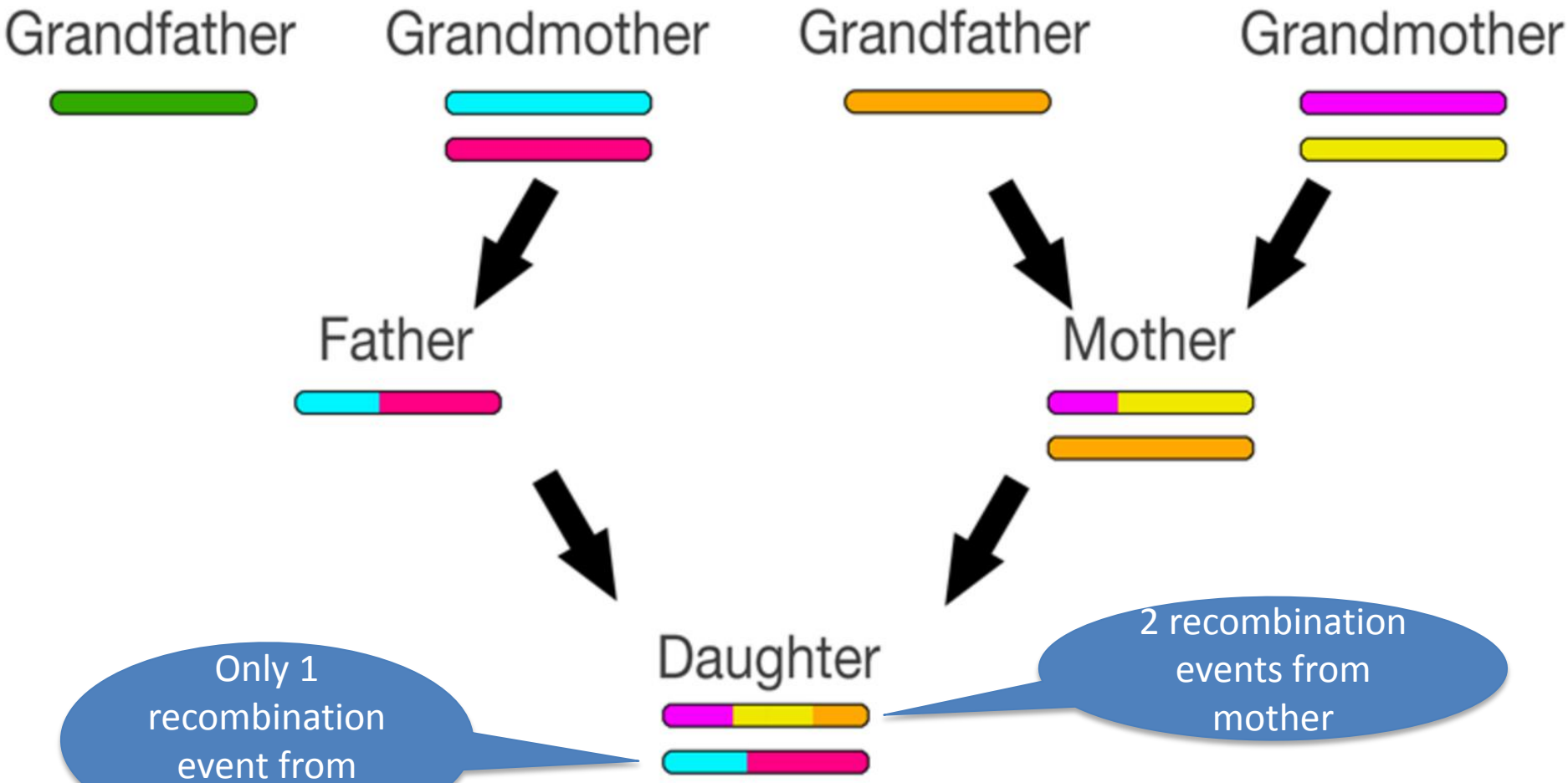


This is only one example, and ONLY the X chromosome is shown
The son has only one X chromosome, composed of parts of the mother's two X's in this case

The daughter has two X chromosomes, she receives the father's X (in blue) and the amalgam of the mother's two X chromosomes as the second X

http://smithplanet.com/stuff/x-chromosome.htm

Just one more generation, and it's already complicated! (Only X Chr shown)



If this daughter has a son, a mix of both chromosomes could become his X

Y-Chromosome

- Who has it? Men only
- Passed on from? Father only
- Where located? In nucleus of cells
- How many are passed down? 1 chromosome to men only
- From whom inherited? Father to son only
- Who can be tested for it? Men only
- What is reported in the most common "consumer" tests? STRs (short tandem repeats) or SNPs in the more recent types of tests

Why is the Y so useful?

- Because the Y is passed down from father to all sons the same way as the surname
- Helpful for genealogists tracing a surname; it can give definitive answers when there isn't paperwork to establish a link
  - Grandson and grandfather will have same Y
  - A nephew will have same Y as his uncle and that man's sons
- **Testing is therefore excellent for identifying distant relationships, as comparison can be done along the entire male line**

Aside -- Who's Your Daddy?

- When the author of this presentation worked at the Broad Institute, we did many disease association studies that incorporated trios – a mom, dad, and kid
  - Trios with affected children were one cohort
  - Trios with unaffected children were the control cohort
  - The two cohorts are statistically compared, to look for variations potentially responsible for disease
- **By running relatedness tests, we concluded that ~11% of the time "daddy wasn't daddy" – so, we had to remove those trios from the study**
- We also eliminated other unknown relatedness through the *identity by descent (IBD)* test
  - This is very common in founder or isolated populations
  - Or places where there is high degree of consanguineous marriage, as in Saudi Arabia

FamilyTreeDNA

What is the Y-DNA 37 test?

- The “37” refers to 37 different marker comparisons
  - A *marker* is a gene or DNA sequence with a known location on a chromosome
- The DAR only accepts the Y-DNA 37 test as DNA evidence, see http://www.dar.org/national-society/genealogy/dna-and-dar-applications
- For a DAR application, women must submit Y-DNA results from two test subjects as specified in the *Guidelines for Using DNA Evidence for DAR applications*

FamilyTreeDNA: Understanding the Y-DNA37 Results

- Test can help you find cousins along your direct paternal line
- Results are reported as STR markers; these values change very slowly from generation to generation
- The exact matches can either be one generation or hundreds of years in the past (!)
- FamilyTreeDNA compares your results with 100's of other men in their database

Your Process: How To's

- First, trace at least two male lineage descendants of one man using traditional genealogical research methods
- The descendants then test their Y-DNA
- If the descendants are exact matches that is evidence that supports the relationship
- Not matching usually disproves the relationship

Genealogical Case Study: Thomas Jefferson

- The most famous Y-DNA case in recent history is the continuing controversy over whether Thomas Jefferson fathered some or all of his slave Sally Hemings's children
- The Monticello website gives both perspectives on the story, but because there were ~25 males in the Jefferson line in the vicinity of Monticello, Y-DNA alone only showed that some Jefferson male fathered the youngest of Hemings's children

Parting Shot – What about Genetic Ancestry Tests?

- Scientists argue that these tests are often so unreliable and inaccurate that they amount to "genetic astrology" http://www.medicaldaily.com/dna-ancestry-tests-are-meaningless-your-historical-genealogy-search-244586
- The same history you get could be given to thousands of others with similar ethnic background; many interpretations are possible
- Once you go back a few generations, specifics are lost and we are all more similar to each other than to a specific ancestor

Population Facts & The Individual

- There is just too much migration and intermixing historically to sort this out
- People descended from more isolated populations (e.g., Scottish highlands) may have minor differences, but not enough to identify a "Scottish gene"
- Nearly every Briton, for example, is "a descendant of Viking hordes, Roman legions, African migrants, Indian Brahmins, or anyone else they fancy"

http://www.medicaldaily.com/dna-ancestry-tests-are-meaningless-your-historical-genealogy-search-244586

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A few supplemental slides…

WHAT ARE THE CAUSES OF GENETIC DISEASE?

What causes disease?

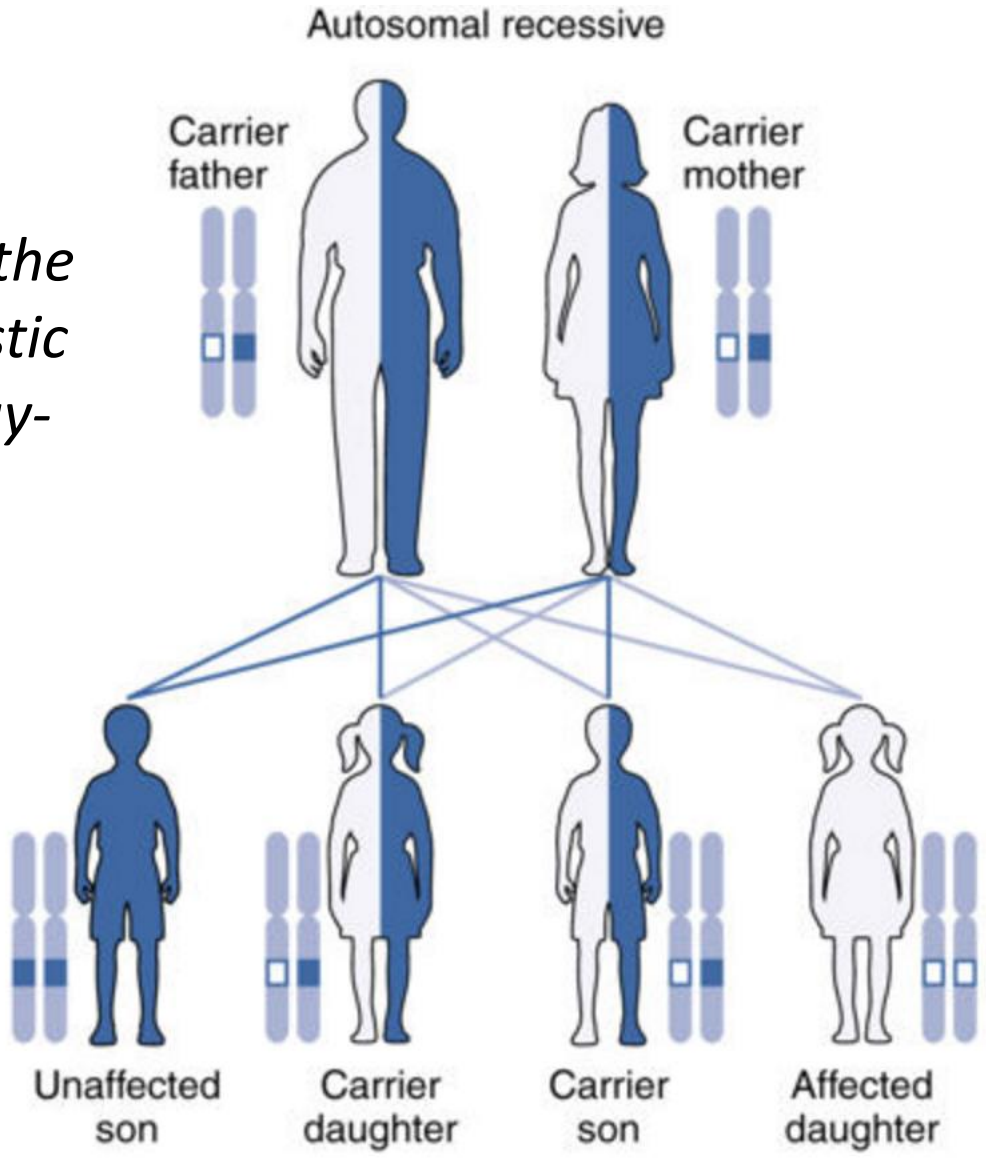
- There are some diseases, like Huntington's, that are due to a particular mutation; if you inherit that mutation from either parent, you are doomed
- Most disease, however, is complex:
  - Potentially many different genomic differences
  - Environmental factors or epigenetic effects
  - Lifestyle, e.g., lack of exercise, obsesty, etc.
  - Just plain bad luck

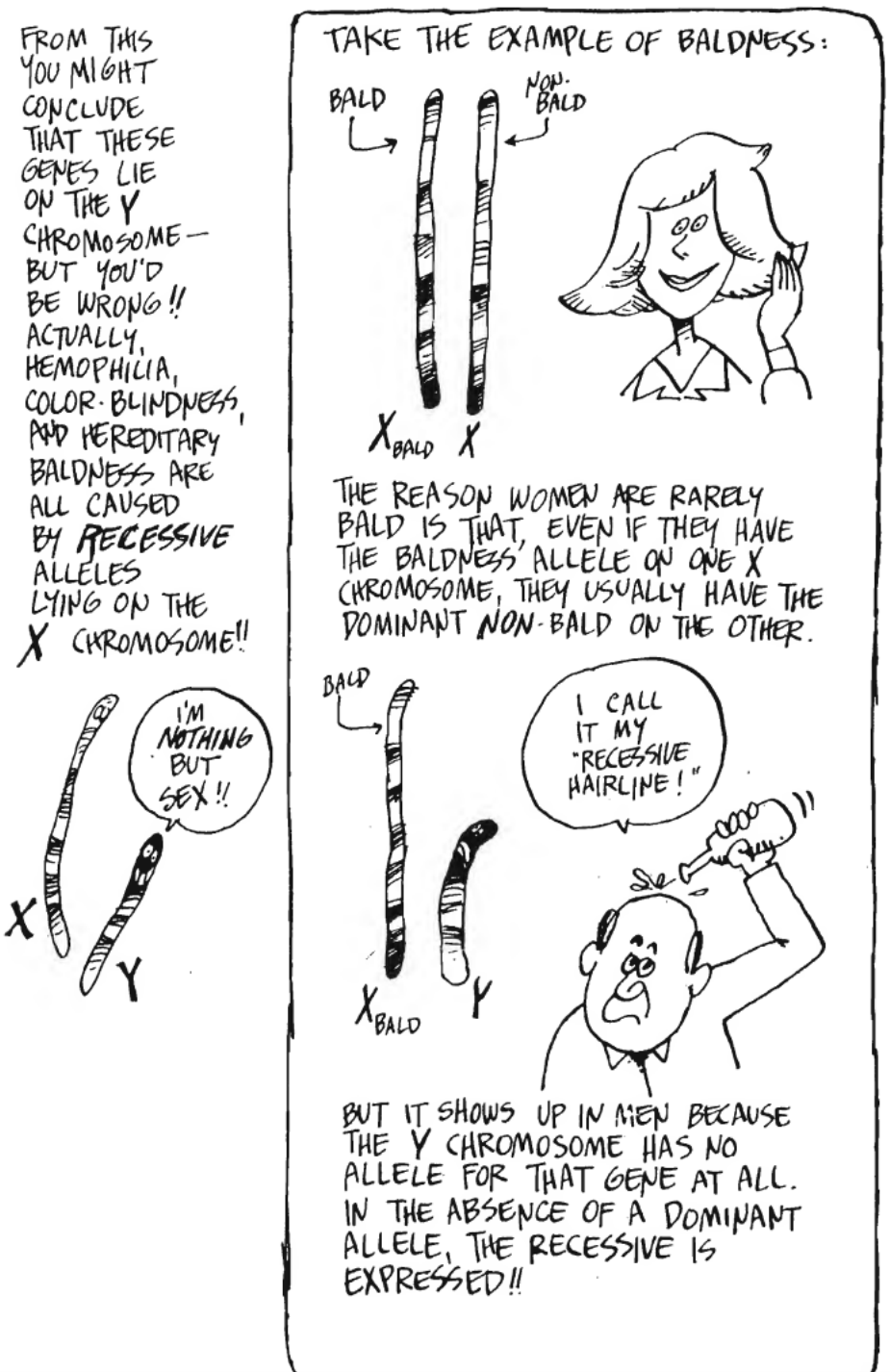
Inheriting Disease

- Understanding one's potential for developing disease is one reason why people want to sequence their genome
- There are two types of inherited Mendelian disease – *dominant* and *recessive*
- With dominant disease, inheriting the defective gene results in the disease. With one affected parent, each offspring has a 50% chance of inheriting the disease (Huntington's is an example)
- Recessive disease requires a causal variant (mutation) from each parent…

Autosomal Recessive Inheritance

Recessive inheritance is the pattern for cystic fibrosis and Tay-Sachs disease





Expression of alleles