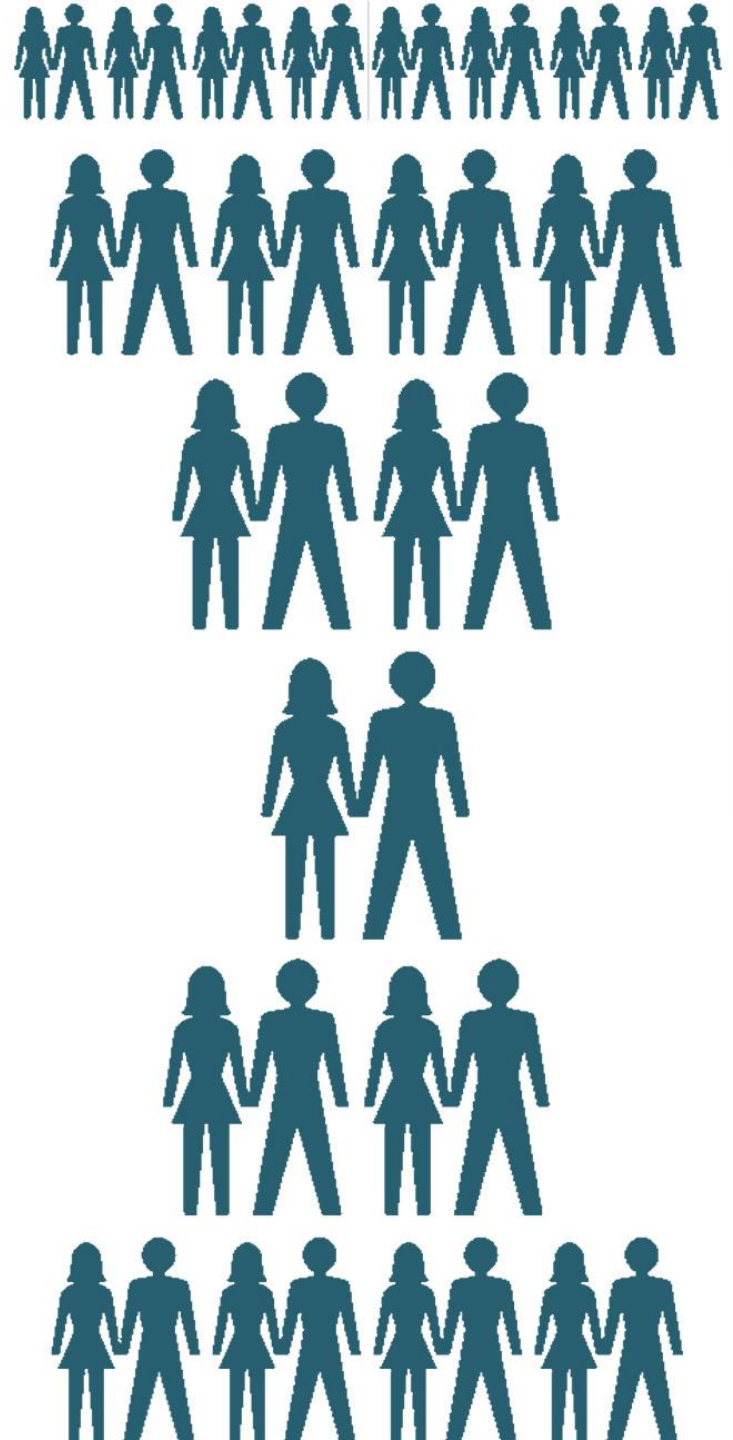


C01 Use computers to research family genealogy and world history

## DNA in Genealogy

Session 6

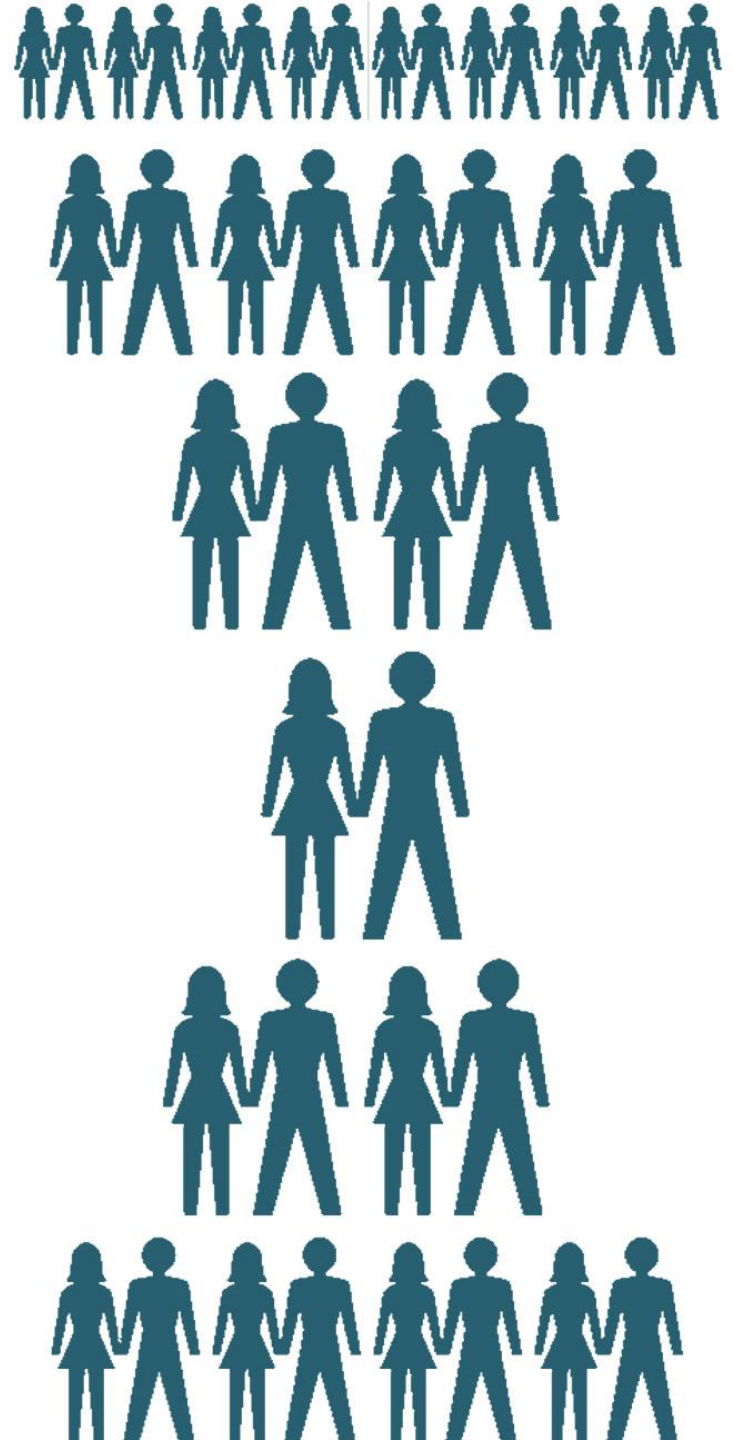
**b4uc.xyz**





# What is DNA?

[b4uc.xyz](http://b4uc.xyz)





# What is DNA?



CTAAAGATGATCTTTAGTCCCGGTTTCGAA  
TCTTTAGTCCCGGTTGATAACACCAACC  
GTAATACCAACCGGGACTAAAGATCCCG  
GGGACTAAAGTCCCACCCCTATATATATG

TTCAAATTTCTTCAAAAAGAGGGGAG  
GTGATTACATACAATCGGAGGTGCCTA  
TTTGTCATACTACATTTGCACCTATGTTTT  
GTAAGTTGATGAGAGAGAAAATGTGTGT

TTTGCTAAACAAGGTTTTATAAAATAGTTG  
AAATAATAGAAAACAACTAAAATGAAAAT  
TATTACTTAACAAATAGTTTTTAAGAATTAT  
AATAAGATATCTTATAATTATTGTATGACT

ACGGTTTTTTTGACTCATGTAGATGGATC  
AGAGTTTATTGACGGCGTGCACTATTTTT  
TTTTATTTGTTGTCCATGCAATAAGTGTA  
TATTCATTTCCACTTGTTTGAGTCGGGGT

# Introduction

- For centuries, genealogists have relied on oral and written records to trace their family trees. But around the year 2000, the age of genealogical DNA testing was launched. This provided genealogists and family historians with an opportunity to use well-established scientific methods to prove relationships and ancestry.
- Compared to paper records, which may be incomplete or inaccurate, DNA testing is precise.
- Our genetic code packs billions of gigabytes into a single gram. A mere milligram of the molecule could encode the complete text of every document in the National Library of Australia and have room to spare for the State libraries.





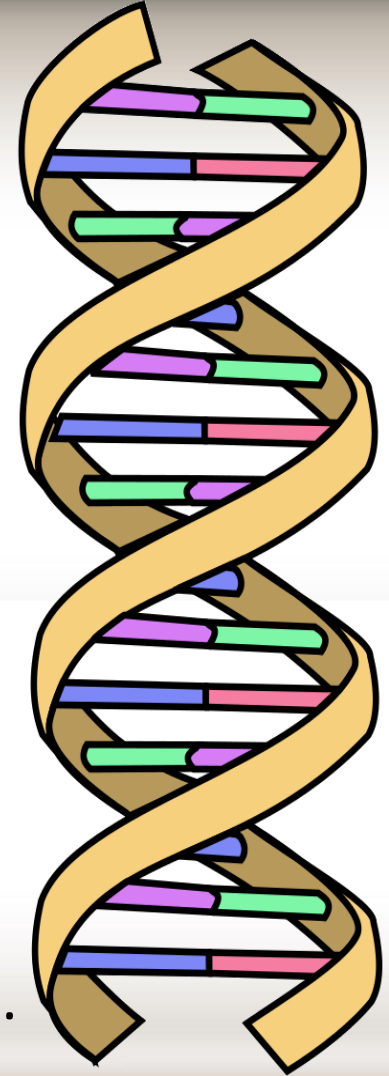
# Definitions

- **Chromosome:** threadlike bodies consisting of chromatin, that carry the genes in a linear order.
- **Chromatin:** the stuff (including DNA, RNA and other proteins) that exists within our cells.
- **DNA (deoxyribonucleic acid):** a long macromolecule that transfers genetic characteristics in all life forms. Strings of chemicals that define us.
- **Gene:** the basic physical unit of heredity; a linear sequence of nucleotides (chemicals) along a segment of DNA.
- **Haplotype:** a combination of closely linked DNA sequences on one chromosome that are often inherited together. (Haplogroup – people who share a haplotype.)



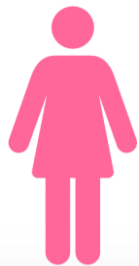
# Deoxyribonucleic acid

- DNA, or deoxyribonucleic acid, is found in every living cell. It is a long chain of chemicals that tells our cells how to grow and act.
- DNA is divided up into chromosomes, or major blocks, which are in turn divided into genes.
- Humans have 23 pairs of chromosomes (46 in all) arranged in a double helix.
- Half our chromosomes come from our mother and half from our father.
- In humans, the 23rd chromosome is either an X-chromosome or a Y-chromosome, and determines if we are male or female.
- Women have two X-chromosomes, while men have one X-chromosome and one Y-chromosome.



# Chromosomes and Genes

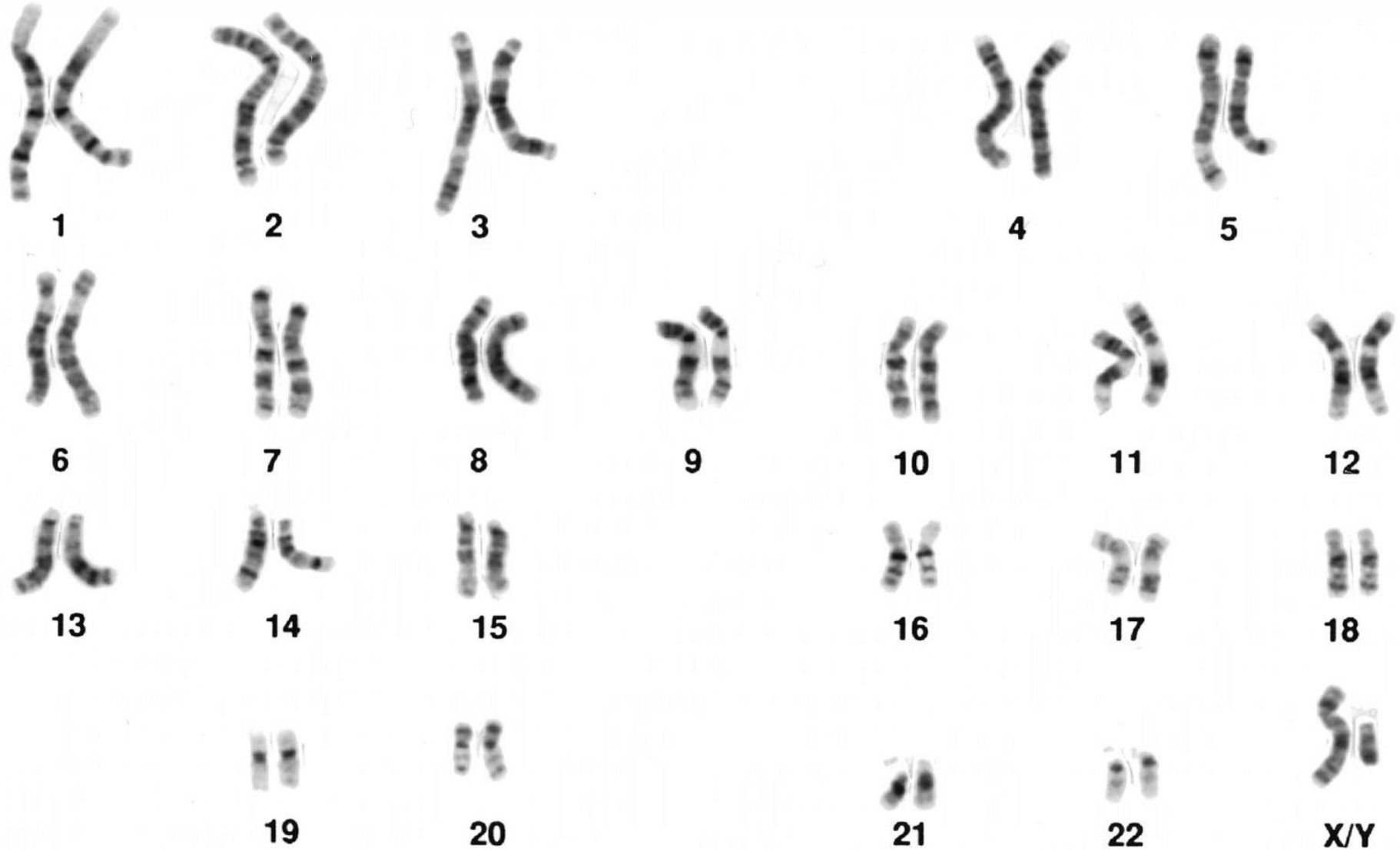
- Within the nucleus of cells, DNA is arranged into structures called chromosomes.
- Humans cells contain 23 pairs of chromosomes, 46 chromosomes per cell.
  - 22 pair are **autosomes** that carry hereditary information.
  - One pair carries **sex chromosomes** (X or Y) that determine gender. Females have a X-X pair; males, a X-Y pair.



XX XY

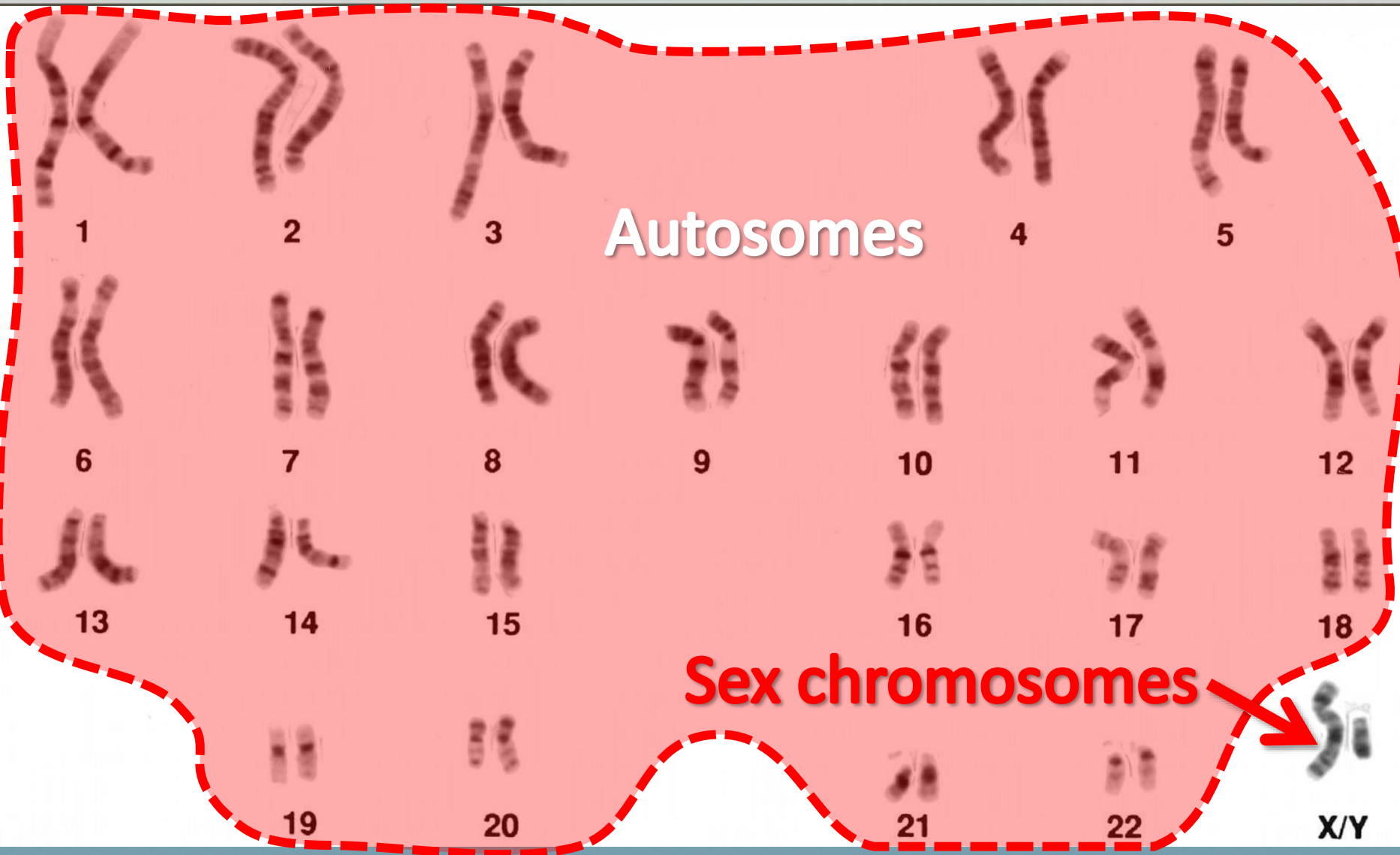
- The mother will pass an X chromosome to her baby.
- If the father passes his X chromosome, the baby will be a girl (X-X).
- If the father passes his Y, the baby will be a boy (X-Y).

# Human cells contain 23 pairs of chromosomes

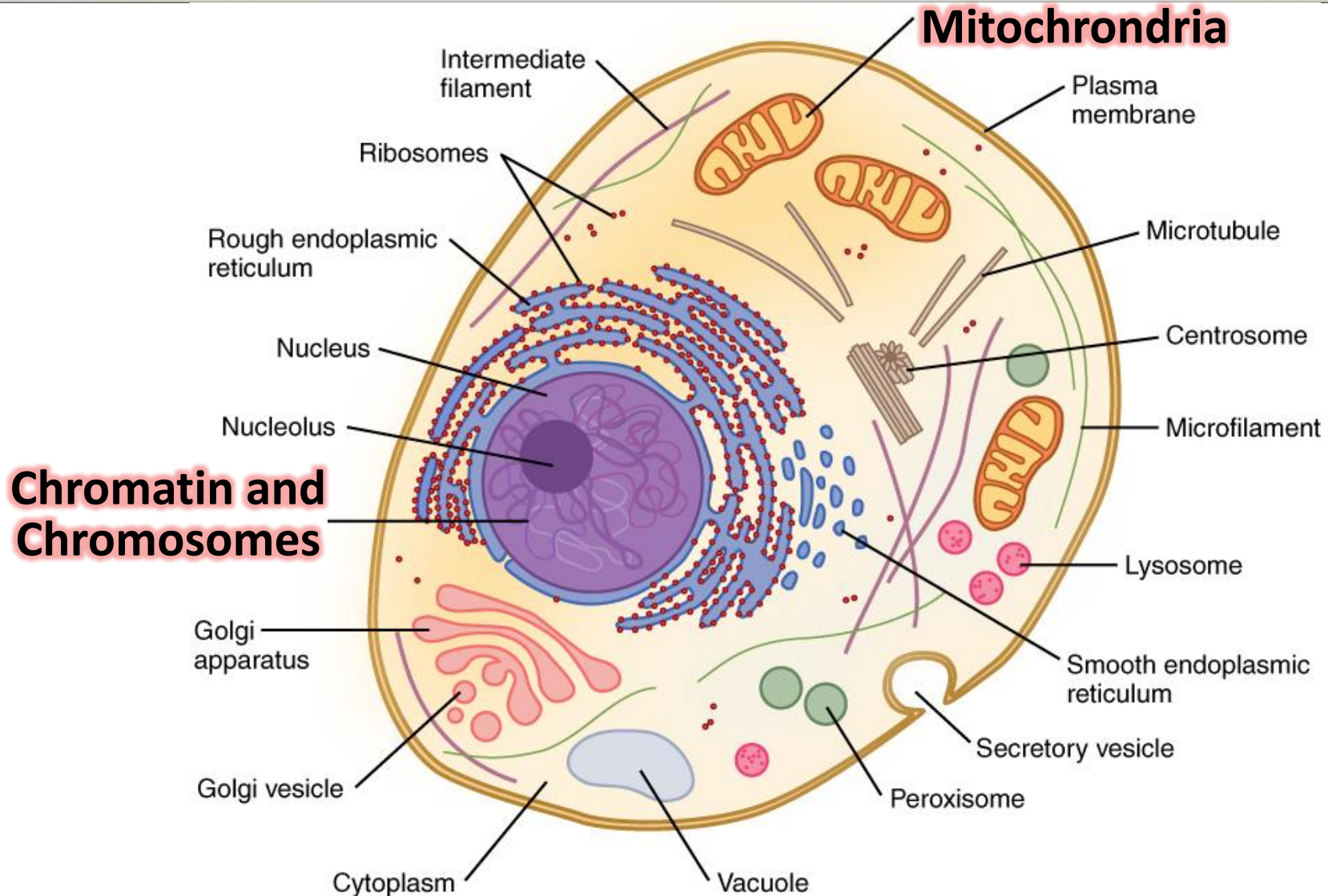




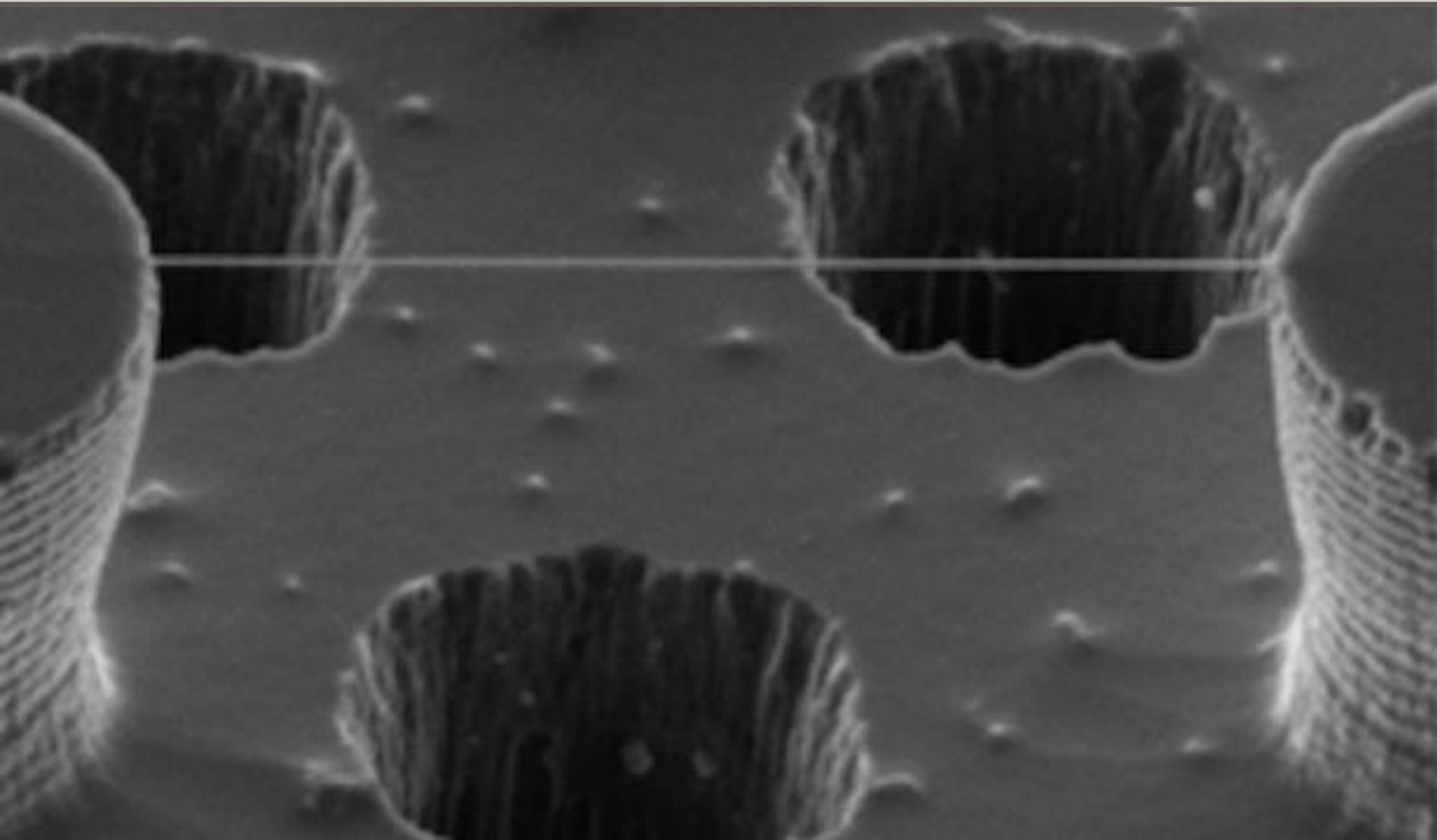
# Human cells contain 23 pairs of chromosomes



# The human cell

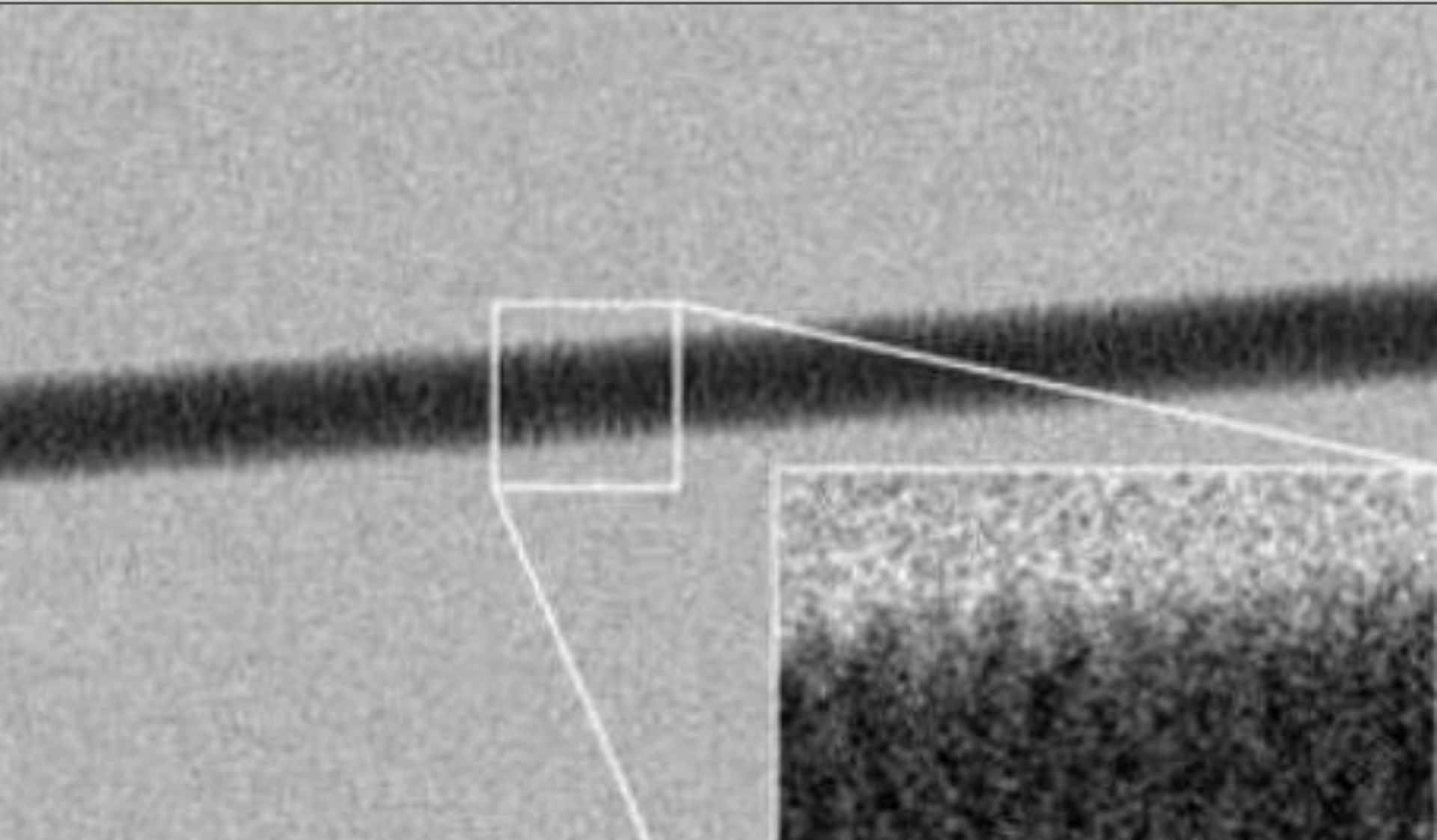


# Actual images of a DNA molecule

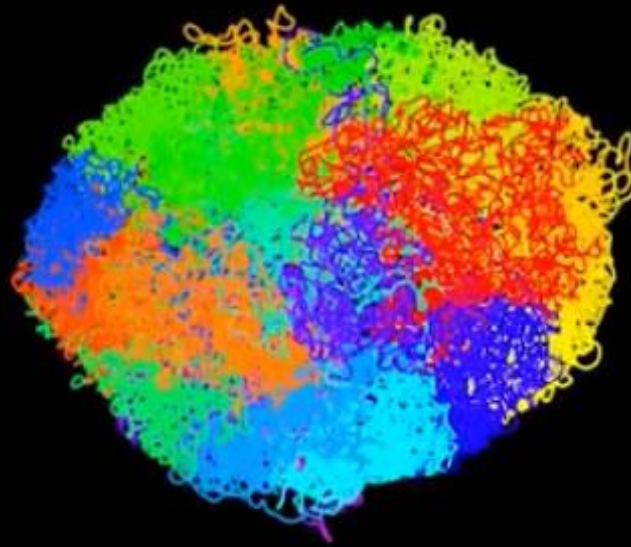




# Close-up of straightened DNA strand



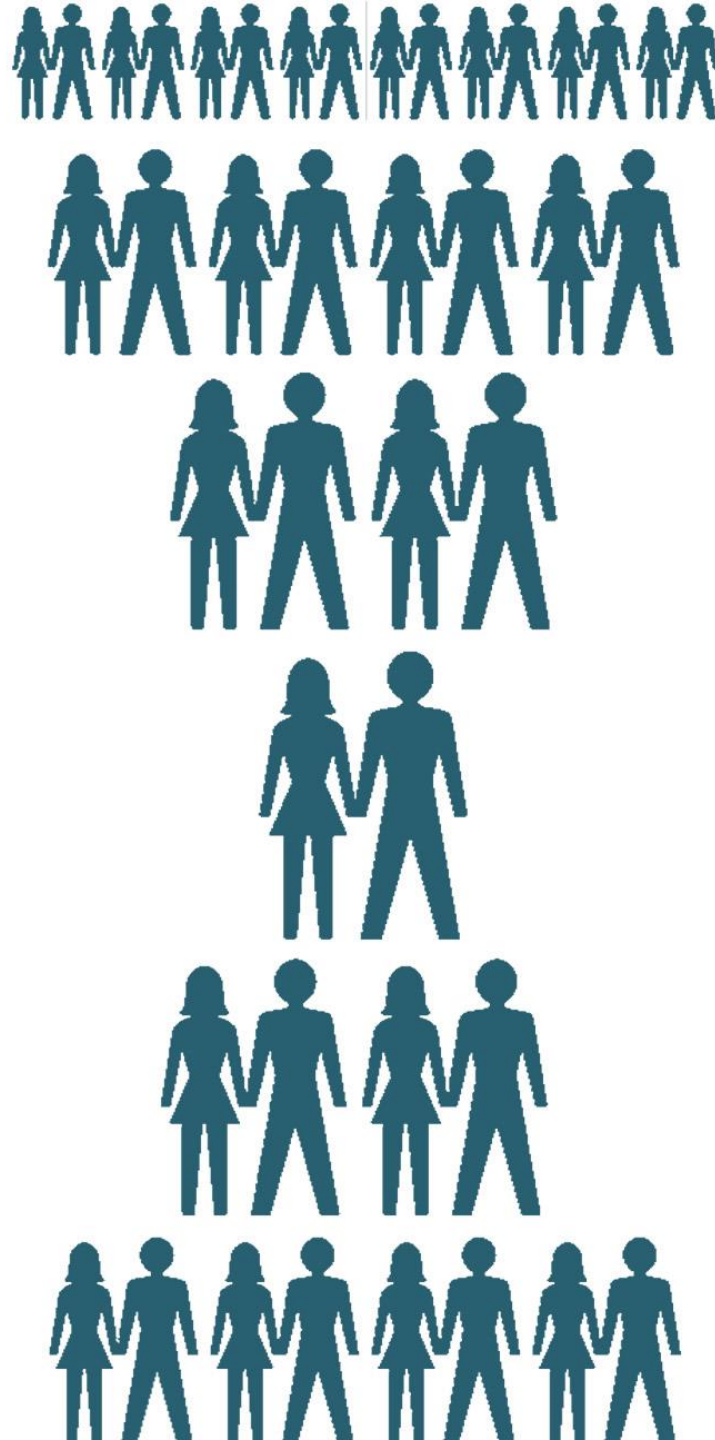
# How the chromosome pairs fit in the nucleus





# DNA Testing for Genealogy

**b4uc.xyz**





# Typical DNA Test Kits



# Introduction to Molecular Genealogy

There are four types of DNA tests used in genealogy. Each one works a little differently, and tells you different things. Therefore, each one has its advantages and disadvantages.

- The Four Types of DNA tests for genealogists:
  - Autosomal DNA (by far the most common).
  - Y Chromosome DNA (Y-DNA) – paternal line.
  - Mitochondrial DNA (mtDNA) – maternal line.
  - Y-DNA and mtDNA – paternal and maternal lines.

<https://learn.genetics.utah.edu/content/basics/molgen/>

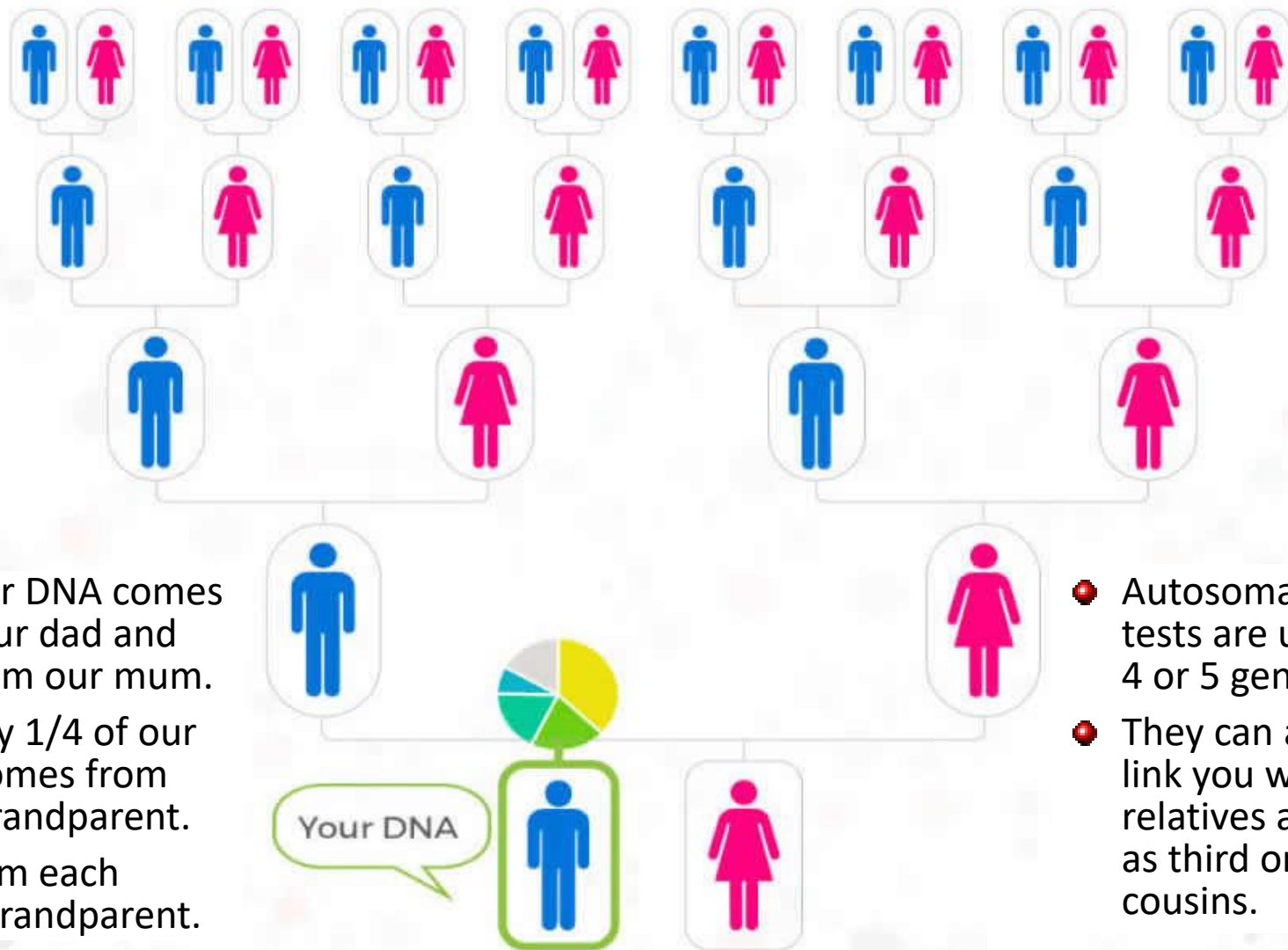
# Autosomal DNA Tests

- Autosomal DNA examines the **first 22 pair of chromosomes**.
- Since it doesn't rely on the 23rd chromosome, autosomal DNA tests can be done in **both men and women** equally.
- Autosomal DNA tests single-nucleotide polymorphisms (SNPs), or the different “shapes” of small chunks of DNA. They check about 700,000 SNPs to determine how closely related you are to someone else.
- The further you go back, the less DNA you inherit from a particular ancestor, so after 5+ generations it is less effective.
- It can also provide an estimate of your ethnicity, or the regions where your ancestors lived within the past few centuries.
- **Every genealogy DNA company** offers autosomal DNA tests.



# Autosomal DNA

The DNA you inherit from ALL of your ancestors.



- Half our DNA comes from our dad and half from our mum.
- Roughly 1/4 of our DNA comes from each grandparent.
- 1/8 from each great-grandparent.

- Autosomal DNA tests are useful for 4 or 5 generations.
- They can accurately link you with relatives as distant as third or fourth cousins.

# Typical Autosomal DNA test kits

Saliva

ancestryDNA<sup>®</sup>  
Where your story grows.<sup>™</sup>



Cheek swab



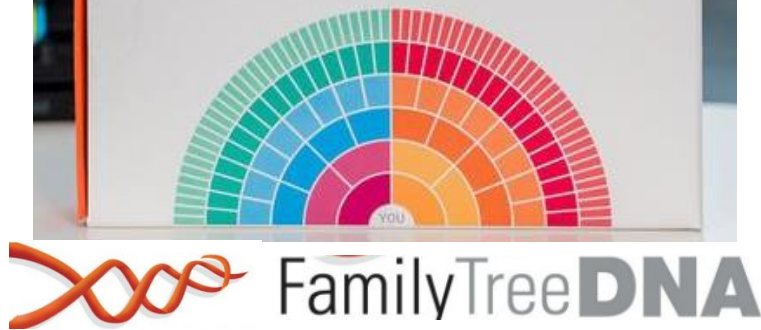
MyHeritage DNA

Cheek swab

Begin Your  
DNA Journey  
Swab Collection Kit



Family Finder Test



Saliva

23andMe<sup>®</sup>

ANCESTRY

Welcome to you<sup>®</sup>

saliva collection kit

23andMe



# Y-DNA Tests = Patriarchal Line ONLY

- Y-DNA tests examine only the Y-chromosome. For men only!
- Because you can only get a Y-chromosome from your father, and he from his father, that means it tends to change very little over time. Tests can go back thousands of years.
- There are actually two sub-tests with Y-DNA testing.
  - The short tandem repeat (STR) test categorizes sections of DNA according to how often a genetic pattern repeats.
  - The single-nucleotide polymorphism (SNP) test works like autosomal DNA testing, but only tests about 30,000 SNPs.
- **Only FamilyTreeDNA offers individual Y-DNA testing.**
- Y-DNA is useful for adoptees as well as Jewish ancestry.



# Y-DNA

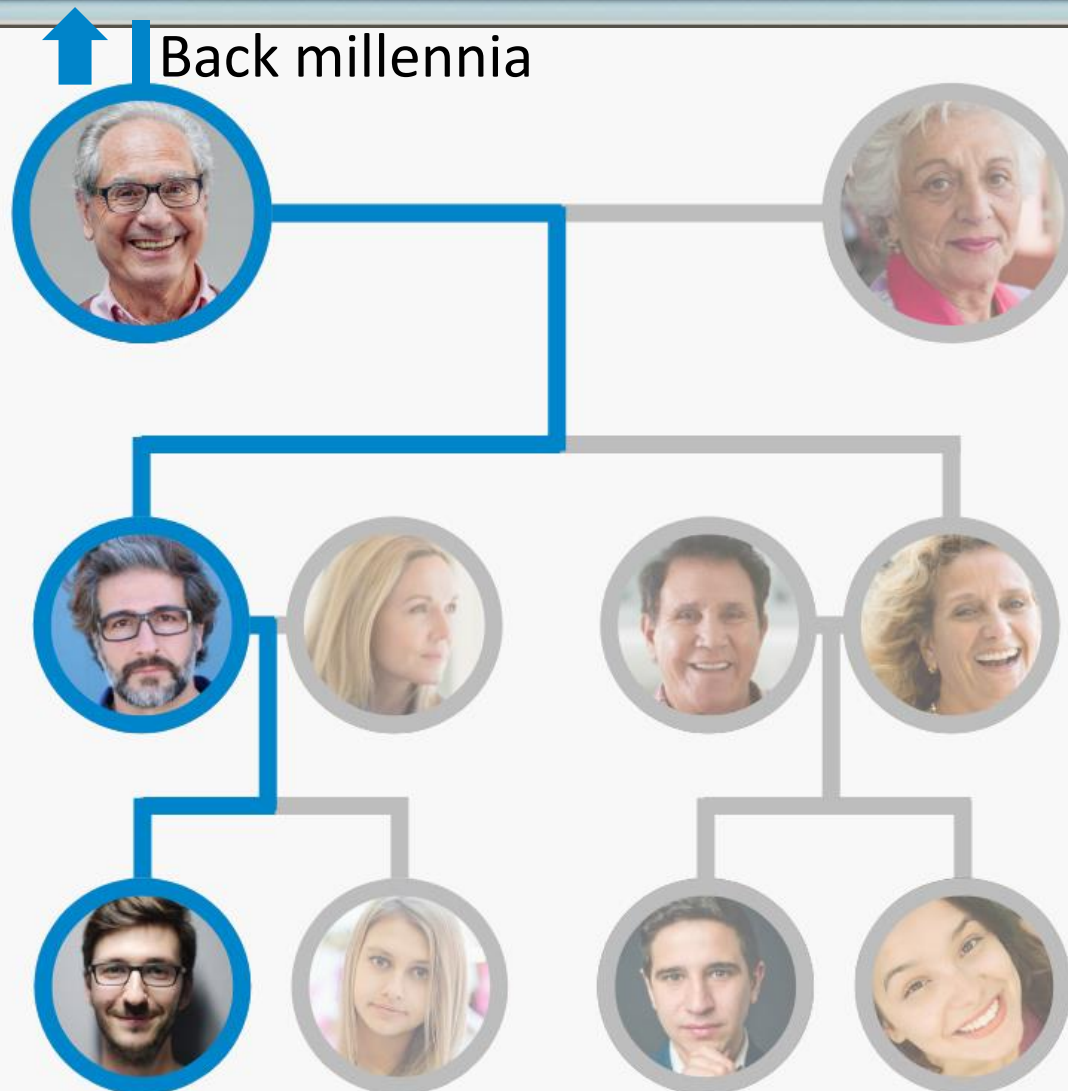
The DNA only males inherit from their direct paternal line.



- Women have two X-chromosomes; men have one X and one Y.
- Y-DNA tests examine only Y-chromosomes.

- Because sons only get Y-chromosomes from their father's line, it tends to change very little over time.
- Results can go back thousands of years.

# What a Y-DNA test shows



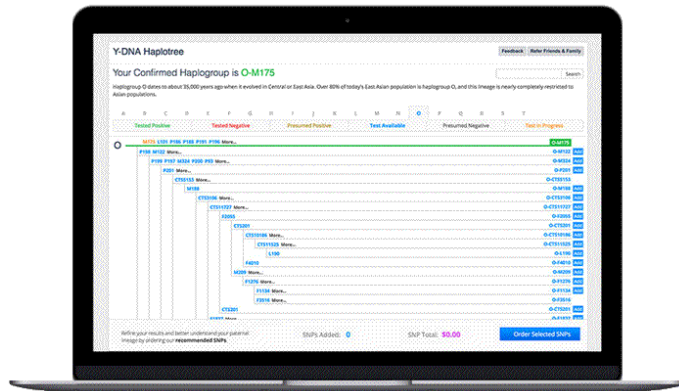
# Y-DNA test kits

Both Bundled



FamilyTreeDNA

The world's largest Y-DNA database



LivingDNA YOUR ANCESTRY

3-in-1

Your autosome DNA  
Your dad's Y-DNA  
Your mum's mtDNA

A GOOD START

Y-37

ONLY \$169 USD

Examines 37 short tandem repeats (STRs) on the Y chromosome

ORDER NOW

Y-67

ONLY \$268 USD

Examines 67 short tandem repeats (STRs) on the Y chromosome

ORDER NOW

Y-111

ONLY \$359 USD

Examines 111 short tandem repeats (STRs) on the Y chromosome

ORDER NOW

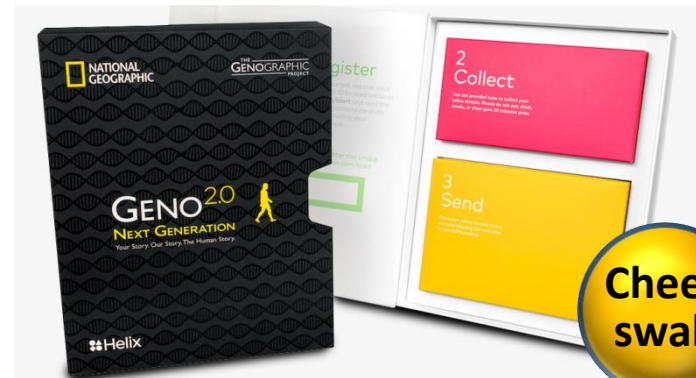
EXPERT LEVEL

Big Y-700

ONLY \$649 USD

Examines 700 short tandem repeats and 100K SNPs on the Y chromosome

ORDER NOW



NATIONAL GEOGRAPHIC

Your autosome DNA  
Your dad's Y-DNA  
Your mum's mtDNA

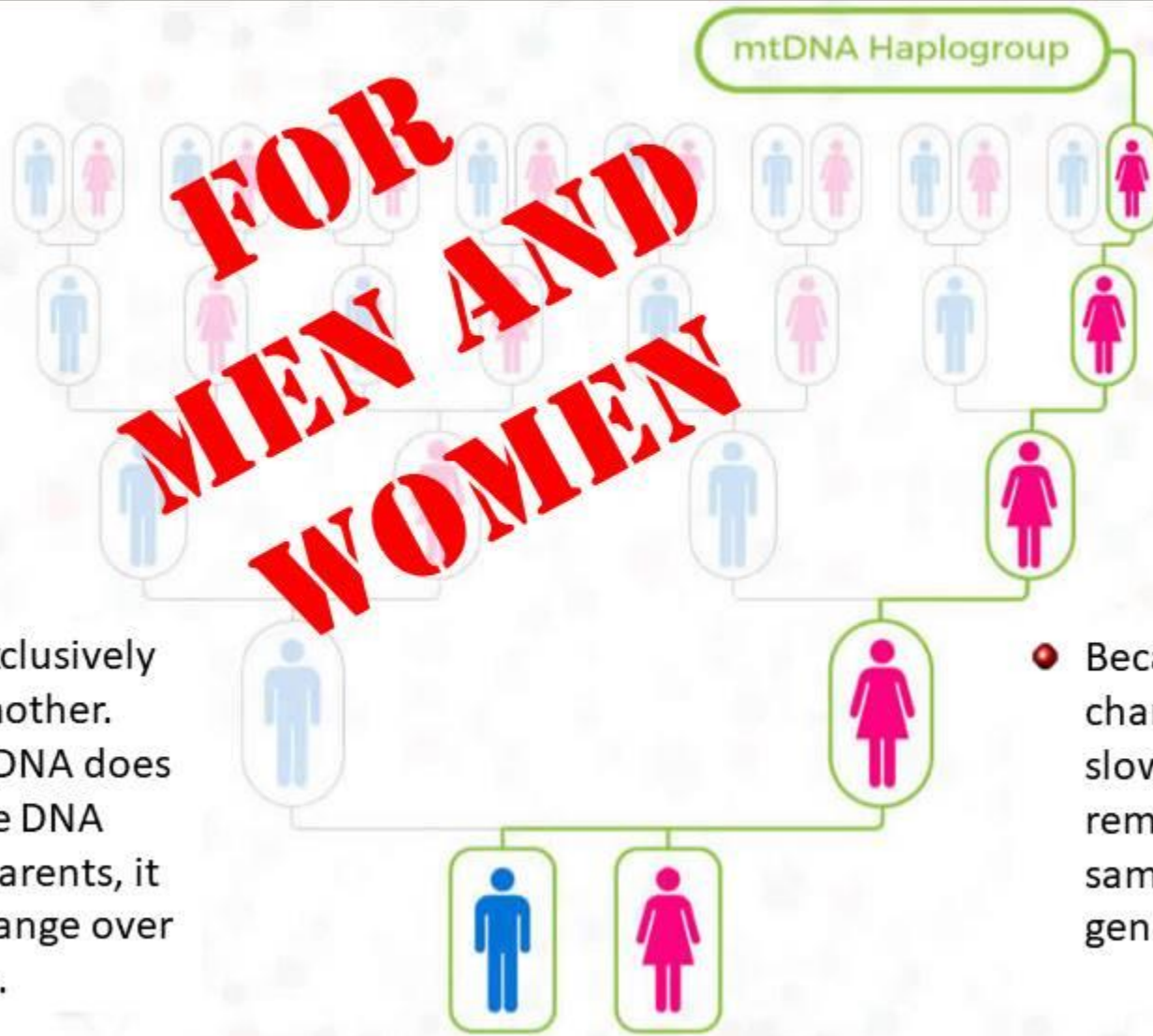


# mtDNA - Mitochondrial DNA Tests

- Mitochondrial DNA, or mtDNA, is genetic material inside mitochondria, small components found inside every cell and which have their own separate DNA strands.
- mtDNA is passed down **almost unchanged** from a mother to her children. Because it doesn't combine with anything, it does not change with every generation and might remain stable for 50 or more generations!
- mtDNA testing ignores the main DNA in a cell, and looks just at the DNA of the mitochondria instead so it only examines about 16,500 genetic base pairs
- mtDNA gives very precise and accurate ancestry results, but only for the maternal line.
- An mtDNA test will identify how closely related you are to a haplogroup (people with a common ancestor). A haplogroup is a group of people with a single common ancestor.

# mtDNA

The DNA both males and females inherit from their maternal line

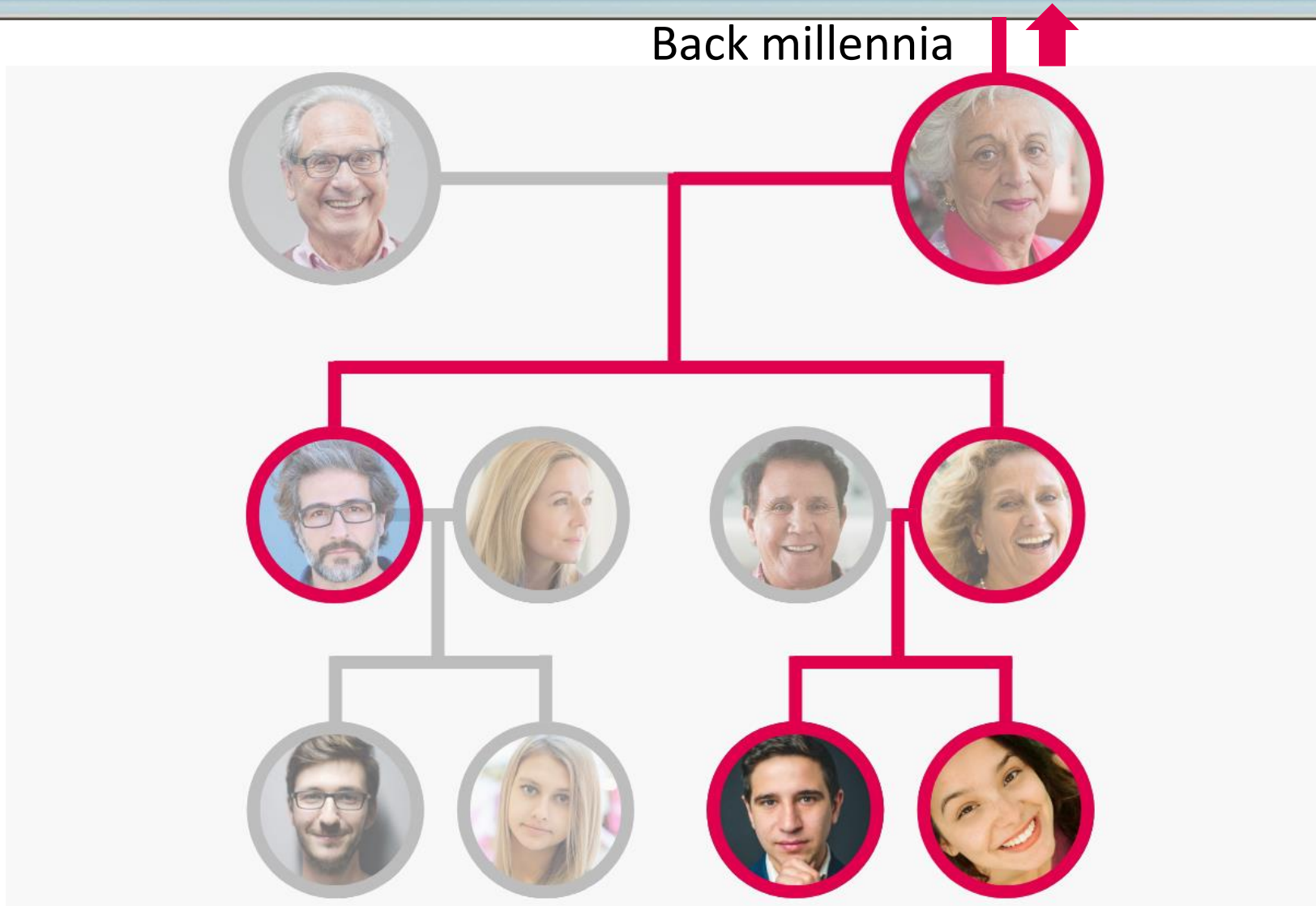


- mtDNA is exclusively from your mother.
- Because mtDNA does not combine DNA from both parents, it does not change over generations.

- Because mtDNA changes extremely slowly, it might remain exactly the same for dozens of generations

# What a mt-DNA test shows

Back millennia





# mtDNA test kits

Both Bundled



FamilyTreeDNA

## The world's largest mtDNA database

Cheek swab



EXPANSION TIMES (YEARS AGO)	
Africa	120,000-150,000
Out of Africa	55,000-75,000
Asia	40,000-70,000
Australia/PNG	40,000-60,000
Europe	35,000-50,000
Americas	15,000-35,000
No-Dene/Esk/Aleuts	8,000-10,000

Family Tree DNA  
mtDNA Migrations Map

mtDNA Plus

ONLY \$89 USD

- Examines two regions of mitochondrial DNA (HVRI and HVRII)
- Identifies basic haplogroup and migration paths

REMOVED FROM SITE

mtFull Sequence

ONLY \$199 USD

- Examines all regions of mitochondrial DNA
- Identifies basic haplogroup and migration paths
- More refined results for



Cheek swab



LivingDNA 3-in-1  
YOUR ANCESTRY

Your autosome DNA  
Your dad's Y-DNA  
Your mum's mtDNA



Cheek swab



NATIONAL GEOGRAPHIC

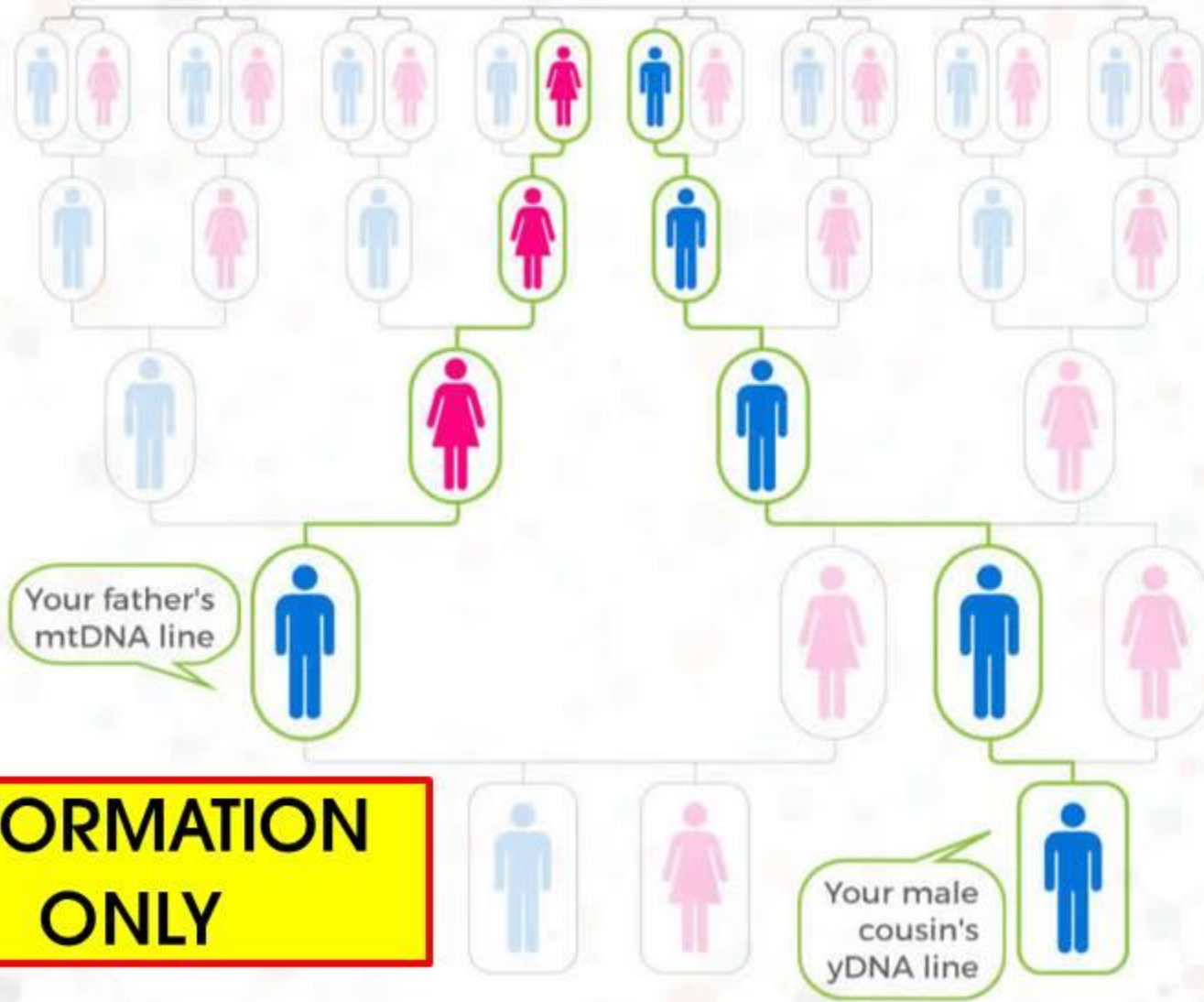
Your autosome DNA  
Your dad's Y-DNA  
Your mum's mtDNA

# Specialised Y-DNA + mtDNA test kits

- In 2007, Ancestry.com introduced genetic genealogy testing by launching paternal Y-DNA and maternal mtDNA tests.
- In 2014, Ancestry.com discontinued both to focus solely on **autosomal DNA testing**.
- While Y-DNA and mtDNA results can have the benefit of tracking back 50 or more generations (in fact, to Adam and Eve), in practical terms family genealogy is mostly concerned with the last five to ten generations.
- For those who have valid reasons for researching deeper ancestry, FamilyTreeDNA offers a range of detailed mtDNA (2 options) and Y-DNA (4 options) testing.

# Y-DNA and mtDNA

Test extended family members to complete a family tree

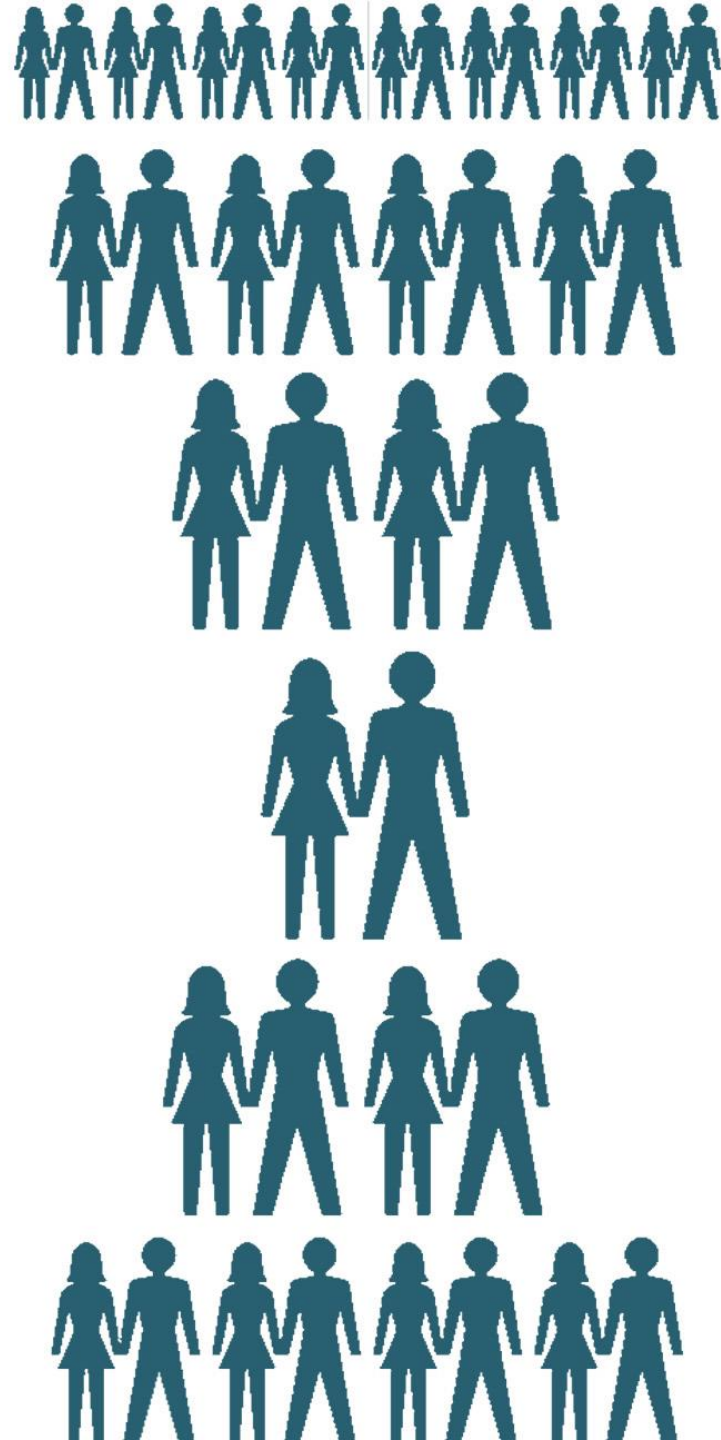






# Taking a DNA Test

[b4uc.xyz](http://b4uc.xyz)



# How is DNA Collected?



Order a DNA kit which is mailed to you.



Return a sample in the prepaid envelope.

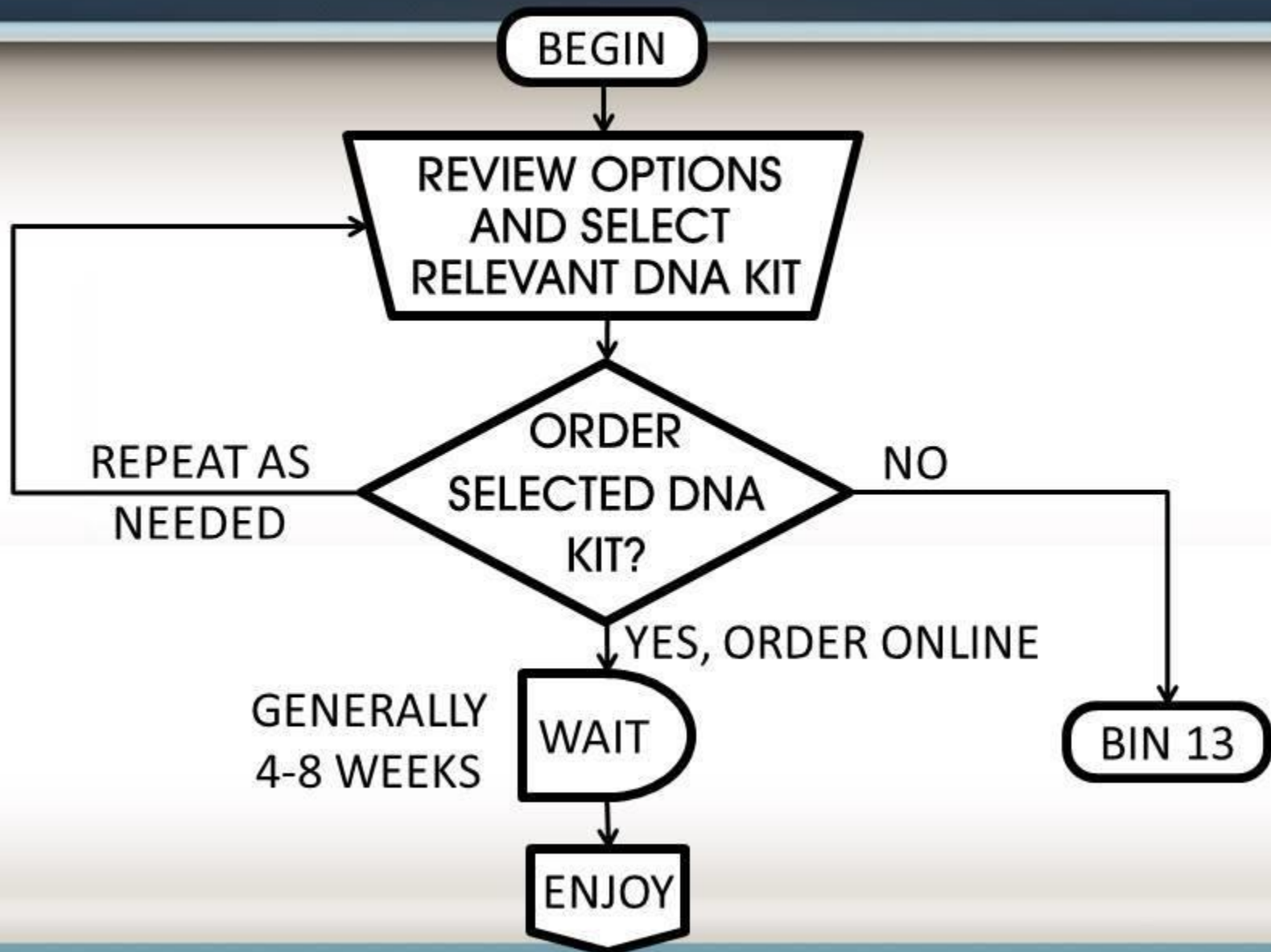


DNA is analysed in a lab.



DNA results are emailed and posted online.

# Select and order a DNA kit online





# Typical DNA test kit (e.g., MyHeritage)

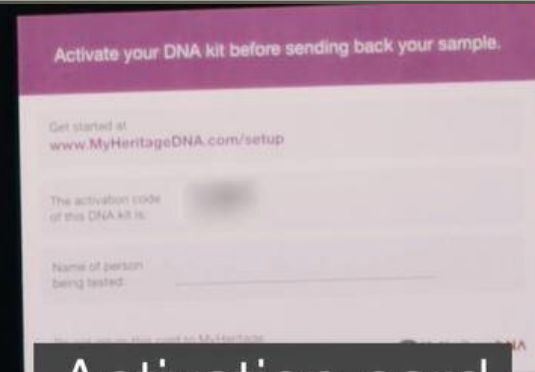
Cheek swab



Return envelope



Instructions



Activation card

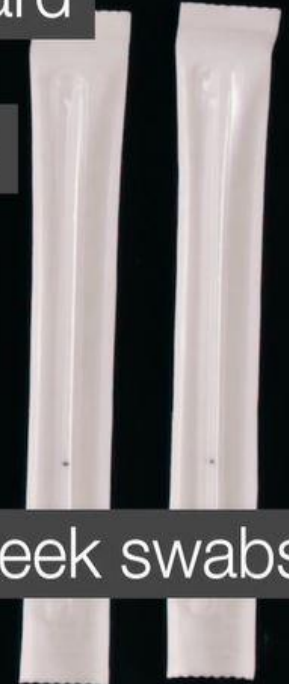


1 plastic bag

2 vials



2 cheek swabs



# How is DNA Collected?

- DNA is collected either with a cheek swab or a saliva sample, depending on which company you use.
- For the most part, there's no advantage to one method over the other.
- However, if the person being tested is very young (too young to be told to spit in the cup) or very old (and can't produce enough saliva), the cheek swab might be easier.
- Right now, AncestryDNA and 23andMe use saliva samples; other companies use cheek swabs.

# What Happens Next?

- Once you've gathered your DNA sample, simply return it to the company for processing.
- It will usually take six to ten weeks for your sample to be processed - but could take longer during and after holidays since DNA tests are a popular gift.
- Results are emailed to you once your test is analysed.
- Depending on the company and the test, your results may include:
  - your raw data
  - ethnicity estimates
  - ways to contact potential relatives



# Keep in Mind

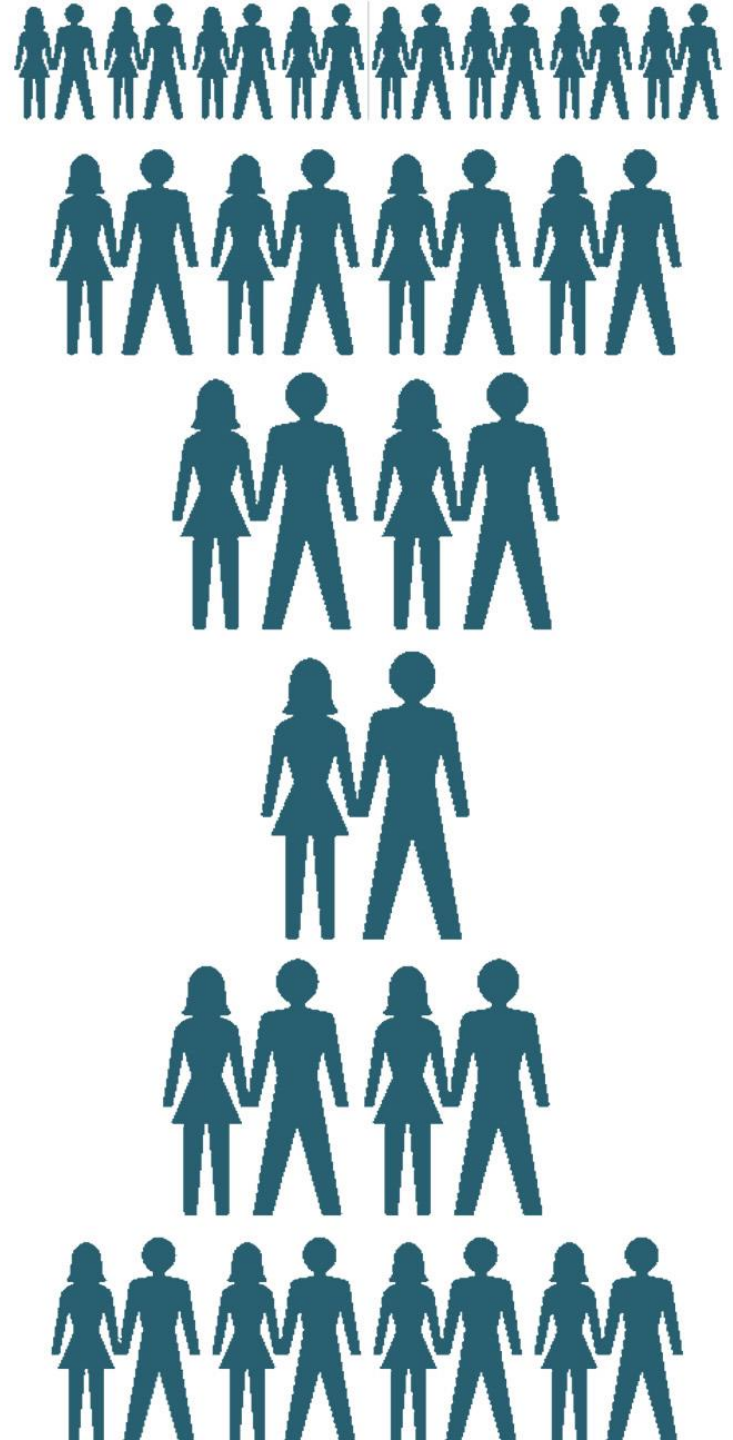
- The vast majority of genealogical DNA testing is based on autosomal DNA. Autosomal DNA is inherited from both parents, and men and women both receive the same service.
- With specialised Y-DNA and mtDNA tests, men can trace both their maternal haplogroup (from mtDNA) and their paternal haplogroup (through Y-DNA), but women can **only** trace their maternal haplogroup (through mtDNA). This is because the paternal haplogroup is traced through the Y chromosome, which women do not inherit. But haplogroups are a **tiny part** of your ancestral analysis.
- If a male relative (e.g., father, brother, paternal uncle or paternal male cousin) is genotyped, women can infer their own paternal haplogroup information from any of them.



What does a  
DNA test show me?

A DNA Case Study

[b4uc.xyz](http://b4uc.xyz)



# Access DNA via drop down menu

View a Photo Discovery that can **add 4 personal photos** for people in your family tree.

[View Discovery](#)



Lady Anne Home,...



Elizabeth Owen (born...



Lady Alice Mary...



1  
Shi

View a Photo Discovery that can **add 2 personal photos** for people in your family tree.

[View Discovery](#)



Lord William King Noel,...

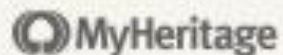


Hon. Anna Middleton...

- Overview
- Ethnicity Estimate
- DNA Matches
- DNA Tools **NEW**
- Manage DNA kits
- Surveys
- Upload DNA data
- Order DNA kits



# Access DNA Overview



Home

Family tree

Discoveries

**DNA**

Research

## DNA results



Ray Sarlin

This is you

[Select another person](#)

### Test additional family members

The more relatives test their DNA, the more you'll discover.

[Get suggestions](#)



Overview

Ethnicity Estimate

DNA Matches

Tools

## Ethnicity Estimate You have 6 ethnicities

[View full estimate](#)

Finnish  
**37.4%**

English  
**29.2%**

+4 others  
**33.4%**



## DNA Matches You currently have 11,586 matches

[View DNA Matches](#)

# My Ethnicity Estimate



Ray Sarlin

This is you

Select another person ▾

Get suggestions



Overview Ethnicity Estimate DNA Matches Tools

All ethnicities

All supported ethnicities

▶ Play Intro

Print

Share

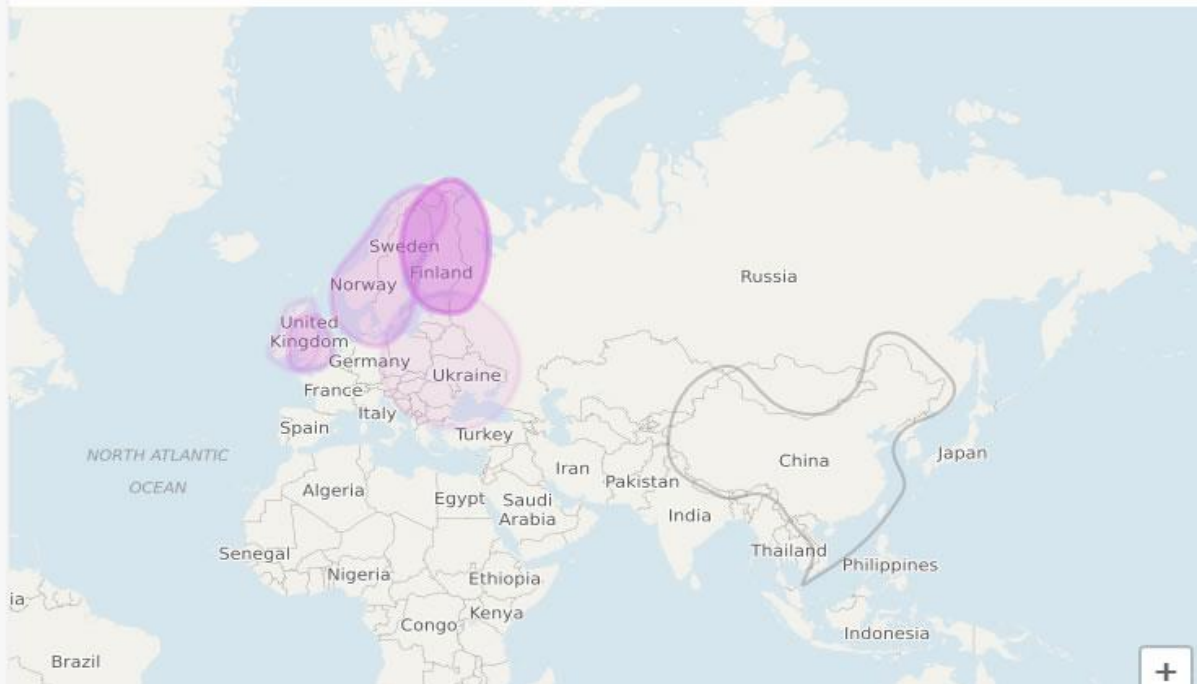


▼	Europe	99.1%
	• North and West Europe	97.6%
	Finnish	37.4%
	English	29.2%
	Scandinavian	23.6%
	Irish, Scottish, and Welsh	7.4%
	• East Europe	1.5%
	East European	1.5%
▼	Asia	0.9%
	• East Asia	0.9%
	Chinese and Vietnamese	0.9%

Ray Sarlin 100.0%

MyHeritageDNA

Show events from your family tree ?



# Scroll down to DNA match summary

DNA Matches You currently have 11,586 matches

[View DNA Matches](#)

 **Relationships**  
Review your DNA Matches by estimated relationship. Select any relationship to see the matches.

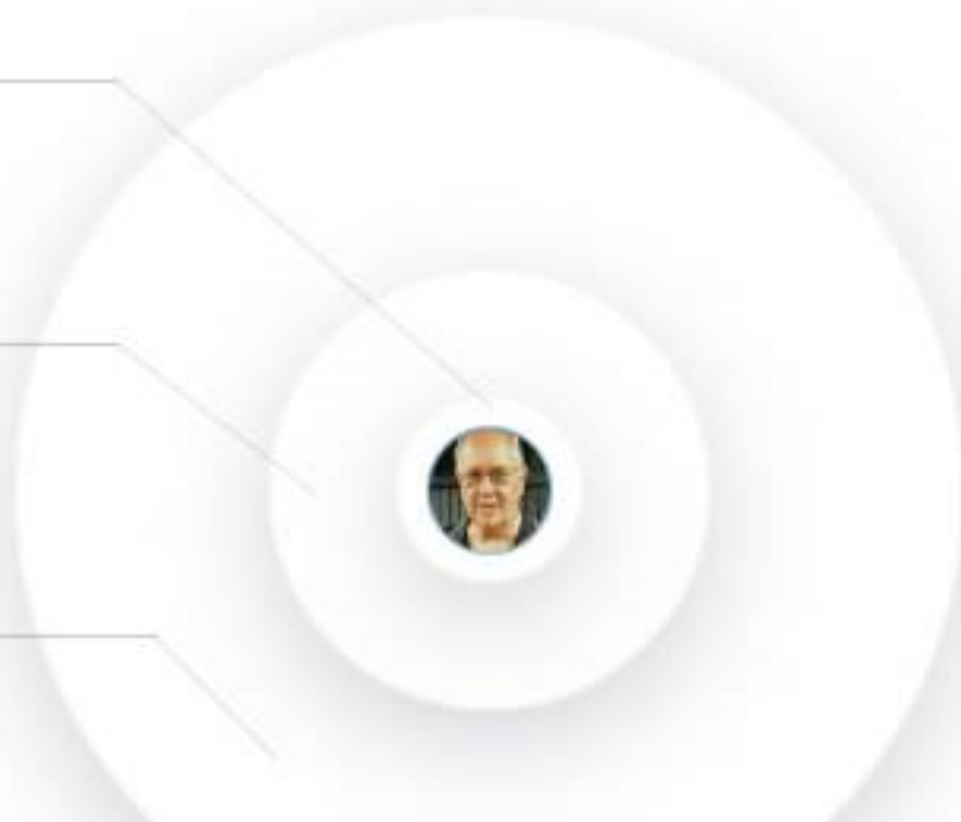
**0** **Close family**  
None of your DNA Matches appear to be a close family.  
Test your family members to get more matches.

[Order DNA kits](#)

**5** **Extended family**



**11,580** **Distant relatives**





# Scroll down to see countries with DNA matches

MyHeritage

Home

Family tree

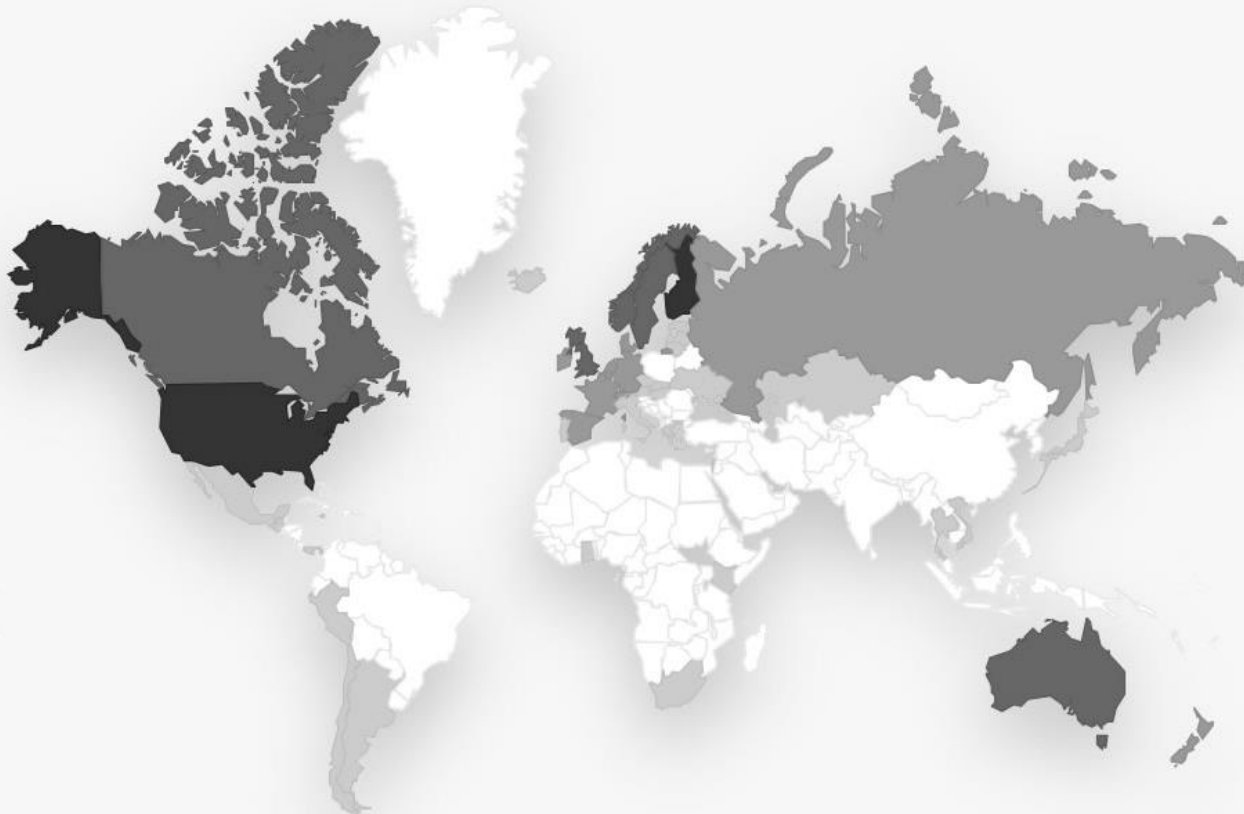
Discoveries

DNA

Research

## Locations

See where your DNA Matches live. Select any country to see the matches.




Country	Number of matches
 Finland	3,807
 USA	2,714
 Sweden	985
 Norway	497
 Great Britain	296
 Canada	163
 Australia	118
 Germany	76

# Click on a country (Ireland) to see matches

Showing 1–10 of 29 DNA Matches

 Filters

 Sort by

 All tree details ▾

 All relationships ▾

 Ireland ▾

 All ethnicities ▾

[Clear filters](#)




**Sampo Koskela**

Age: 30's

From: Ireland 

[Contact Sampo](#)

Estimated relationships

3rd - 5th cousin 

DNA Match quality 


Shared DNA: 0.5% (32.0 cM)

Shared segments: 3

Largest segment: 17.1 cM

[Review DNA Match](#)

[View tree](#)

 Appears in a family tree with one person that he manages




**Gerard O'Neill**

Age: 60's

From: Ireland 

[Contact Gerard](#)

Estimated relationships

3rd - 5th cousin 

DNA Match quality 

Shared DNA: 0.4% (31.1 cM)

Shared segments: 1

Largest segment: 31.1 cM

[Review DNA Match](#)

[View tree](#)

 Appears in a family tree with 264 people that he manages

 Ancestral places common to you and Gerard O'Neill include [Great Britain](#) and [Ireland](#).




**Veronica Malone**

Age: 50's

From: Ireland 

Estimated relationships

3rd cousin - distant cousin 

Low confidence

DNA Match quality 

Shared DNA: 0.3% (20.6 cM)

Shared segments: 3

Largest segment: 8.1 cM

[Review DNA Match](#)

# Ethnicities

[Home](#)[Family tree](#)[Discoveries](#)[DNA](#)[Health NEW](#)[Research](#)

## Ethnicities

See the ethnicity distribution of your DNA Matches. Select any ethnicity to see the matches.

Ethnicity	Your ethnicity results	Number of matches
Finnish	37.4%	7,503
English	29.2%	2,807
Scandinavian	23.6%	5,215
Irish, Scottish, and Welsh	7.4%	3,261
East European	1.5%	948
Chinese and Vietnamese	0.9%	10
North and West European	0%	2,016



# Scroll to bottom of index page

Eskimo/Inuit	0%	<div style="width: 0%;"></div>	3
Thai and Cambodian	0%	<div style="width: 0%;"></div>	3
Mongolian	0%	<div style="width: 0%;"></div>	2
Papuan	0%	<div style="width: 0%;"></div>	2
Somali	0%	<div style="width: 0%;"></div>	1
Ethiopian Jewish	0%	<div style="width: 0%;"></div>	1
Yemenite Jewish	0%	<div style="width: 0%;"></div>	1

## Need help understanding your results?

Hire a DNA expert who will help you make the most of your DNA Matches and Ethnicity Estimate.

[Learn more](#)



LegacyTree  
Genealogists

## Family Tree DNA analysis

Explore more about your DNA with our partner FTDNA

[Go to FTDNA>](#)



FamilyTreeDNA

# Click drop down to select DNA results



Home

Family tree

Discoveries

DNA

Research

## DNA results



Ray Sarlin

This is you

Select another person ▾

### Test additional family members

The more relatives test their DNA, the more you'll discover.

[Get suggestions](#)



Overview

Ethnicity Estimate

DNA Matches

Tools

### Theory of Family Relativity™ NEW

MyHeritage has found theories that may explain how you and some of your DNA Matches are related.

[View theories](#)



Showing 1–10 of 11,586 DNA Matches

Filters

Sort by



# Select individual match to explore

219-71 Beeston Street Ten... SSA Apply for Benefits, Social ... Bet on International Lotter... Alinta Customer - My Det...



Home

Family tree

Discoveries

DNA

Research

Overview

Ethnicity Estimate

**DNA Matches**

Tools

Showing 1–10 of 10 DNA Matches

Filters

Sort by



**Has Theory of Family Relativity™** ▾

All relationships ▾

All locations ▾

All ethnicities ▾

[Clear filters](#)



**Evan Custer**

Age: 20 or below

From: USA

[Contact Evan](#)

Estimated relationships

**3rd - 5th cousin**

DNA Match quality

Shared DNA: 0.7% (48.1 cM)

Shared segments: 4

Largest segment: 24.5 cM



[Review DNA Match](#)

[View tree](#)

Appears in a family tree with 1,413 people that he manages

- Evan Custer is your 3rd cousin once removed according to the Theory of Family Relativity™ (+1 more theory). [View theory](#)
- Ancestral surnames common to you and Evan Custer include [Anderson](#); [McClay](#); [Robinson](#) and [3 more](#).
- Your family tree has 39 Smart Matches™ with this tree. [View Smart Matches](#)
- Ancestral places common to you and Evan Custer include [Great Britain](#); [Minnesota, USA](#) and [8 more](#).



# Explore Theory of Family Relativity

MyHeritage found 2 theories that may explain how Evan Custer is related to you.

Theory 1: Evan Custer is your 3rd cousin once removed on your mother's side (via your great-great-grandmother)

[View another theory >](#)

**Path 1** Path 2 Path 3 Path 4 Path 5

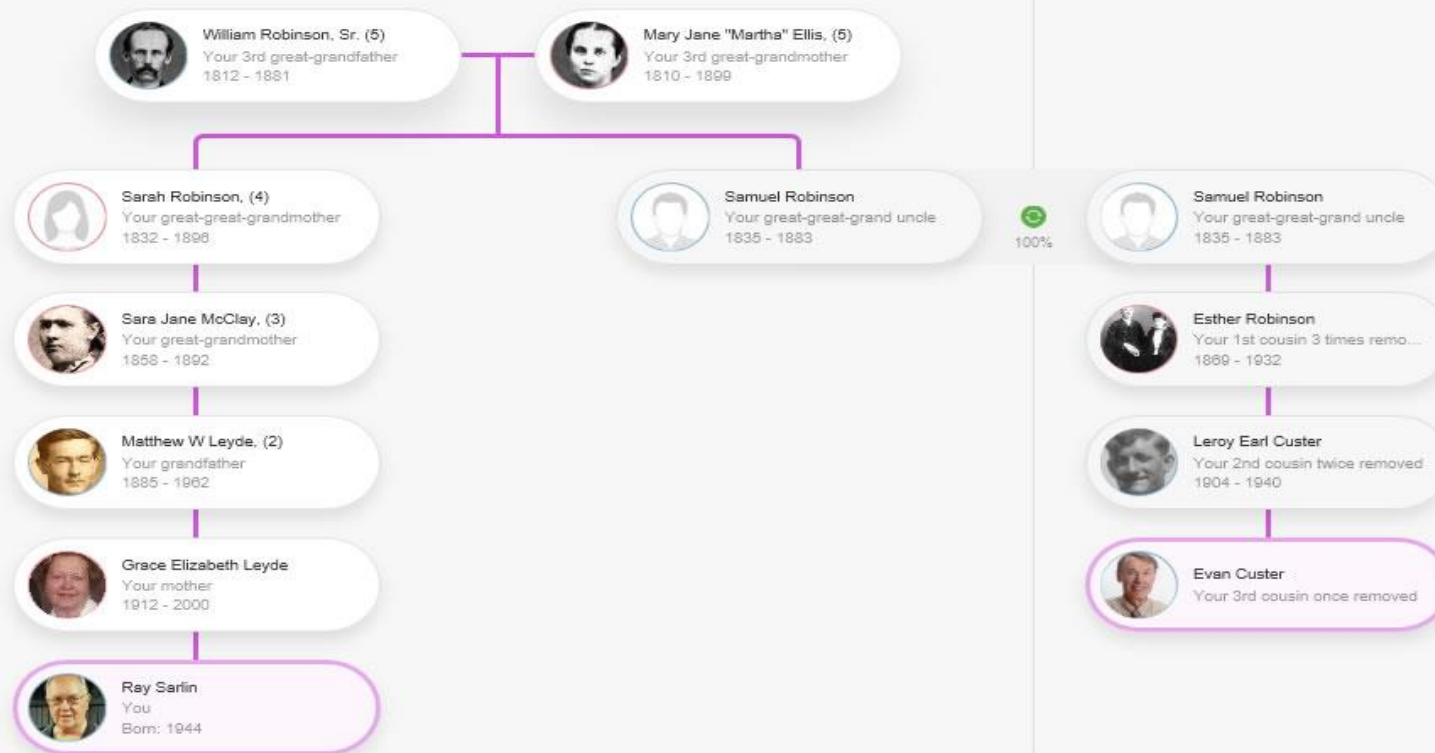
This path is based on 2 MyHeritage family trees, with 100% confidence



Sarlin+ managed by you



Custer Family Site (23andMe) managed by Evan Custer from USA



# Normal DNA match information



Ray Sarlin

This is you | Showing 1–10 of 11,586 DNA Matches



Janet Finnigan

Age: 70's

From: USA

[Contact Janet](#)

Estimated relationships

**2nd cousin - 3rd cousin once removed**

DNA Match quality

Shared DNA: **1.5%** (104.2 cM)

Shared segments: 8

Largest segment: 32.2 cM

[Review DNA Match](#)



Janet Finnigan is currently not associated with a family tree on MyHeritage. You can contact her for more information.



Rauli Mäntyvaara

Age: 80's

DNA managed by [Leena Mäntyvaara](#)

[Contact Leena](#)

Estimated relationships

**1st cousin twice removed - 3rd cousin once removed**

DNA Match quality

Shared DNA: **1.3%** (93.4 cM)

Shared segments: 7

Largest segment: 34.7 cM

NEW

[Review DNA Match](#)

[View tree](#)



Appears in a family tree with 2 people, managed by [Leena Mäntyvaara](#) from Finland



Lawrence McKenzie

Age: 80's

From: USA

[Contact Lawrence](#)

Estimated relationships

**3rd - 4th cousin**

DNA Match quality

Shared DNA: **1.2%** (84.4 cM)

Shared segments: 5

Largest segment: 51.3 cM

[Review DNA Match](#)



Lawrence McKenzie is currently not associated with a family tree on MyHeritage. You can contact him for more information.



Linda Evangelisti

Age: 80's

From: USA

[Contact Linda](#)

Estimated relationships

**1st cousin twice removed - 4th cousin**

DNA Match quality

Shared DNA: **1.2%** (81.5 cM)

Shared segments: 5

Largest segment: 35.8 cM

[Review DNA Match](#)



Linda Evangelisti is currently not associated with a family tree on MyHeritage. You can contact her for more information.

# My DNA Raw Data as an Excel file

MyHeritage\_raw\_dna\_data.csv - Microsoft Excel non-commercial use

File Home Insert Page Layout Formulas Data Review View

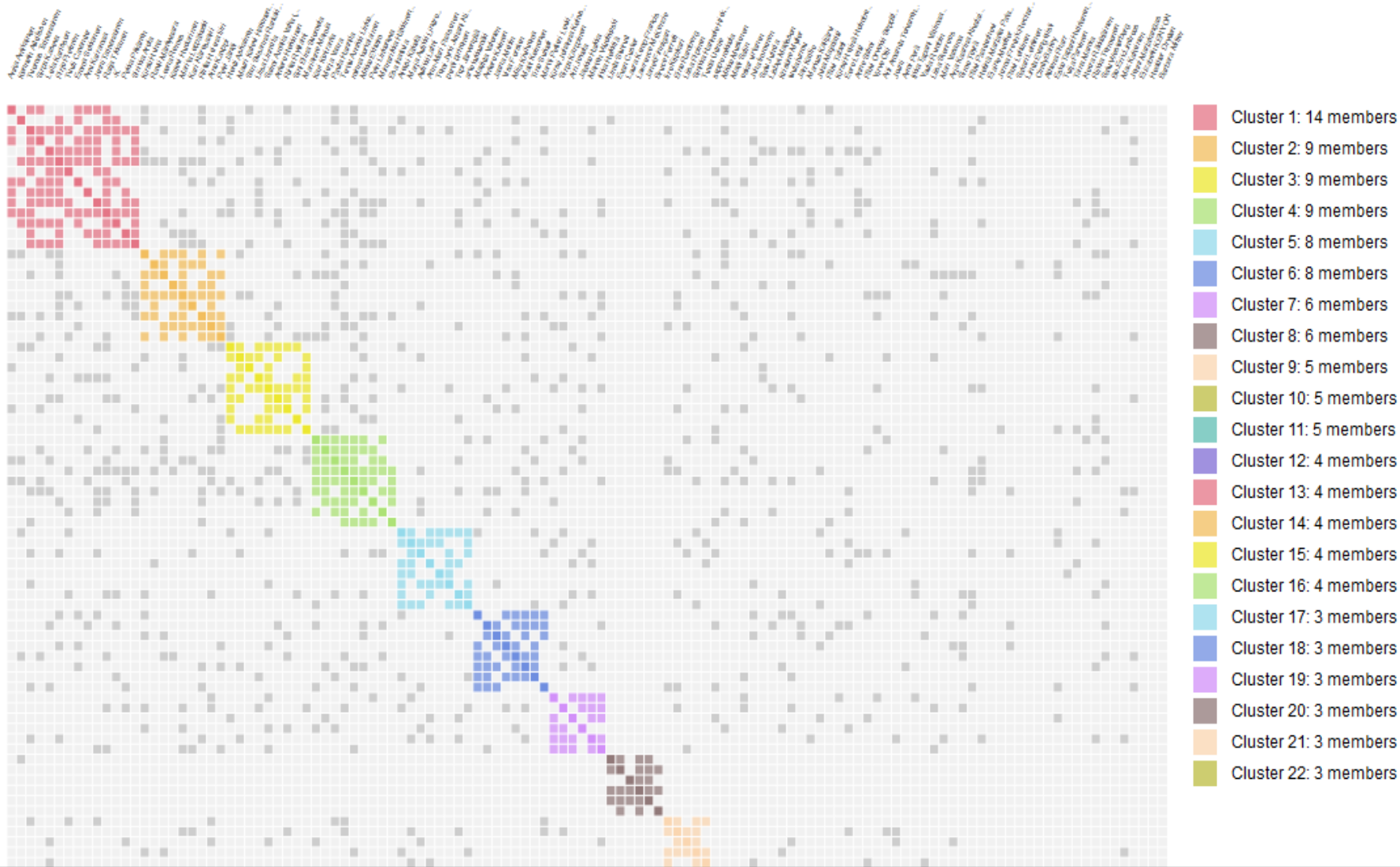
Paste Cut Copy Format Painter Clipboard Font Alignment Number Conditional Formatting Format as Table Styles

D1

	A	B	C	D	E	F	G	H	I	J	K	L	M
1	# MyHeritage DNA raw data.												
2	# This file was generated on 2019-03-02 09:27:35												
3	# For each we provide chromosomal base pair position and genotype. The genotype is reported on the forward (+) strand with respect to the												
4	# THIS INFORMATION IS FOR YOUR PERSONAL USE AND IS INTENDED FOR GENEALOGICAL RESEARCH												
5	# ONLY. IT IS NOT INTENDED FOR MEDICAL OR HEALTH PURPOSES. PLEASE BE AWARE THAT THE												
6	# DOWNLOADED DATA WILL NO LONGER BE PROTECTED BY OUR SECURITY MEASURES.												
7	RSID	CHROMOS	POSITION	RESULT									
8	rs4477212	1	82154	AA									
9	rs3094315	1	752566	--									
10	rs3131972	1	752721	GG									
11	rs1256203	1	768448	--									
12	rs1212481	1	776546	--									
13	rs1124077	1	798959	GG									
14	rs6681049	1	800007	--									
15	rs4970383	1	838555	AC									
16	rs4475691	1	846808	CC									
17	rs7537756	1	854260	AG									

**CONTINUES DOWN TO LINE 720,823.**

# My DNA clustered by DNA matches



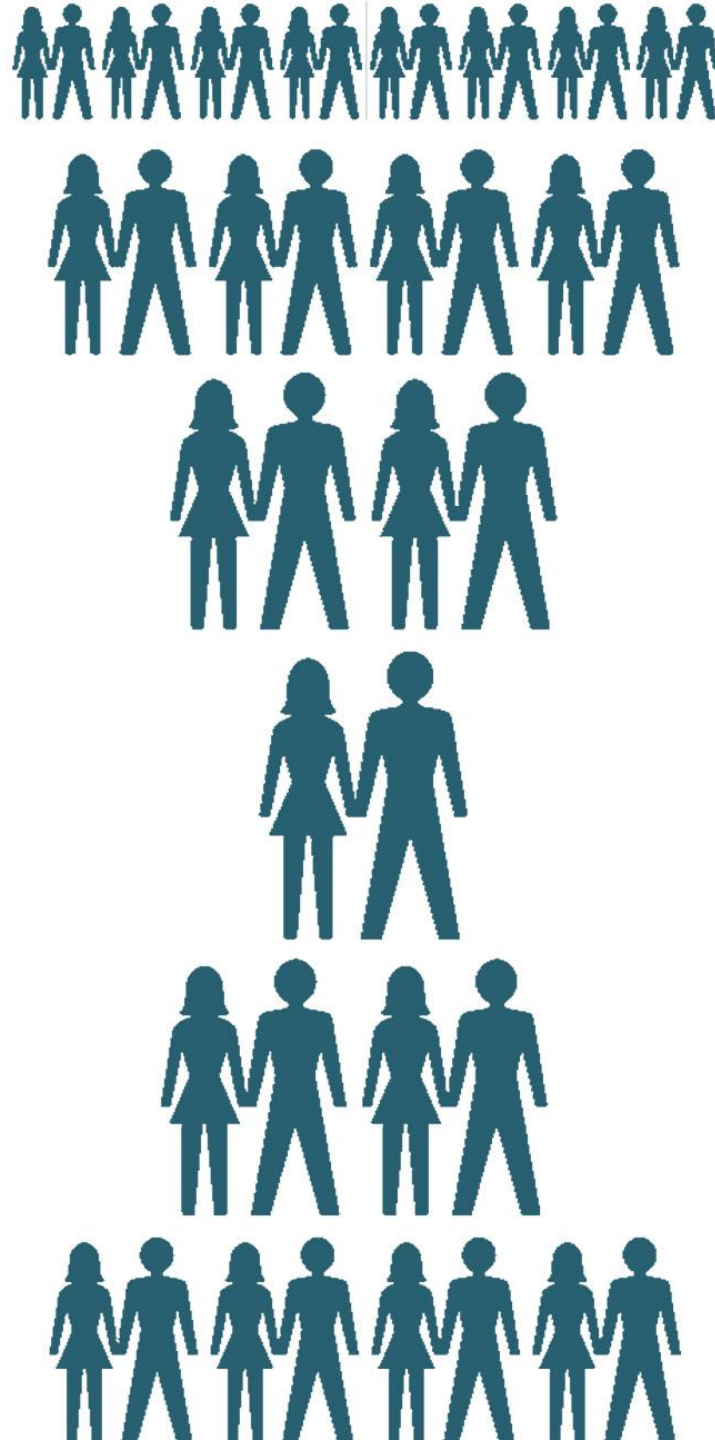




# What does a DNA test show me?

## Case Study extension

**b4uc.xyz**



# An apparent DNA anomaly



Ray Sarlin

This is you

Select another person ▾

Get suggestions

Overview Ethnicity Estimate DNA Matches Tools

All ethnicities

All supported ethnicities

▶ Play Intro

Print

Share

▼	Europe	99.1%
•	North and West Europe	97.6%
	Finnish	37.4%
	English	29.2%
	Scandinavian	23.6%
	Irish, Scottish, and Welsh	7.4%
•	East Europe	1.5%
	East European	1.5%
▼	Asia	0.9%
•	East Asia	0.9%
	Chinese and Vietnamese	0.9%
Ray Sarlin		100.0%

MyHeritageDNA

Show events from your family tree ?





# A little internet research

- When trying to run down this apparent anomaly, I came across a fascinating 2012 study in the *American Journal of Human Genetics* entitled The Genetic Legacy of the Mongols.

## Report

*Am. J. Hum. Genet.* 72:717–721, 2003

### The Genetic Legacy of the Mongols

Tatiana Zerjal,<sup>1</sup> Yali Xue,<sup>1,2</sup> Giorgio Bertorelle,<sup>3</sup> R. Spencer Wells,<sup>4</sup> Weidong Bao,<sup>1,3</sup> Suling Zhu,<sup>1,3</sup> Raheel Qamar,<sup>1,4</sup> Qasim Ayub,<sup>1,6</sup> Aisha Mohyuddin,<sup>1,6</sup> Songbin Fu,<sup>2</sup> Pu Li,<sup>2</sup> Nadira Yuldashева,<sup>4,7</sup> Ruslan Ruzibakiev,<sup>7</sup> Jiujun Xu,<sup>8</sup> Qunfang Shu,<sup>8</sup> Ruofu Du,<sup>2</sup> Huanming Yang,<sup>3</sup> Matthew E. Hurles,<sup>8</sup> Elizabeth Robinson,<sup>1,2</sup> Tudevdayva Gerelsaikhan,<sup>1,1</sup> Bumbein Dashnyam,<sup>9</sup> S. Qasim Mehdi,<sup>3</sup> and Chris Tyler-Smith<sup>1</sup>

<sup>1</sup>Department of Biochemistry, University of Oxford, Oxford; <sup>2</sup>Department of Medical Biology, Harbin Medical University, Harbin, China; <sup>3</sup>Department of Biology, Università di Ferrara, Ferrara, Italy; <sup>4</sup>Wellcome Trust Centre for Human Genetics, University of Oxford, Headington, United Kingdom; <sup>5</sup>Institute of Genetics, Chinese Academy of Sciences, Beijing; <sup>6</sup>Biomedical and Genetic Engineering Lab, Islamabad; <sup>7</sup>Institute of Immunology, Academy of Sciences, Tashkent, Uzbekistan; <sup>8</sup>McDonald Institute, University of Cambridge, Cambridge, United Kingdom; and <sup>9</sup>Institute of Biotechnology, Mongolian Academy of Sciences, Ulaanbaatar, Mongolia

We have identified a Y-chromosomal lineage with several unusual features. It was found in 16 populations throughout a large region of Asia, stretching from the Pacific to the Caspian Sea, and was present at high frequency: ~8% of the men in this region carry it, and it thus makes up ~0.5% of the world total. The pattern of variation within the lineage suggested that it originated in Mongolia ~1,000 years ago. Such a rapid spread cannot have occurred by chance; it must have been a result of selection. The lineage is carried by likely male-line descendants of Genghis Khan, and we therefore propose that it has spread by a novel form of social selection resulting from his behavior.

The patterns of variation found in human DNA are usually considered to result from a balance between neutral processes and natural selection. Among the former, mutation, recombination, and migration increase variation, whereas genetic drift decreases it. Natural selection can act to remove deleterious variants (purifying selection), maintain polymorphism (balancing selection), or produce a trend (directional selection), or produce the latter are rare in humans, but probable examples of those associated with resistance to malaria (Hamblin and Di Rienzo 2000) or unidentified pathogens (Stephens et al. 1998), can be recognized by the "signature" they leave in the genome. The rapid increase in frequency of the selected allele and its linked sequences results in a haplotype that is found at higher frequency than would be expected from its degree of variation. We have now

identified such a haplotype on the Y chromosome, but we suggest that its spread results not from a biological advantage, but from human activities recorded in history.

In surveys of DNA variation in Asia, we typed 2,123 men with ≥32 markers to produce a Y haplotype for each man; these included 1,126 individuals described elsewhere (Qamar et al. 2002; Zerjal et al. 2002). Over 90% of the haplotypes showed the usual pattern (Mohyuddin et al. 2001); most males had a unique code, and the few haplotypes present in more than one individual were generally found within the same population. However, we also saw one pattern that was novel in two respects. First, there was a high frequency of a cluster of closely related lineages, collectively called the "star cluster" (fig. 1, shaded area). Second, star-cluster chromosomes were found in 16 populations throughout a large geographical area extending from Central Asia to the Pacific (fig. 2); thus, they do not result from an event specific to any single population. We can deduce the most likely time to the most recent common ancestor (TMRCA) and place of origin of this unusual lineage from the observed genetic variation. To do this, it is first necessary to distinguish star-cluster chromosomes from the remainder. For this, we used the criterion that haplotypes linked to the central one in the shaded area of the network without gaps would be included (fig. 1).

Received September 27, 2002; accepted for publication November 25, 2002; electronically published January 17, 2003.  
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0002-9297/2003/7217-717\$15.00

# A little internet research

- Y-DNA studies show that 1 in 200 Finnish men are direct line descendants of Genghis Khan (1162-1227), who had the most male children of anyone in history (>2,000).
- In addition, some 8-10% of men from lands covered by his empire have his Y-chromosome. In Asiatic societies, descent from Genghis Khan became a mark of prestige even within Islamic societies, almost essential for would-be warlords.
- In a separate study, genetic clusters unique in Europe to the Sami Peoples indigenous to northern Finland appear to have migrated from Asia at about the time of the Mongols.
- These studies and other indicators (including tracing ancestors to the edge of the area of the Mongol Golden Horde) enable me to form a hypothesis about the seeming 0.9% Asian DNA anomaly.



# Testing the Hypothesis

To test the hypothesis that my 0.9% East Asian comes from Genghis Khan, I took a Y-DNA test.



A screenshot of the FamilyTreeDNA dashboard. The top navigation bar includes the FamilyTreeDNA logo, 'HOME', 'myDNA', 'myTREE', 'myPROJECTS', 'ADD ONS &amp; UPGRADES', a bell icon, and a shopping cart icon. Below the navigation bar, there is a 'Welcome to myFTDNA' message. On the right side, there are two buttons: 'N-M231 Y-DNA Haplogroup' and 'Get Yours! mtDNA Haplogroup'. The main content area is divided into several sections: 'Your Account' (with a profile card for Ray Sarlin), 'Family Tree' (with 'myFamilyTree' and 'myFamilyTree Beta' options), 'Family Finder' (with 'Matches', 'Chromosome Browser', 'Linked Relationships', 'myOrigins', and 'ancientOrigins' options), and 'Y-DNA' (with 'Matches', 'Ancestral Origins', 'Haplotree &amp; SNPs', 'Matches Maps', 'Migration Maps', 'SNP Map', 'Haplogroup Origins', 'Y-STR Results', and 'Print Certificates' options). A 'Cookie Policy' update notification is visible in the top left of the dashboard area.

My Y-DNA dashboard

# Results



## Ray's updates

Refund Policy updated on October 21, 2019 **IMPORTANT**

## myProjects

T2 mtDNA

MANAGE PROJECTS

JOIN A PROJECT

## Badges



Family Ancestry

## Family Finder

Transfer Unlock

See the percentage breakdown of your origins as well as your ancient origins, and connect with your autosomal DNA relatives on all of your ancestral lines within the last 5 generations.

Results Completed: April 11, 2019



Matches



myOrigins



Matrix



Data Download



Chromosome Browser



ancientOrigins

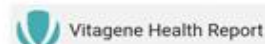


Advanced Matches



Learn More

## Partner Services



Vitagene Health Report



Paternal Ancestry N-Y7795

## Y-DNA

Y12 Y25 Y37 Y67 Y111 Big Y

Follow the migration paths of your paternal line's ancestors, and connect with your Y-DNA matches.

Results Completed: September 13, 2019



Matches



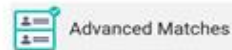
Ancestral Origins



Matches Maps



Data Download



Advanced Matches



Haplogroup Origins



Migration Maps



Y-STR Results



Haplotree & SNPs



SNP Map



Print Certificates



Learn More

# Where is Y-DNA Haplogroup N-M231 found?



## Y-DNA - SNP Map





# How did the haplogroup spread?

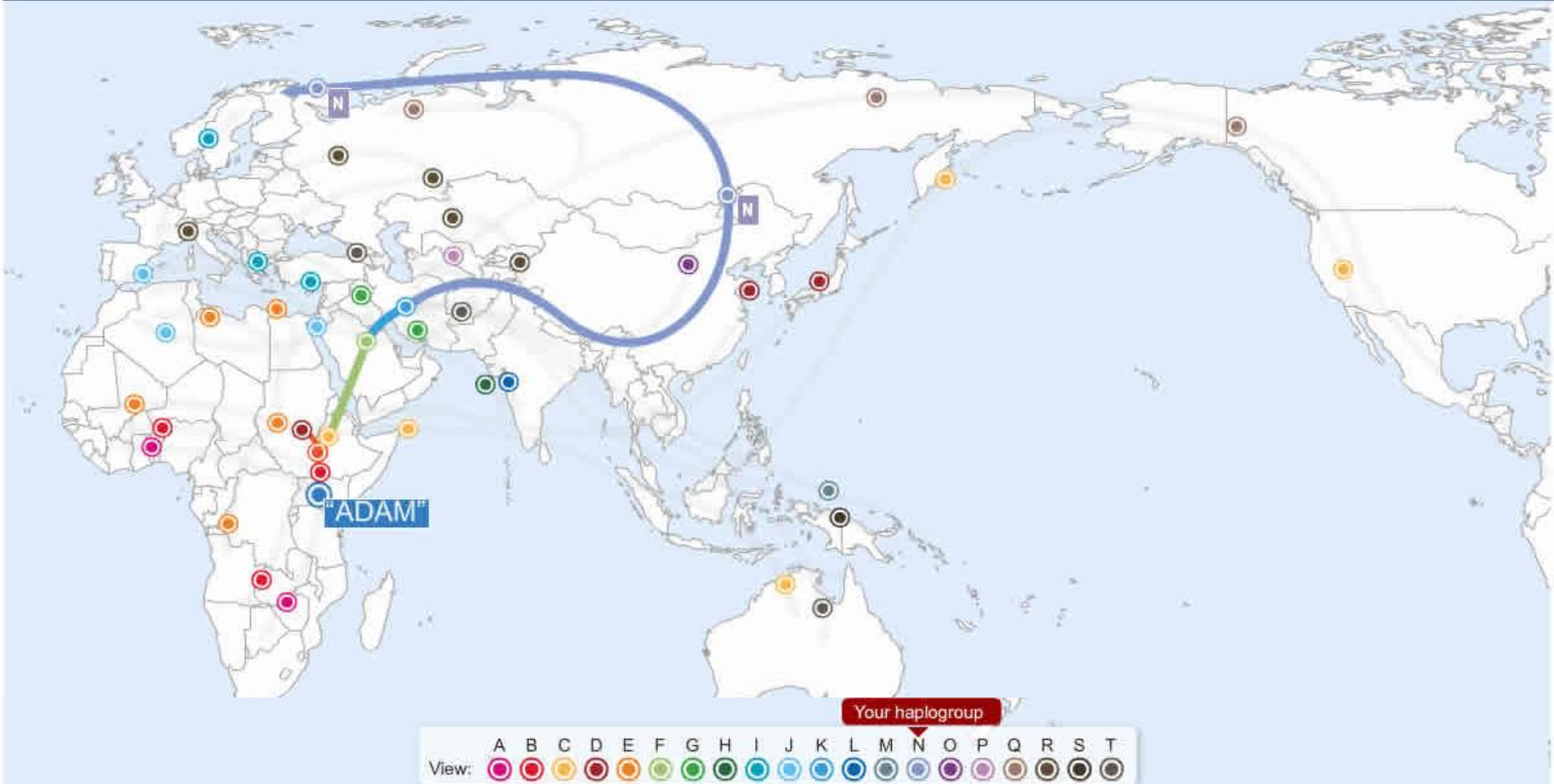


## Y-DNA - Migration Maps

Migration Map

Frequency Map

### MIGRATION MAP





# Among the 4,459 individual matches

## Family Finder - Matches













[Advanced Search](#)

Chromosome Browser  In Common With  Not In Common With [Reset Filter](#)

1-30 of 4459 [«](#) [<](#) [>](#) [»](#) Page 1 / 149 [Go](#)

All (4459)  Paternal (44)  Maternal (0)  Both (0)

[Calculating Family Matching](#)

	Name	Match Date	Relationship Range	Shared cM	Longest Block	X-Match	Linked Relationship	Ancestral Surnames	
<input type="checkbox"/>	 Leena Marja Blomvall	04/11/2019	2nd Cousin - 3rd Cousin	169	40		2nd Cousin 1R	Kujala (Tammela) / Lehtinen (Kalanti) / Lahomaa (Lokalahti) / Vahtonen (Lokalahti)	
<input type="checkbox"/>	 Richard J Roberts	04/11/2019							
<input type="checkbox"/>	 Samuli Röynä								
<input type="checkbox"/>	 Esko Keskitalo							/ Cajanus (Finland) / Heikkinen (Finland) / Keskitalo (Finland) / Liisa Sigfridintytär	
<input type="checkbox"/>	 Mr. Timo Heikki Tamminen	04/11/2019							
<input type="checkbox"/>	 Mirva Ranta	05/30/2019	2nd Cousin - 4th Cousin	90	19	X-Match			

Nestled among the 4,459 matches are three Y-DNA markers to the “Golden Family” of Genghis Khan – the man himself, his first son Jochi, and his first son Batu, my direct ancestors.

# Welcome to the family, Mr. Chinggis Khan.

Or would you prefer me to call you, "Oh Mighty Conqueror of the World?"

The greatest joy for a man is to defeat his enemies, to drive them before him, to take from them all they possess, to see those they love in tears, to ride their horses, and to hold their wives and daughters in his arms.



# Questions???

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[www.glasbergen.com](http://www.glasbergen.com)



**“You don’t look anything like the long haired, skinny kid I married 25 years ago. I need a DNA sample to make sure it’s still you.”**