

# **Objectives**



- Use Y-DNA results to research the Patrilinear line.
  - Identify father's father's (etc.) ancestral heritage.
- Y chromosome basics, types of markers, and how results help find male relatives.
- BigY 700 test (Rolls Royce of Y-DNA tests).
  - Y-DNA in genealogically relevant timeframes.
  - New "Discover" tools at FTDNA.
  - Tools for surname projects/paternal research.
- Convince Jim Cross to take the BigY test.

### Y Chromosome

- The Y chromosome is the sex-determining chromosome in humans
- Y-chromosome has about 59 million base pairs
- Y does not have matching chromosome, so it escapes recombination every generation
- Y-DNA is passed down through the male line with only random mutation

### Y-DNA Tests

- Y-DNA Test in use longest, best track record
- Only males inherit the Y chromosome, so can only be used to trace the direct paternal line
- Great genealogical value since Y-DNA has same inheritance pattern as surnames
- Uses 2 types of markers: STRs and SNPs
  - Short Tandem Repeat
  - Single Nucleotide Polymorphism

### **Y-DNA** Test Evolution

### STR testing

- Number of markers increased
- -12 > 25 > 37 > 67 > 111 > 500 > 700

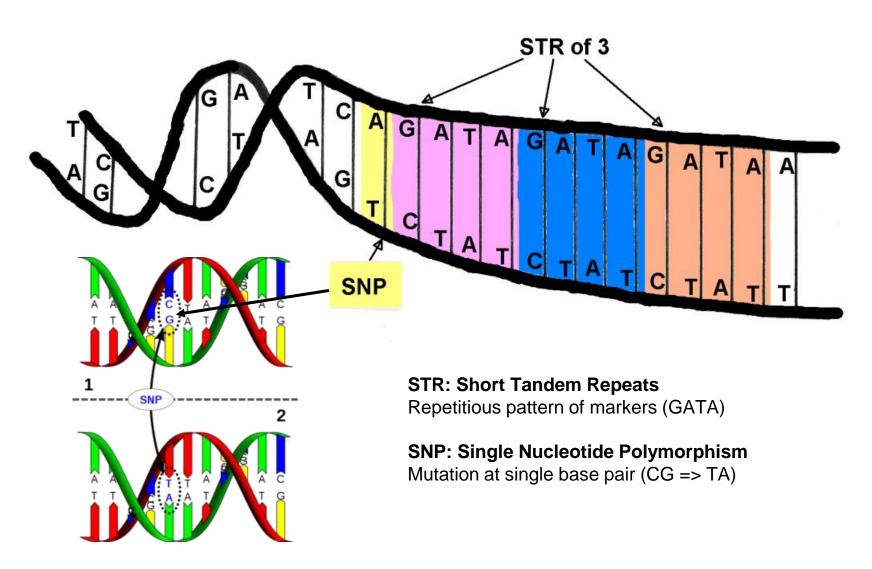
### SNP testing

- Expanding number of SNP discoveries
- New Terminal Branches on Y Tree

### Big Y

- Walk through the Y
- Big Y advancement caused SNP "tsunami"

## STRs & SNPs

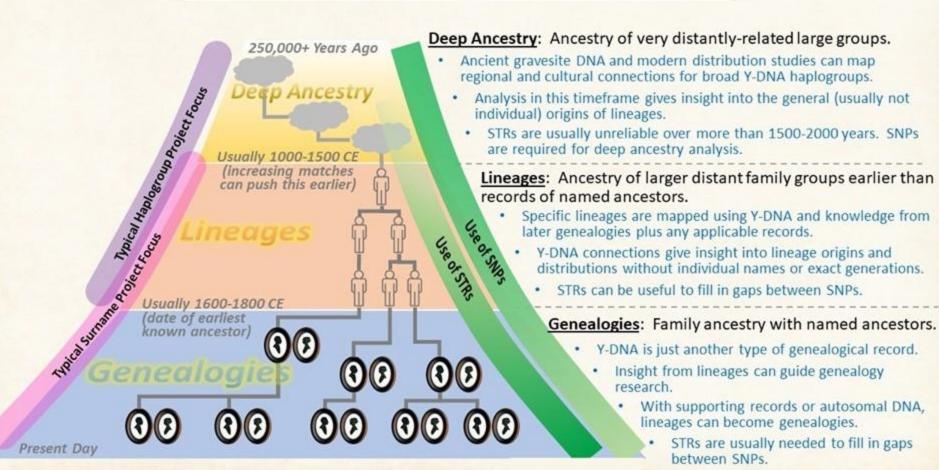


### Differences of STRs & SNPs

- STRs represent Haplotypes
  - Determine matches in recent times
  - Past 500 years
  - STR changes happen often
  - Can differ between father and son
- SNPs define Haplogroups
  - Ancient Origins thousands of years
  - SNPs happen infrequently
  - SNPs inherited from same ancestor
  - Everyone sharing a particular Y-SNP is related

# Y-DNA in Genealogy

### **Y-DNA: Three Periods of Ancestry**

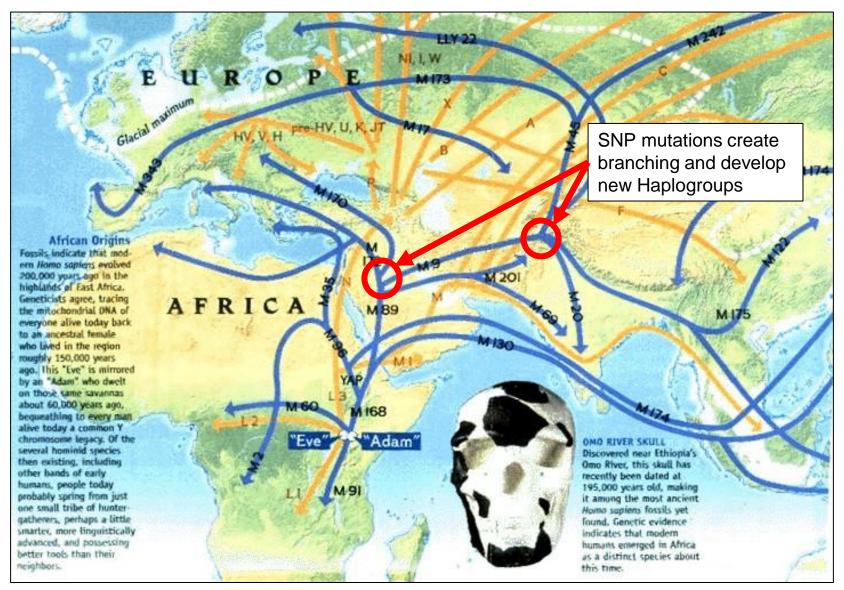


Note: Borrowed with permission from David Vance

## Haplogroups

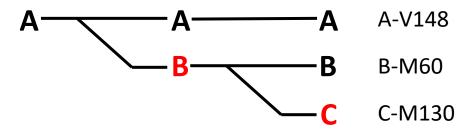
- Genetic group of people who share a common ancestor on patrilineal or matrilineal line
- Haplo (haploid) = single (refers to single copy)
- Y-DNA & mtDNA Haplogroups are different
- Haplogroups are defined by SNPs (mutations)
  - All descendants will carry that mutation
  - Main branch has sub-branches or "subclades"
- Y-DNA phylogenic tree
  - History of human relatedness and migration.
  - Unlike autosomal DNA, which mixes within a population, Y-DNA unambiguously traces a family's male line.

## **SNP Branching of Haplogroups**

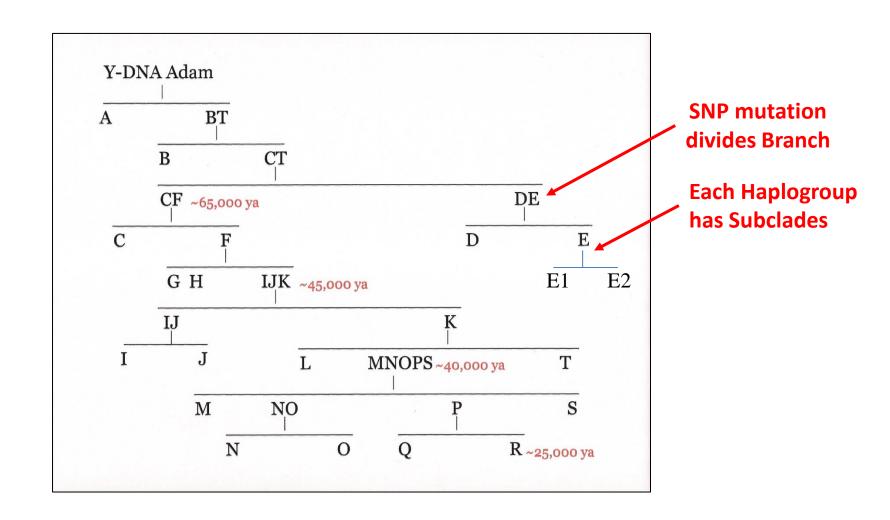


### Y-DNA SNP Tests

- Single Nucleotide Polymorphism (SNP)
- Also called Y Haplogroup Test
- Used for Patrilineal Deep Ancestry
- Each subclade has a SNP of its own
- First mutation split Haplogroup A into A and B
- Each mutation divided tree into finer branches
- Defined by Terminal SNPs



## Y-DNA Haplogroups

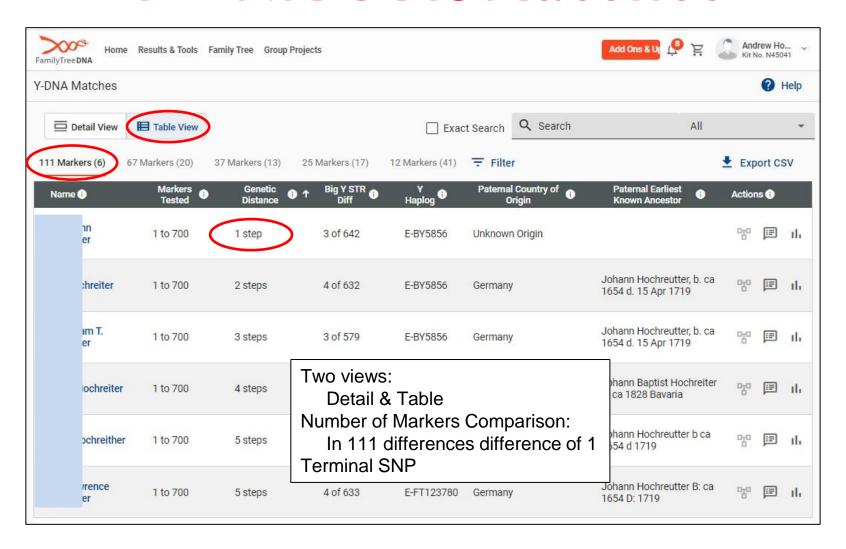


## Y DNA STR Matching

- STRs were used to determine male relatedness
- Increased from 12 to 11, now 700 in BigY
- Relationships determined by the first 111 STR markers, not the additional STR markers
- Results above 111 markers not available on the public project pages
- "Every family group who is participating in Y DNA testing wants to discover markers that delineate between various male lines descending from a specific progenitor."\*

<sup>\*</sup>The Big Y Test Increases Again to Big Y-700, DNAeXplained – Genetic Genealogy, Posted on January 30, 2019 by <u>robertajestes</u>

## **Y-DNA STR Matches**



## Big Y Results

- Big Y introduced in 2014
  - Provided deep SNP testing
- Big Y 500 introduced in 2017
  - Provided +389 STRs beyond 111 markers
  - Free Upgrade to Big Y testers
- Big Y 700 introduced in 2019
  - Provided +200 more STRs
  - Must pay and retest for additional STRs

# Big Y Test

- Big Y introduced in 2014
- GOAL: Read all of the Y chromosome that was useful for genealogical purposes
- Huge Impact on Y-DNA Science

Chromosome Y has roughly 57,200,000 nucleotides or base pairs (bp).



Roughly 23.6 Mbp of chromosome Y (white) are genealogically relevant Genealogically relevant regions (white) are those that are passed intact from father to son with high fidelity Other regions are either a) highly repetitive sequence (black: inaccessible to NGS sequencing technology) or, b) subject to recombination with chrX (grey; pseudo autosomal regions PAR1 and PAR2) and therefore of limited utility for genealogical applications.\*

<sup>\*</sup>Big Y-700 White Paper, Family Tree DNA, March 22, 2019

### **New SNP Research**

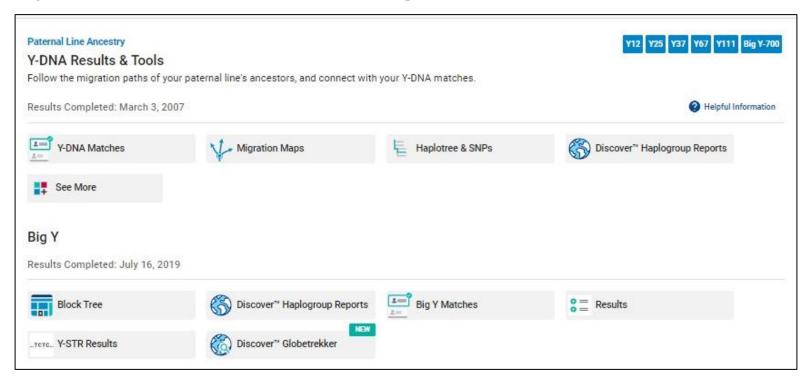
- Revealing thousands of new SNPs
- Y-DNA Tree has finer details (more subclades)
- In past, SNPS represented mutations that happened thousands of years ago
- New SNPS in the past 100-500 years
- Currently 25,000 SNPs, could reach 250,000
- More geographic granularity
- Possible "Family SNPs"

## Big Y 700 Improvements

- New Chemistry
  - More uniform coverage of Y chromosome
  - Quality reads of regions previously unavailable
  - Provides more consistent results
  - Provides better coverage, fewer no-reads
  - Allows for more STRs to be accessed and reliably read

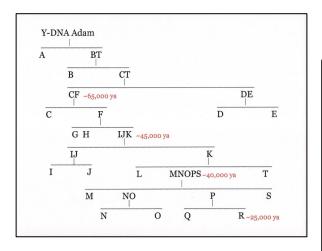
## Big Y Tools

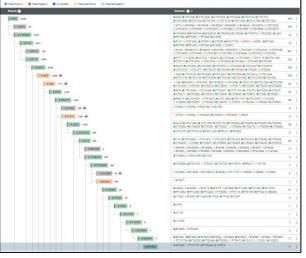
 These tools provide the means to compare your results to others who took Y-DNA tests to explore your terminal SNP and ages of various mutations.

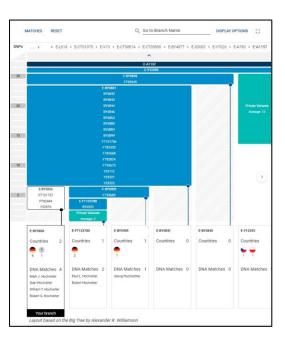


# New Big Y Block Tree

 Personalized Tree showing all your SNPs from ancestral (original value) to derived (mutated state)

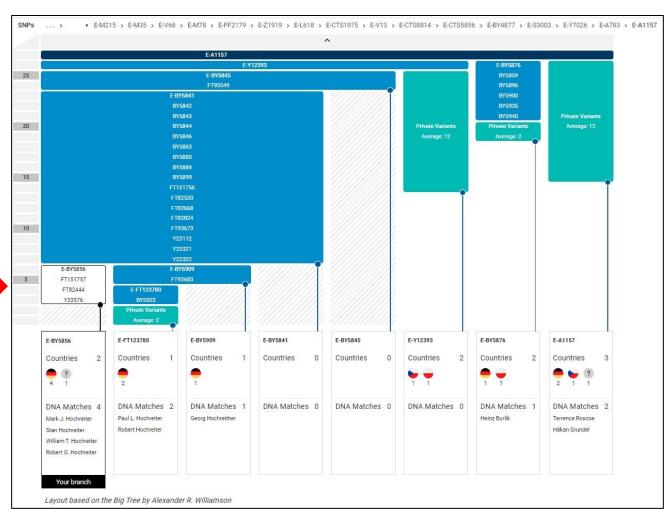




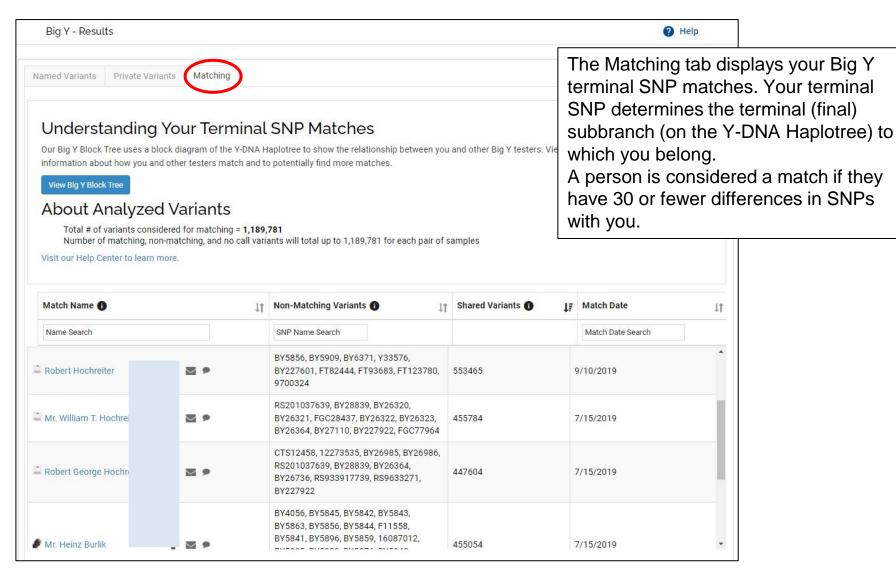


## Big Y Block Tree Features

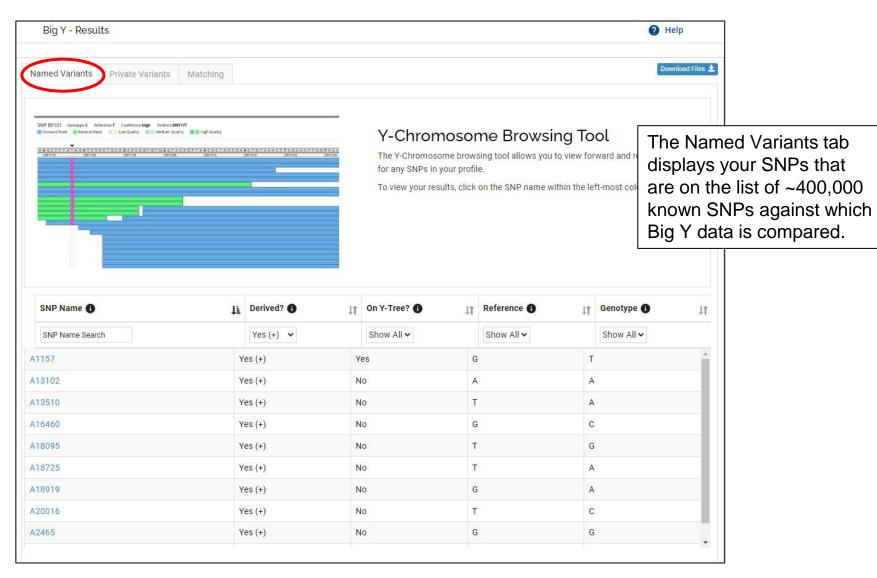
Distance, Years & SNPs
The general consensus is that a SNP generation is someplace, on average, between 80 to 140 years.
Each SNP represents one grey block



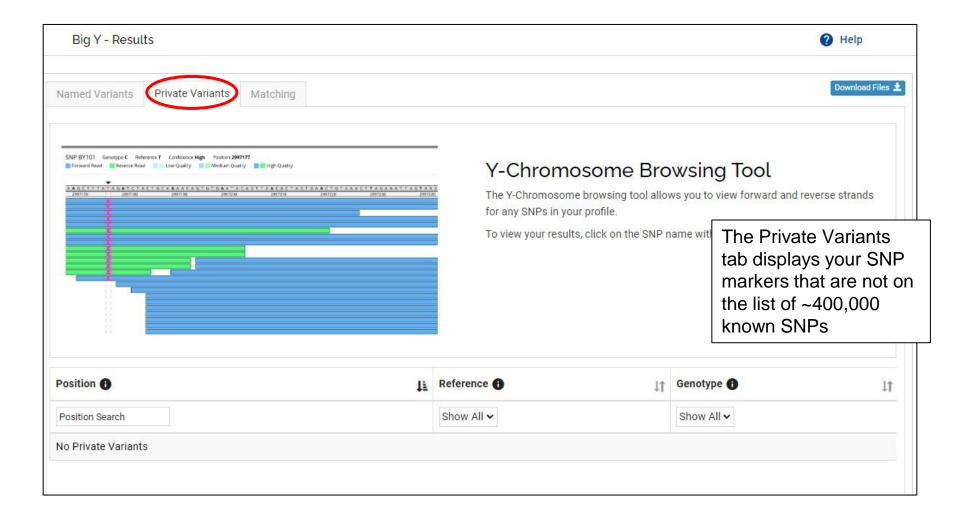
# Big Y SNP Matching



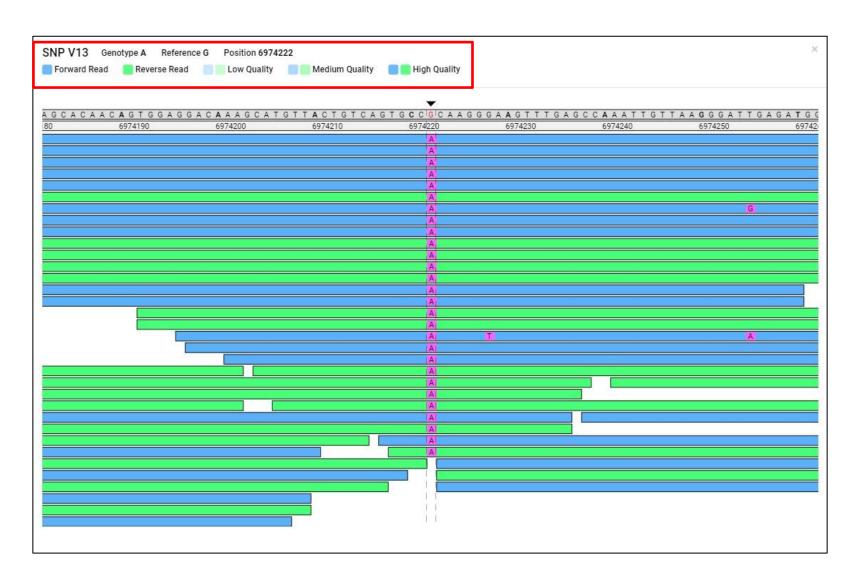
## **SNP Named Variants**



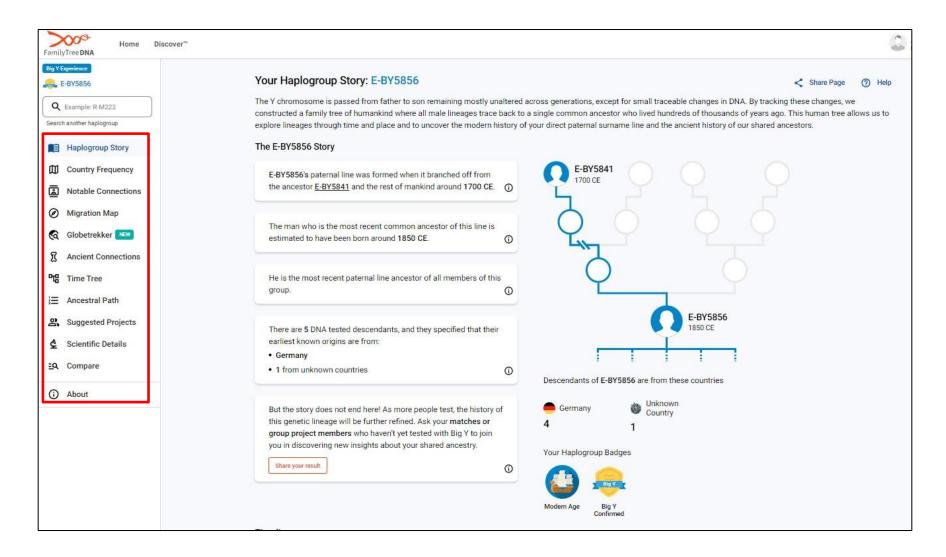
## **SNP Private Variants**



### Y-Chromosome Browser Tool



## FamilyTreeDNA Discover



### Discover Sources

#### You already have the Big Y test!

Your haplogroup information will change and be refined over time as more people test their Y-DNA. We are always adding more connections and features so check back often to see what new information is available:

You can also discover more about your other ancestral paternal lineages by testing the Y-DNA of your maternal grandfather, uncles, and genetically male cousins.

#### Where does this information come from?

#### FamilyTreeDNA Y-DNA Database

The longest-running, largest, and most comprehensive Y-DNA database with hundreds of thousands of testers from 200 different countries around the world.

#### **Population Genetic Studies**

Thousands of genomes from hundreds of studies of present-day individuals from around the world, including many populations from parts of the world that are otherwise underrepresented in DNA testing databases.

#### **Carlos Quiles**

Dr. Carlos Quiles runs the Indo-European.eu website and has collected and curated a large list of ancient DNA samples and associated metadata, some of which has been used for this website.

#### **Phylogenetics**

The FamilyTreeDNA Y-DNA Haplotree is the largest and most comprehensive phylogenetic Tree of Humankind based on the largest database of high-coverage Y-DNA sequences. It is curated by phylogenetic specialist Michael Sager.

#### TMRCA Algorithm

The state-of-the-art FamilyTreeDNA algorithm for inferring age estimates for the Y-DNA Haplotree. Developed together with Iain McDonald.

#### Genographic Project

National Geographic's global, citizen science project that helped map ancient human migration. The Project team employed dozens of international scientists and data from over one million people. Although the Project ended in 2020, the research continues.

#### **Ancient DNA**

Thousands of ancient DNA samples spanning tens of thousands of years of human history from archaeological remains from all corners of the world.

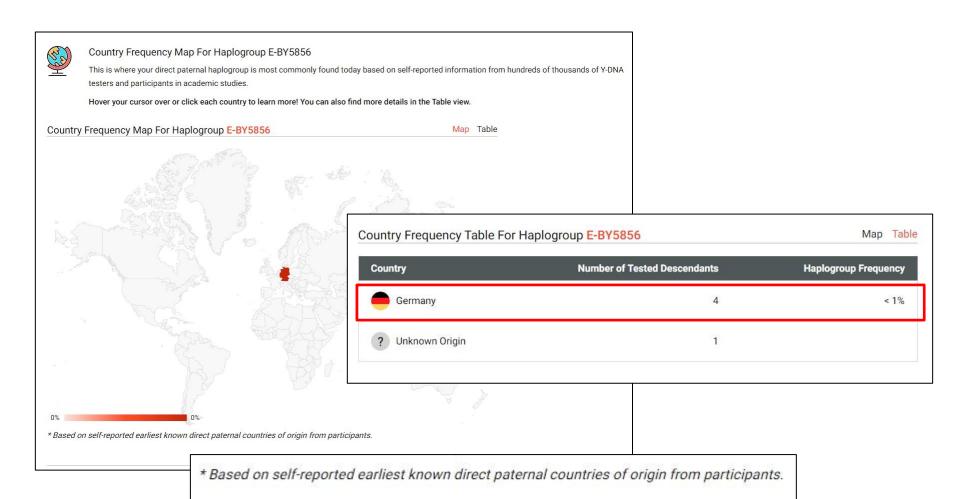
#### FamilyTreeDNA Group Projects

Citizen science research performed by volunteers for thousands of different projects, each specializing in research of different lineages, surnames, and regions of the world.

#### The Community

This project would not be possible without all the testers, each one providing one more piece to the great puzzle of human history.

## Discover Country Frequency



### Discover: Other SNPs



#### Your Y-DNA Haplogroup Report for E-V13

The Y chromosome is passed from father to son remaining mostly unaltered across generations, except for small traceable changes in DNA. By tracking these changes, we constructed a family tree of humankind where all male lineages trace back to a single common ancestor who lived hundreds of thousands of years ago. This human tree allows us to explore lineages through time and place and to uncover the modern history of your direct paternal surname line and the ancient history of our shared ancestors.

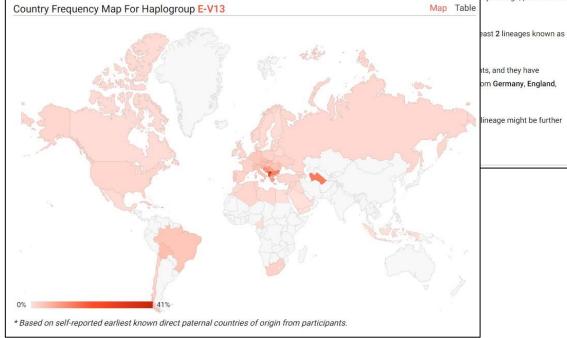
#### The E-V13 Story

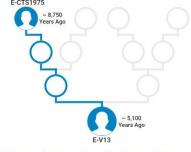
Haplogroup E-V13 represents a man who is estimated to have been born around 5,100 years ago, plus or minus 750 years.

That corresponds to about 3050 BCE with a 95% probability he was born between 3809 and 2401 BCE.

E-V13's paternal line was formed when it branched off from

9 750 years ago, plus or minus





#### E-V13 descendants are from these countries: L England

293 Testers

Other Countries 77 Countries

\* Based on self-reported earliest known direct paternal countries of origin from participants.

# Discover Migration



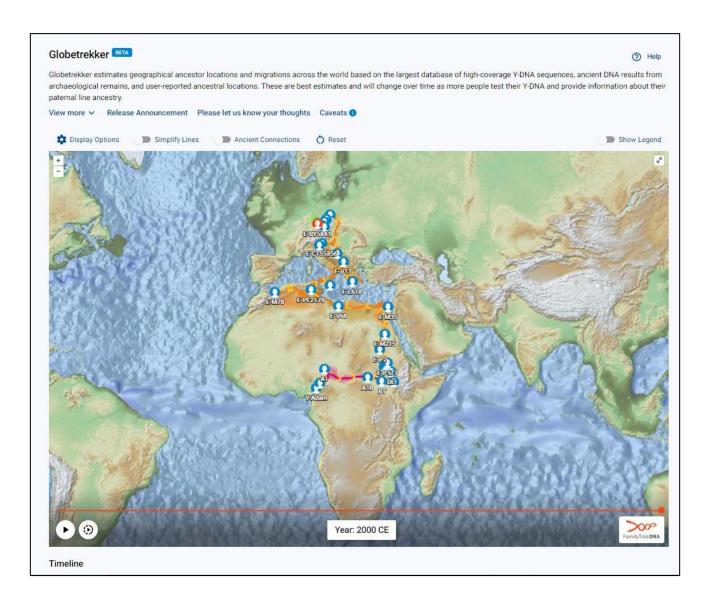
#### The Estimated Migration Route

All human male lineages can be traced back to a single common ancestor in Africa who lived around 230,000 years ago, nicknamed Y-Adam. Here we show the estimated migration route from Y-Adam to your ancestral haplogroup E-V13 (estimated to 5,100 years ago) and his descendants found in ancient DNA from archaeological remains.

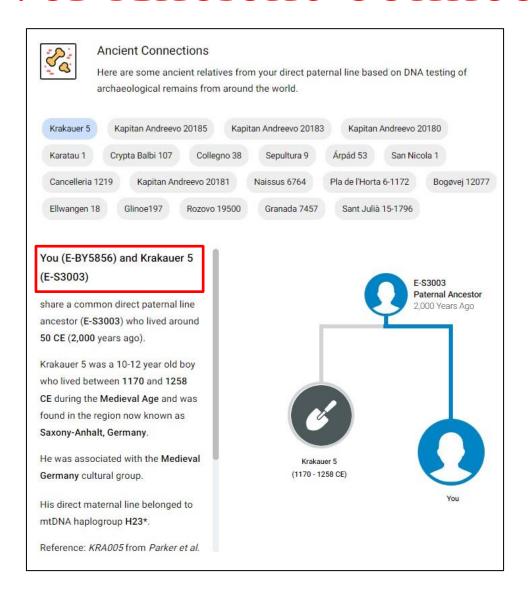
Click the icons on the map to learn more!



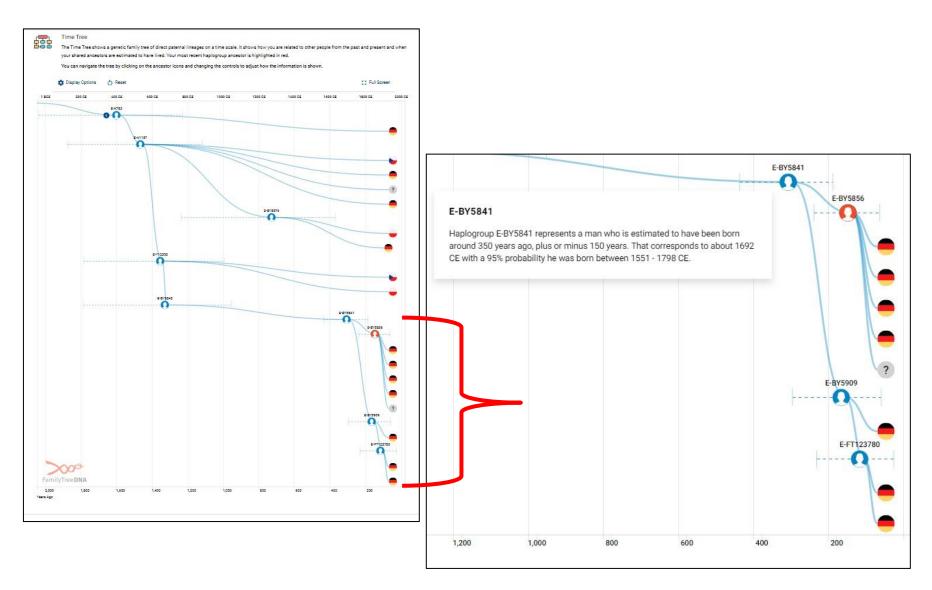
## Globetrekker



### **Discover Ancient Connections**



## Discover Time Tree

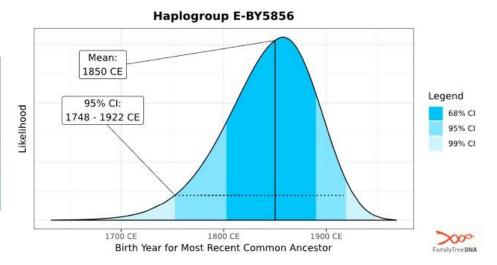


# Discover Age Estimate

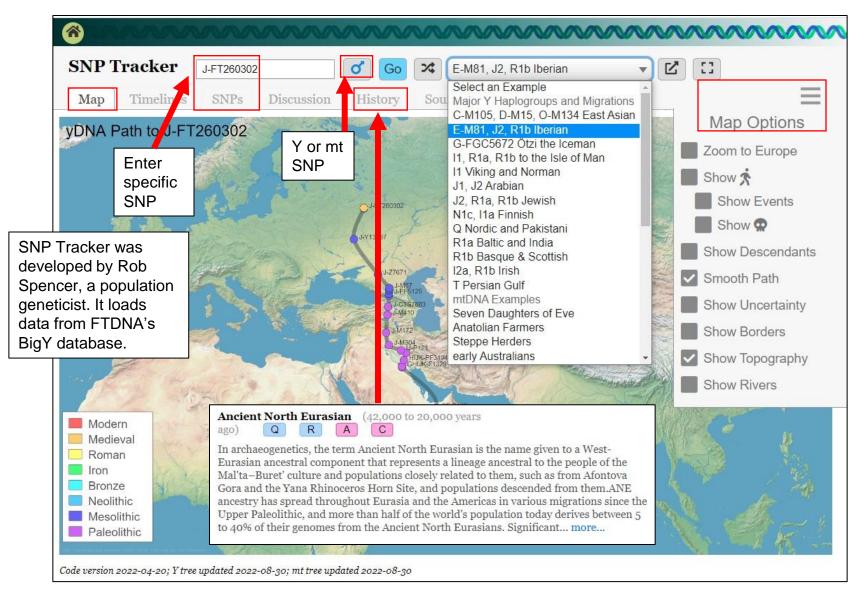
#### Age Estimate Variants

The FamilyTreeDNA Time to Most Recent Common Ancestor (TMRCA) estimate (Beta) is calculated based on SNP and STR test results from many present-day DNA testers. The uncertainty in the molecular clock and other factors is represented in this probability plot, which shows the most likely time when the common ancestor was born amongst the other statistical possibilities.

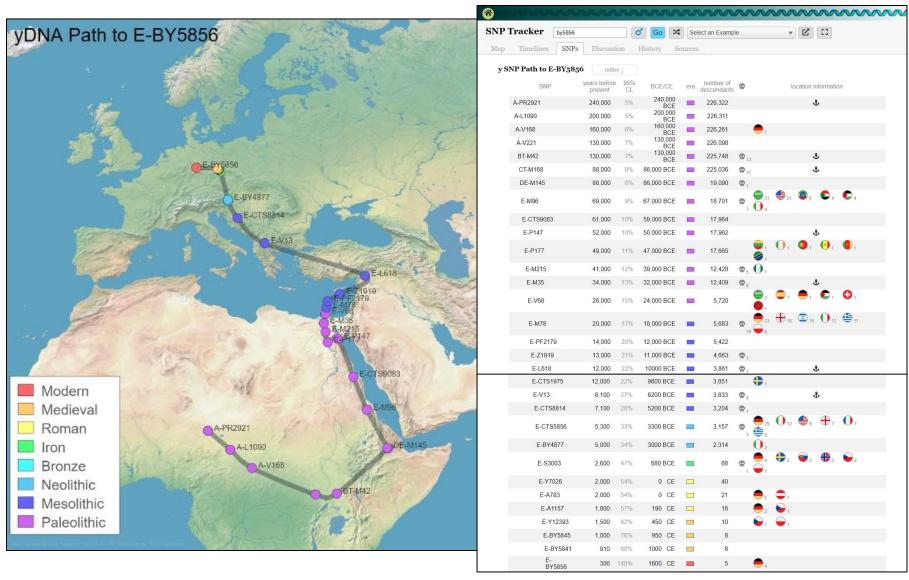
Confidence Interval	Years Before Present	Calendar Date
99%	338 - 72	1684 - 1950 CE
95%	274 - 100	1748 - 1922 CE
68%	219 - 132	1803 - 1890 CE



# Rob Spencer's SNP Tracker



## Rob Spencer's SNP Tracker

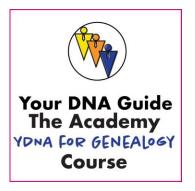


# Y-DNA Continuing Education

YDNA for Genealogy (Diahan Southard)

Your DNA Guide: The Academy

https://www.yourdnaguide.com/ydna-course



for Genetic Genealogy

 The Genealogist's Guide to Y-DNA Testing for Genetic Genealogy

by David Vance