Genetic Genealogy

Using Autosomal DNA to find your cousins

SURVEY

Thinking about an Autosomal DNA test?

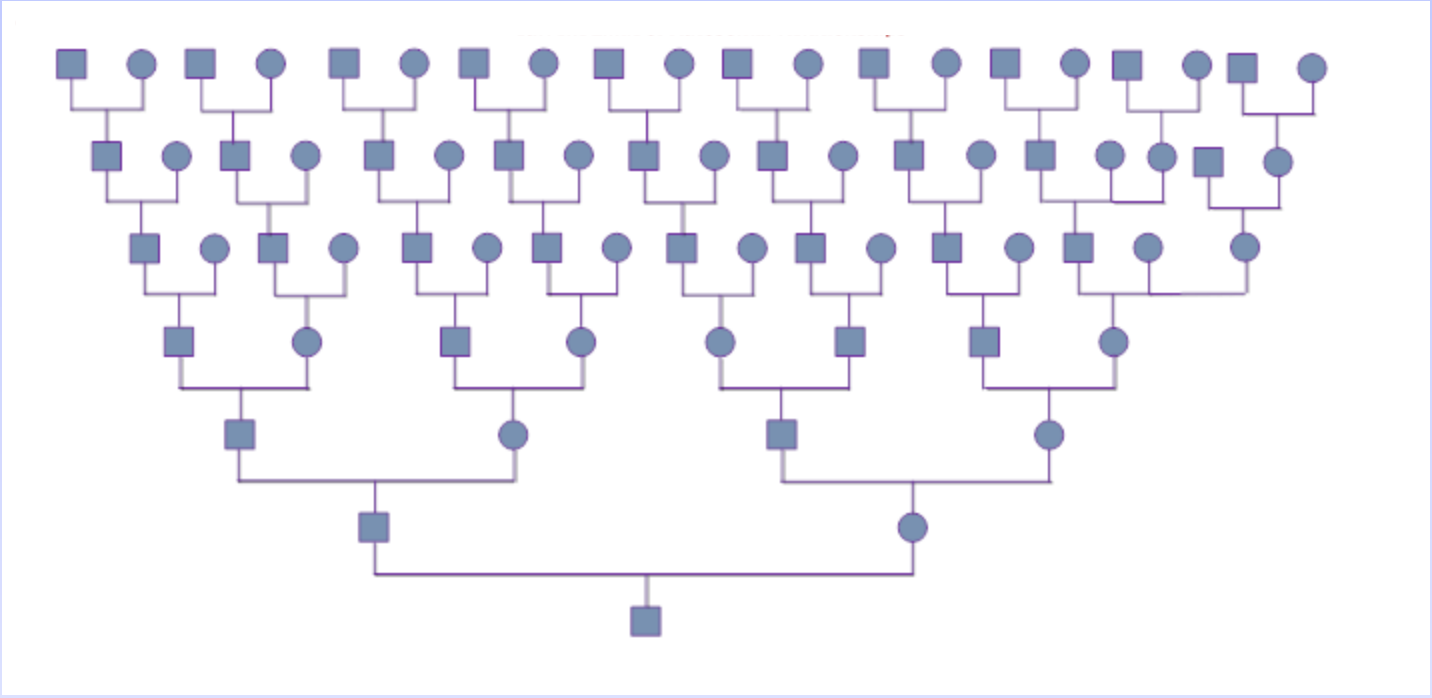
Already taken an Autosomal DNA test?

Taken a test with multiple companies?

Taken a Y-chromosome test? mtDNA?

Company other than 23&Me, FTDNA, AncDNA?

Genealogy – identifying family



Goals

- Discuss the genetics behind autosomal testing
- Identify its main uses for genealogical research
- Compare companies
- Review the format of your results

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- Identify its main uses for genealogical research
- Compare companies
- Review the format of your results

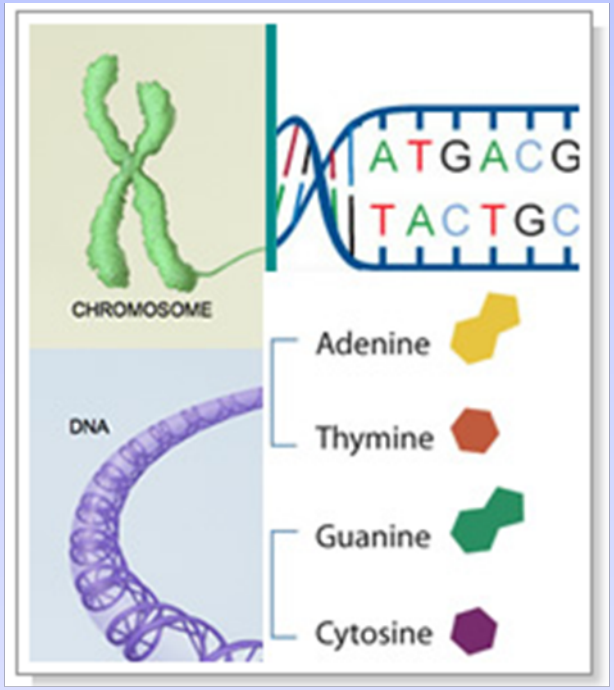
Should you test? Which company?

Each Cell's 23 Chromosome Pairs

Karyogram of Human Male Cell

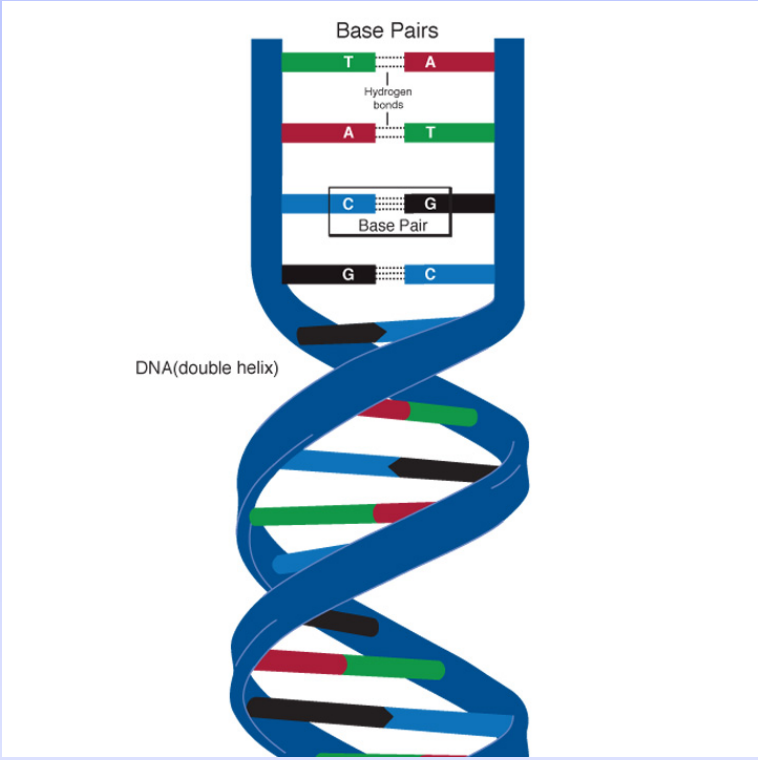
Source: *Wikipedia.org (en.wikipeida.org/wiki/Karyotype)*

DNA Molecule



Source: *National Human Genome Research Institute (NHGRI)*

DNA Molecule



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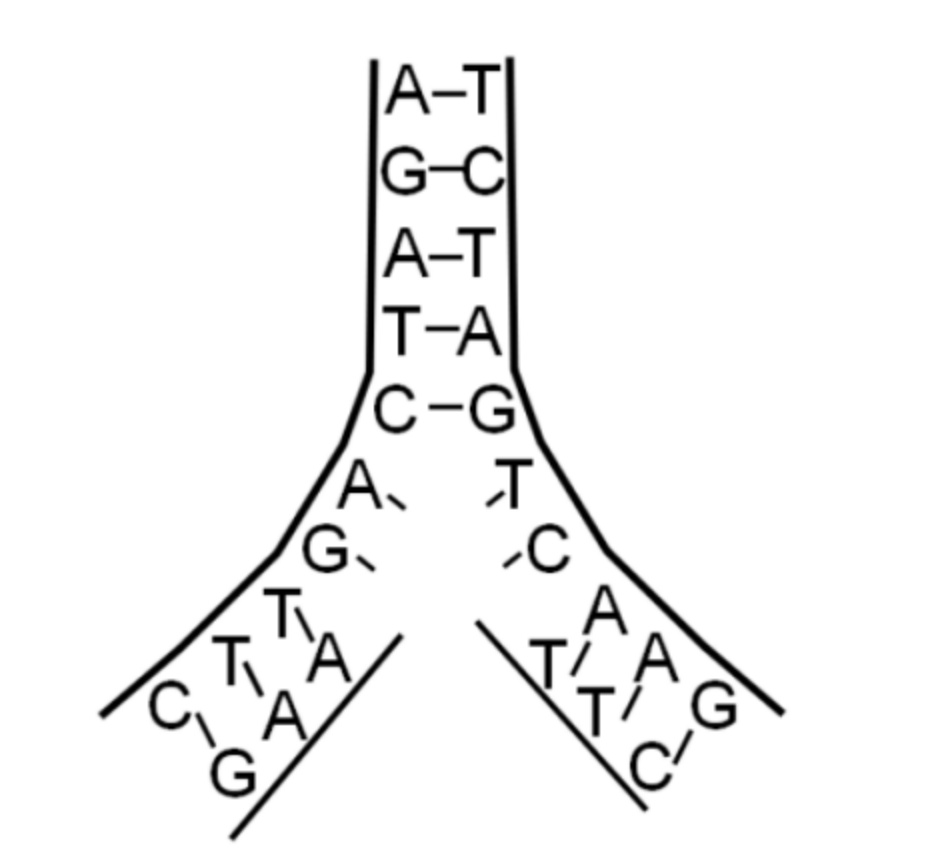
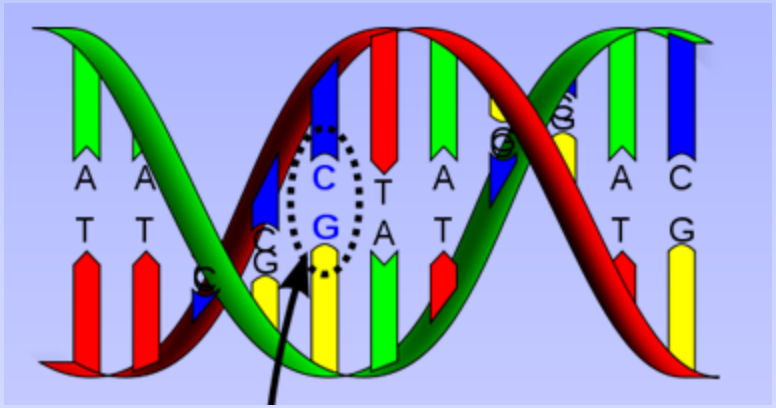


Source: *National Genealogical Society: Genetic Genealogy The Basics*

DNA Mutations



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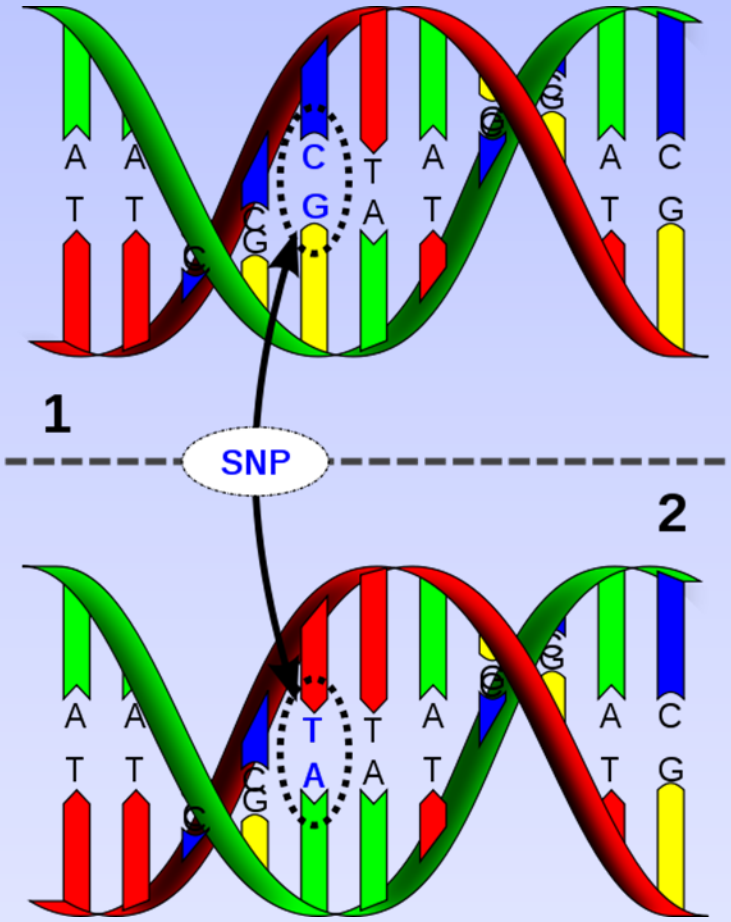
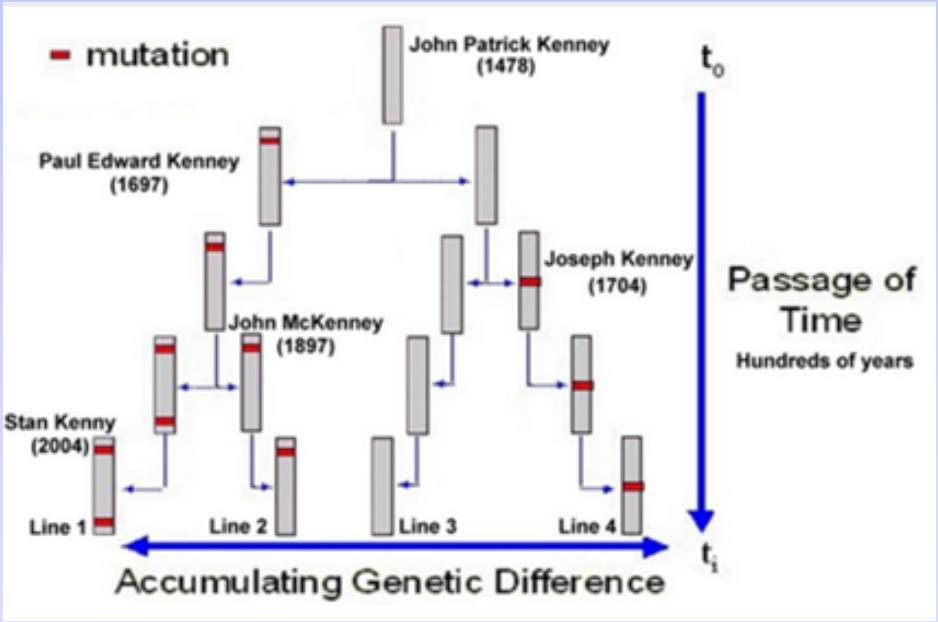


SNP = Single Nucleotide Polymorphism

DNA Mutations



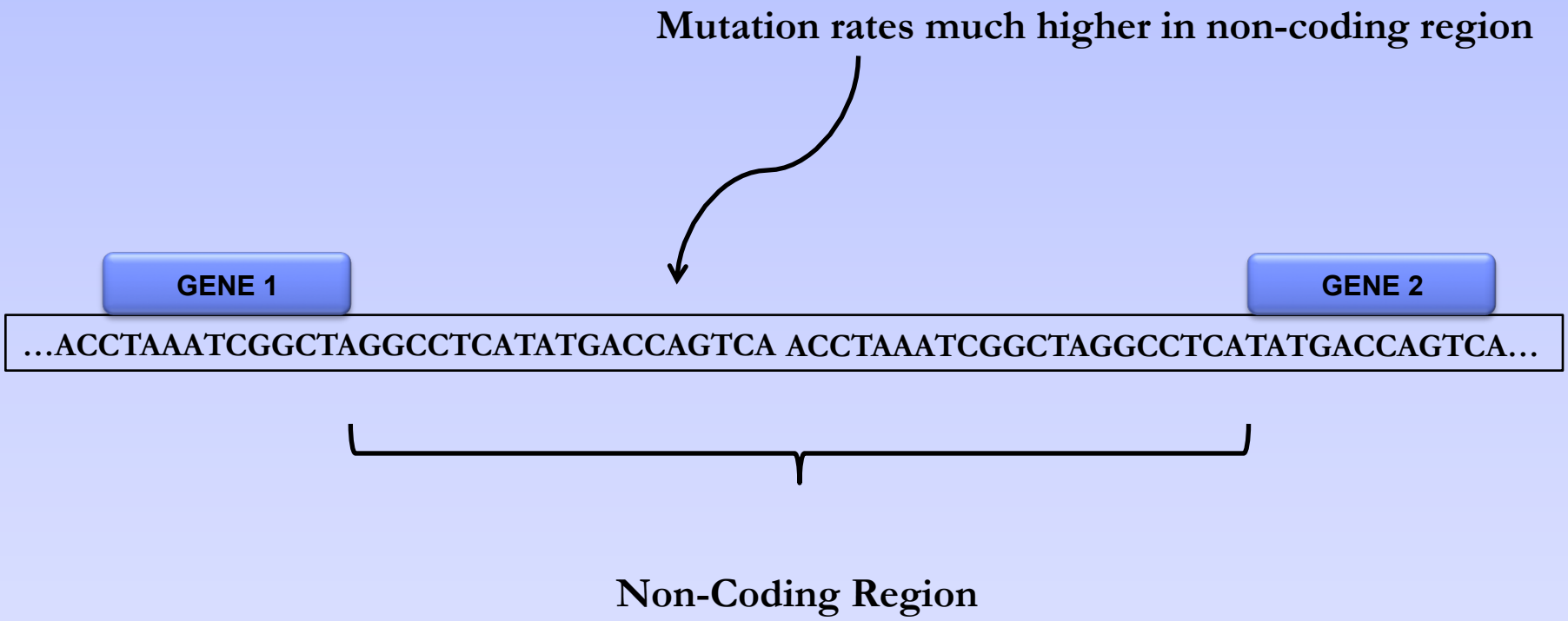
Source: Roberta Estes, explained.com

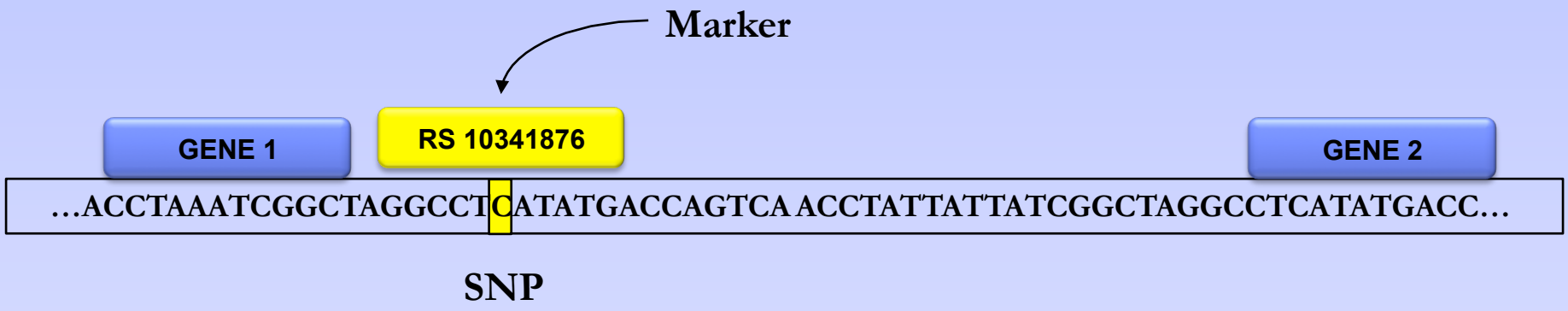
(dna-explained.com/2013/10/06/dna-testing-for-genealogy-101/)

Humans have about 20,000 genes

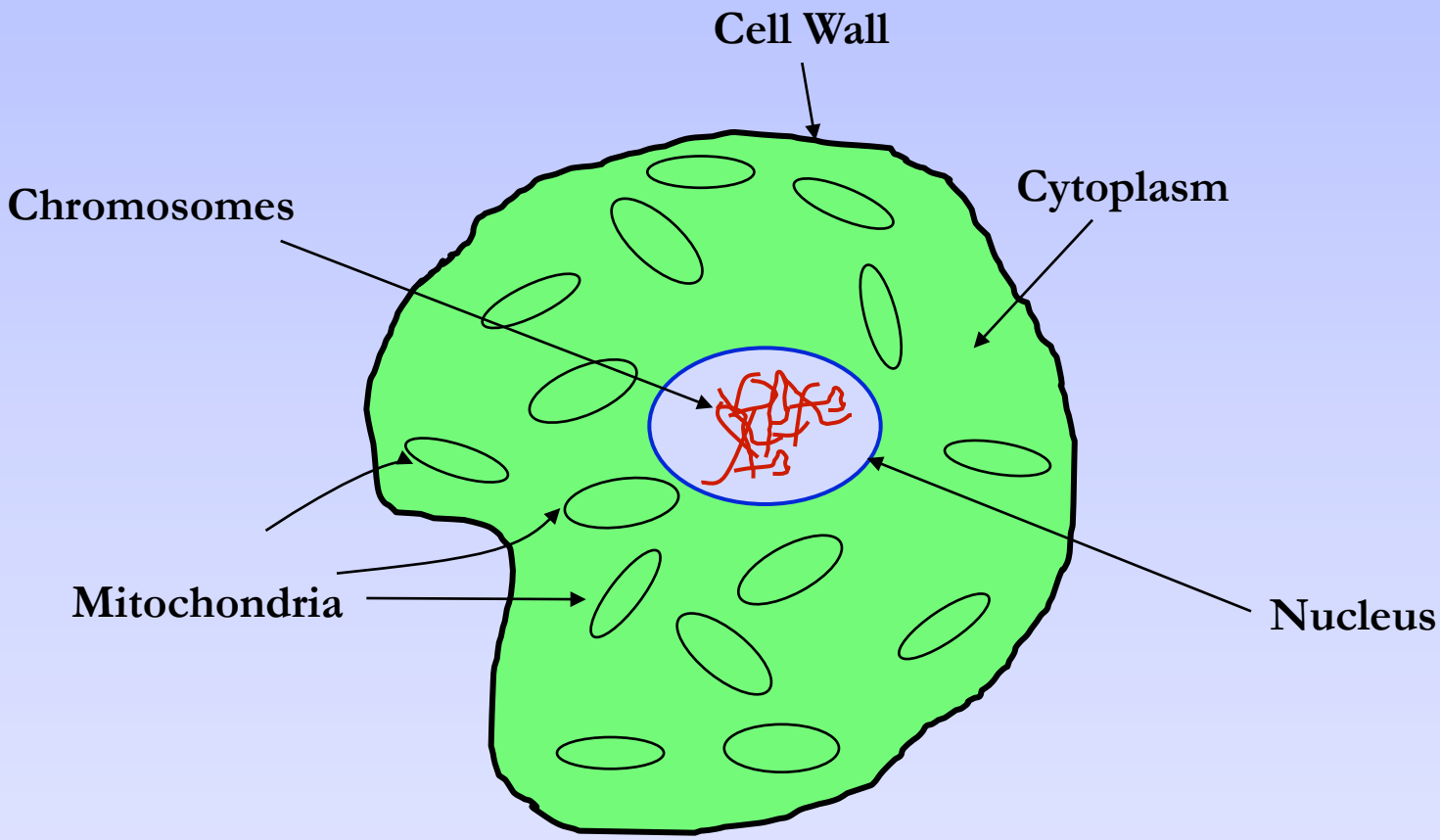
Genes comprise 1.5% of our DNA

Non-Coding Region


Mutation rates much higher in non-coding region
GENE 1
GENE 2
…ACCTAAATCGGCTAGGCCTCATATGACCAGTCA ACCTAAATCGGCTAGGCCTCATATGACCAGTCA…
Non-Coding Region

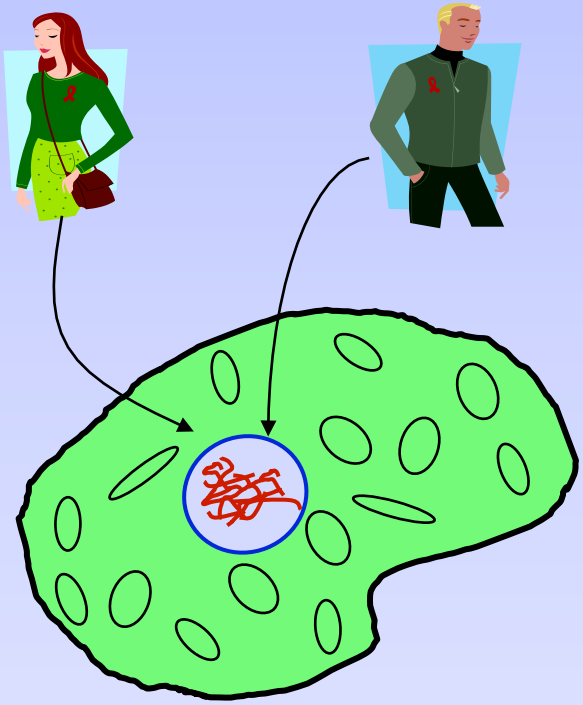


A ‘marker’ is a location where we already know mutations have occurred in the past.



Hominid Cell

Parental Contributions to Autosomal DNA



Variation in the Inheritance of Autosomal DNA

Random Assortment of Chromosomes

1	2	3	4	5
M	M	M	M	M
D	D	D	D	D

1	2	3	4	5
M	M	M	M	M
D	D	D	D	D

Mom = Adams

Dad = Jones

Mom = Meyer

Dad = Provost

(Simplified example with 5 chromosome pairs instead of full 22 pairs)

Random Assortment of Chromosomes

1	2	3	4	5
M	M	M	M	M
D	D	D	D	D

1	2	3	4	5
M	M	M	M	M
D	D	D	D	D

Mom = Adams

Dad = Jones

Mom = Meyer

Dad = Provost

CHR 1	CHR 2	CHR 3	CHR 4	CHR 5
Meyer	Adams	Meyer	Jones	Provost
Jones	Meyer	Adams	Provost	Jones

$(2^5 = 32 \text{ possibilities})^2$

$(2^{22} = 4M+ \text{ possibilities})^2$

Autosomal DNA and Recombination

Thomas Hunt Morgan's illustration of crossing over (1916)

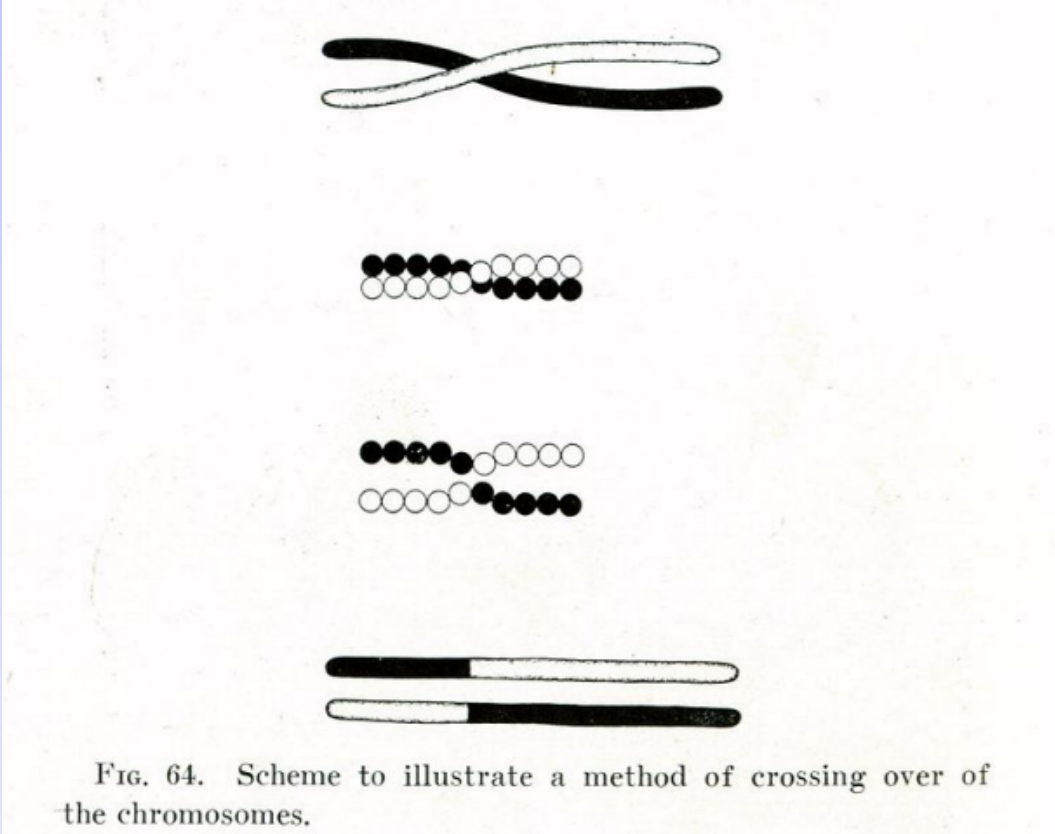


FIG. 64. Scheme to illustrate a method of crossing over of the chromosomes.

http://en.wikipedia.org/wiki/File:Morgan_crossover_1.jpg

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Thomas Hunt Morgan's illustration of crossing over (1916)

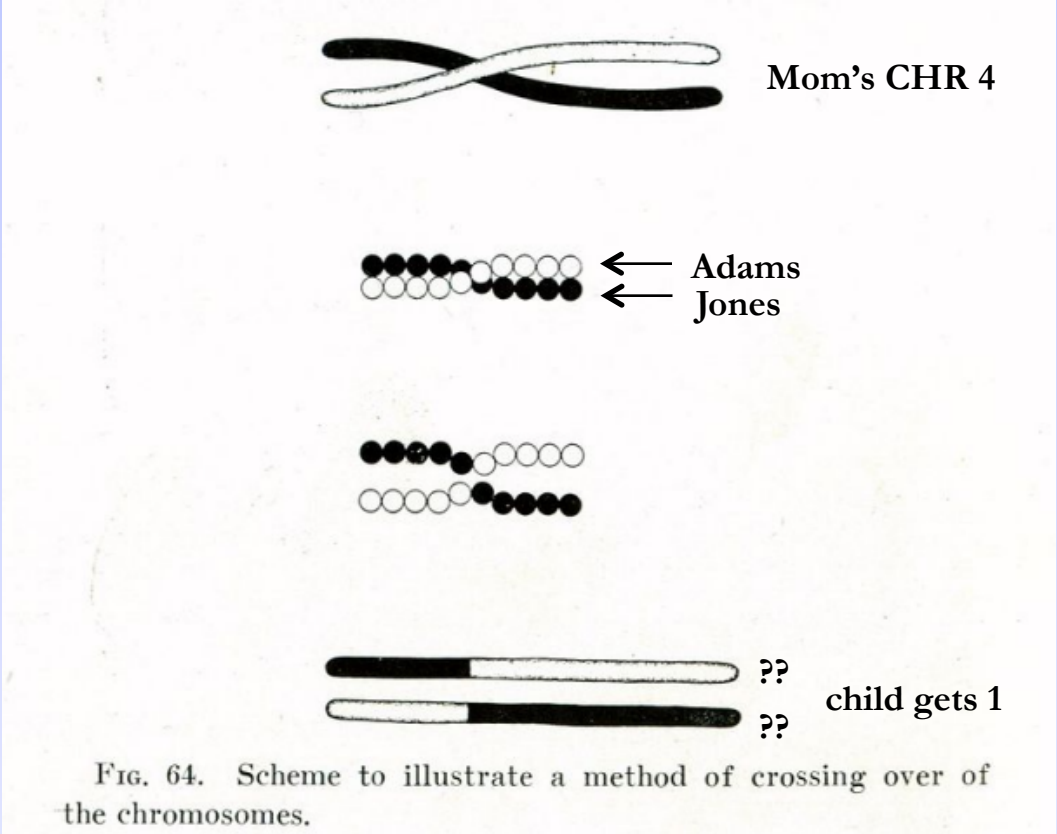


FIG. 64. Scheme to illustrate a method of crossing over of the chromosomes.

http://en.wikipedia.org/wiki/File:Morgan_crossover_1.jpg

What Do We Inherit?

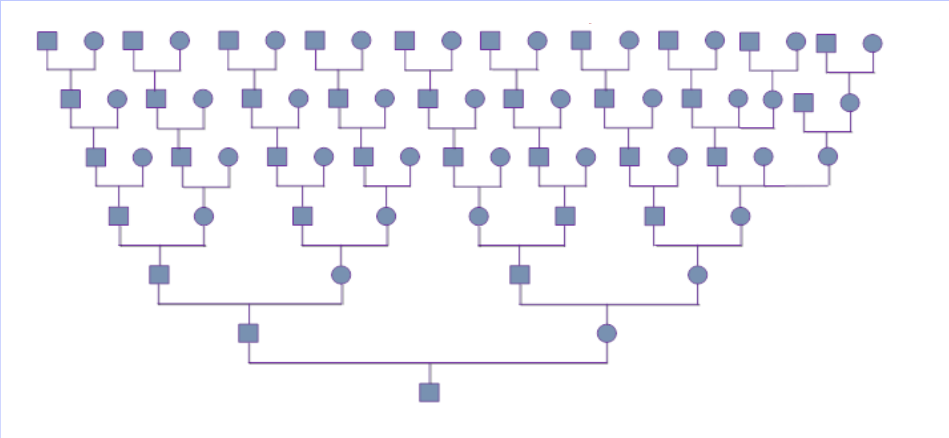
- Parents have total of 44 chromsome pairs (12 billion bases)
- Grandparents have total of 88 pairs (24 billion bases)
- Children only have room for 22 pairs (6 billion bases)

We must have less and less DNA from older ancestors

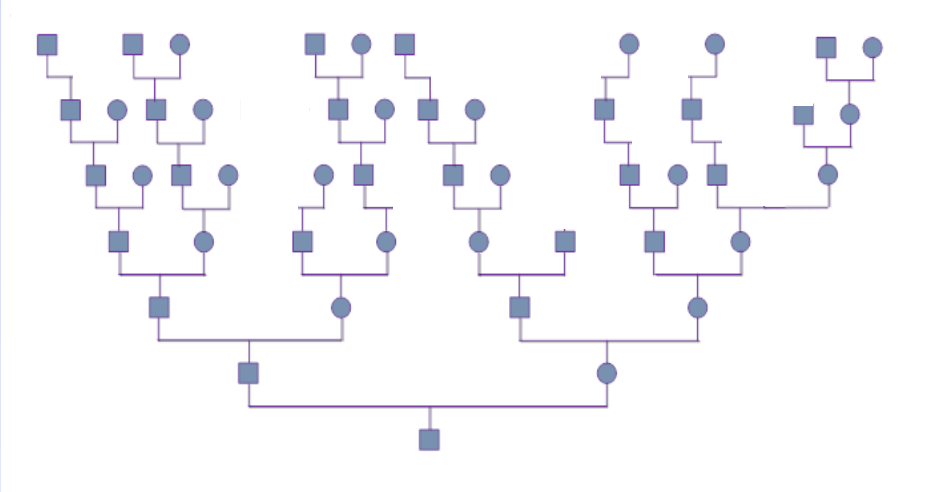
NET: Our oldest ancestors start to disappear from our genetic tree !!

What Do We Inherit?

Genealogical Tree

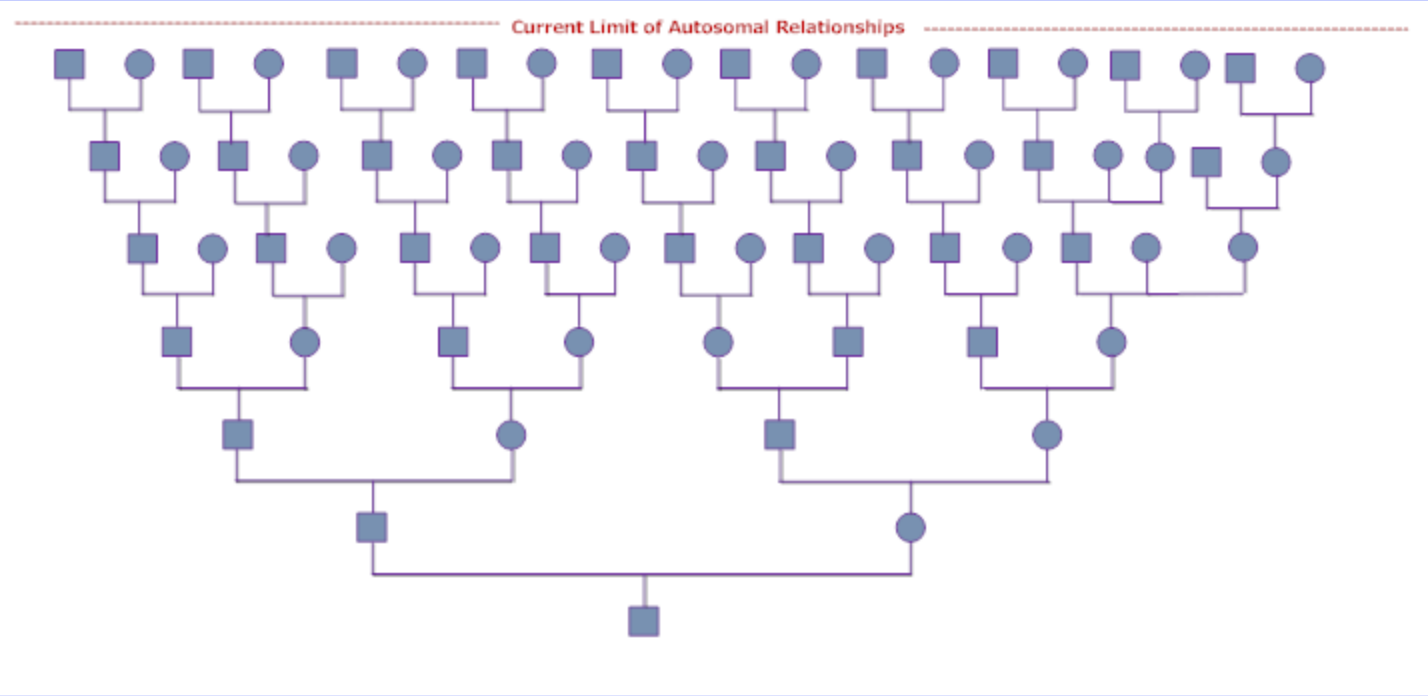


Genetic Tree



Autosomal DNA Testing Finds More Relationships

Limitation Is 5 to 6 Generations



Genetic Genealogy Testing Companies

23andMe

ancestryDNA BETA

FamilyTreeDNA
HISTORY UNEARTHED DAILY

Various DNA Sample Kits

DNA Tests & Test Kits

FamilyTreeDNA

DNA Tests & Test Kits

AncestryDNA & 23andMe

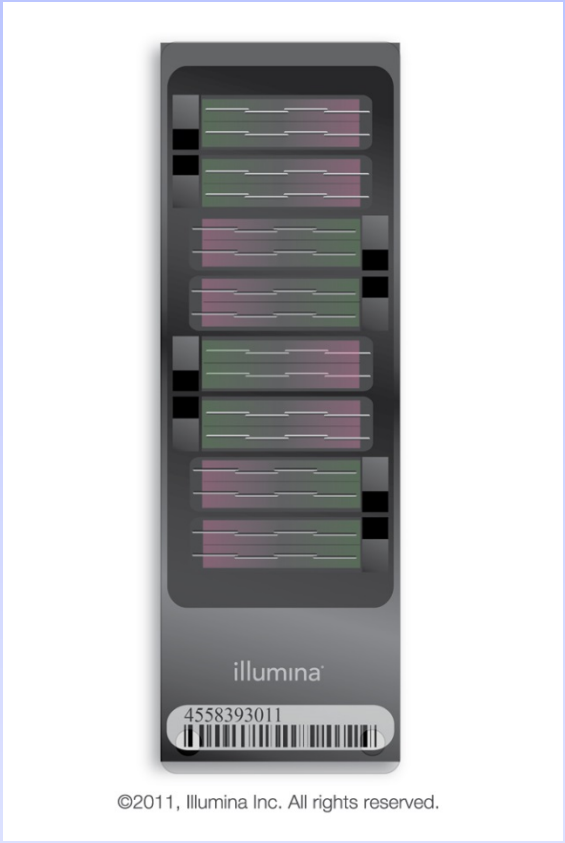
Samples Mailed to Companies' Testing Labs

23andMe

Autosomal DNA Testing Process

1. Receive & enter sample into inventory
2. Separate & purify DNA
3. Amplify DNA many times
4. Add your DNA sample & reagents to the Illumina beadchip

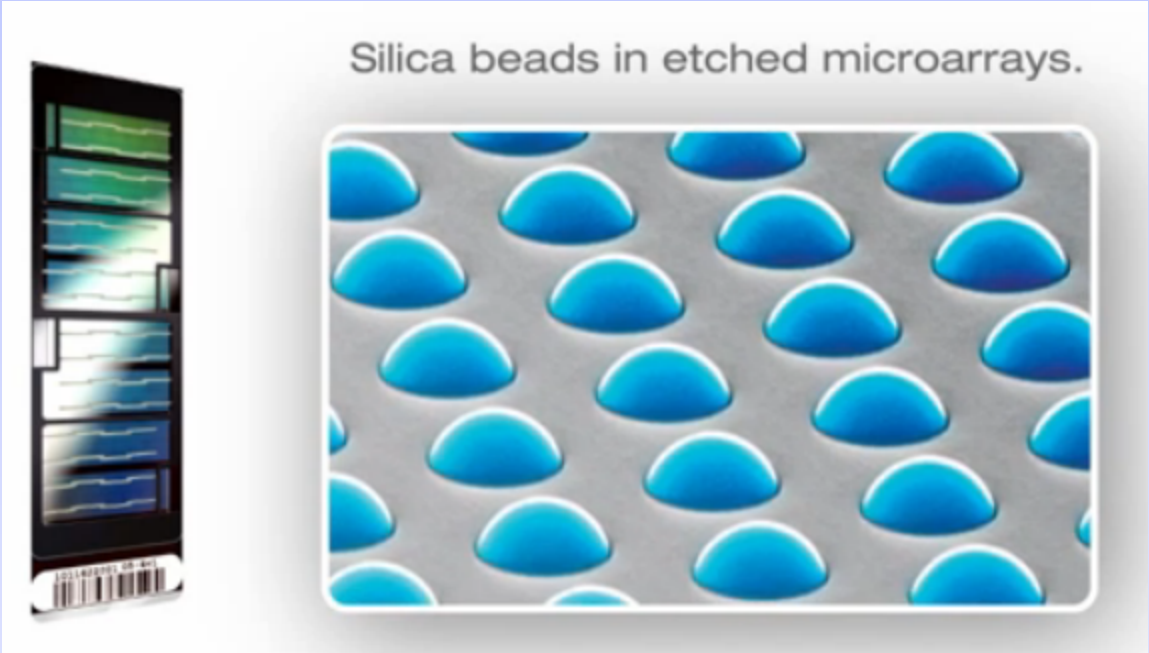
Autosomal DNA Testing Process



Illumina OminiExpress Bead Chip

Courtesy of Illumina, Inc.

Autosomal DNA Testing Process



Courtesy of Illumina, Inc.

Autosomal DNA Testing Process

1. Receive & enter sample into inventory
2. Separate & purify DNA
3. Amplify DNA many times
4. Add sample, markers & reagents to Illumina beadchip

5. DNA fragments attach to specific beads
6. Fluorescent tags identify individual bases
7. Beadchip "read" by high-resolution scanner

Autosomal DNA Testing Process

Illumina HiSeq 2500 Array Reader

Courtesy of Illumina, Inc.

Autosomal DNA Test Results

Testing results in a list of 700,000 SNPs*

Analysis of your SNPs compared to → **company's database**

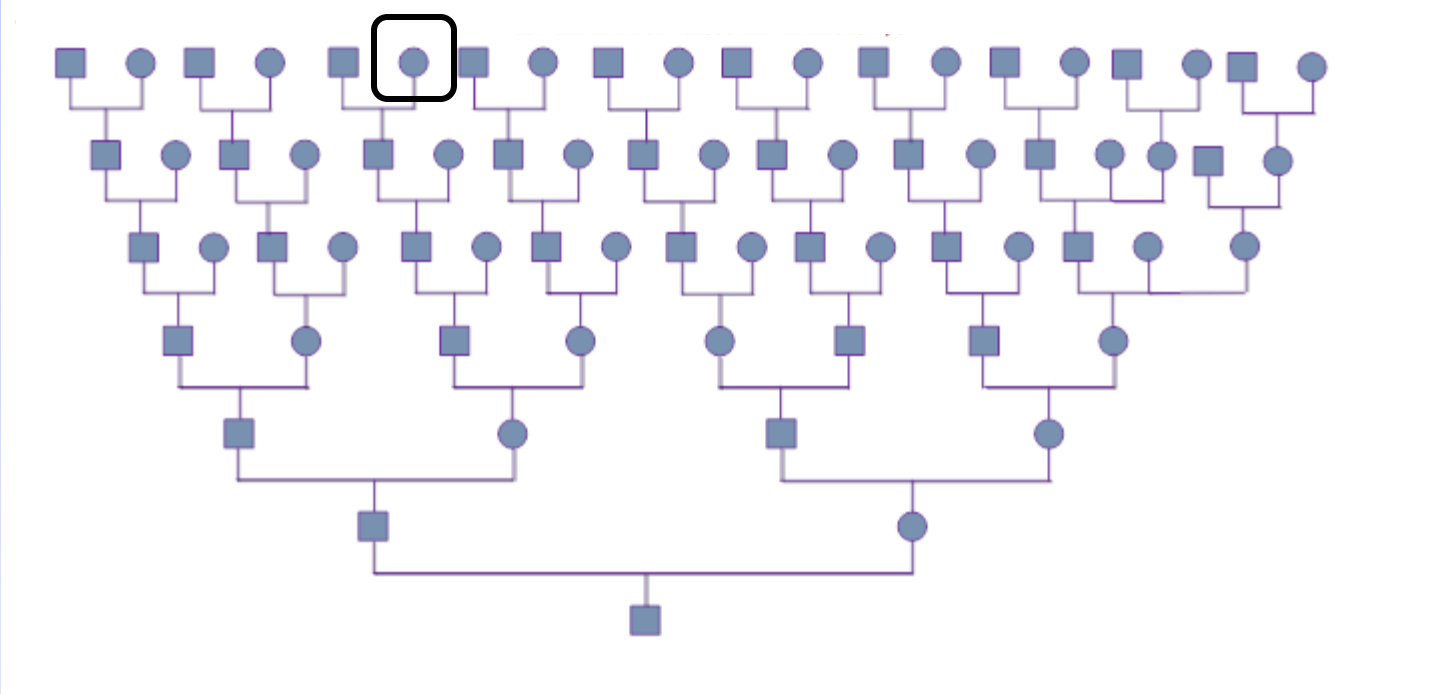
Result: list of your genetic matches

(* 23andMe v4 chip looks at ~600K SNPs)

How Do Our DNA Tests & Genealogies Work Together ?

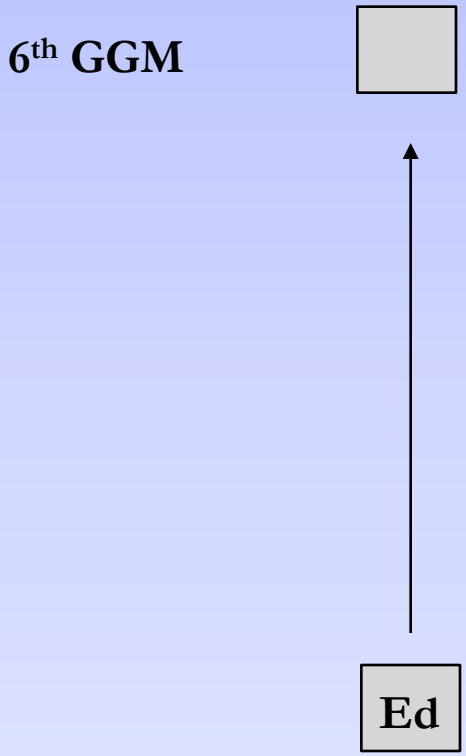
How Your DNA Tests & Genealogies Work Together

Hundreds of Ancestors with Thousands of Living Descendants



Traditional Genealogy

How Your DNA Tests & Genealogies Work Together



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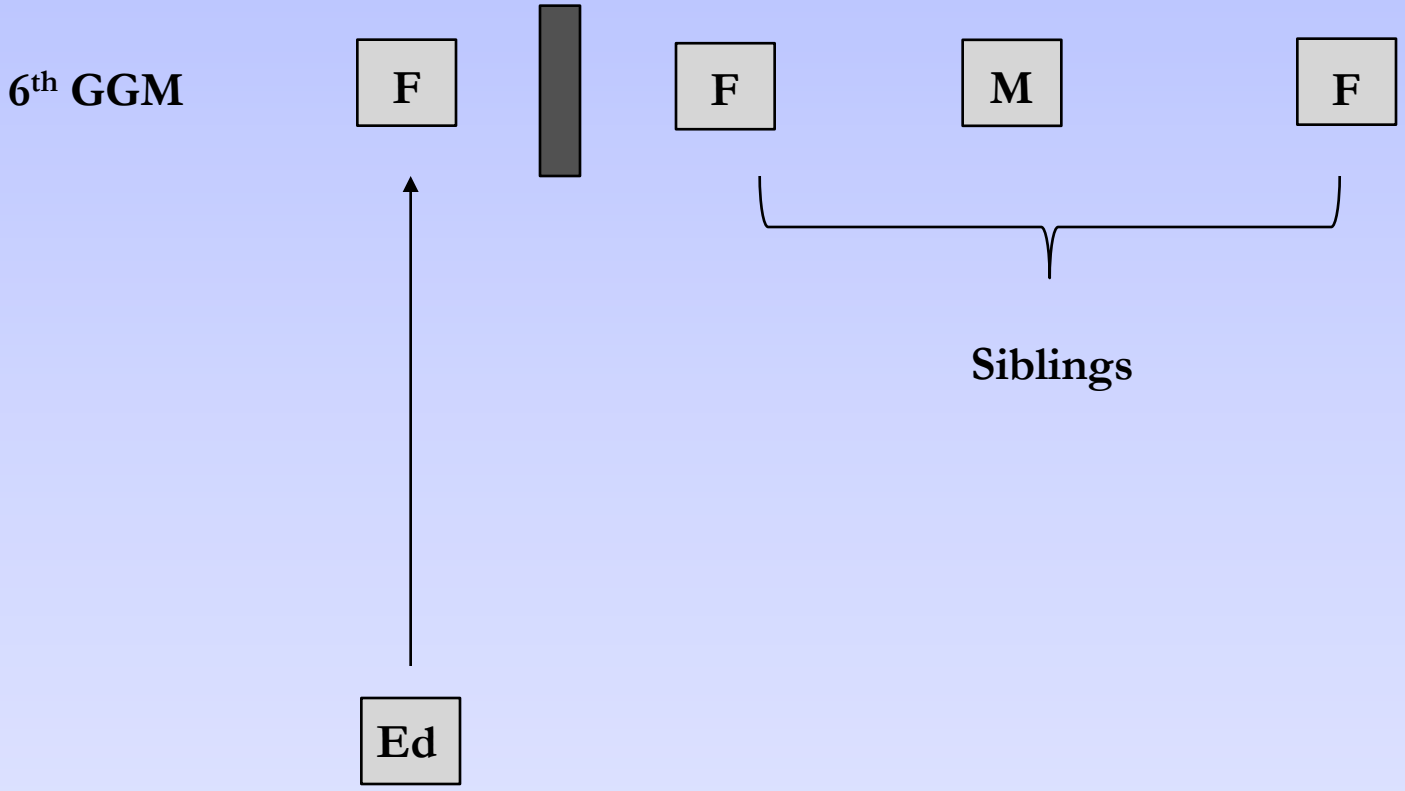

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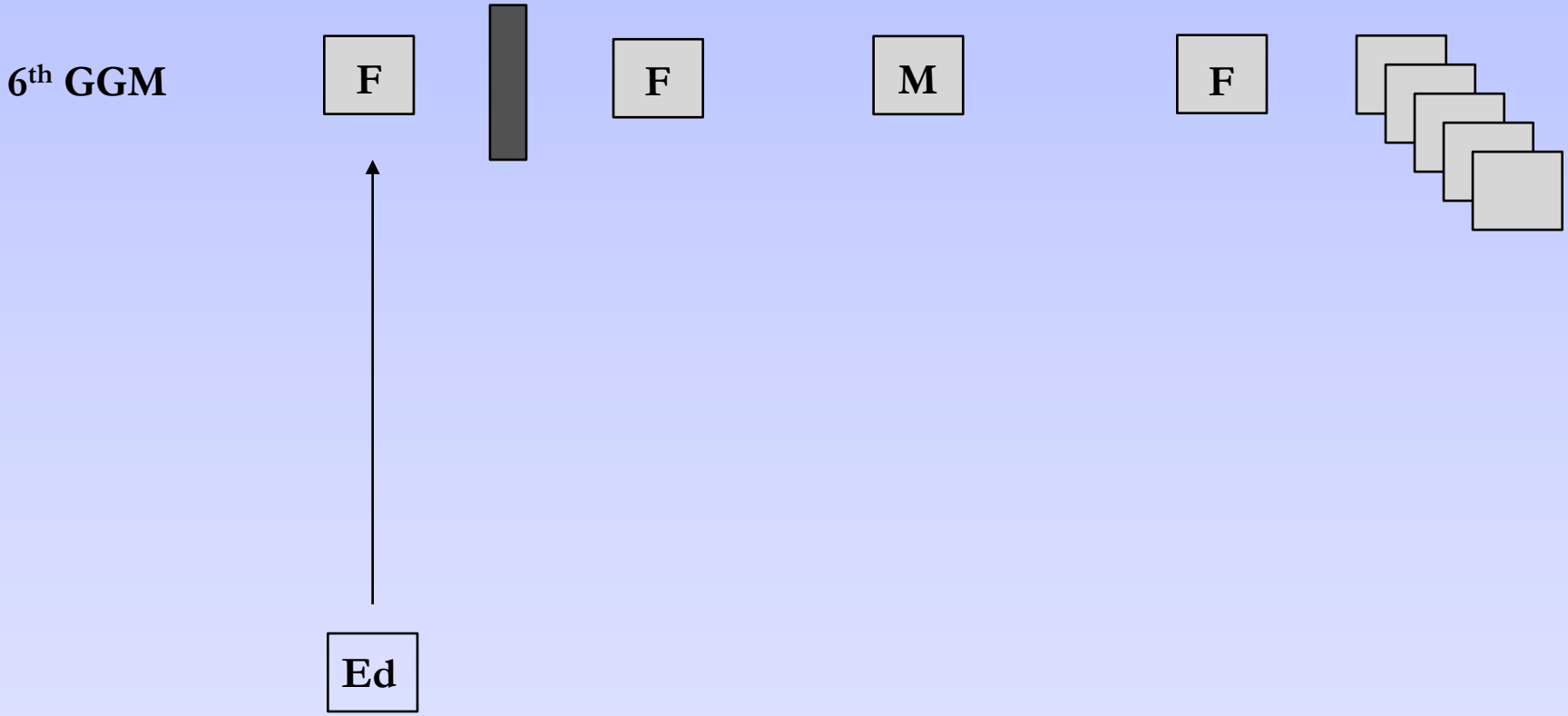

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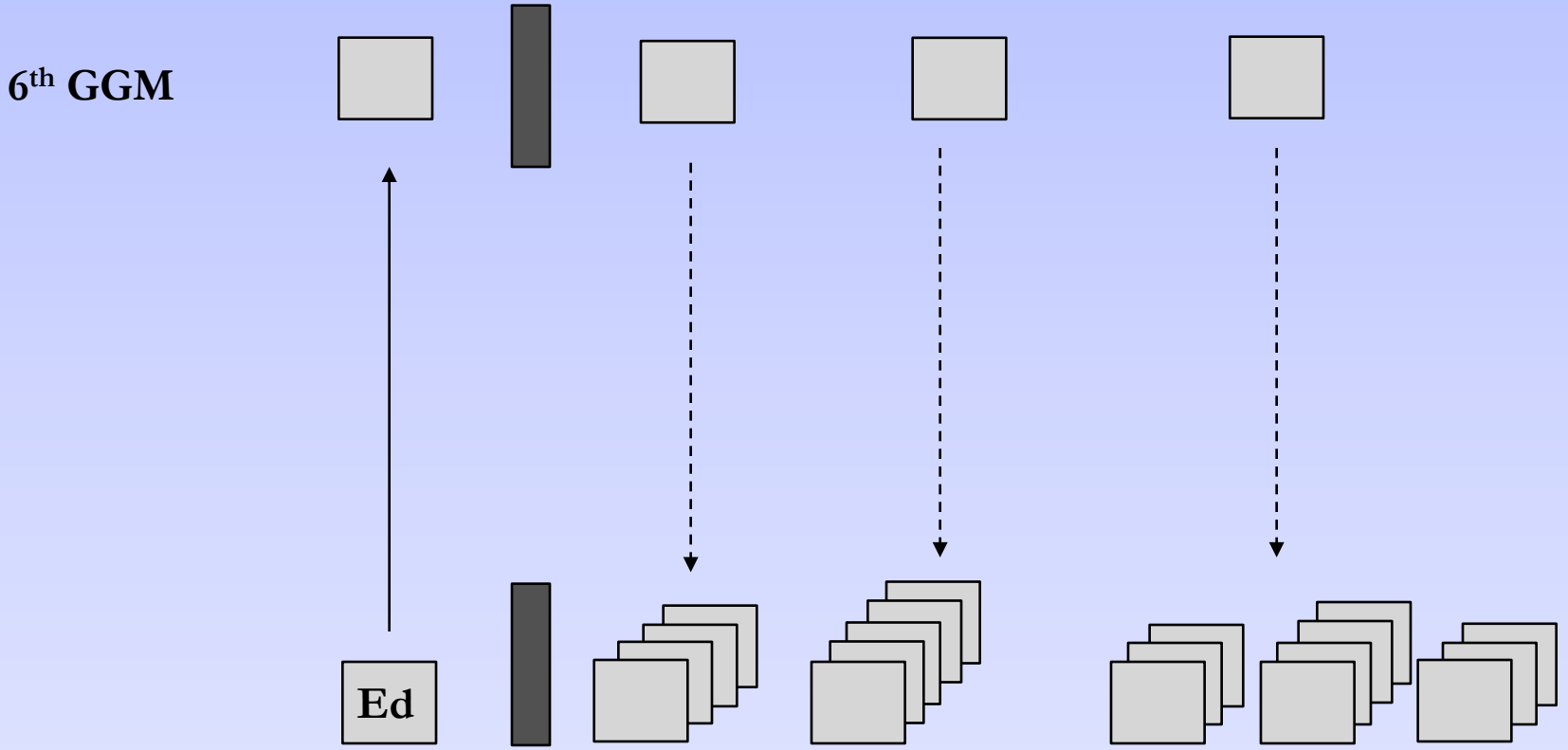
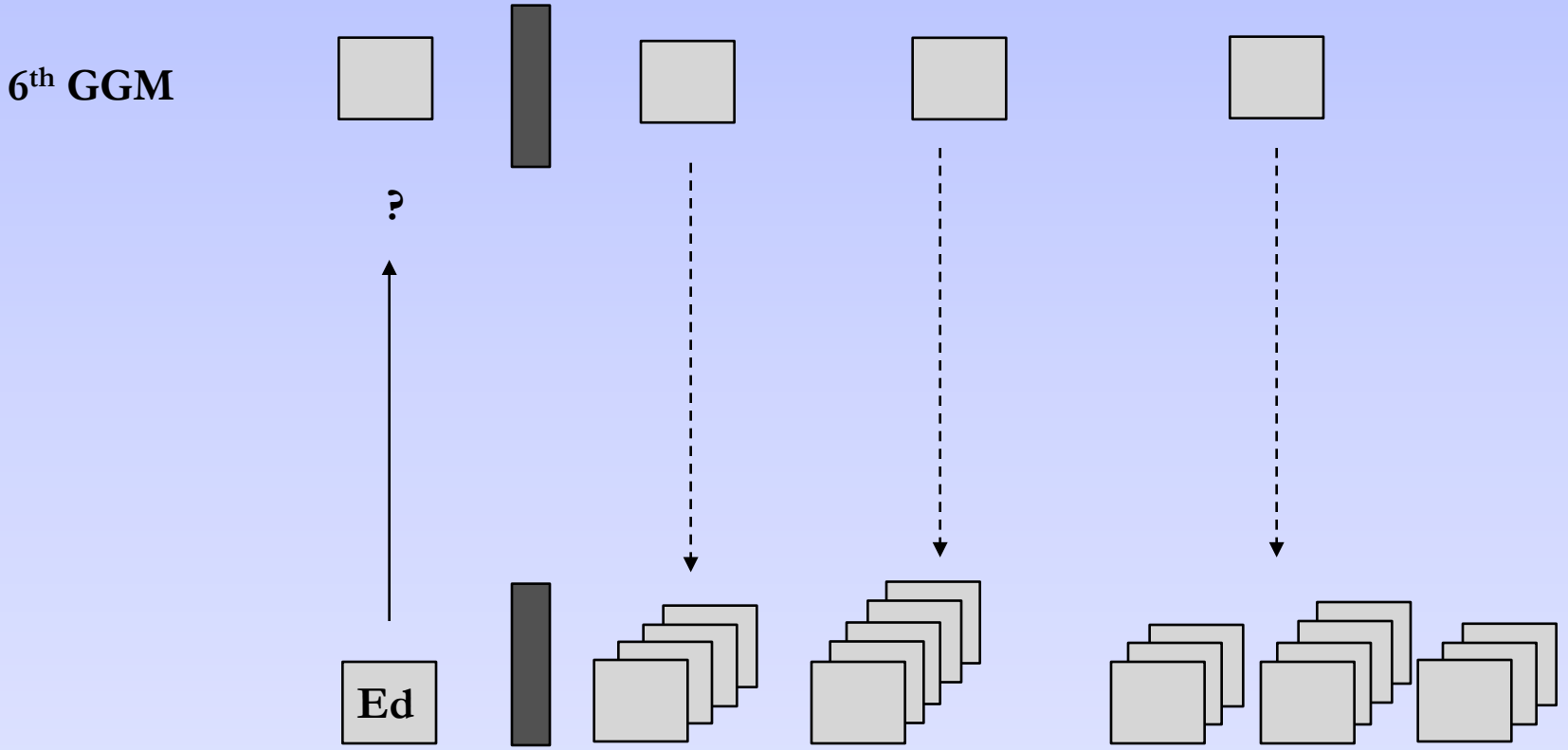


Genealogy – it's all about finding family



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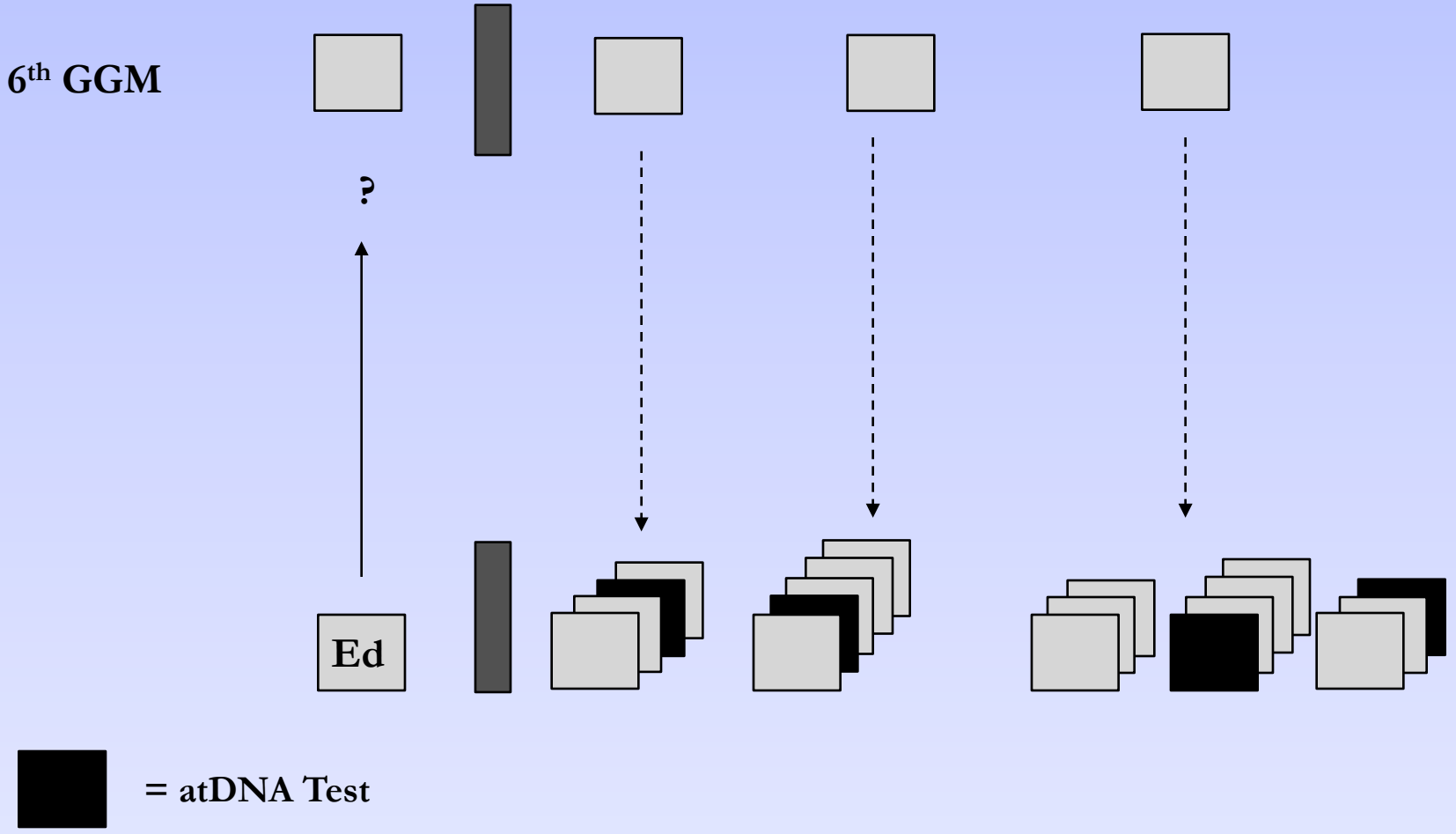

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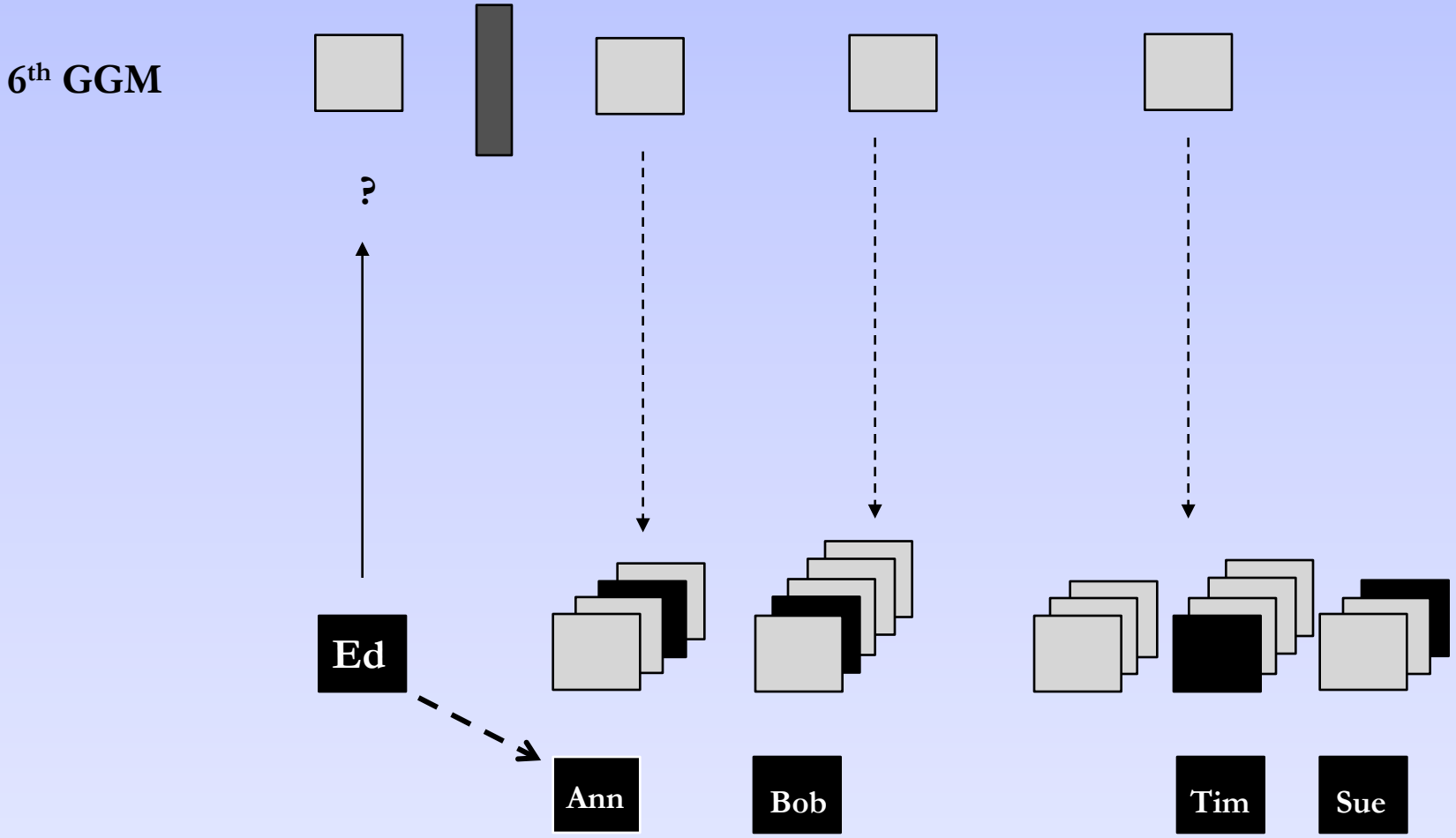

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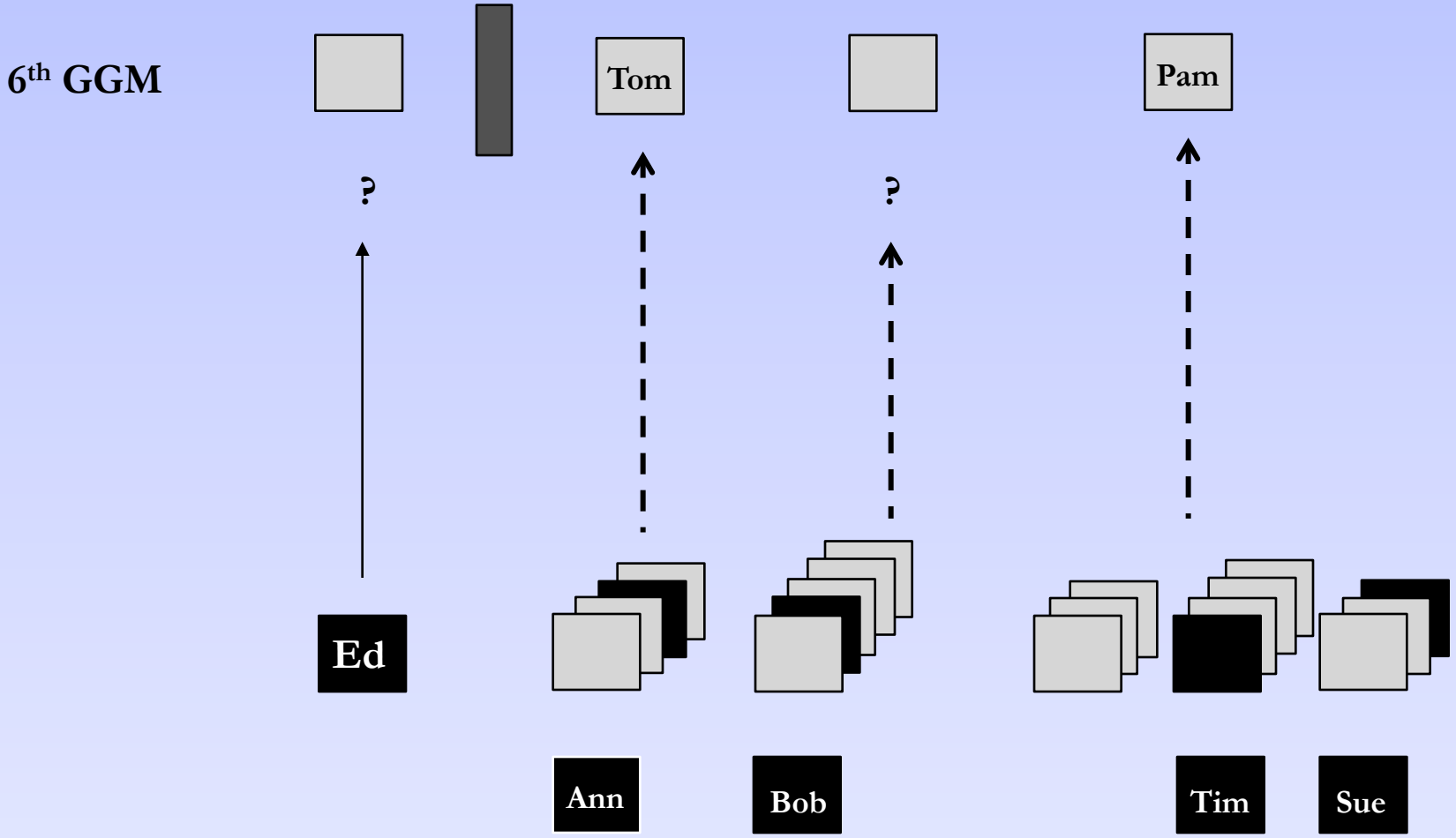

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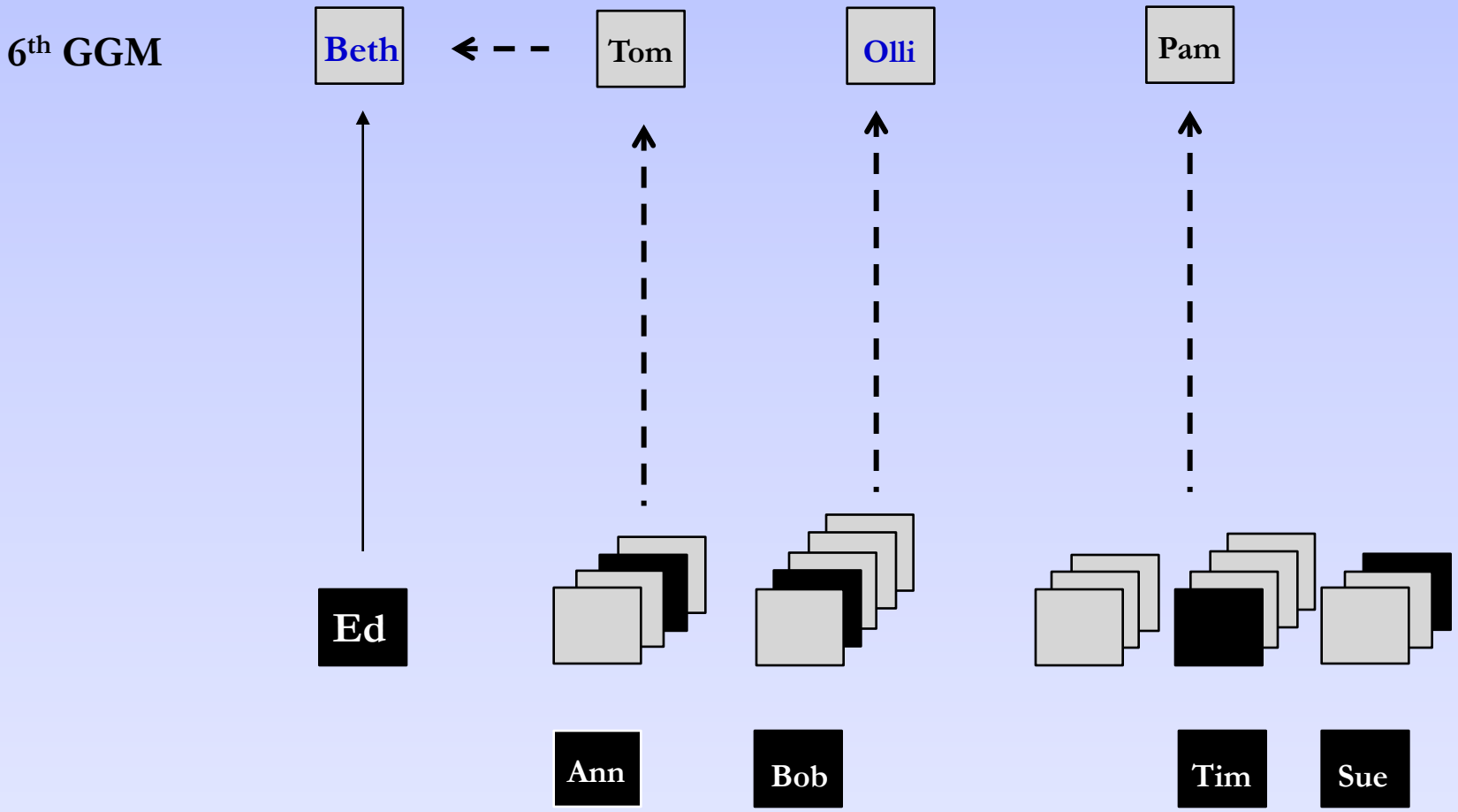


The Research Process

- Ideally – you find several 'cousins' for a given piece of shared DNA
- One (or more) need to get back to that (6th GGM) family unit
- You all share your family trees, surnames & locations
- Your data needs to be within 1 (or 2) generations of that 'family unit'
- Deep genealogies (and/or luck) are the key to success

Autosomal DNA Test Results

Three key test outputs:

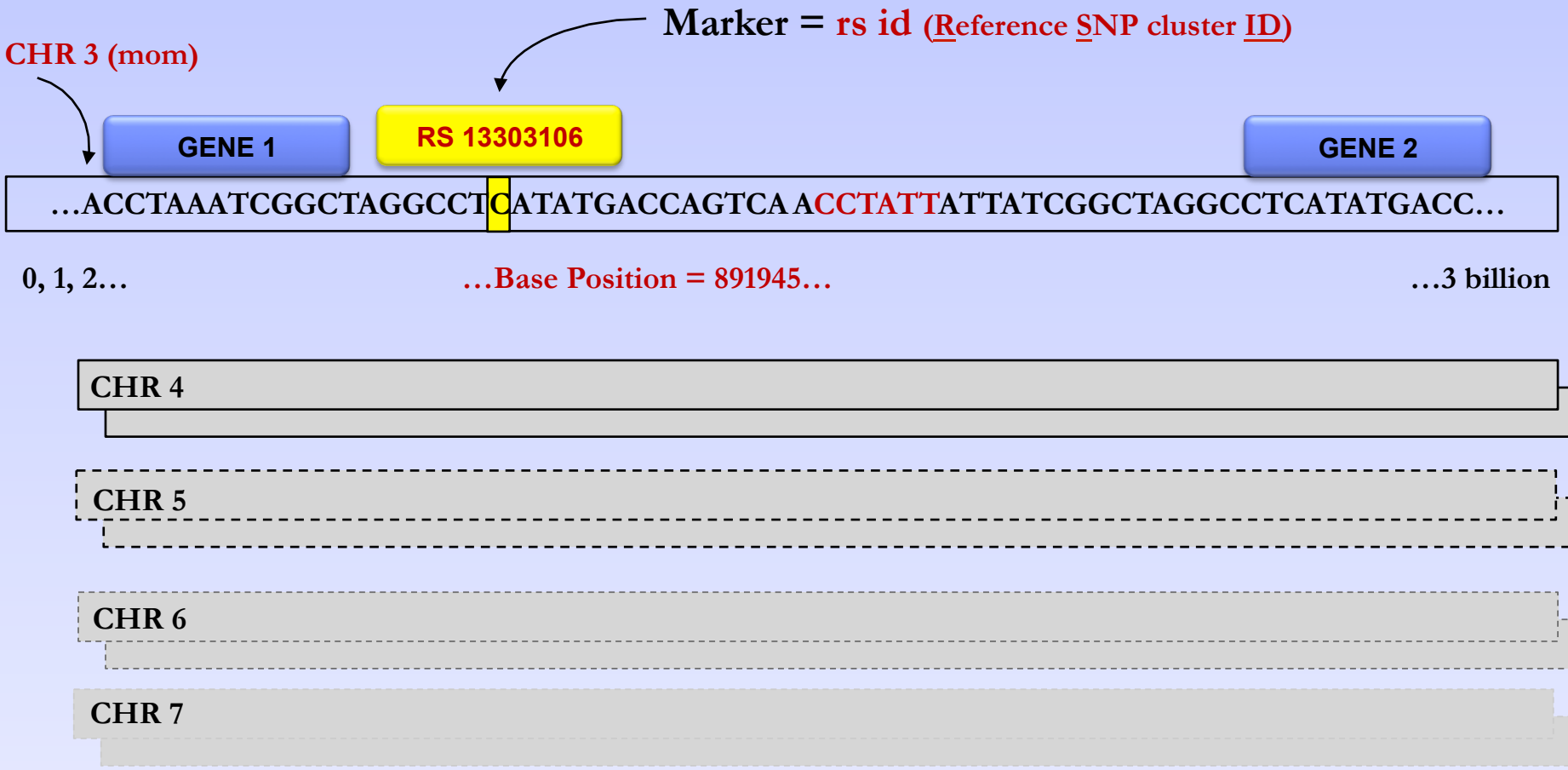
1. Separate comparison used for '**Admixture**'
2. Your SNPs compared to other test samples → your **matches**
3. Specialized tools provided by test companies → Chromosome browsers, etc

Autosomal DNA Raw Data

Ancestry DNA Download

RSID	CHRS	POSITION	RESULT
rs13303106	1	891945	AG
rs28415373	1	893981	CC
rs13303010	1	894573	AA
rs6696281	1	903104	CC
rs28391282	1	904165	GG
rs2340592	1	910935	GG
rs13303118	1	918384	TT
rs2341354	1	918573	GG
rs6665000	1	924898	AA
rs2341362	1	927309	CC
rs9777703	1	928836	TT
rs1891910	1	932457	GG
rs9697457	1	934345	GG

SNP Number — Base Position — Maternal & Paternal Values


Marker = rs id (Reference SNP cluster ID)
CHR 3 (mom)
GENE 1
RS 13303106
GENE 2
…ACCTAAATCGGCTAGGCCTCATATGACCAGTCAACCTATTATTATCGGCTAGGCCTCATATGACC…
0, 1, 2…
…Base Position = 891945…
…3 billion
CHR 4
CHR 5
CHR 6
CHR 7

Autosomal DNA Test Results

Ancestry DNA Download

RSID	CHRS	POSITION	RESULT
rs13303106	1	891945	AG
rs28415373	1	893981	CC
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Ancestry DNA – 700K

FT-DNA – 700K

23andMe – ~~1000~~K 577K

Autosomal DNA Test Results

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FT-DNA – 700K

23andMe – 577K

Total Genome = 3+ billion bases

Autosomal DNA Test Results

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Total Genome = 3 billion bases

Only 10 Million SNPs (<0.1%)

Autosomal DNA Test Results

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Non-medical SNPs

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Autosomal DNA Test Results

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rs2340592	1	910935	GG
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rs6665000	1	924898	AA
rs2341362	1	927309	CC
rs9777703	1	928836	TT
rs1891910	1	932457	GG
rs9697457	1	934345	GG

Family Tree DNA

RSID	CHRS	POSITION	RESULT
rs13303106	1	881808	AG
rs28415373	1	883844	CC
rs13303010	1	884436	AA
rs6696281	1	892967	CC
rs28391282	1	894028	GG
rs2340592	1	900798	GG
rs13303118	1	908247	TT
rs2341354	1	908436	GG
rs6665000	1	914761	AA
rs2341362	1	917172	CC
rs9777703	1	918699	TT
rs1891910	1	922320	GG
rs9697457	1	924208	GG

End of Technical Background

Any questions on this?

Autosomal DNA Applications

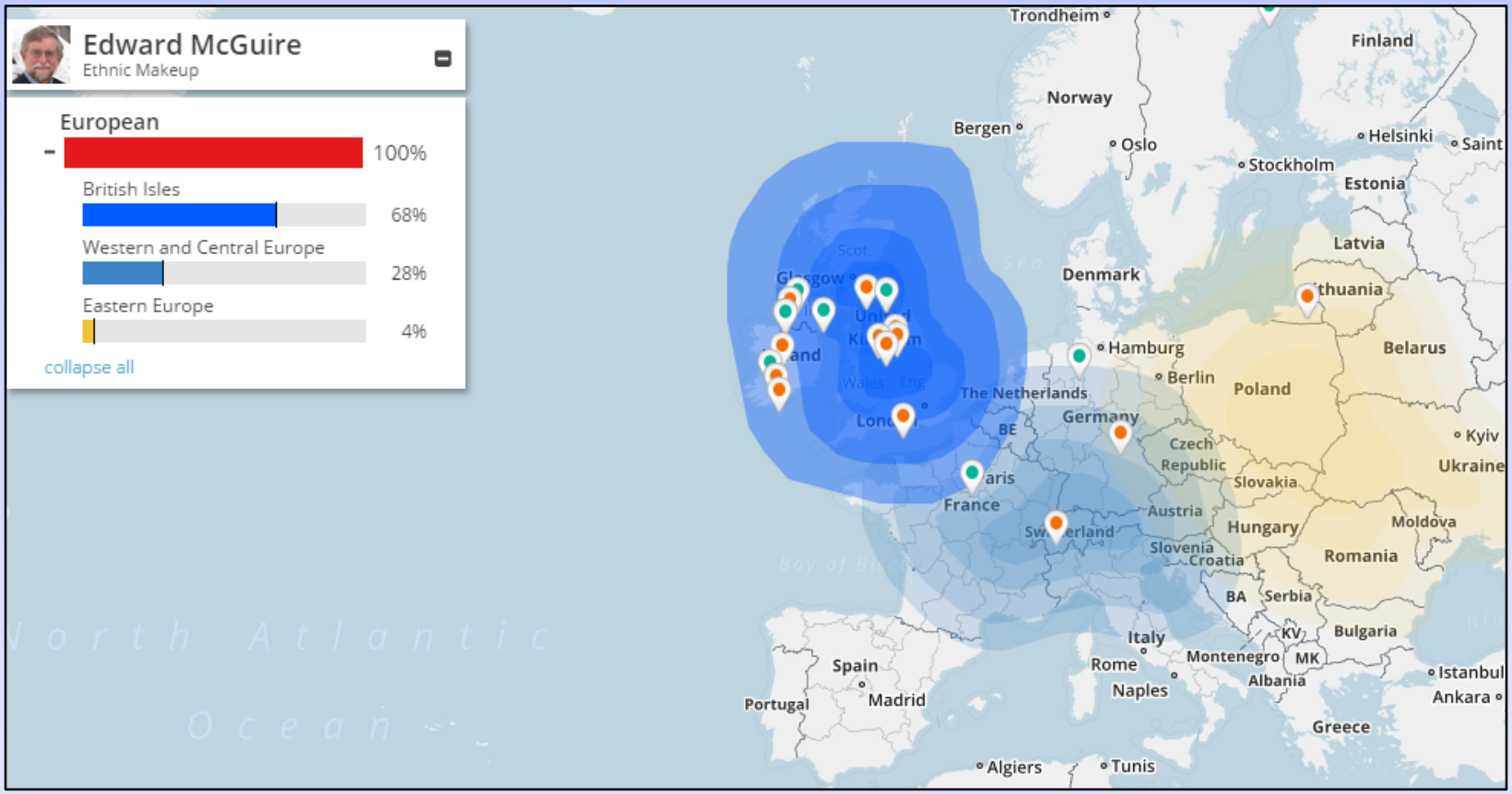
Application	Objective
Ethnicity	Estimates of ethnic percentage in your DNA
DNA Matches	Identify matches & do the work to find the relationship
Test Hypothesis	2d cousins Sally & Beth are really step-sisters not sisters
Crossing The Pond	Locate 'recent' O'REILLY cousins in County Mayo
Unknown Parentage	Adoptees, slave ancestors, unknown paternity

Autosomal DNA Tools

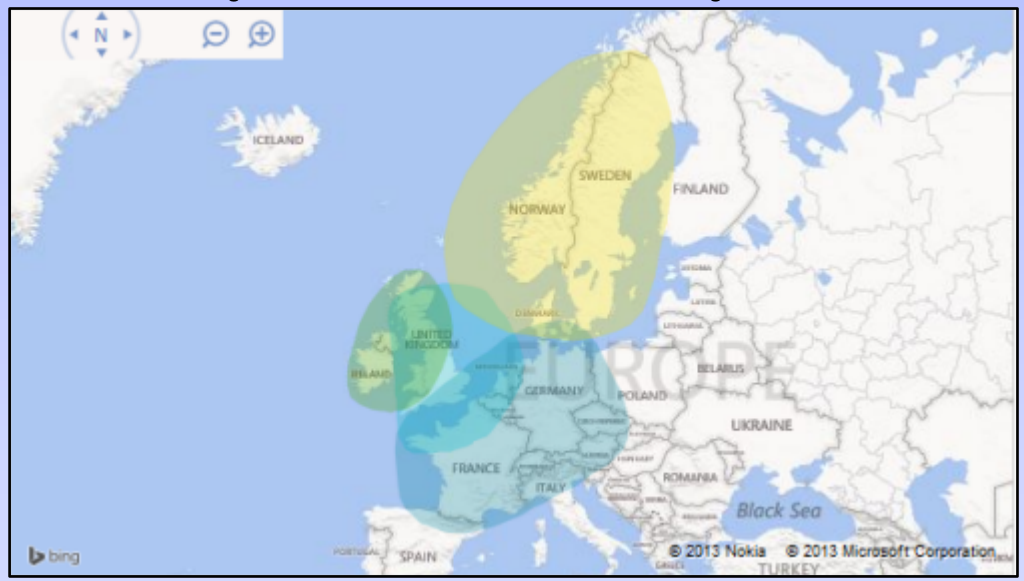
- Admixture (or 'Ethnic Percentages')
- Matches
- DNA Segment Tools
  - Chromosome browsers
  - 'In Common With' Analysis

Admixture

FT-DNA Population Finder

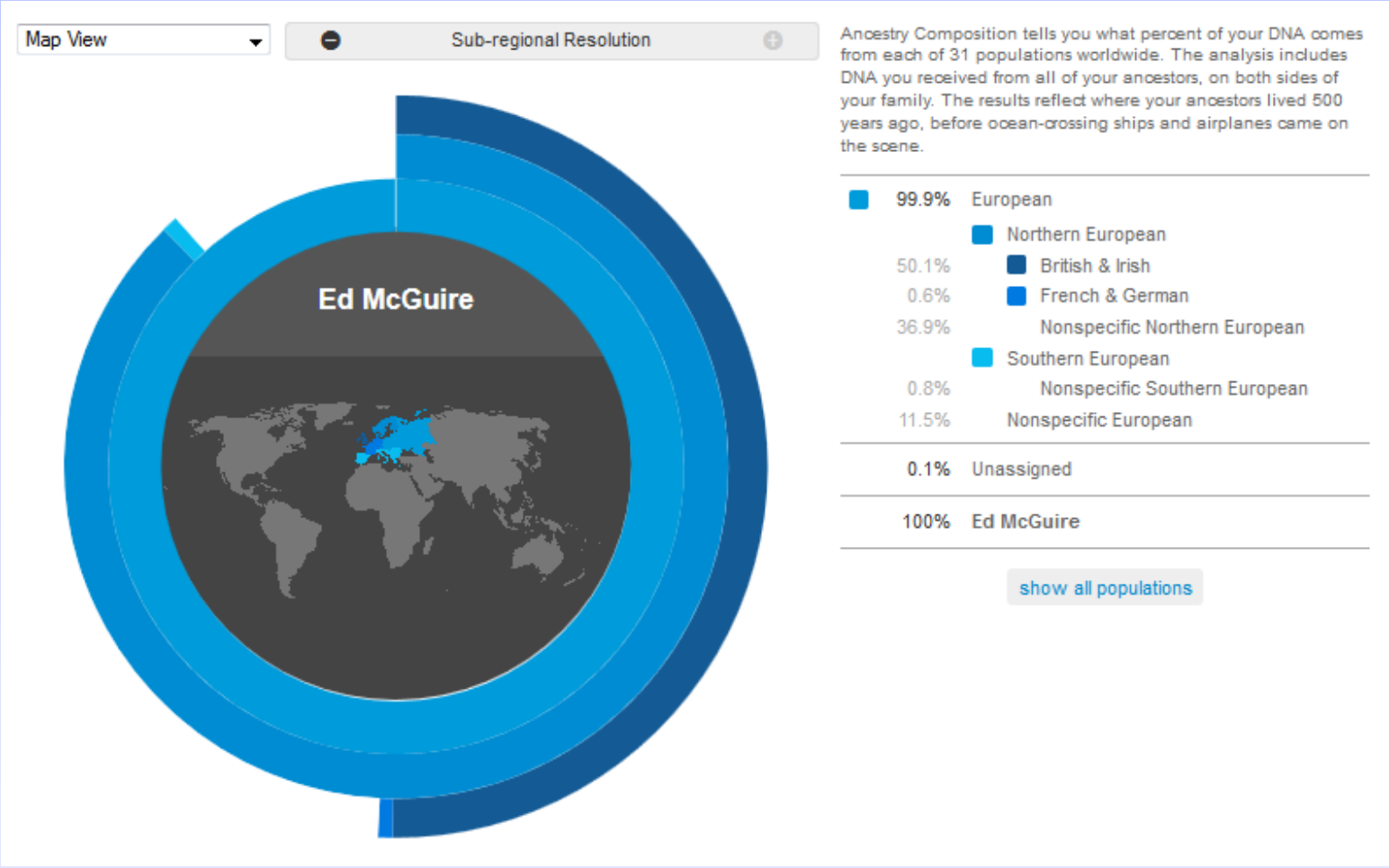


AncestryDNA Ethnicity Estimate

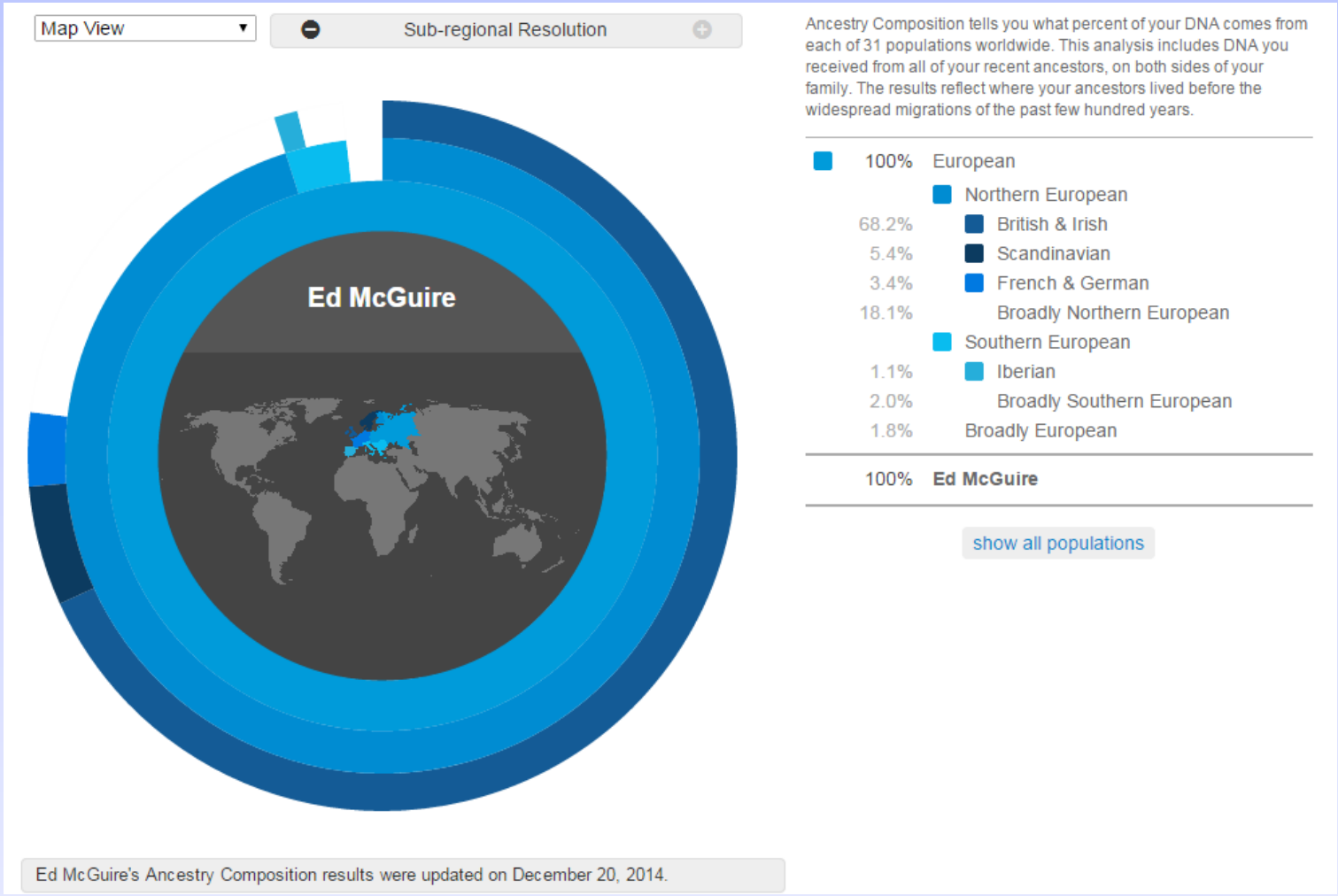


Europe	96%
Europe West	35%
Great Britain	24%
Ireland	18%
Scandinavia	14%
Iberian Peninsula	4%
Italy/Greece	1%
Caucasus	2%

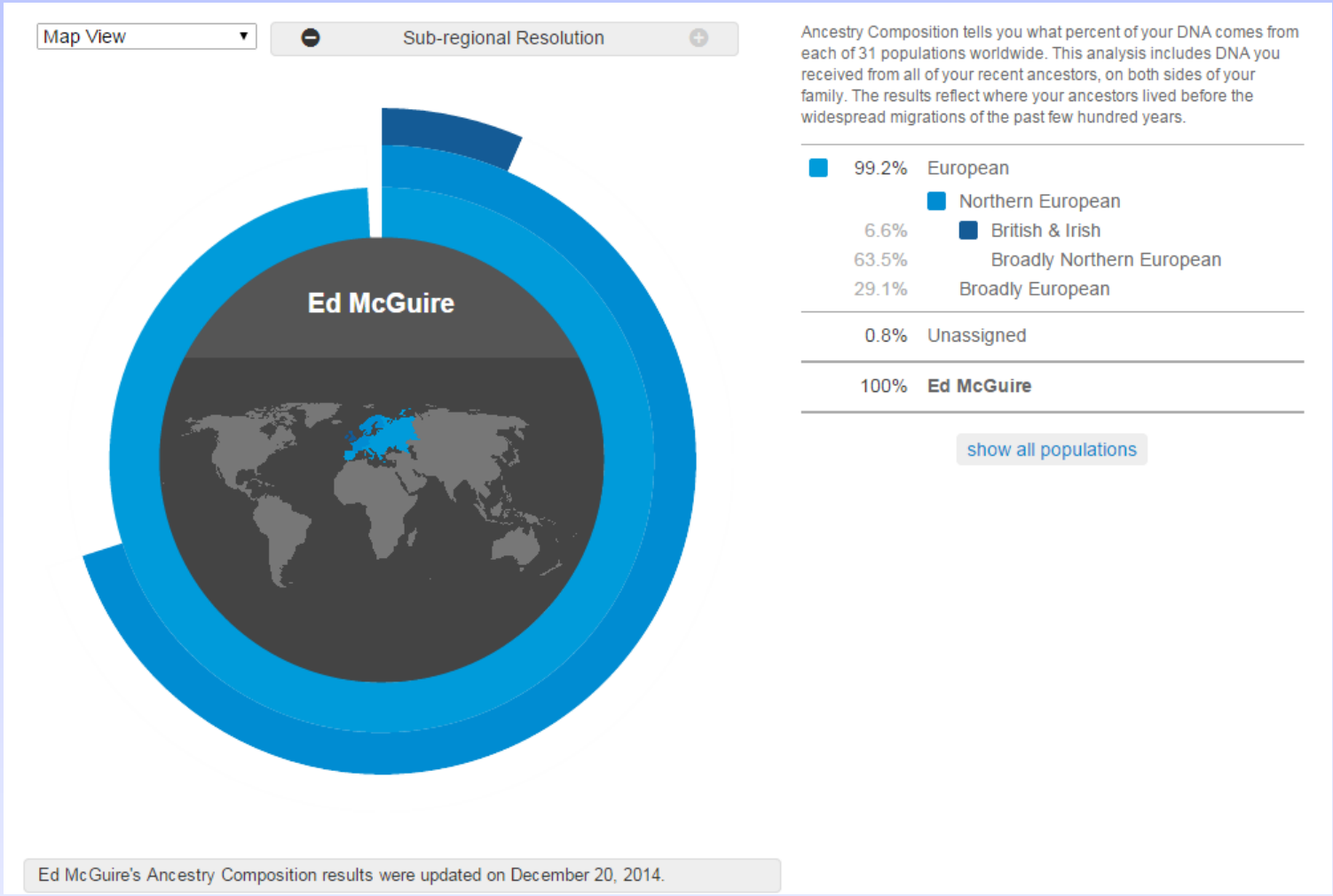
23andMe Ancestry Composition – Standard Model



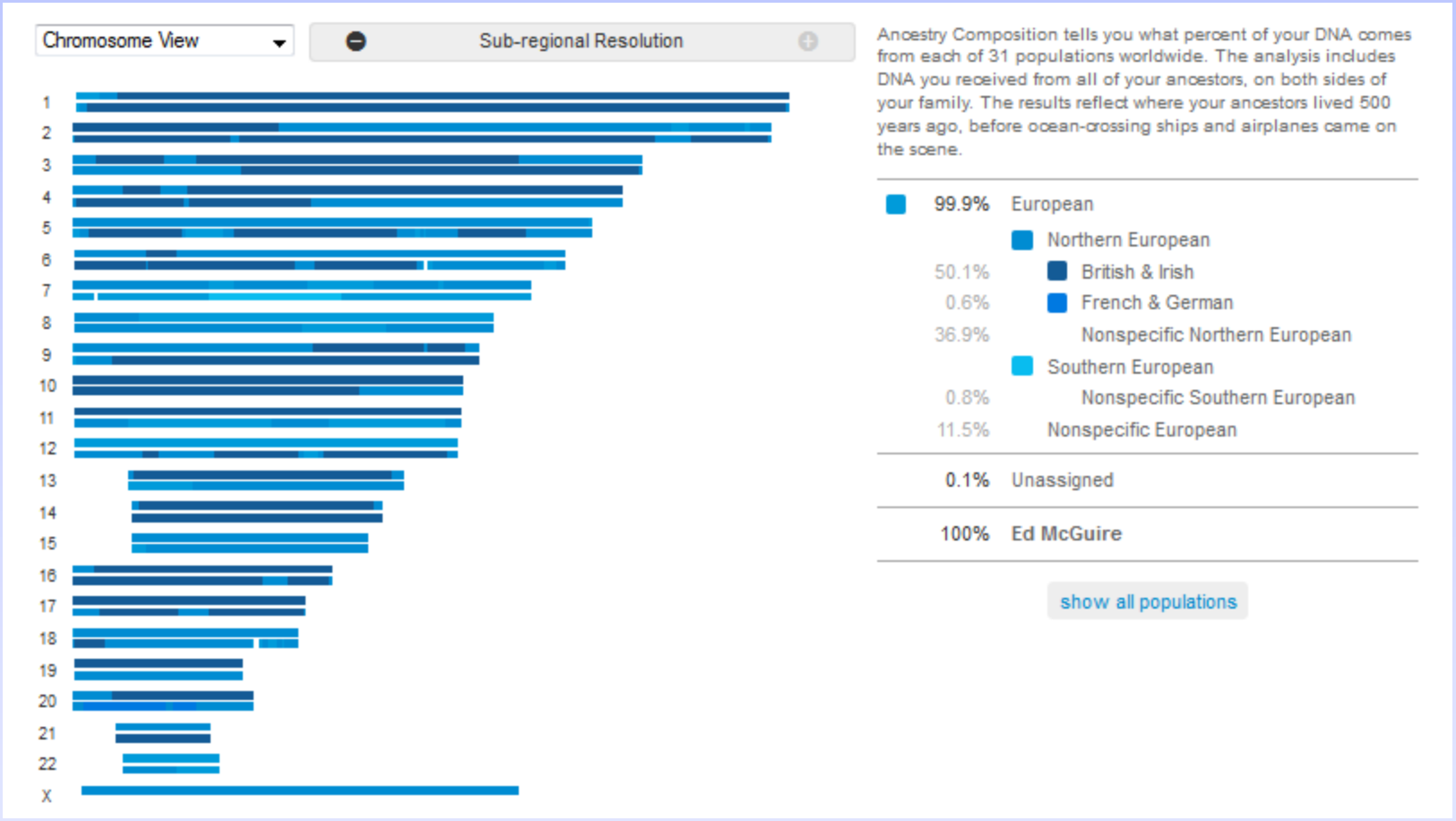
23andMe Ancestry Composition – Speculative Model



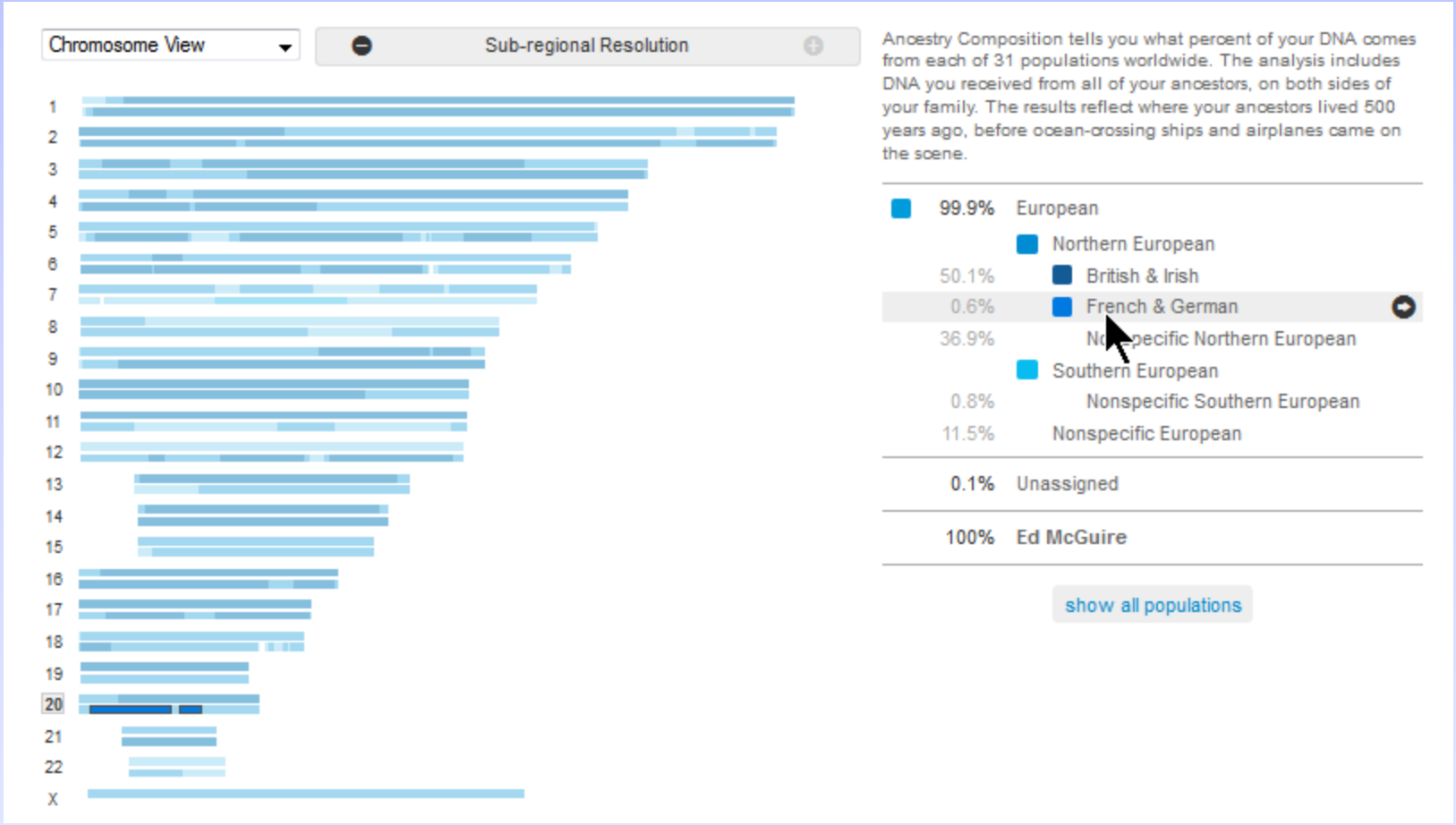
23andMe Ancestry Composition – Conservative Model



23andMe Ancestry Composition



23andMe Ancestry Composition



Comparison: Admixture Estimates

(Ed's Results)

	FT-DNA	Anc-DNA	23andMe	
British/Irish	55.5	42.0	50.1	
Europe	44.5	49.0	37.5	
Other Europe	0.0	5.0	11.5	
West Asia	0.0	2.0	0.0	

Comparison: Admixture Estimates

(Ed's Results)

	FT-DNA	Anc-DNA	23andMe	Pedigree
British/Irish	55.5	42.0	50.1	62.5
Europe	44.5	49.0	37.5	37.5
Other Europe	0.0	5.0	11.5	0.0
West Asia	0.0	2.0	0.0	0.0

Admixture: "Not ready for prime time"

"The bottom line is that ethnicity tools are not well understood by consumers in terms of the timeframe that is being revealed…"

"…it's not consistent between vendor, nor are the results"

"In some cases, they are flat out wrong, as with Ancestry, and can be proven."

"This does note engender a great deal of confidence"

- Roberta Estes (http://dna-explained.com/category/admixture/)

Matches

Matches

- **The more DNA you share the closer you are to your match**
- **The more DNA you share in large segments the closer you are**
- **Shared segments are often measured in centiMorgans (cM)**

> *"A centiMorgan is equal to a 1% chance that a marker at one genetic locus on a chromosome will be separated from a marker at a second locus due to crossing over in a single generation"*

- **centiMorgans are statistical estimates of equal distance (genetically) from one point to another – 'DNA yardsticks'**

Working with Cousins Who Match Your DNA

Here is where the work begins (and pays off)

Matches displayed highest to lowest relationship (default)

Make it easy on your matches:

- **up-to-date email address**
- **oldest known paternal/maternal ancestor, date, location**
- **detailed list of ancestral surnames, dates, locations**
- **pedigree chart (GEDCOM or PDF) for them to use**

Working with Cousins Who Match Your DNA

Response rates (user reported):

- **23andMe – least likely to reply**
- **AncestryDNA – much better**
- **FamilyTreeDNA – highest level**

Working with Cousins Who Match Your DNA

Response rates (user reported):

- 23andMe – least likely to reply
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- FamilyTreeDNA – highest level

Develop friendly note asking if they'd like to share data

- include some basic info from your family research
- not generic; make it specific to them

Working with Cousins Who Match Your DNA

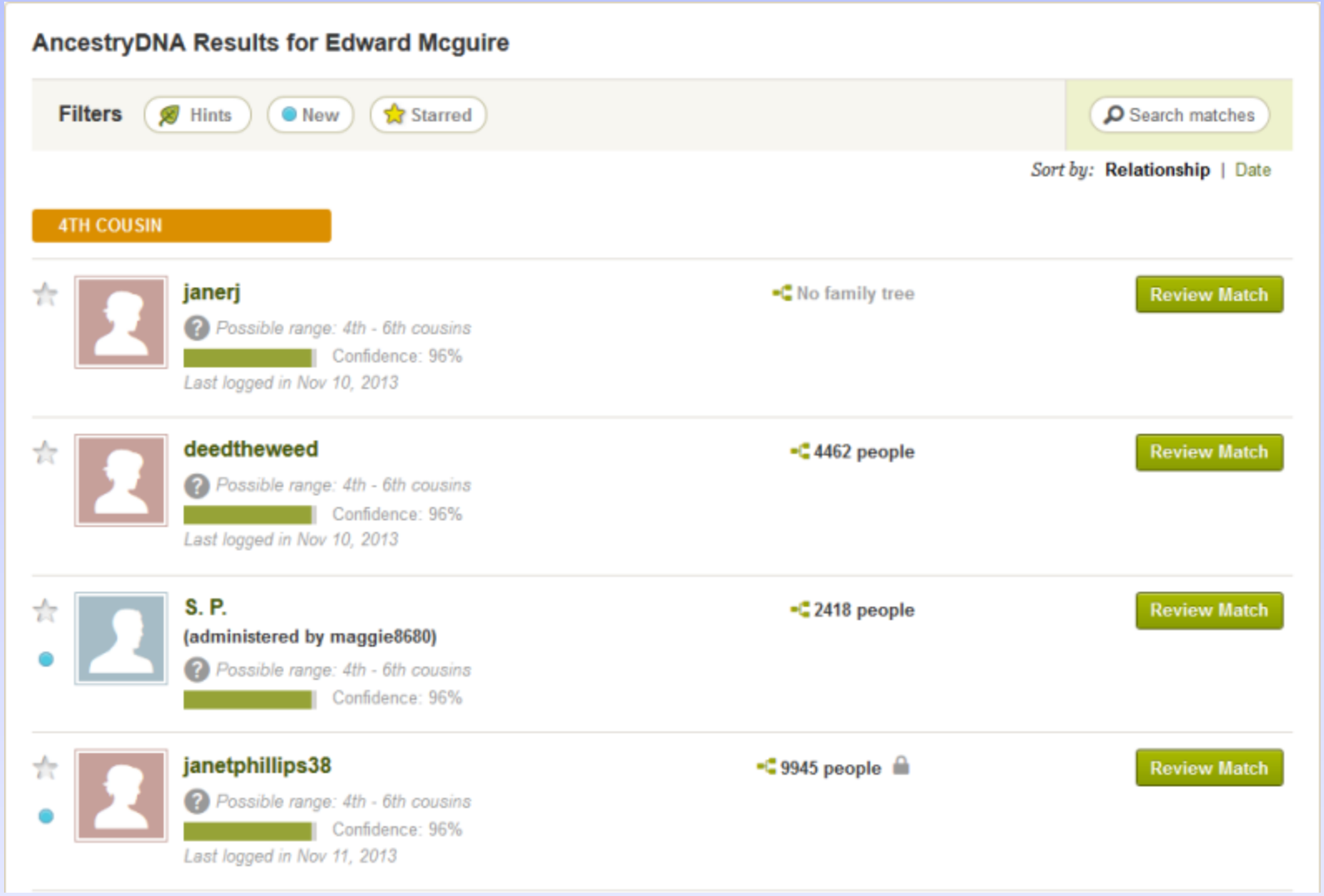
Estimates of relatedness:

- **FT-DNA estimates are closest (2d – 4th cousins)**
- **AncestryDNA seem to start at 4th – 6th cousins**
- **23andMe often '3rd to Distant' cousins**

Messaging systems differ:

- **AncestryDNA – uses internal Ancestry mail system**
- **23andMe – most controlled due to medical data involved**
- **FamilyTreeDNA – uses your normal email (required)**

AncestryDNA Matches Page



Effective Ancestral Range of Autosomal Testing

Chances of finding a match:

Relationship	Match Probability
2nd cousins or closer	> 99%
3rd cousin	> 90%
4th cousin	> 50%
5th cousin	> 10%
6th cousin and more distant	Remote (typically less than 2%)

Effective Ancestral Range of Autosomal Testing

Most experts suggest matches to cousins probable out 5-6 generations

Reasons for the 5-6 generation 'cliff':

- we inherit only 50% of each parent's DNA; 25% G-parents…

- we begin to lose contributions from more distant ancestors

- segments inherited from distant ancestors more fragmented

Our matches beyond 5-6 generations tend to share << 10 cM limit

- much harder to find MRCA

Testing Companies

Which DNA Testing Company Do I Use?

Genetic Genealogy Testing Companies

Top Picks

23andMe

ancestryDNA BETA

FamilyTreeDNA
HISTORY UNEARTHED DAILY

Genetic Genealogy Testing Companies

Critical Attributes

Expertise

Experience

Size of existing DNA databases

Proven track record

Support

Cost

DNA Testing Companies

	Y-DNA	mt-DNA	at-DNA	Medical
23andMe	Y*	Y*	Y	~~Y~~
AncestryDNA	N	N	Y	N
FamilyTreeDNA	Y	Y	Y	N

All 3 companies use versions of the Illumina chip for autosomal testing

(* 23andMe test for Y-chromosome & mtDNA limited to defining major Haplogroups)

DNA Testing Companies

Recommendations

Autosomal Test Results

Each testing company has it's own database of customer DNA results

AncestryDNA customers →

FamilyTreeDNA customers →

23andMe customers →

Autosomal Test Results

You can move your results into FamilyTreeDNA for ~$59

ancestryDNA → FamilyTreeDNA HISTORY UNEARTHED DAILY

Company Comparisons - AncestryDNA

Strengths:

- *Easier to locate 'possible' matches w/ their trees
- *Biggest user database (> 800K)
- Good African-American data
- Good for colonial America ancestry
- DNA Circles tool is nice improvement

Weaknesses:

- *Exclusive US focus (no overseas sales until 2015 - UK/Irl)
- *Lack of any 'segment analysis' tools
- *Don't commit to save DNA
- Testers not normally versed in DNA basics
- *Trees have well-documented accuracy issues

* = significant factor

Company Comparisons - FamilyTreeDNA

Strengths:

- *High overseas test percentage
- *Very good 'segment' tools
- *Y-DNA & mtDNA data integrated w/ atDNA
- *Committed to 25 years storage of DNA
- Accept raw data transfers (AncDNA & old 23andMe data)
- Very connected to DNA community & 3rd party tools
- Strong DB for Jewish research

Weaknesses:

- *Smallest atDNA database (~ 150K)

* = significant factor

Company Comparisons – 23andMe

Strengths:

***Very good 'segment' tools**

***Large user database (> 700K)**

Strong DB for Jewish research

Best Admixture tool ('Ancestry Composition')

Chromosome View in 'Ancestry Composition'

Will store DNA for years

Weaknesses:

***Main focus is medical applications**

Test medically significant SNPs

***Matches are least responsive (10-20%)**

Site is poorly laid out & confusing

***Unnecessarily difficult to communicate**

*** = significant factor**

AutosomalDNA Testing Recommentations

#1 choice is FamilyTreeDNA (in my opinion)

If looking for African-American cousins & origins – AncestryDNA

If not intending to ever work with segment data - AncestryDNA

If also doing medical testing – 23andMe

If mainly interested in ethnicity percentages – 23andMe

If adoptee / unknown parentage – need to test with all three

If someone in your family tested w/ company A – you might follow lead

Autosomal Test Results

You can search for those common ancestors in a **bigger** pond!!

ancestryDNA

FamilyTreeDNA
HISTORY UNEARTHED DAILY

23andMe

→ GEDmatch

How to Save Money on DNA Tests

- Join a Surname Project or Haplogroup / Location Project first.
- Holiday Discounts at FT-DNA (~25 Nov – 31 Dec)
- Remember 'DNA Day' (April 25th)
- Ancestry.com gives discounts to members
- Ancestry & 23andMe offer discounts for multiple kits
- Watch company websites for random offers
- Look for promotions at conferences - like NERGC (?)

Advice and Warnings

- Ethnicity Percentages are speculative
- Using these tools requires more work than advertised

  - sending in the kit is the easy part

- The key is enter your ancestral data and seek out matches
- Data security – if you download results protect them
- Be prepared for NPEs – or don't test

Genetic Genealogy and Privacy Concerns

Our DNA is unique and a linked with our identity

- **23andMe autosomal test includes medically significant mutations**
- **Full Mitochondrial Sequence (FMS) has some medical implications**

Extensive data protections are used by all 3 companies

Before you test read the Terms & Conditions of the testing company

If you aren't happy after reading T's & C's of a site – don't test

Goals

- Discuss the genetics behind autosomal testing
- Identify its main uses for genealogical research
- Compare companies
- Review the format of your results

Should you test? Which company?

Genetic Genealogy At Our Library

4th Tuesday is 'DNA Night' for members

- DNA forum for discussions, questions and answers
- A focus on tools, tips and deeper understanding of the technology

"We're Here To Help"

Special Interest Groups

Interest Group	Week of the Month
Scottish	1st Tuesday (6:30 pm)
Quebec Research	2nd Tuesday (6:30 pm)
Irish	3rd Tuesday (6:30 pm)
Genetic Genealogy	4th Tuesday (6:30 pm)

Upcoming Classes

14 Feb	Ed McGuire	DNA Tests: Maternal & Paternal Lines
21 Feb	Bill Craig	Resources & Tools at VTGENLIB.ORG
28 Feb	Ed McGuire	Finding Cousins with Autosomal DNA (Part 1)
7 Mar	Erenst Anip	*Chronicling America* : Using Historic Newspapers in Genealogical Research
10 Mar*	Irish History Festival	Researching Your Irish Ancestors
14 Mar	Library Staff	Recording Your Ancestry with *Family Tree Maker*
21 Mar	Ed McGuire	Finding Cousins with Autosomal DNA (Part 2)
28 Mar	Gloria & Anastasia Pratt	Genealogy Research in New York State
4 Apr		**--- Closed for Easter Holiday ---**
11 Apr	Carol Schwenk	Daughters of the American Revolution: Their History & the DAR Research Process
18 Apr	Tom Devarney	Using Drouin's Lafrance website & its databases
25 Apr	Joanne Polanshek	Finding your ancestors in print at *GenealogyBank.com*
2 May	Connie Reik	Beyond Pension Files! Discover Your Civil War Veterans in Federal Publications
9 May	Lynn Johnson	Maximizing Your Use of Census Records
16 May	Sheila Morris	Tracing Your Scottish Ancestors

Don't Forget!

- Use our online forum on DNA testing (in Members Only area)
- Best books on genetic genealogy on sale here
- NEW: DNA testing laminated guides (6 topics)
- Big selection of other genealogy books & laminate guides

  - we do take credit cards & cash & checks

- Memberships ($30)