

**2023 BLACK HISTORY MONTH
VIRTUAL GENEALOGY CONFERENCE**

“NO LONGER FORGOTTEN”

Introduction to DNA

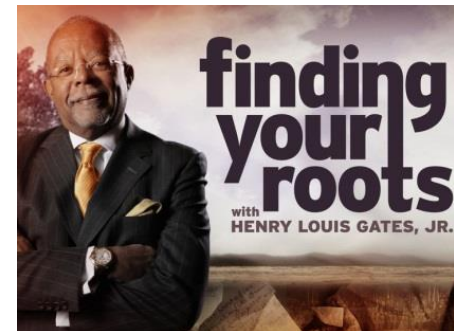
February 11, 2023
Andrew Hochreiter



“No Longer Forgotten” by DNA

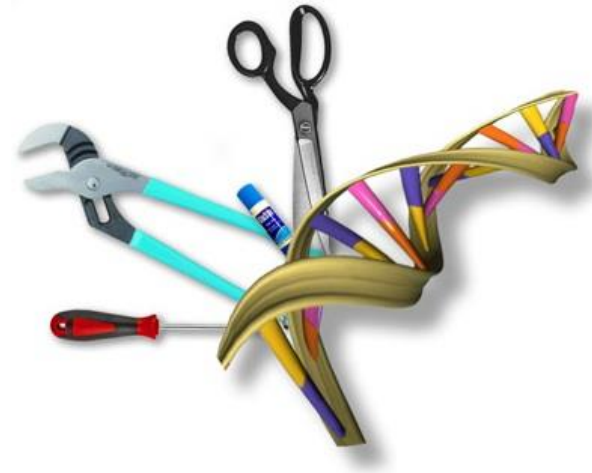
- “Wherever you go in the history of America, there have been Black people making contributions, but their contributions have been **obscured, lost, buried.**”
- “If you don’t tell your stories, other people will tell their story about you. It’s important that we **nurture and protect these memories.** Things change. Existence means change.”
- “My goal is to get everybody in America to **do their family tree.**”
- “There are just so many stories that are **buried on family trees.**”

— Henry Louis Gates Jr.



Powerful Tool for Genealogy

- **Genealogy research:**
 - Study of Family History
 - Identify Kinships & Pedigrees
 - Traditional Research Tools:
 - Records & Documentation
 - Oral Interviews
- **Genetic Genealogy is latest tool**
 - **Genetic genealogy** is the application of genetics to traditional genealogy.
 - Genetic genealogy uses genealogical **DNA testing** to determine the level and type of the **genetic relationship** between individuals

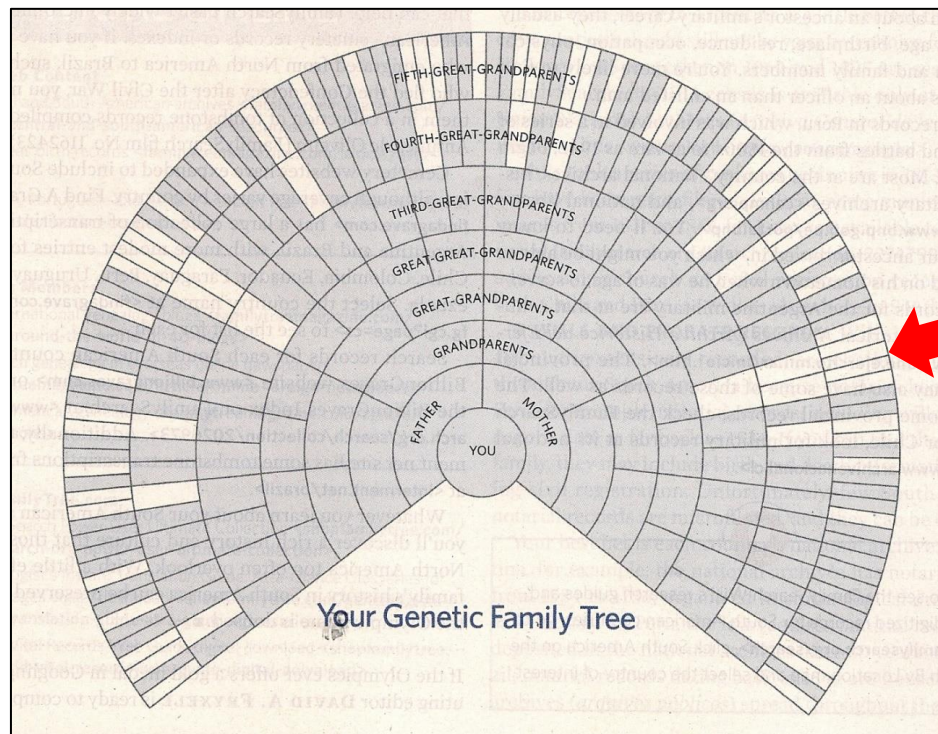


Your 2 Family Trees

Genealogical Tree: all your ancestors

Genetic Tree: ancestors whose DNA you inherited

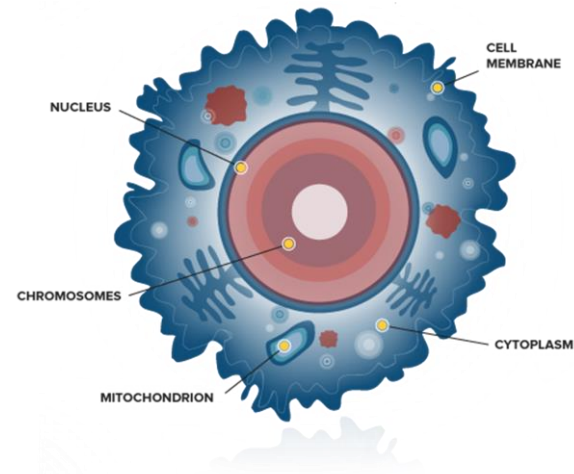
- Your Genetic Tree is a sub-set of your Genealogical Tree
- Siblings have same Genealogical Tree but different Genetic Trees



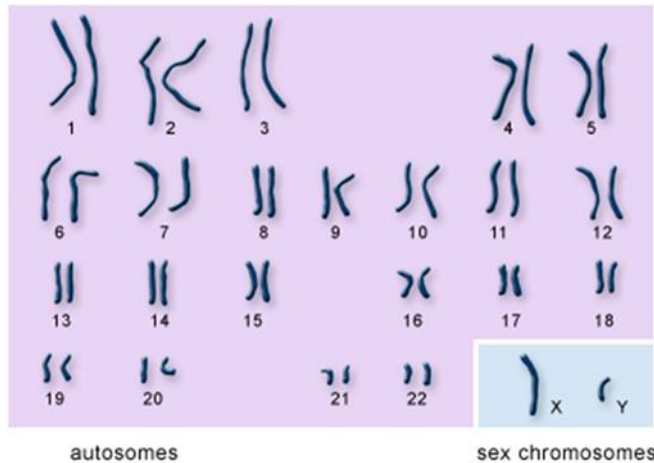
Ancestors
“drop off” the
Genetic Tree

Cells & Chromosomes

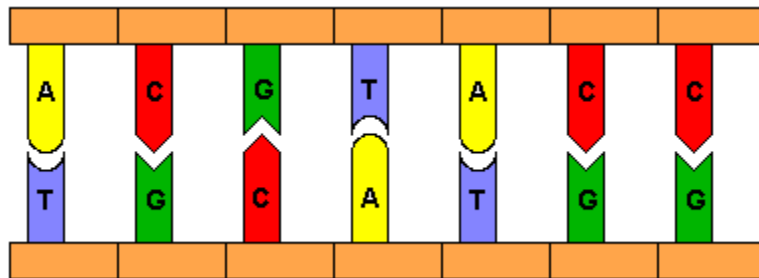
- Human Body contains ~ 37.2 trillion cells
- Humans are **99.9%** genetically identical
 - All variations from **.1%** difference
- Cell Nucleus (chromosomes) and Mitochondria
- Four Tests for DNA
 - **Autosomal Chromosomes**
 - **Y Chromosome**
 - **X Chromosome**
 - **Mitochondrial**



DNA Base Pairs



U.S. National Library of Medicine

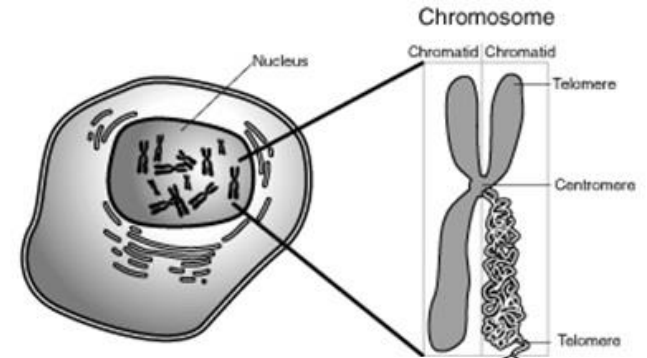


There are four bases:

Adenine & **T**hymine

Guanine & **C**ytosine

Only connect with their partner



Use of DNA in Genealogy

- **DNA tests** can be used by genealogists to:
 - **Link specific individuals** - Test to see whether you and another person may be cousins who descend from a common ancestor
 - **Prove or disprove the ancestry of people sharing the same last name (or NOT)** - Test to see if males carrying the same surname are related to each other
 - **Map the genetic origins of large population groups** - Test to see what geographical origins or ancestry you have
 - **Determine Admixture** – Test to see what Ethnicity percentages you have



DNA Test Concerns

- DNA Testing and Research can reveal:
 - **Family Secrets** (illegitimacy/Adoption)
 - Unexpected **Relationships**
 - Unexpected **Ethnic Makeup**
 - **Medical** Conditions/Preconditions
- Sharing can expose **Personal data**
- Your DNA results can **Inform on Others**



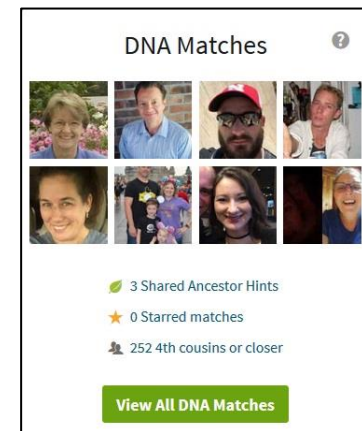
What You Get from DNA

- **Raw Data**

- Your base pair values

rsid	chromosome	position	allele1	allele2
rs4477212	1	82154	T	T
rs3131972	1	752721	G	G
rs12562034	1	768448	G	G
rs11240777	1	798959	A	G
rs6681049	1	800007	C	C
rs4970383	1	838555	C	C
rs4475691	1	846808	C	C
rs7537756	1	854250	A	A

- **Ethnicity / Admixture / Deep Ancestry**



- **Relative Connections (Matches)**

- Relationships back along any Family Tree branch
 - Access to contact information for genetic relatives in the company database (usually up to 6Cs)



Testing Companies

There is **NOT A BEST** testing company:



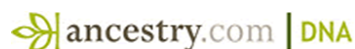
FTDNA

Good for serious genetic genealogists, Y-DNA and mtDNA tests, and X-DNA matching



23andMe

Good for genetic health screening and comparing results with useful analysis tools



AncestryDNA

Good for cousin matching, comparing family trees and assessing many geographic regions for ethnicity



MyHeritage

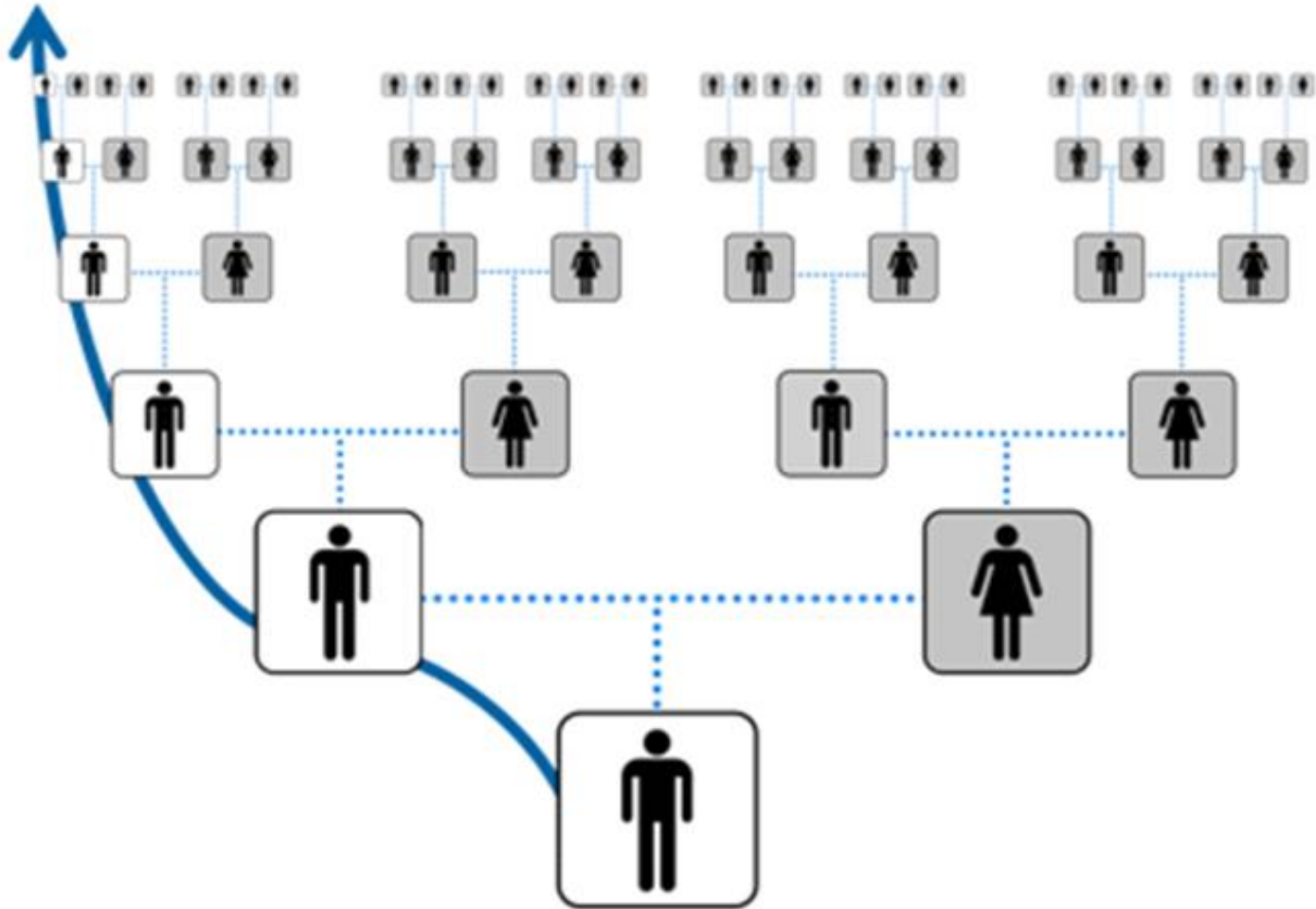
Good for European roots & records, excellent triangulation & autosomal tools



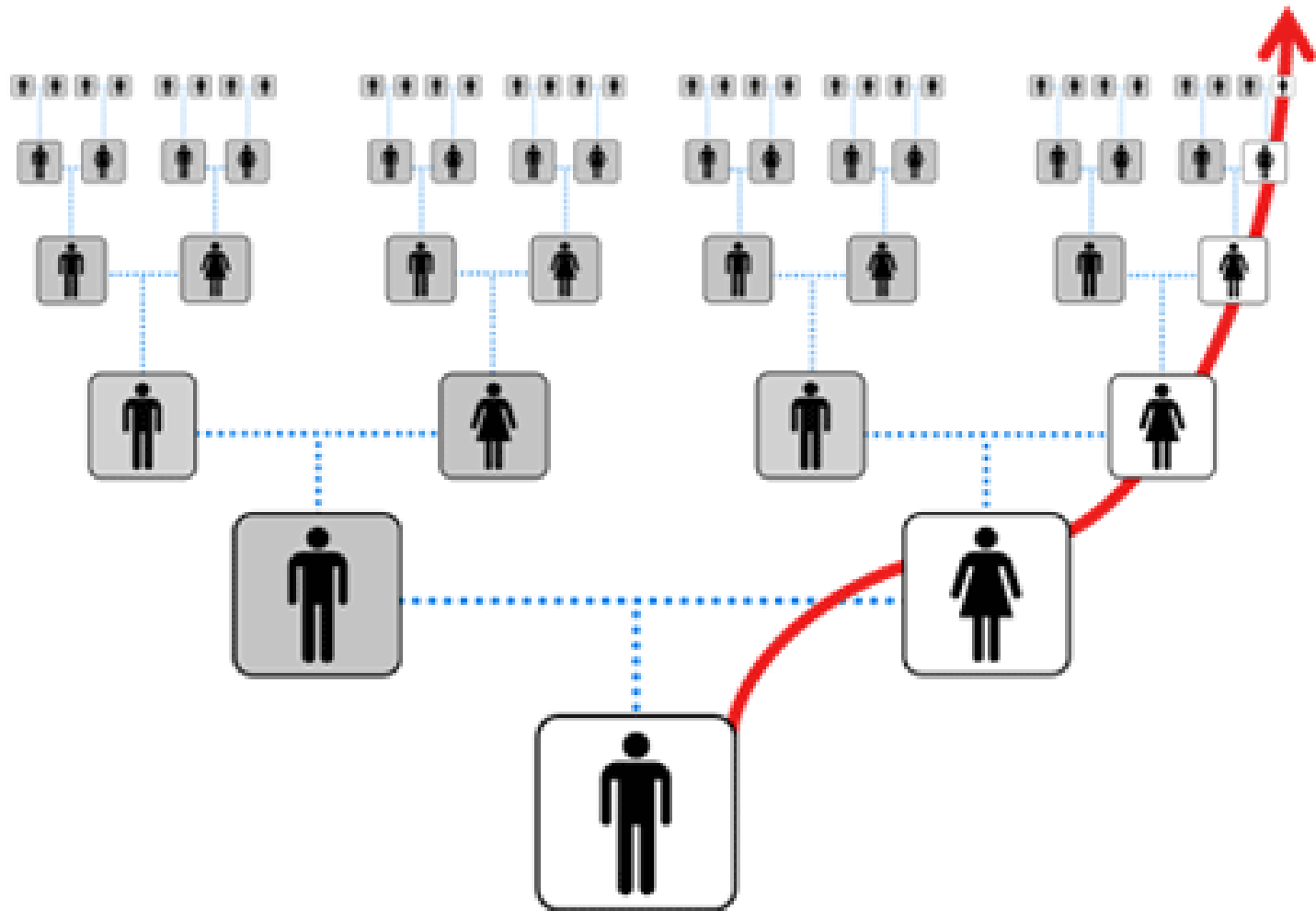
LivingDNA

Good for British ancestry, Ethnicity based on POBI Project

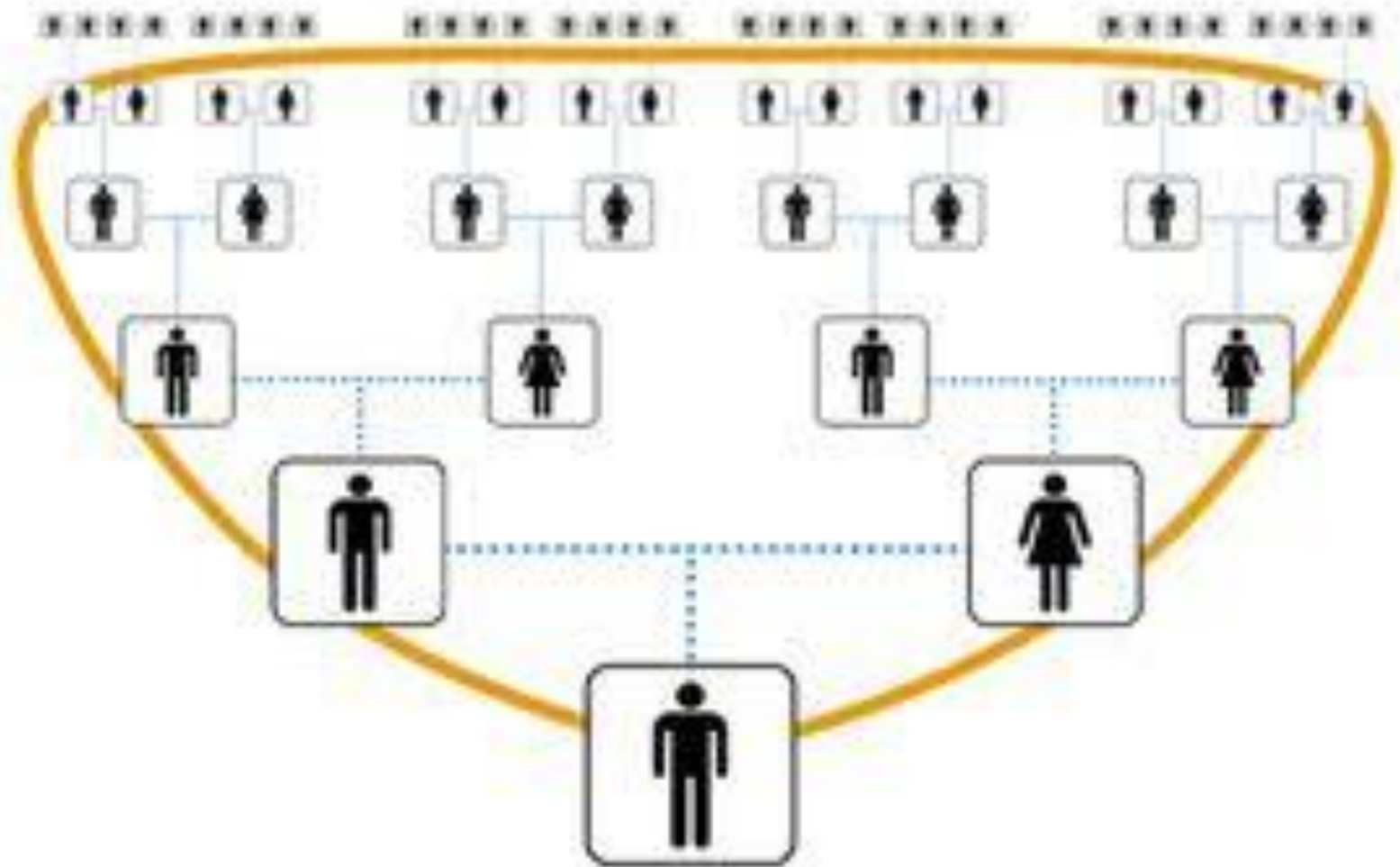
Y- DNA Pattern



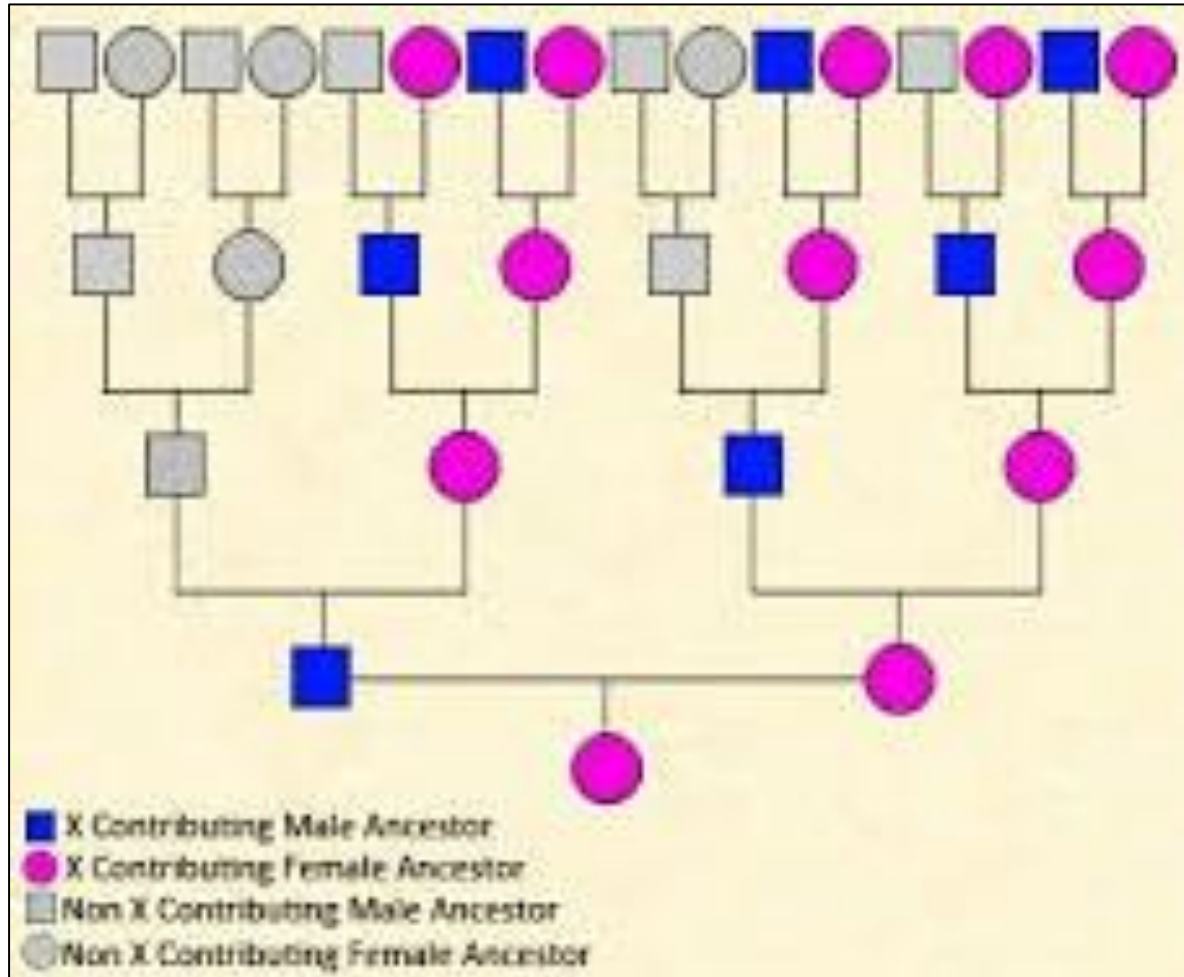
mtDNA Pattern



atDNA Pattern



X-DNA Pattern



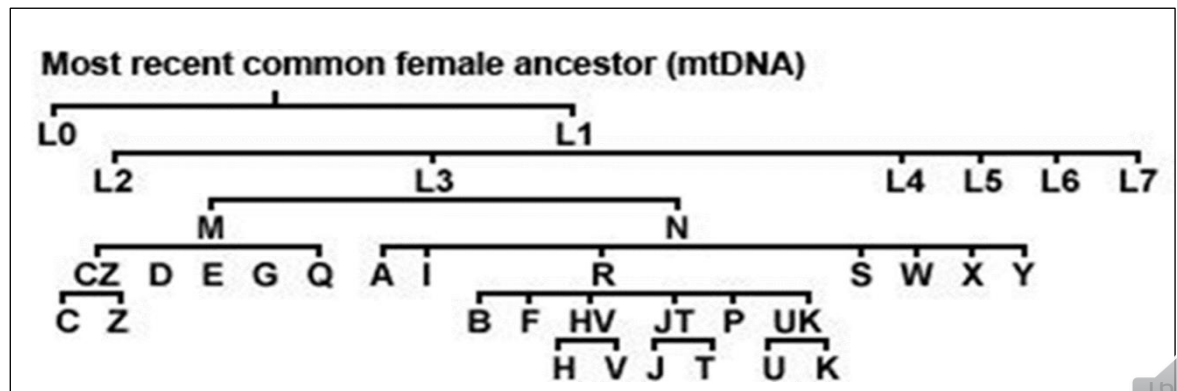
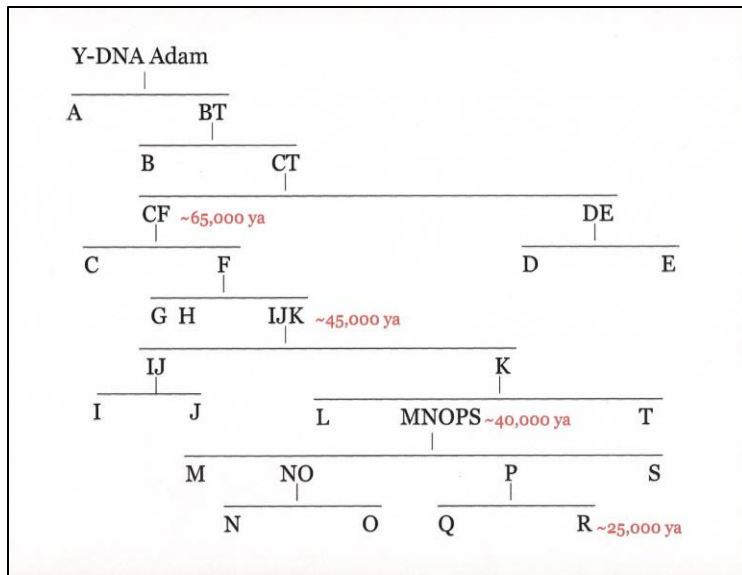
Haplogroups

- Genetic group of people who share a **common ancestor** on patrilineal or matrilineal line
- **Y-DNA & mtDNA Haplogroups are different**
- Haplogroups defined by **Mutations**
 - All descendants will carry that mutation
 - Main branch has sub-branches or “**subclades**”
- Haplogroups reveal **ancient origins**



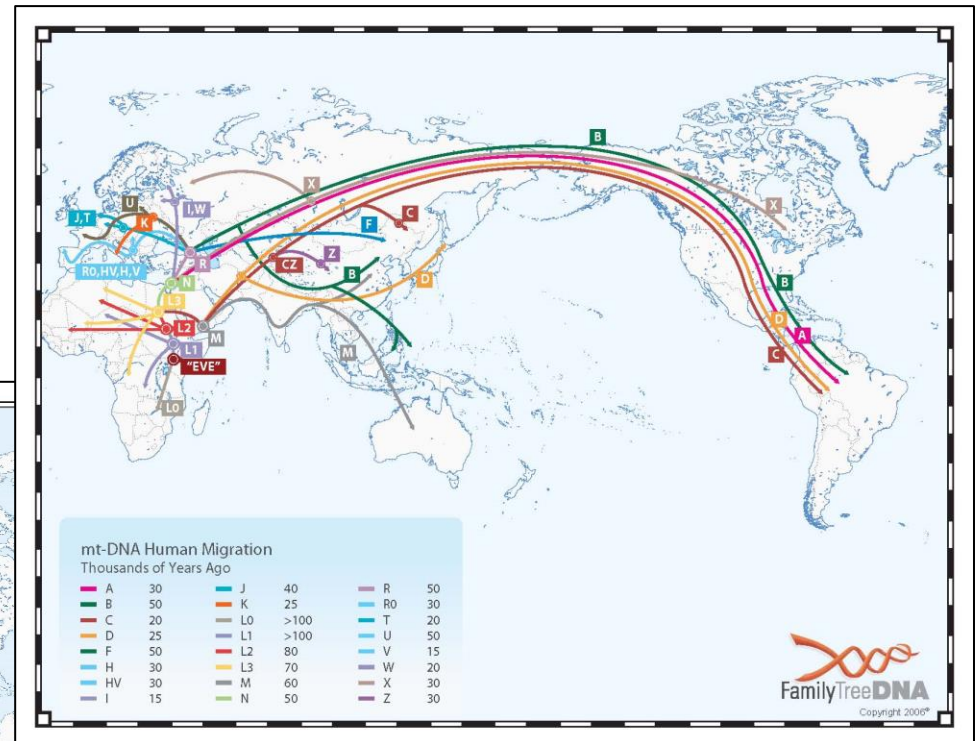
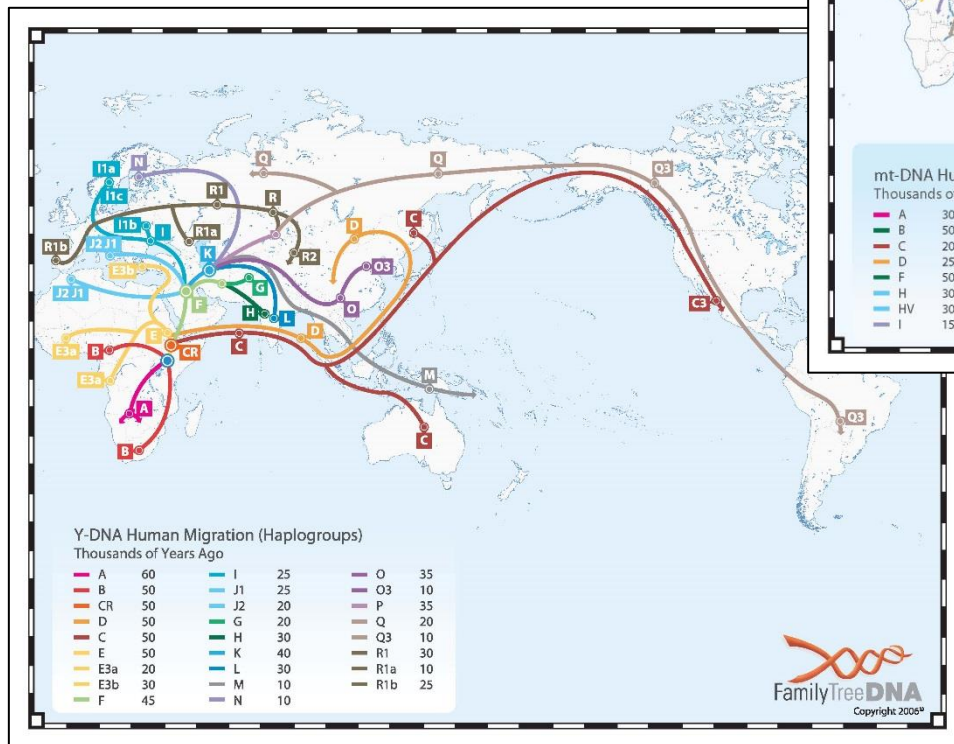
Y-DNA & mtDNA Haplogroups

Patrilinear (Y-DNA) & Matrilinear (mtDNA) Haplogroups are different



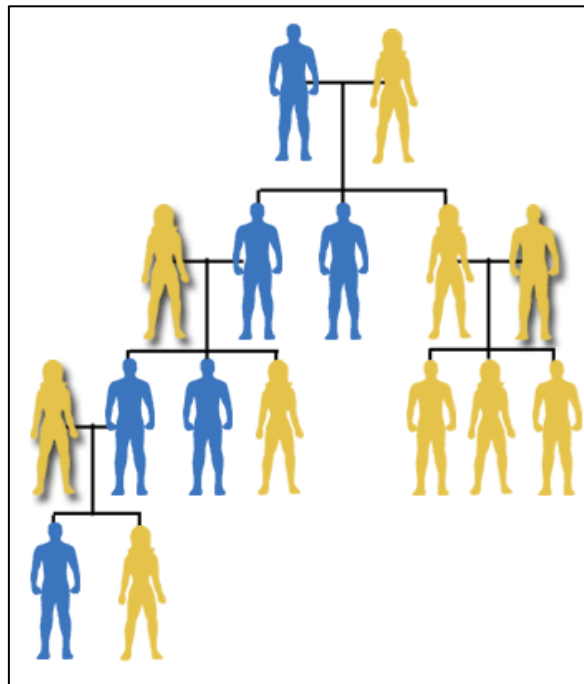
Ancient Origins & Human Migration

Y-DNA Haplogroup Migration



mtDNA Haplogroup Migration

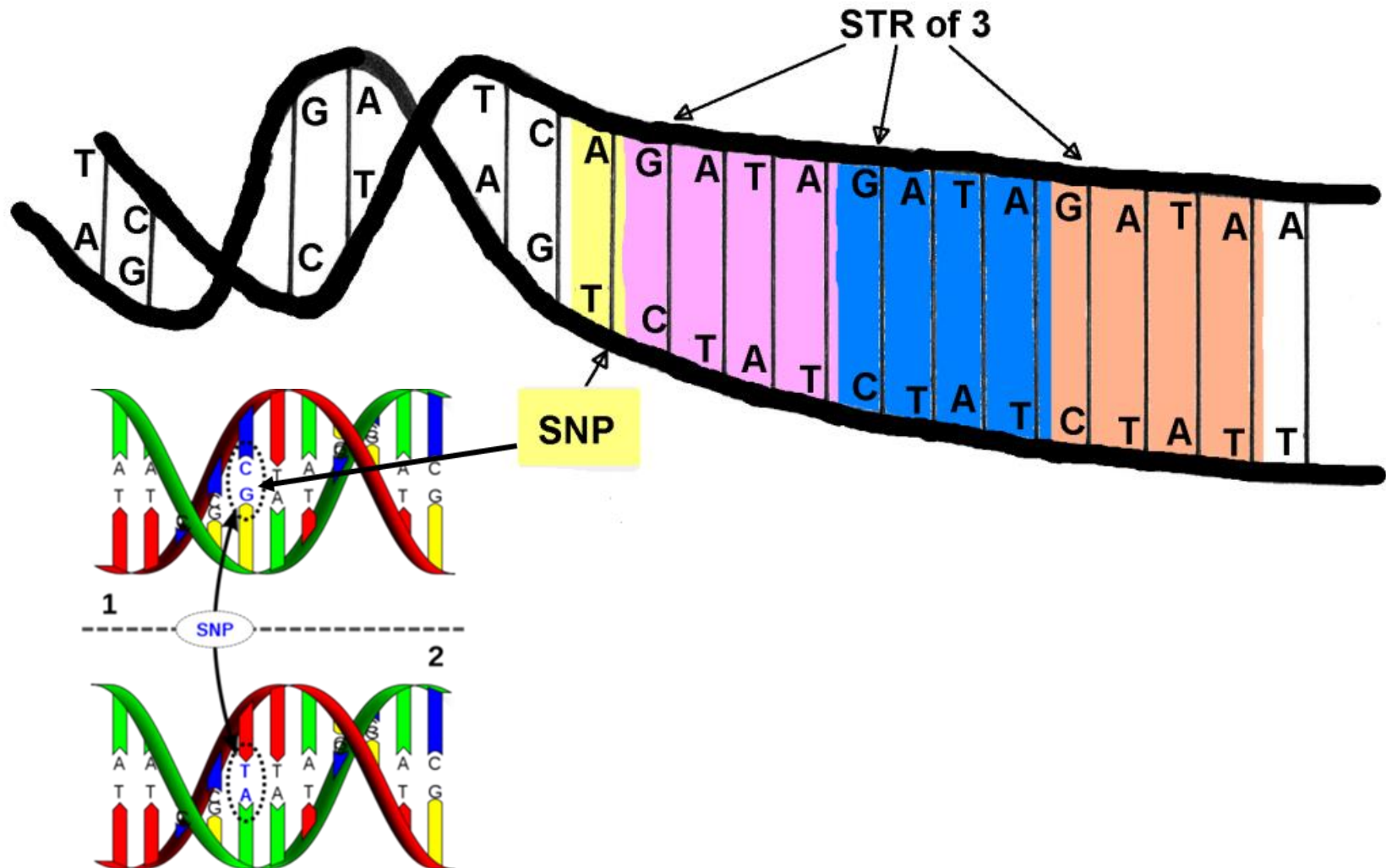
Y- Chromosome DNA Tests



Y-Chromosome

- Y-chromosome has about **59 million base pairs**
- Only **males** inherit the Y chromosome, so it is **only** used to trace **direct paternal line**
- Same inheritance pattern as **surnames**
- Y does not have a matching chromosome, so it **escapes recombination** every generation
- Y-DNA is passed down through the male line with **only random mutational events**
- Uses 2 types of markers: **STRs** and **SNPs**
 - Short Tandem Repeat
 - Single Nucleotide Polymorphism

STRs & SNPs



STRs & SNPs

LOOKING AT THE "Y" CHROMOSOME

SNP short tandem repeat (STR)



Man 1 GTACT**T**AGACTACTACTACTACTACTGGTG...
5 repeats

Man 2 GTAC**A**AGACTACTACTACTACTACTACTGGTG...
6 repeats

Man 3 GTAC**A**AGACTACTACTACTACTACTACTACTGGTG...
7 repeats

Y-DNA Tests

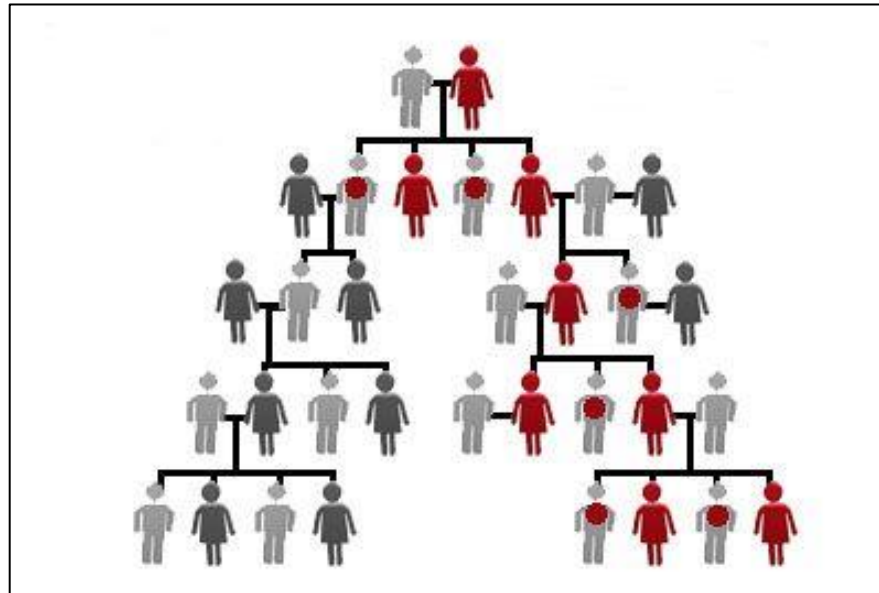
Advantages:

- **Inheritance pattern** provides powerful technique
 - Invaluable for Surname and Male Line projects
- Reveals **ancient origins** and **genetic relationships**
- Can identify NPEs (“**non-paternal events**”)

Limitations:

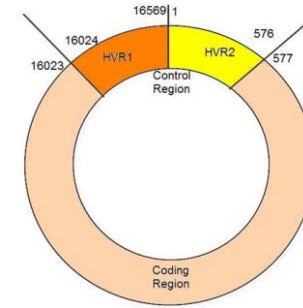
- Can identify **NPEs**
- Must test a male
- Lines “**daughter out**”

Mitochondrial DNA Tests



mtDNA Structure

- Mitochondrial DNA has
 - **A Round Structure**
 - **with three Regions**
 - **16,569** base pair locations
- Mitochondria are passed down **solely** from **mother** to **all** children (female and male)
- **Fathers do not pass** on mtDNA to children
- Does not recombine so is **inherited intact**
- mtDNA **mutates very slowly** so is passed down virtually **unchanged for thousands** of years



Mitochondrial Tests

Advantages

- **Full mtDNA sequence test** provides full haplogroup designation automatically
- Unlike Y, no additional SNP test required to fully determine mtDNA haplogroup
- **Both men and women** can take this test

Disadvantage

- mtDNA may be identical to that of **very distant** direct maternal ancestors

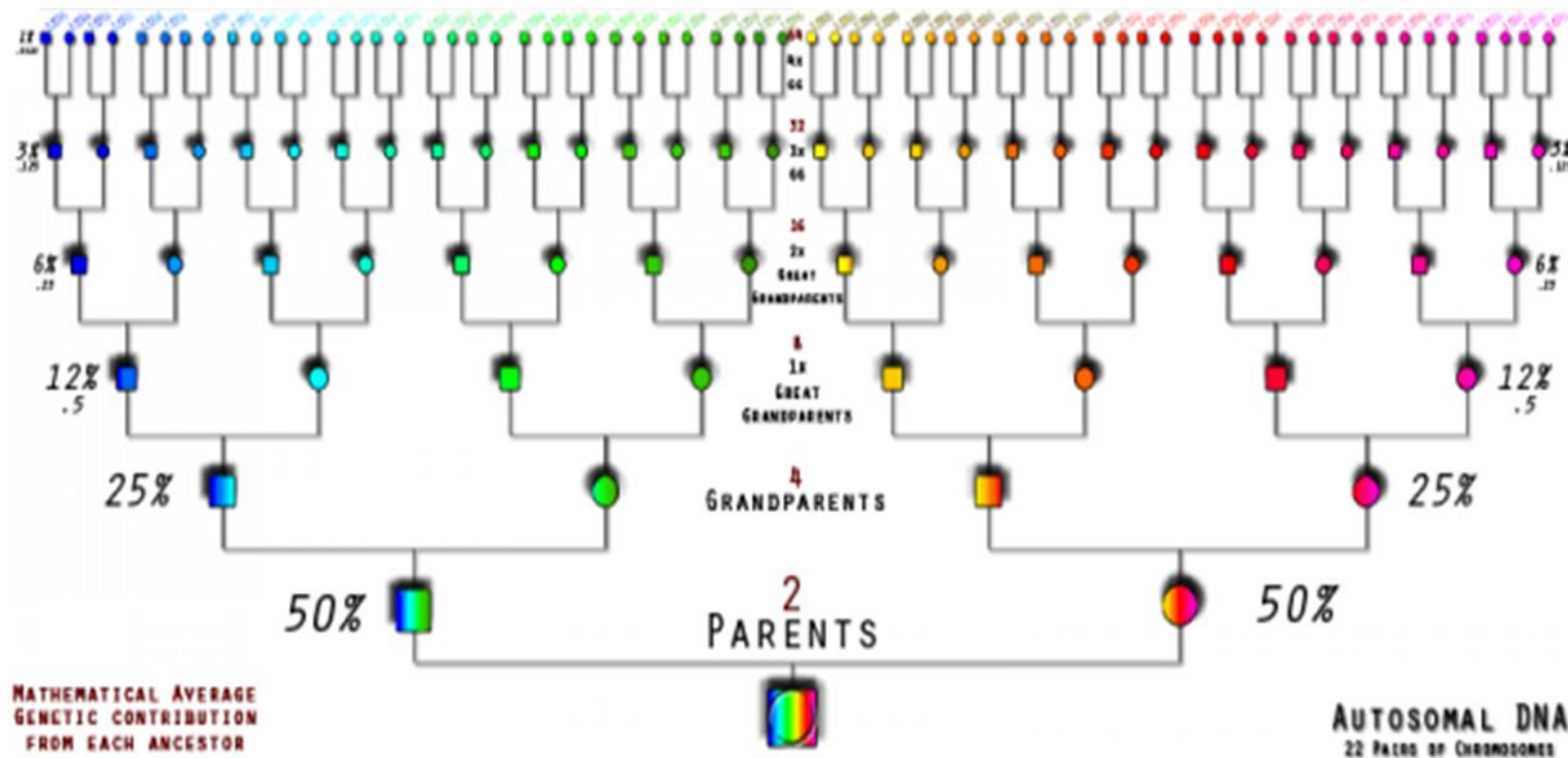
Autosomal DNA Tests



Autosomal Chromosomes

- **DNA in Chromosomes 1-22 (Autosomes)**
- 95% of our DNA is Autosomal
- atDNA divides every generation by about half
- **50% from each parent**
- ~25% from each grandparent
 - **% is variable**
 - Farther back, % is difficult to predict
- atDNA undergoes **recombination** each generation
 - Usefulness quickly dissipates after **5-6 generations**
 - Distant ancestor atDNA is gradually lost

Autosomal Recombination



Autosomal DNA

Advantages:

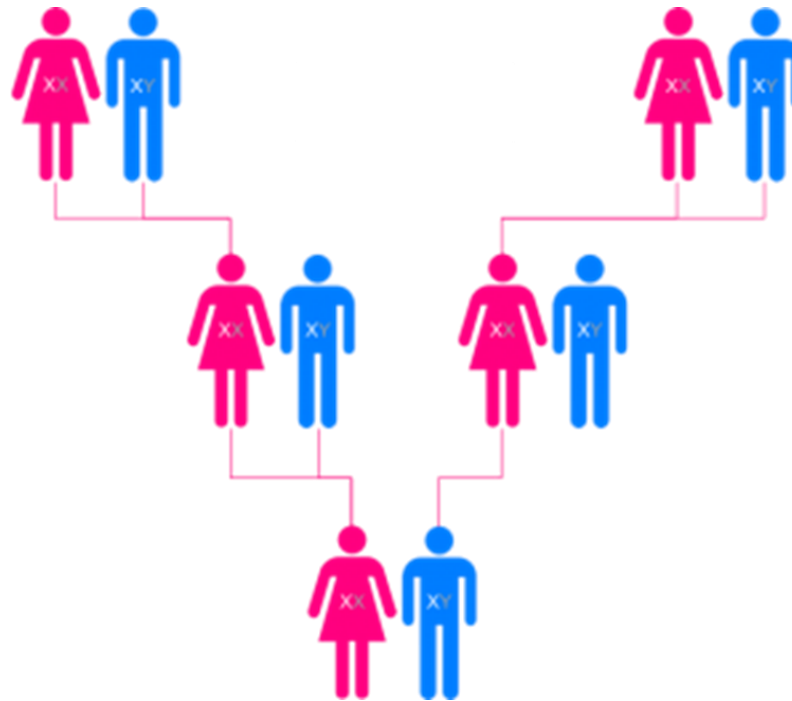
- **Widest Range of Connections** to Potential Cousins
- Offered by **All** test companies
- Both **Men and Women** can test

Disadvantages

- Inherited **Randomly**
- **Dissipates** over generations
- Test companies cannot tell **paternal or maternal contribution** (New advances are being made)



X Chromosome DNA Tests



X Chromosome

- Looks at **markers on X-chromosome(s)**
 - Males have **one X from Mother**
 - Females have **2 Xs, one from each Parent**
- **X chromosome & mtDNA tests are different**
 - Men inherit Mother's **mtDNA** but do not pass
 - Men inherit Mother's **X-DNA**, pass to daughter but not to son
 - **X-DNA** includes Mother's Paternal lines
 - **X-DNA** potential to trace more ancestors than mtDNA

X-DNA Inheritance - male

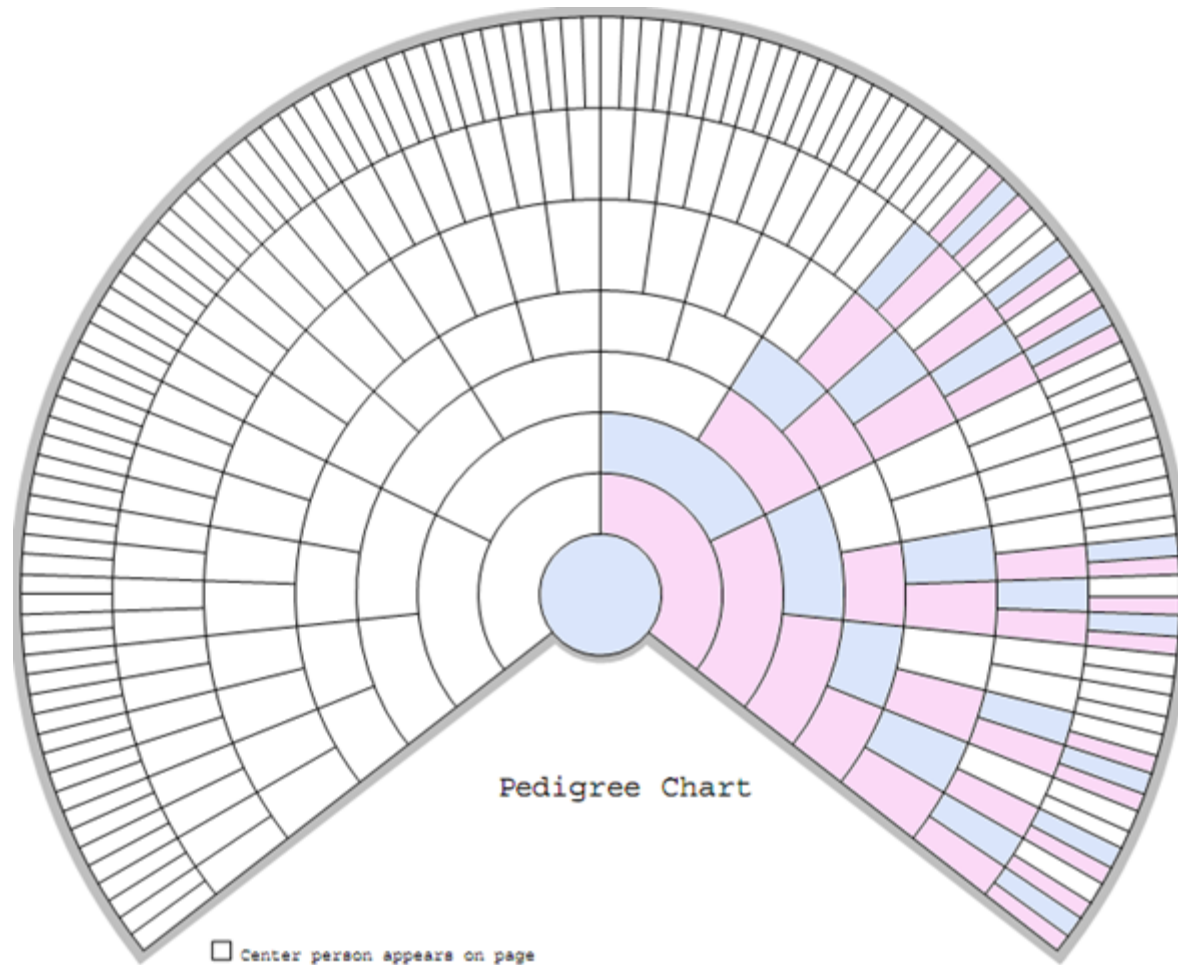
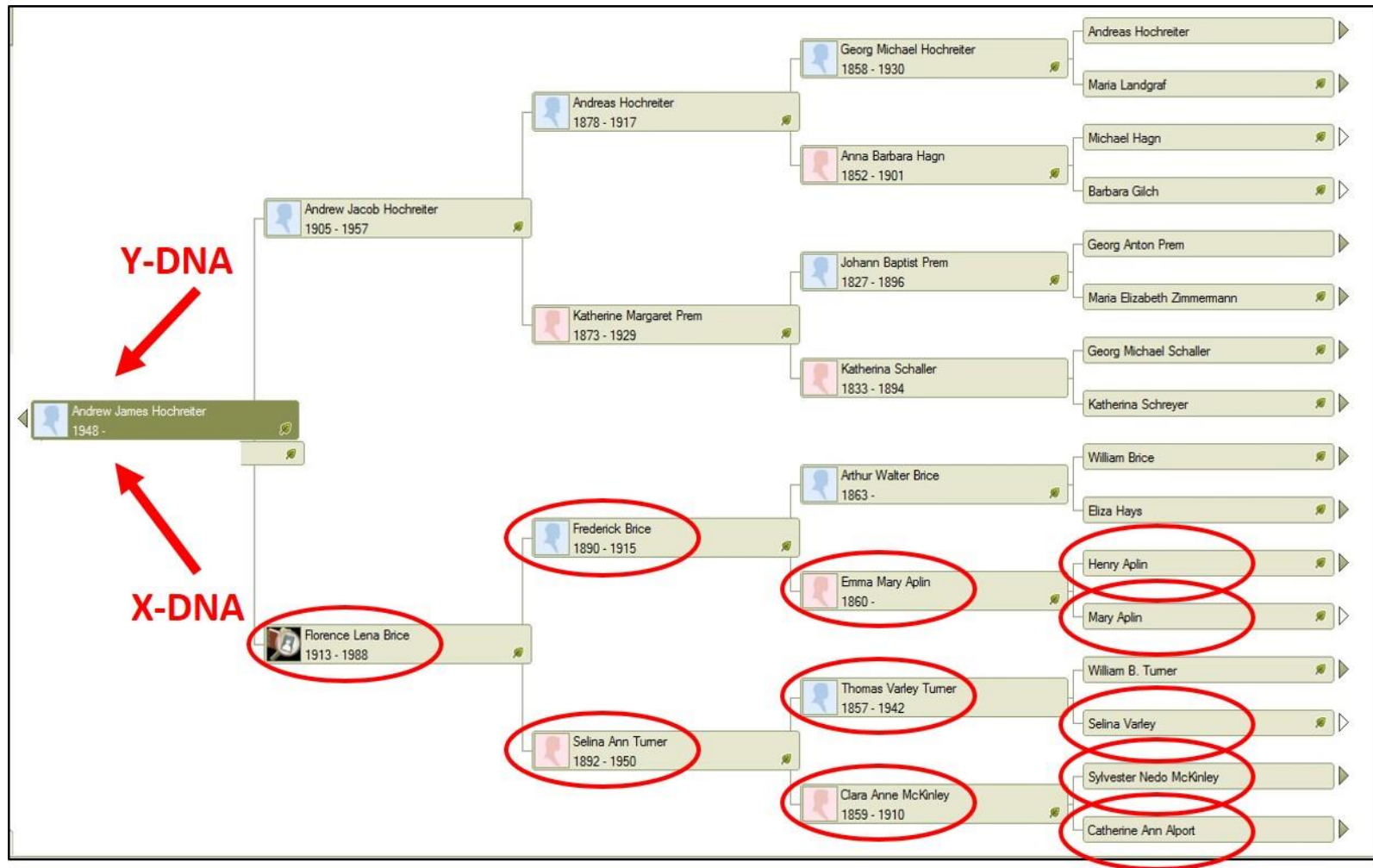


Figure 2: Male X-DNA Inheritance Chart

Male X Pedigree Example



X-DNA Inheritance - female

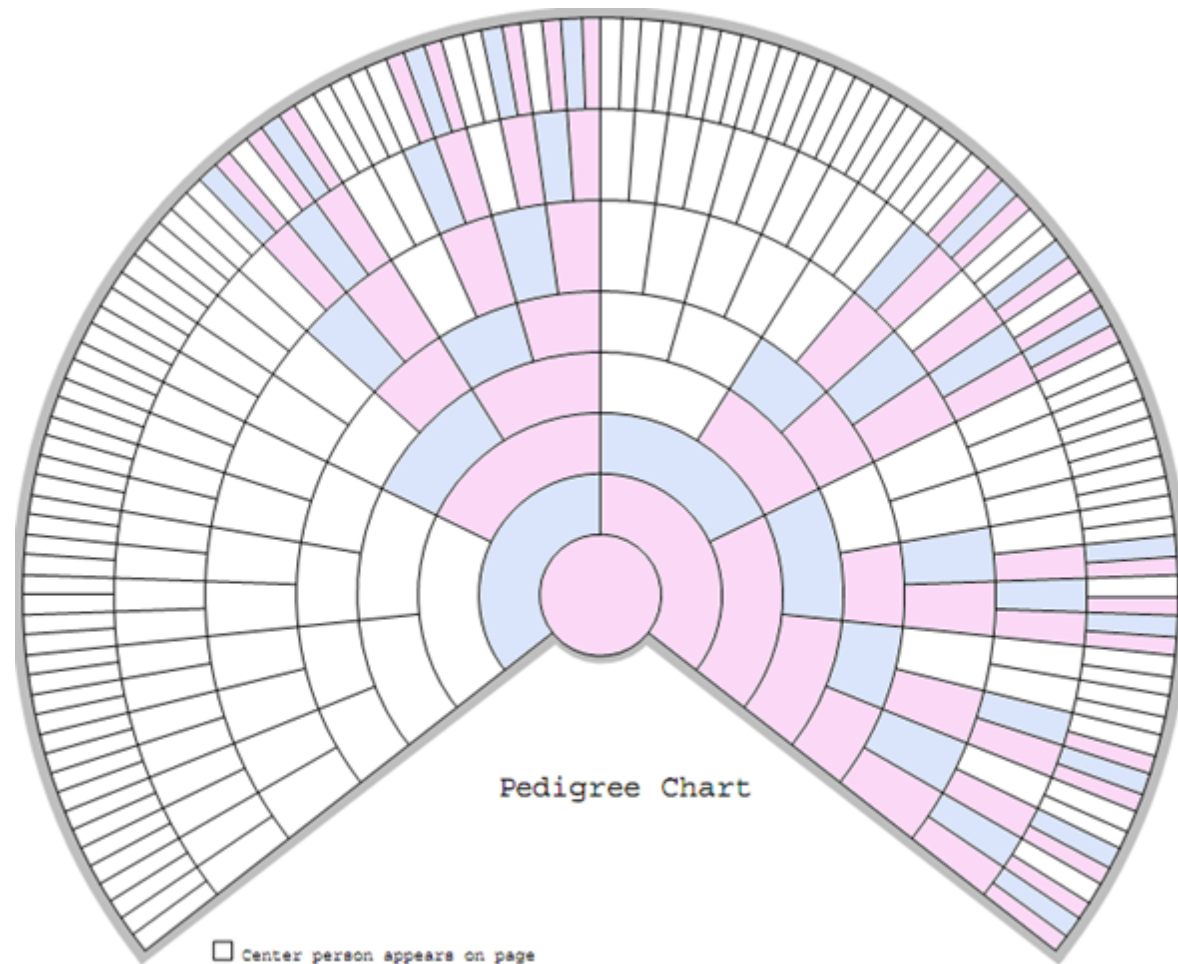
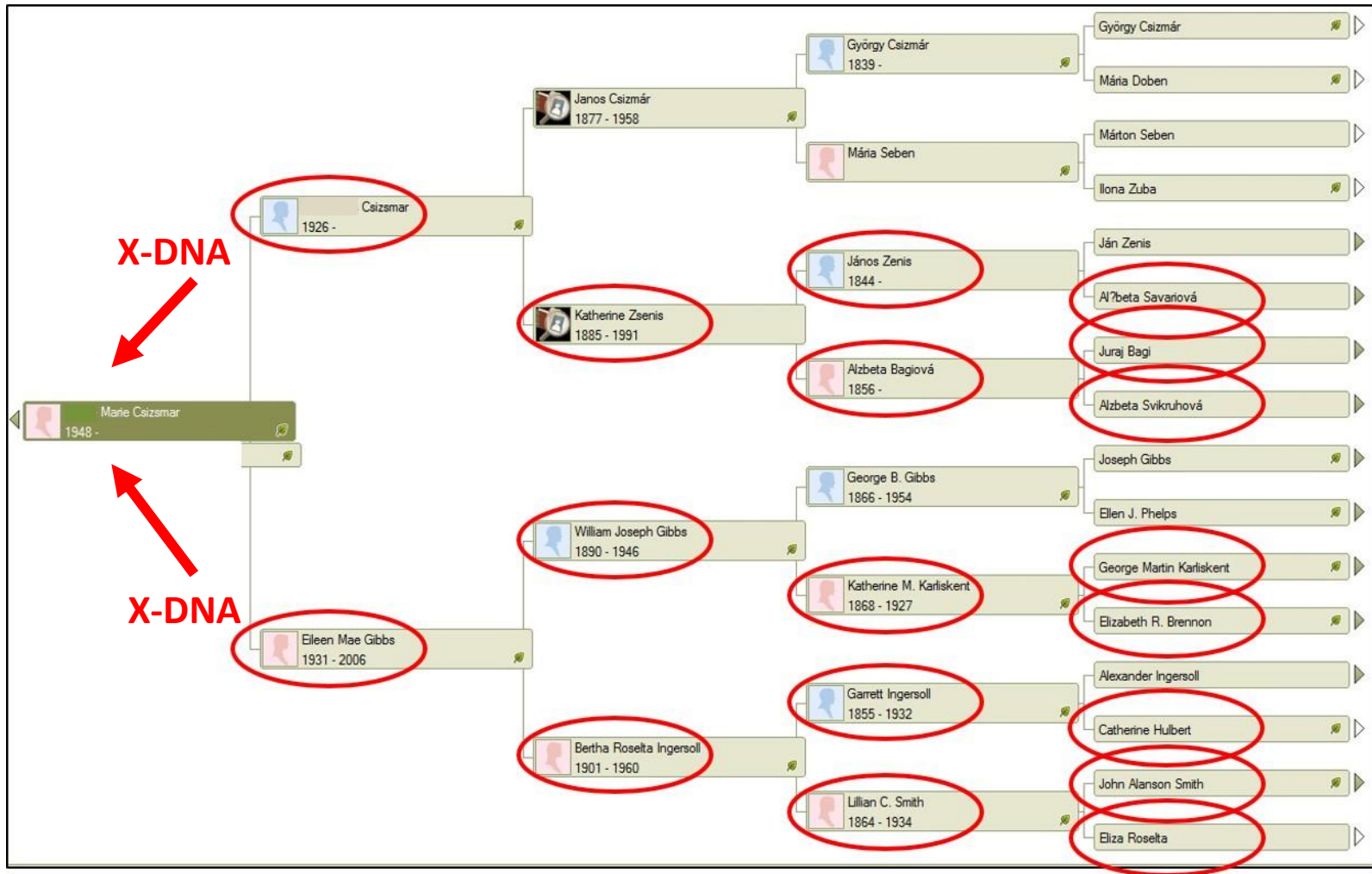


Figure 1. Female X-DNA Inheritance Chart

Female X Pedigree Example



Ethnicity

- **Nearly impossible to predict**
 - Cannot represent entire ancestors' ethnicity
 - Some DNA inheritance is lost forever
- Test companies **estimate Ethnicity differently**
 - Different **reference populations**
 - Different **matching thresholds**
 - Different **algorithms**



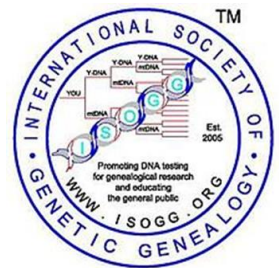
Maximize the Outcome

Example:

- Test at Ancestry or 23andMe first
- Transfer raw data to FTDNA, MyHeritage, LivingDNA
 - Ancestry & 23andMe do not accept other tests
- Test Yourself, Next test oldest generations
- Add other tests: Y-DNA, mtDNA, BigY-700

Continue Education:

- International Society of Genetic Genealogy
 - ISOGG website: <https://isogg.org/>
 - Comprehensive explanations of Genetic Genealogy



- DNA helps to **restore** forgotten:
 - **History**
 - **Identities**
 - **Relationships**
- Use it to pursue your **Family Story**
- Help to let their stories be heard and

"No Longer Forgotten"



Thank You!

