DNA:
Expectations and Realizations

Disclaimers:

- If you were expecting a presentation on DAR's DNA policy or the use of DNA on an application paper those then expectations will not be realized.

- If you were expecting to hear that now you have taken a DNA test, that you will no longer have to visit libraries, courthouses, etc. , then those expectations will not be realized.

Overview

- The Science—Biology and Statistics (sorry I have to)
- The three principal tests—yDNA, mtDNA, and atDNA
- The companies—Family Tree DNA, 23andMe, and ancestry.com
- The people—they're just human
- The final story

You have tested with a genetic genealogy company, have your results and are still not making any progress.

- Did you take the right test?
- Did you test with the right company?
- Can DNA testing possibly answer your questions?
- Do you believe that the person or persons who might help answer your questions were also tested?

Deoxy ribose Nucleic Acid

- Anything with “ose” is a sugar. These sugars along with phosphates form the backbone of DNA.
- The nucleic acids or bases are held in place by the sugars and phosphate backbone.
- Think of a ladder where the rungs are the nucleic acids and the sides are the sugars and phosphates.

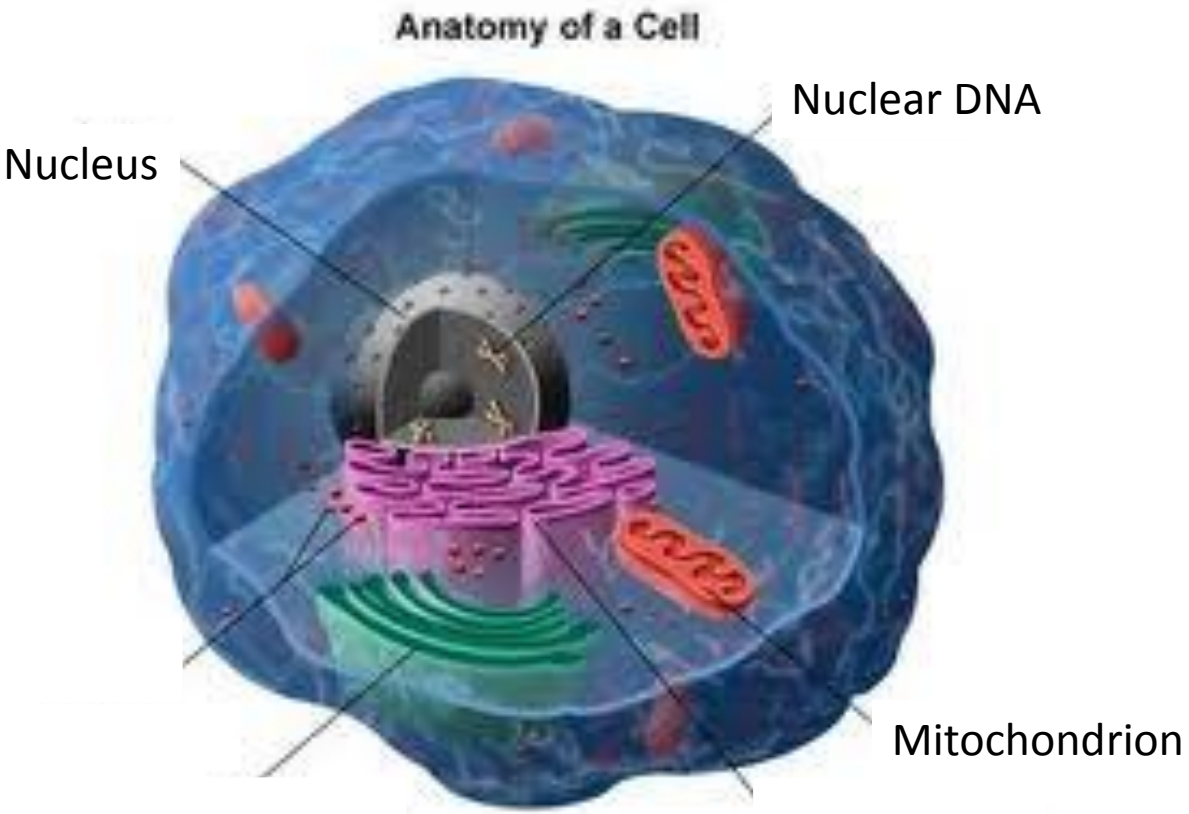
The Nucleic acids or bases

- There are four—A, C, G, and T
- They come in pairs A & T and C & G
- Chemically A cannot connect with another A, a C or a G. Similarly for the others. Think of a zipper that no longer is usable.
- The order of the bases is a code—the Genetic Code
- Who we were, who we are and who might become

DNA

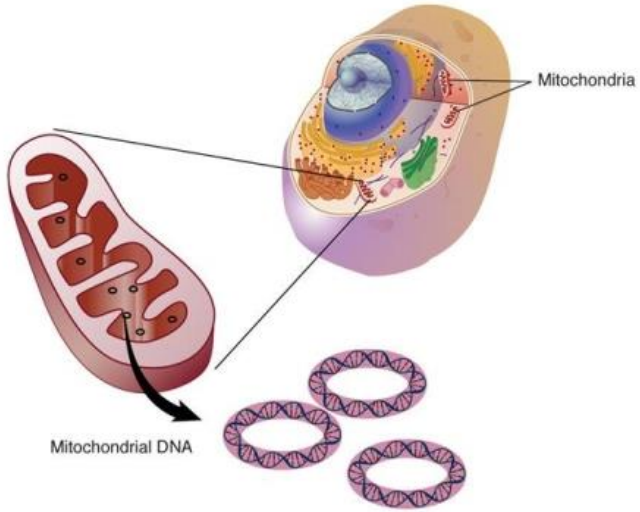
- Forms long strands
- Found in two locations in every cell
- In the mitochondria—mitochondrial DNA/mtDNA
- In the nucleus—nuclear DNA (both yDNA and atDNA)

Genealogy 101—DNA: Expectations and Realizations



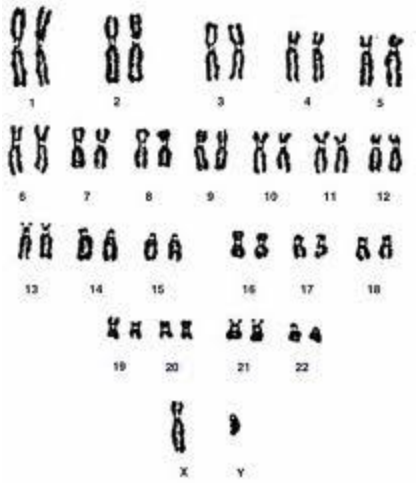
Mitochondrial DNA

- The ends of the long strands join forming a loop or ring
- 16,000+ base pairs long
- Your mitochondrial (mtDNA) comes from your mother



Nuclear DNA

- 23 pairs of long strands of DNA
- Totaling nearly 3,000,000,000 base pairs
- Numbered chromosomes 1 - 22 and X and Y



Chromosomes 1 - 22

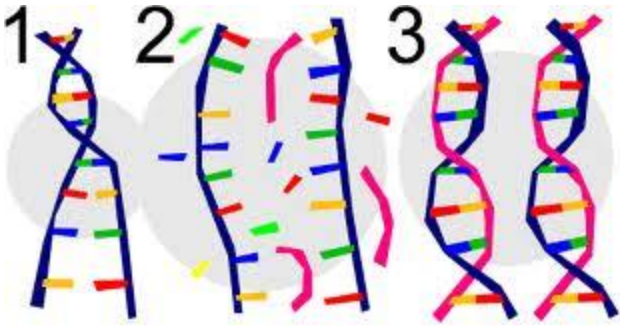
- Autosomal DNA (atDNA)
- One chromosome #1 comes from our father and one comes from your mother—making a pair

X and Y

- The sex chromosomes
- XY is a male. The X from the mother and the Y from the father.
- XX is a female. One X from each parent.

Growth and Division

- A cell must divide in two it cannot continue to grow in size.
- All structures in the cell including the DNA must be replicated prior to forming the new cell.
- To replicate the DNA strands unzip, are replicated, and then zipped up again.



Mistakes or Mutations

- As the DNA is copied on rare occasions mistakes occur
- There are editors within the cell, but still some mistakes get through
- These mistakes are called mutations and are the basis of genetic genealogy
- Scientist can measure the rate of time between mutations.
- The greater the number of accumulated mutations between two samples the greater the time since the first mutation.

Genetic Testing for Genealogy

- Since mtDNA only comes from our mother this can determine connections on our umbilical line.
- Since only males have yDNA this can determine connections on our paternal line.
- The atDNA is a collection of portions of the DNA of all of our ancestors. Statistical analysis of chromosomes 1 -22 can begin to identify cousins of varying degrees.

mtDNA testing

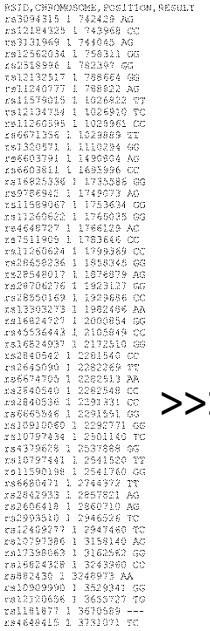
- Usually only test a few hundred locations.
- Determines which base pair is present at that location.
- Time frame is wrong for genealogy.
- Tests of all 16,000+ base pairs are available
- A perfect match suggests a common ancestor within 550 years.

yDNA testing

- Much of the Y Chromosome is considered “junk DNA.”
- Consists of groups of repetitive sequences called single tandem repeats (STR’s). I prefer to call them stutters.
- The number of repeats for each of these STR’s changes through time just like a mutation.
- Can determine how closely related two male lines are.
- Satisfactory for genealogical purposes. Tracks with the family surname in most western cultures.

atDNA testing

- Determines which base pair is present at a large number of locations (500,000 or more) covering all of the 22 non-sex chromosomes.
- Two bases at every location—one from your mother, one from your father. Do not know which is which.
- A statistical analysis is necessary to sort out the voluminous data.



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rs12184325 1 743968 CC
rs3131969 1 744045 AG
rs12562034 1 758311 GG
rs2518996 1 782197 GG
rs12132517 1 788664 GG
rs11240777 1 788822 AG
rs11579015 1 1026852 TT
rs12134789 1 1026910 TC
rs11260595 1 1029561 CC
rs66671356 1 1029889 TT
rs1320571 1 1110294 GG
rs6603791 1 1490904 AG
rs6603811 1 1685996 CC
rs16825336 1 1735585 GG
rs9786942 1 1749073 AG
rs11589067 1 1753634 GG
rs11260622 1 1765035 GG
rs4648727 1 1766129 AC
rs7511905 1 1783646 CC
rs11260624 1 1799369 CC
rs28658236 1 1858345 GG
rs28548017 1 1876879 AG
rs28706276 1 1923127 GG
rs28550169 1 1923686 CC
rs13303273 1 1962496 AA
rs16824727 1 2000854 GG
rs45586443 1 2105549 CC
rs16824937 1 2172510 GG
rs2840542 1 2281540 CC
rs2645090 1 2282269 TT
rs6674705 1 2282513 AA
rs2840540 1 2282548 CC
rs2840536 1 2291931 CC
rs6665566 1 2291951 GG
rs10910060 1 2292771 GG
rs10797434 1 2301140 TC
rs4379628 1 2537888 GG
rs10797541 1 2541520 TT
rs11590198 1 2541760 GG
rs6680471 1 2744972 TT
rs2842933 1 2857821 AG
rs2606418 1 2860710 AG
rs2903510 1 2946526 TC
rs12409277 1 2947460 TC
rs10797586 1 3158140 AG
rs17398063 1 3160562 GG
rs16824328 1 3243980 CC
rs882430 1 3248973 AA
rs10909990 1 3529341 GG
rs12300656 1 3535727 TG
rs1181877 1 3670589 ---
rs4648415 1 3731071 TC
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>>>>>>14,000+ pages more>>>>>

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"rs9616906","22","49451546","GG"
"rs2341010","22","49486246","TC"
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"rs3810648","22","49522492","AA"
```

Parental Warning:

Some viewers may consider the following slides offensive or obscene.

Science is not an exact Science

- No experiment or measurement is without error.
- Repeated measurements can only give you an average.
- Scientists deal with errors by using probability and statistics.
- Measuring mutation rates is a scientific experiment and therefore subject to uncertainty and statistical analysis

The mass of an electron

0.510 998 928 ± 11 MeV

Average value

Statistical uncertainty

The true value is between
0.519 998 917 and 0.519 998 939
with 68.2 % certainty

If we take double the error the value becomes
0.519 998 906 and 0.519 998 950
with 95.4 % certainty

In the second example we are more sure we have the value within the given range but the range is larger so we are less sure of the actual value.

Probability and Statistics for Genetic Genealogists

- No rigorous mathematics.
- Odds are based on millions and millions of chances.
- Simple examples-flipping a coin, rolling a die, drawing a card.
- Complicated example—a lottery ticket

Flipping a coin

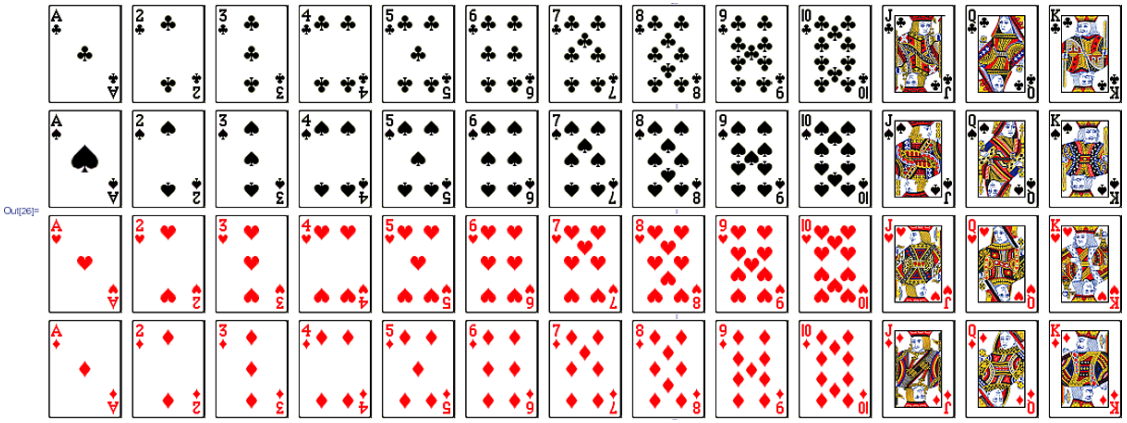
- Usually two possibilities.
- Heads or Tails.
- 50-50 chance, one out of two.

Rolling a die

- Six possibilities.
- Side with 1, or 2, 3, 4, 5, or 6 pips facing up.
- About 16.7 % chance of any side facing up, one out of six.

Drawing a card

- 52 possibilities, but take out one-eyed Jacks—50 posssibilities.
- Numbers 2-10, A, K, Q, and J for four different suits.
- 2 % chance of drawing any card with replacement.



"Complicated" Example

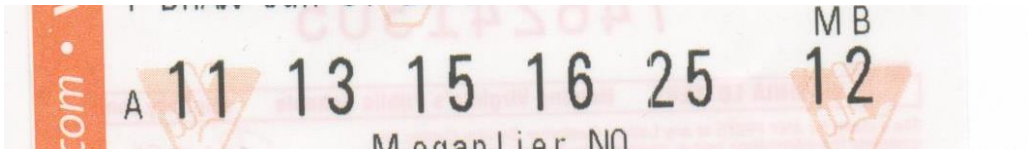
- We want "heads," to roll one on the die, and to draw the Ace of spades.
- One out two multiplied by one out of six multiplied by one out of 50.
- One out of 600 chance of this combination occurring.

Playing the lottery

- Choose six numbers
- Five numbers from 1-75 without replacement.
- One number from 1-22.

Playing the lottery (part 2)

- 1 in 258,890,850 chance of matching all six numbers.
- Five numbers drawn-one chance out of 75, times one chance out of 74, etc.
- Times one chance out of 22.
- In this case the order does not matter.



Playing the lottery (part 3)

- Change to a spreadsheet
  - Place my name at the left with an explanation.
  - Give each column a heading name.

	Ball #1	Ball #2	Ball #3	Ball #4	Ball #6	Ball #7
Tom's Lottery Picks	11	13	15	16	25	12

Playing the lottery (part 4)—kind of

- Changes to the spreadsheet
  - Change the explanation next to my name.
  - Change the column heading names.

The lottery results have now morphed into yDNA results

- These are the actual values of six of the first seven markers on my yDNA
- The fifth value was the same as one of the first four and could not be used (without replacement)
- We know how to calculate the probabilities if we know the mutation rate

	DYS391	DYS393	DYS385b	DYS19	DYS390	DYS426
Tom's—yDNA	11	13	15	16	25	12

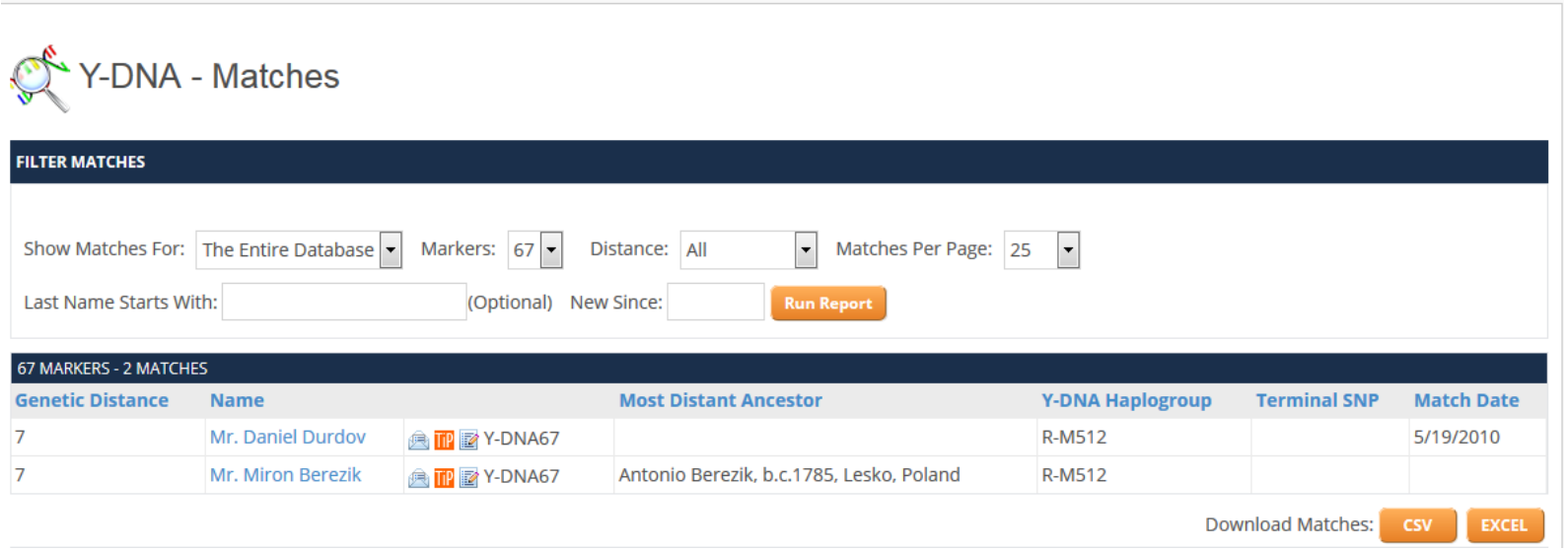
Genealogy 101—DNA: Expectations and Realizations

My yDNA results as reported in a project on the Family Tree DNA website.

Row Number	Kit Number	Name	Paternal Ancestor Name	Country	Haplogroup	DYS393	DYS390	DYS19	DYS391	DYS385	DYS426	DYS388	DYS439	DYS389I	DYS392	DYS389II	DYS458	DYS459	DYS455	DYS454	DYS447	DYS437	DYS448	DYS449	DYS464	DYS460	Y-GATA-H4	YCAII	DYS456	DYS607	DYS576	DYS570	CDY	DYS442	DYS438	DYS531	DYS578	DYF395S1
E-M96>P147																																					
1	187637			England	E-M183	13	24	13	9	13-15	11	12	10	14	11	30	18	9-9	11	11	23	14	20	33	14-14-16-16	11	11	19-22	16	13	18	22	38-38	12	10	10	8	15-
I-M170>M253																																					
2	249925			Poland	I-S6402	13	22	14	10	13-15	11	14	11	12	12	28	15	8-9	8	11	23	16	20	28	12-14-14-15	10	10	19-21	14	14	16	20	36-38	12	10	11	8	15-
3	297473			England	I-M253	13	22	14	10	13-16	11	14	11	12	11	28	16	8-9	8	11	23	16	20	28	12-14-15-16	10	10	19-22	14	14	17	19	32-37	12	10			
J-M304>M267																																					
4	83579			Unknown Origin	J-ZS1559	12	23	14	10	13-18	11	15	11	13	11	30	18	8-9	11	11	25	14	20	24	12-14-16-17	11	10	22-22	15	14	15	19	32-33	13	10	11	8	15-
Q-M242																																					
5	324491	Pedro Alfonso Sosa Garavito	Indalesio Sosa	Colombia	Q-L472	13	23	13	10	14-16	12	12	12	13	14	30	16	8-9	11	11	26	14	20	29	14-15-15-15	10	12	19-23	15	16	19	17	34-38	12	10			
R-M207>M173>M420																																					
6	243564			Greece	R-M512	13	24	15	11	11-14	12	12	10	13	11	30	15	9-10	11	11	24	14	20	32	12-15-15-16	12	12	19-25	15	15	16	20	36-39	12	11	11	8	17-
7	225976			Unknown Origin	R-CTS11962	13	25	16	10	11-14	12	12	11	13	11	30	17	9-10	11	11	23	14	19	33	12-15-15-16	11	10	19-23	15	16	18	19	34-40	15	11			
R-M207>M173>M420>Z283>Z282																																					
8	179780	Thomas John Ragusin		Croatia	R-Z282	13	25	16	11	11-15	12	12	10	14	11	31	15	9-10	11	11	24	14	20	32	12-15-15-16	10	11	19-23	16	15	18	21	36-42	12	11	11	8	17-

My two "closest" yDNA matches

- Genetic distance of 7 is not very good
- Both are different
- But how close are we?



Y-DNA - Matches

FILTER MATCHES

Show Matches For: The Entire Database | Markers: 67 | Distance: All | Matches Per Page: 25

Last Name Starts With: (Optional) New Since: Run Report

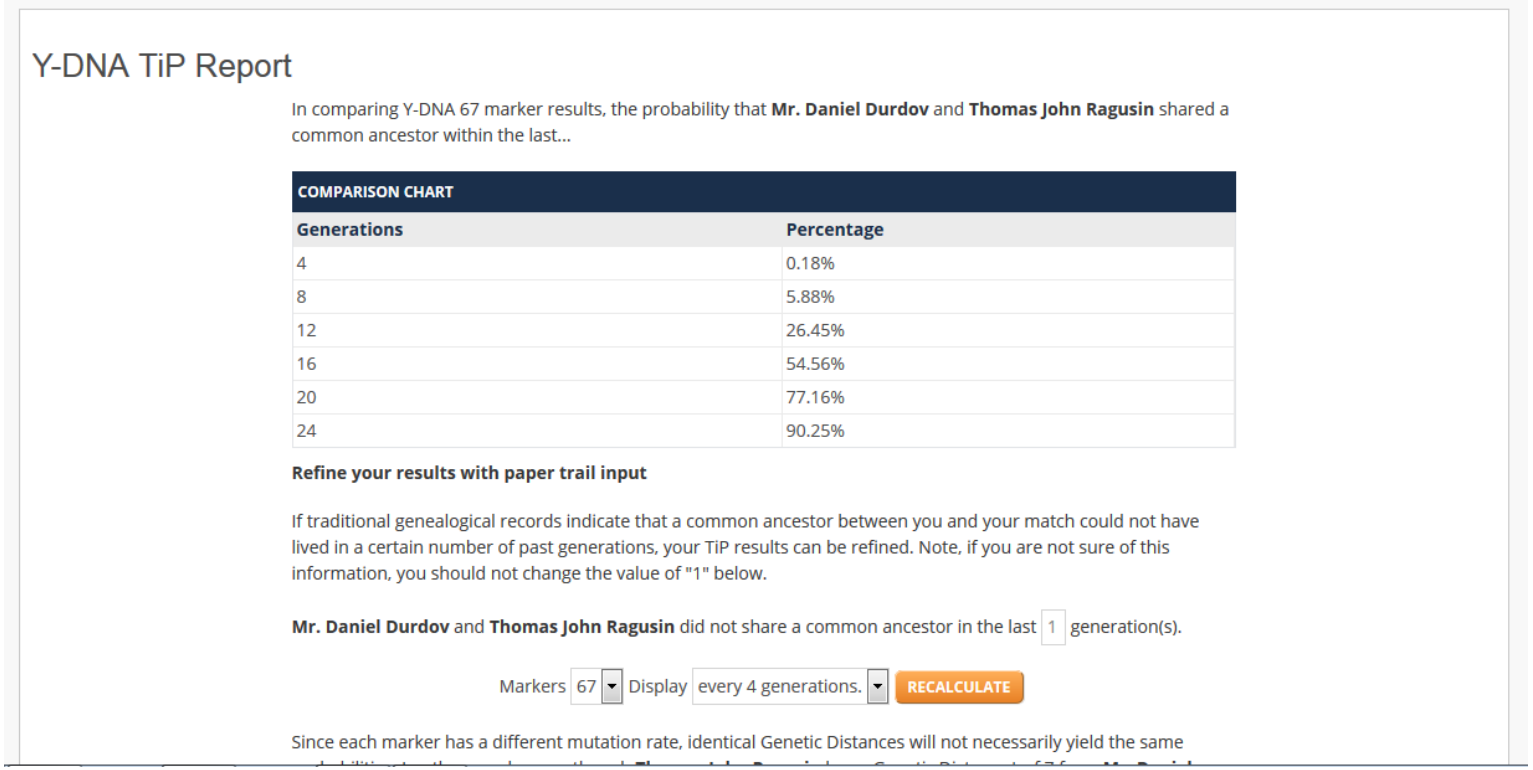
67 MARKERS - 2 MATCHES

Genetic Distance	Name		Most Distant Ancestor	Y-DNA Haplogroup	Terminal SNP	Match Date
7	Mr. Daniel Durdov	Y-DNA67		R-M512		5/19/2010
7	Mr. Miron Berezik	Y-DNA67	Antonio Berezik, b.c.1785, Lesko, Poland	R-M512		

Download Matches: CSV EXCEL

The closeness of the match

- Generations do not exist!
- Think of years instead with 25 years to a generation



Y-DNA TiP Report

In comparing Y-DNA 67 marker results, the probability that **Mr. Daniel Durdov** and **Thomas John Ragusin** shared a common ancestor within the last...

COMPARISON CHART	
Generations	**Percentage**
4	0.18%
8	5.88%
12	26.45%
16	54.56%
20	77.16%
24	90.25%

Refine your results with paper trail input

If traditional genealogical records indicate that a common ancestor between you and your match could not have lived in a certain number of past generations, your TiP results can be refined. Note, if you are not sure of this information, you should not change the value of "1" below.

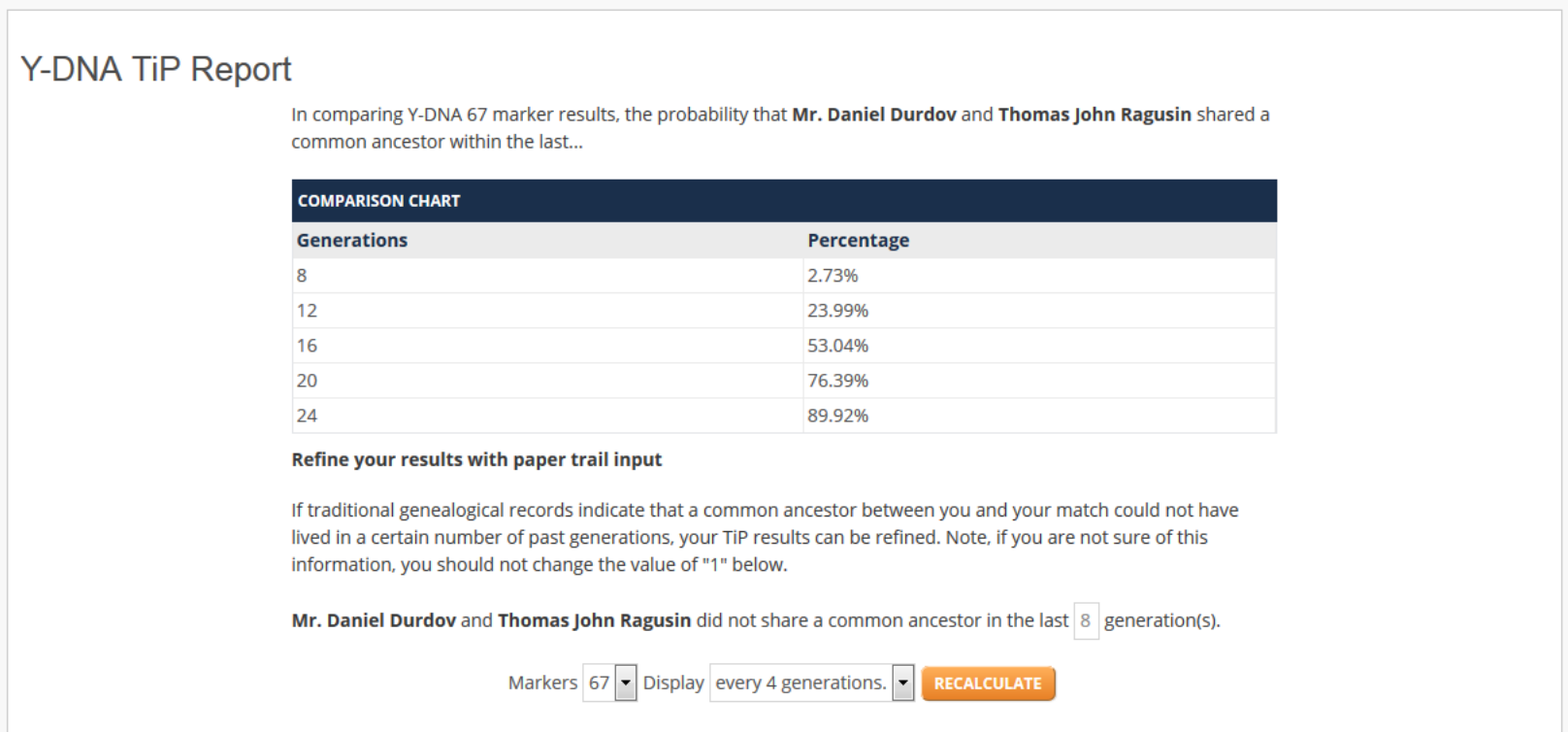
Mr. Daniel Durdov and **Thomas John Ragusin** did not share a common ancestor in the last 1 generation(s).

Markers 67 Display every 4 generations. RECALCULATE

Since each marker has a different mutation rate, identical Genetic Distances will not necessarily yield the same

Traditional Genealogy considered

- We are not related within eight generations
- More of a chance of being more than 24 generations



Y-DNA TiP Report

In comparing Y-DNA 67 marker results, the probability that **Mr. Daniel Durdov** and **Thomas John Ragusin** shared a common ancestor within the last...

COMPARISON CHART	
Generations	**Percentage**
8	2.73%
12	23.99%
16	53.04%
20	76.39%
24	89.92%

Refine your results with paper trail input

If traditional genealogical records indicate that a common ancestor between you and your match could not have lived in a certain number of past generations, your TiP results can be refined. Note, if you are not sure of this information, you should not change the value of "1" below.

Mr. Daniel Durdov and **Thomas John Ragusin** did not share a common ancestor in the last 8 generation(s).

Markers 67 Display every 4 generations. RECALCULATE

My mtDNA results

- Compared to a reference sequence, list of differences
- Formerly CRS, now RSRS

Haplogroup - W1

Your Origin

Haplogroup W is derived from the N superhaplogroup, which dates to approximately 65,000 years ago. The origin of haplogroup W dates to approximately 25,000 years ago, and it is mainly found distributed in west Eurasia (or Europe). It is likely that individuals bearing this lineage participated in the expansion into the bulk of Europe following the Last Glacial Maximum. Future work, including obtaining more samples from central Asia, will further refine the historical distribution of this haplogroup and better determine the role it played in the peopling of Europe.

USAGE POLICY: Use of the above Haplogroup description requires written permission from Gene by Gene.

Your Results

You are logged in as a GAP Administrator. This user has changed his/her settings so the mtDNA FASTA file **cannot** be downloaded by GAP Administrators.

RSRS Values | rCRS Values

Extra Mutations	315.1C G513c 522.1A 522.2C G16145A
Missing Mutations	

HVR1 DIFFERENCES FROM RSRS				
A16129G	G16145A	T16187C	C16189T	G16230A
T16278C	C16292T	C16311T		

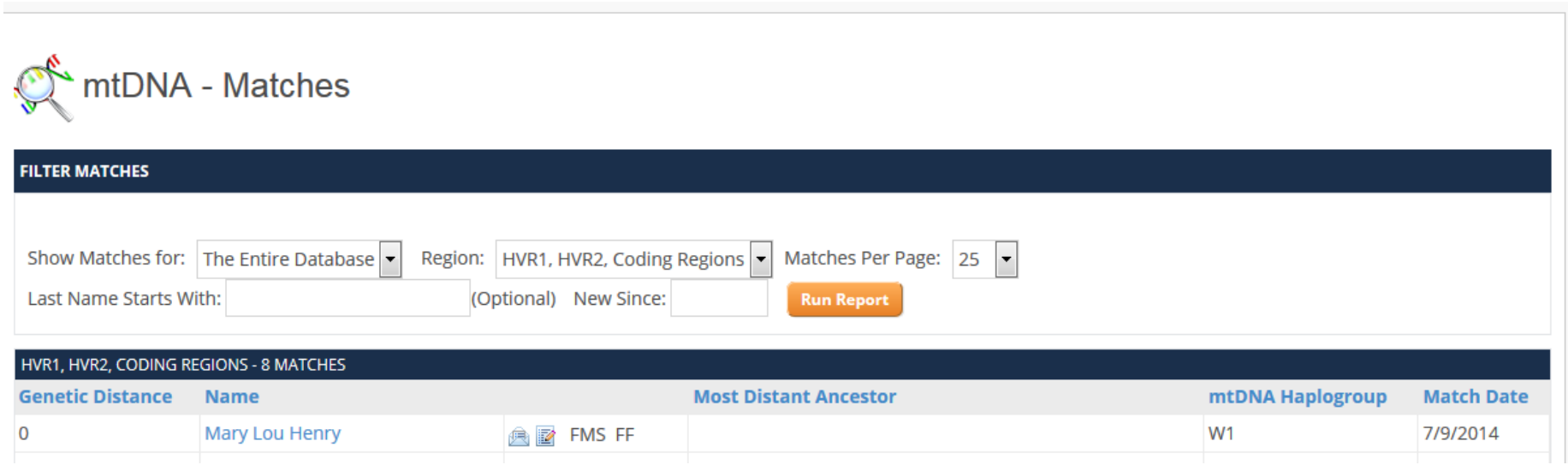
HVR2 DIFFERENCES FROM RSRS				
C146T	C152T	A189G	T204C	G207A
A247G	315.1C	G513c	522.1A	522.2C

Only the primary user of this account can view these results. Please sign in with the kit number and password to access this information.

To learn more about RSRS click here.

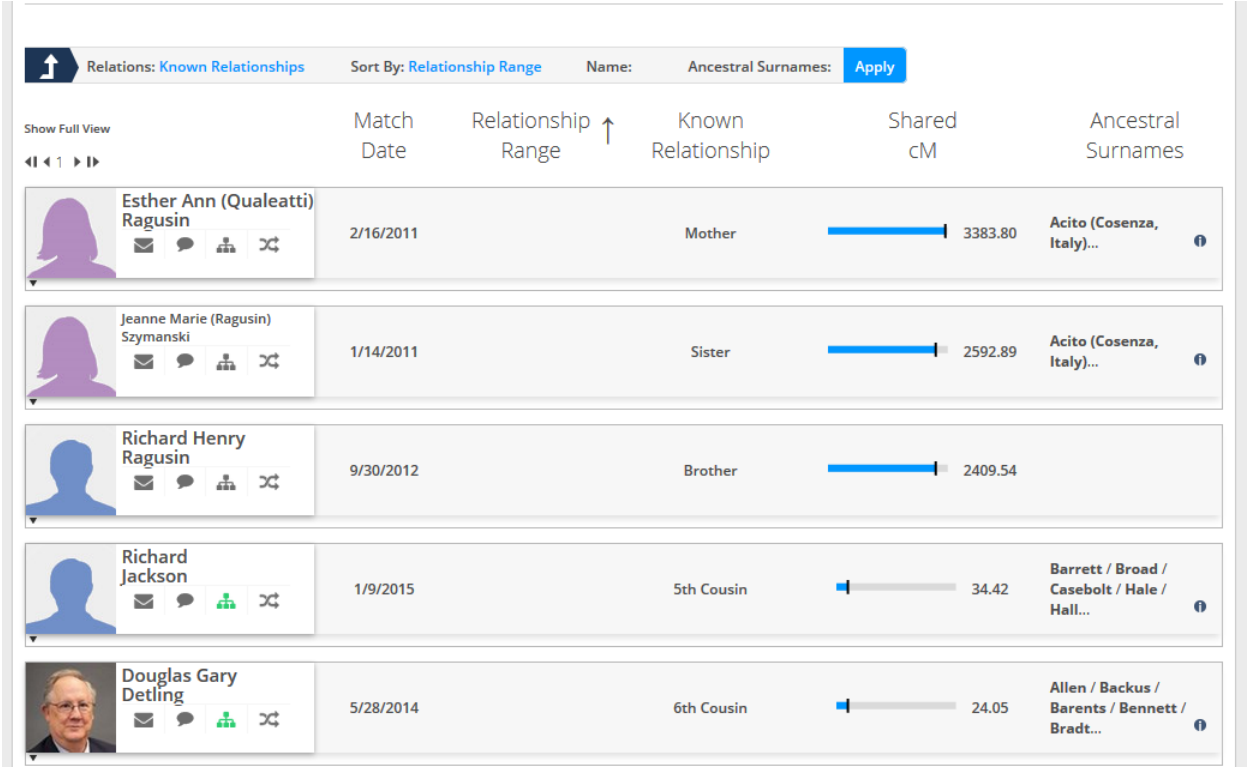
mtDNA example

- A perfect match--all 16,000+
- 50 % within 125 years, 95 % within 550
- She's adopted



My atDNA results-known relationships

- I have 131 matches
- I have identified only five relationships including my mother, sister, and brother



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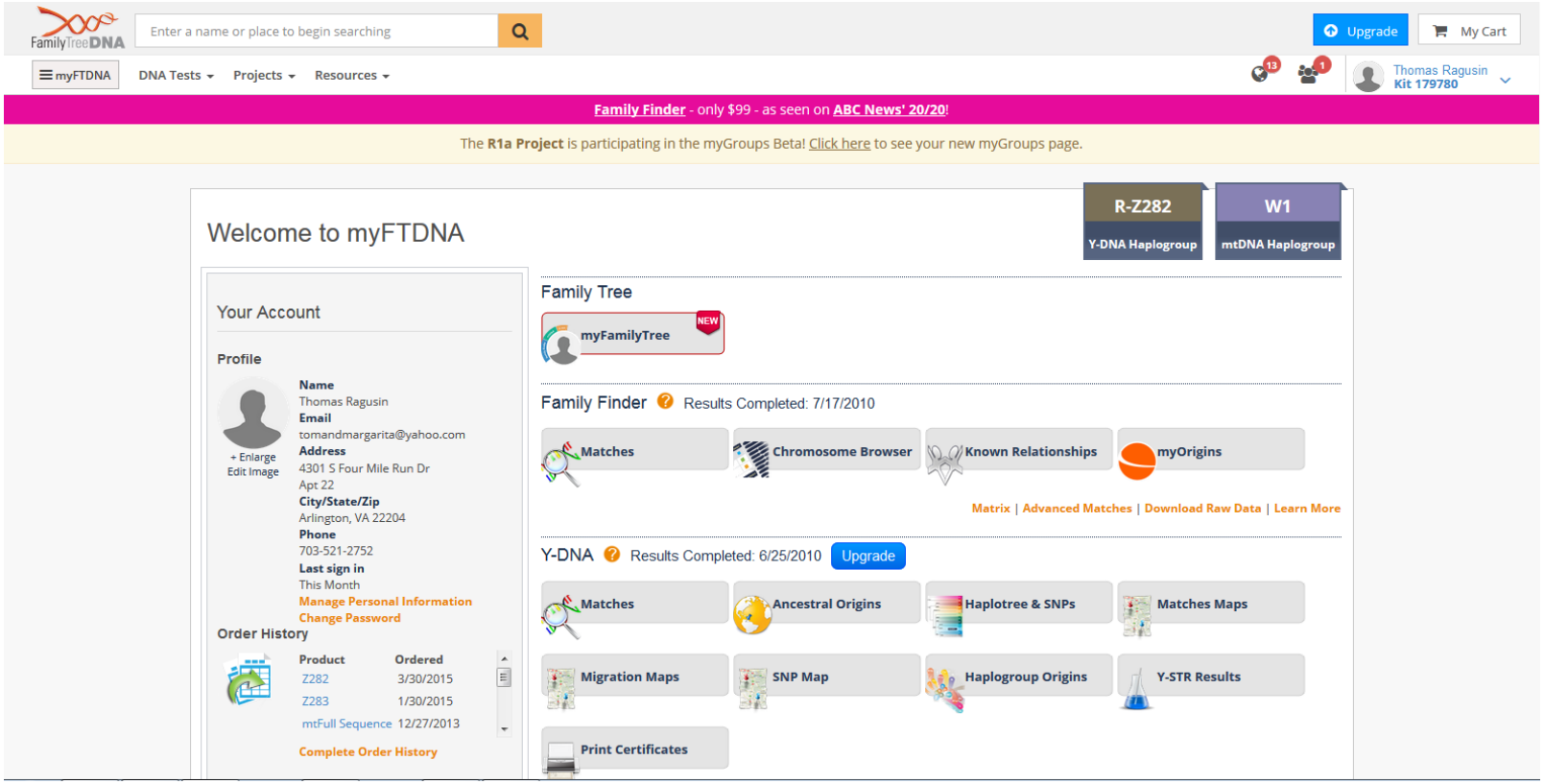
My Family Tree DNA homepage—the “dashboard”



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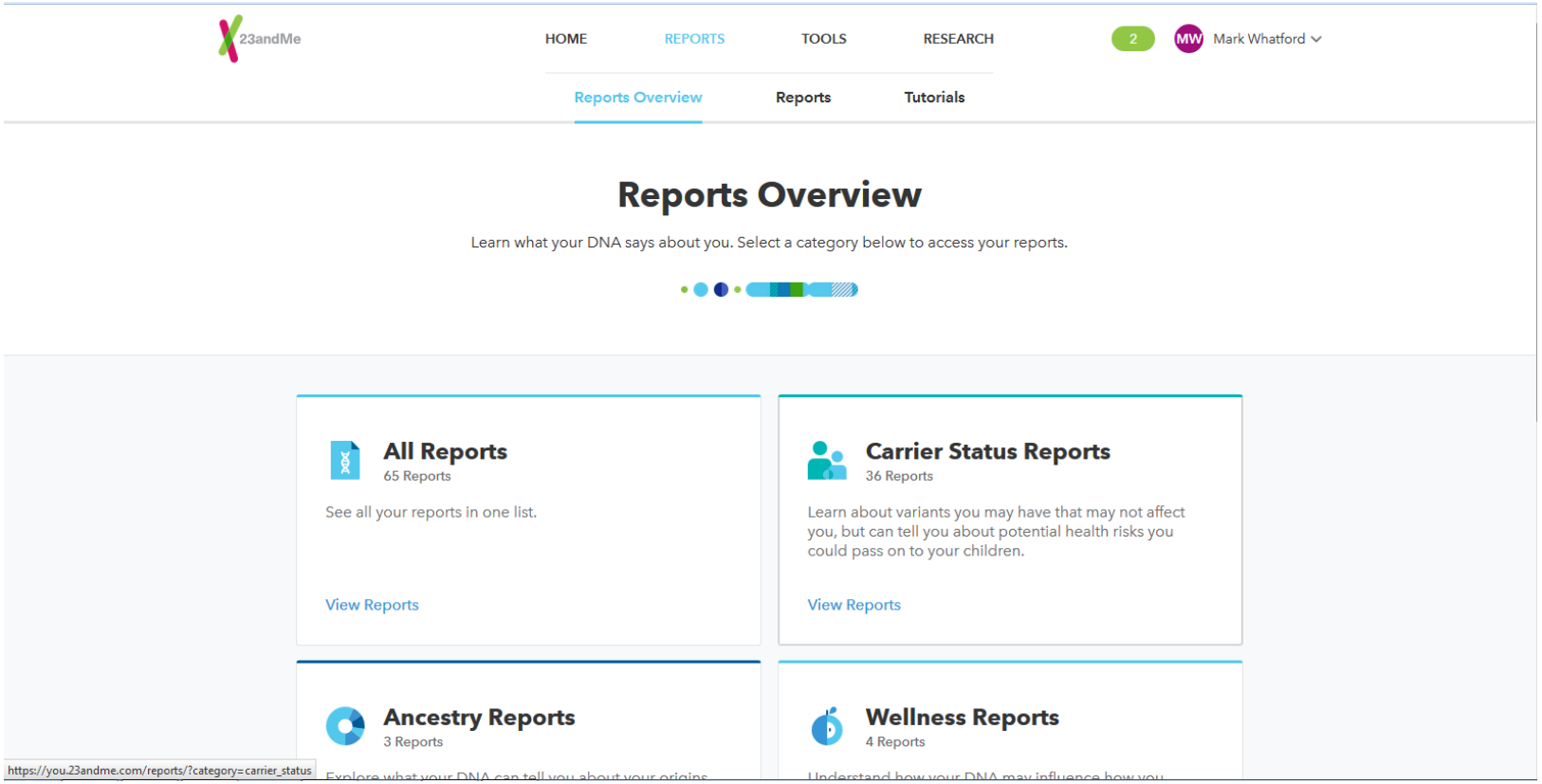
Mark Whatford's report page on 23andMe website.



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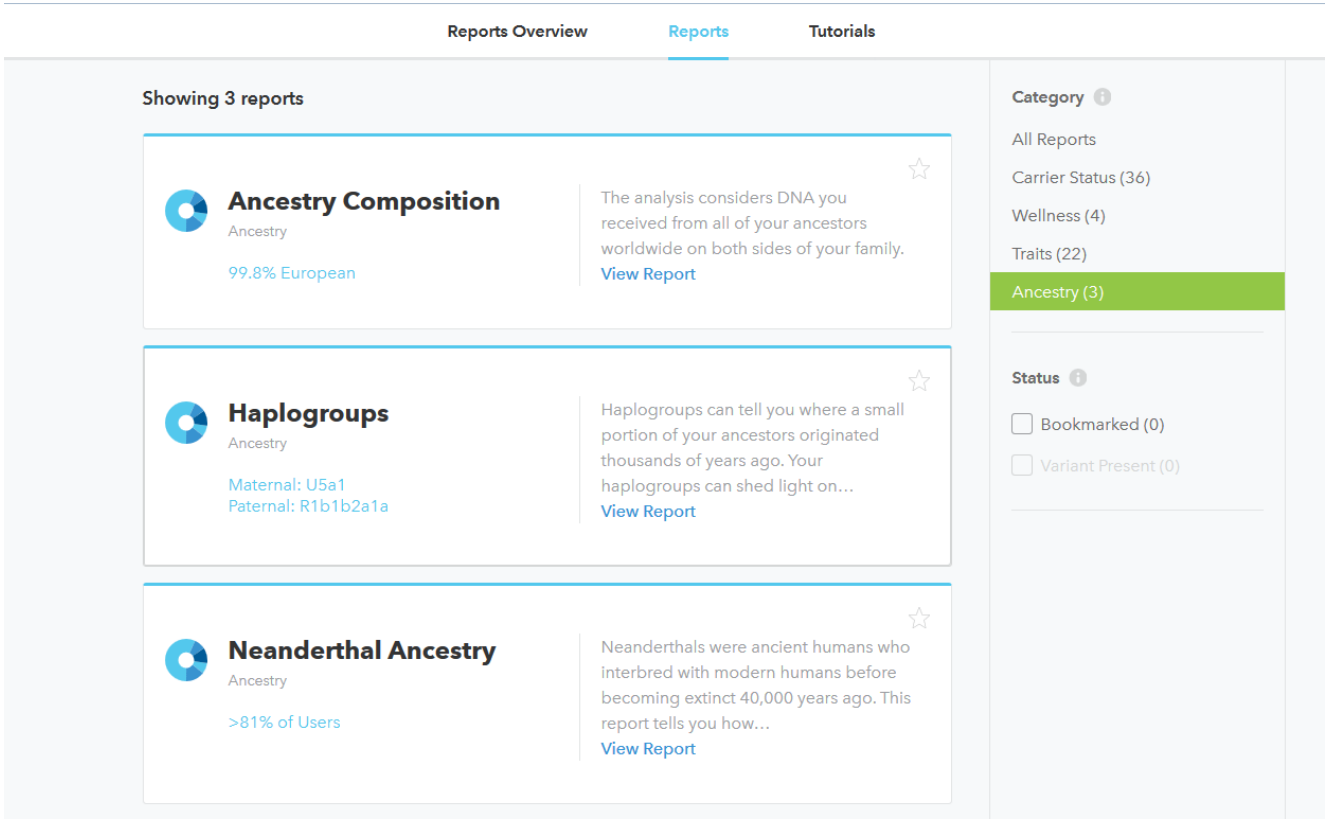
Ancestry Reports page of the 23andMe website



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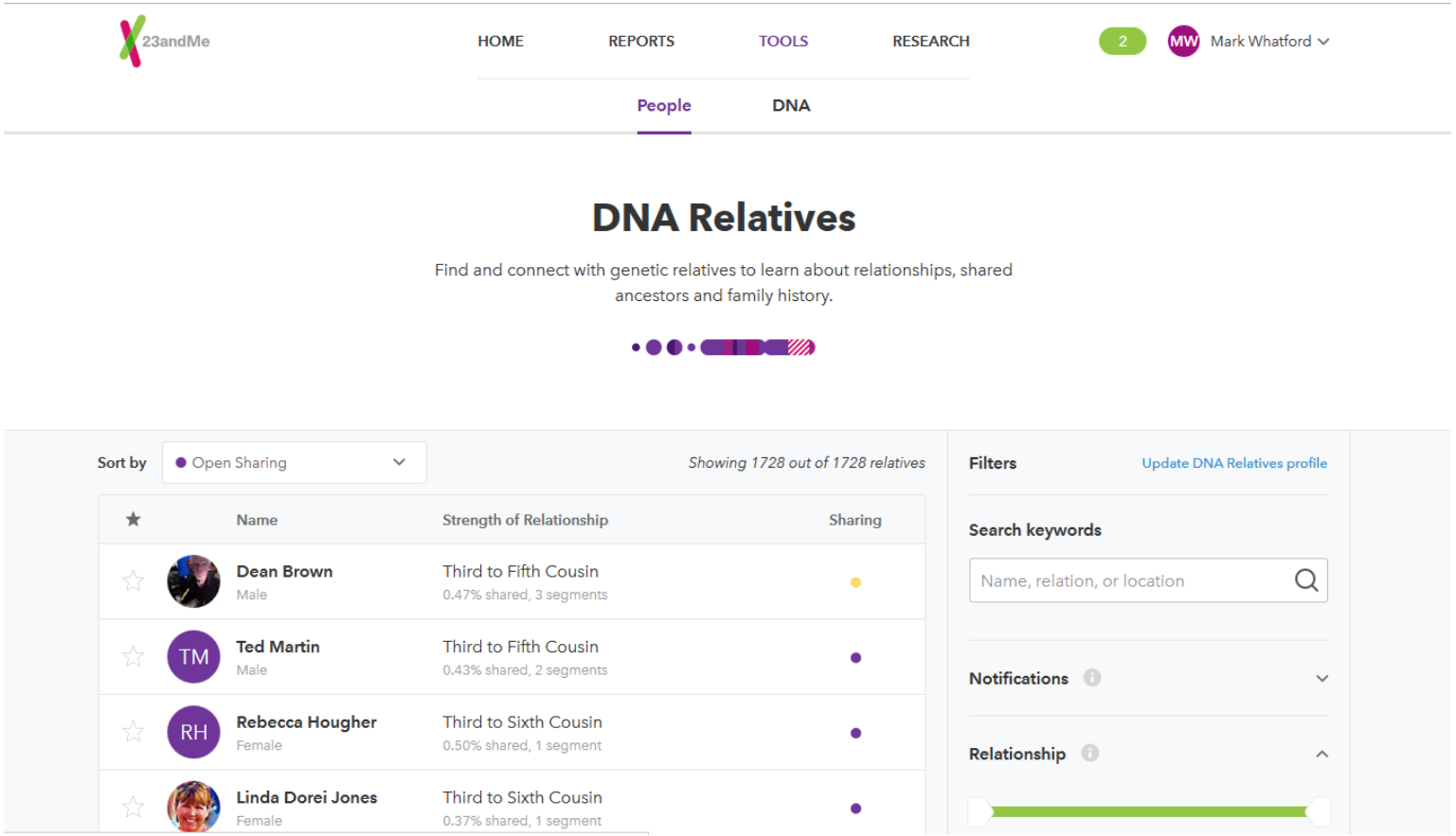
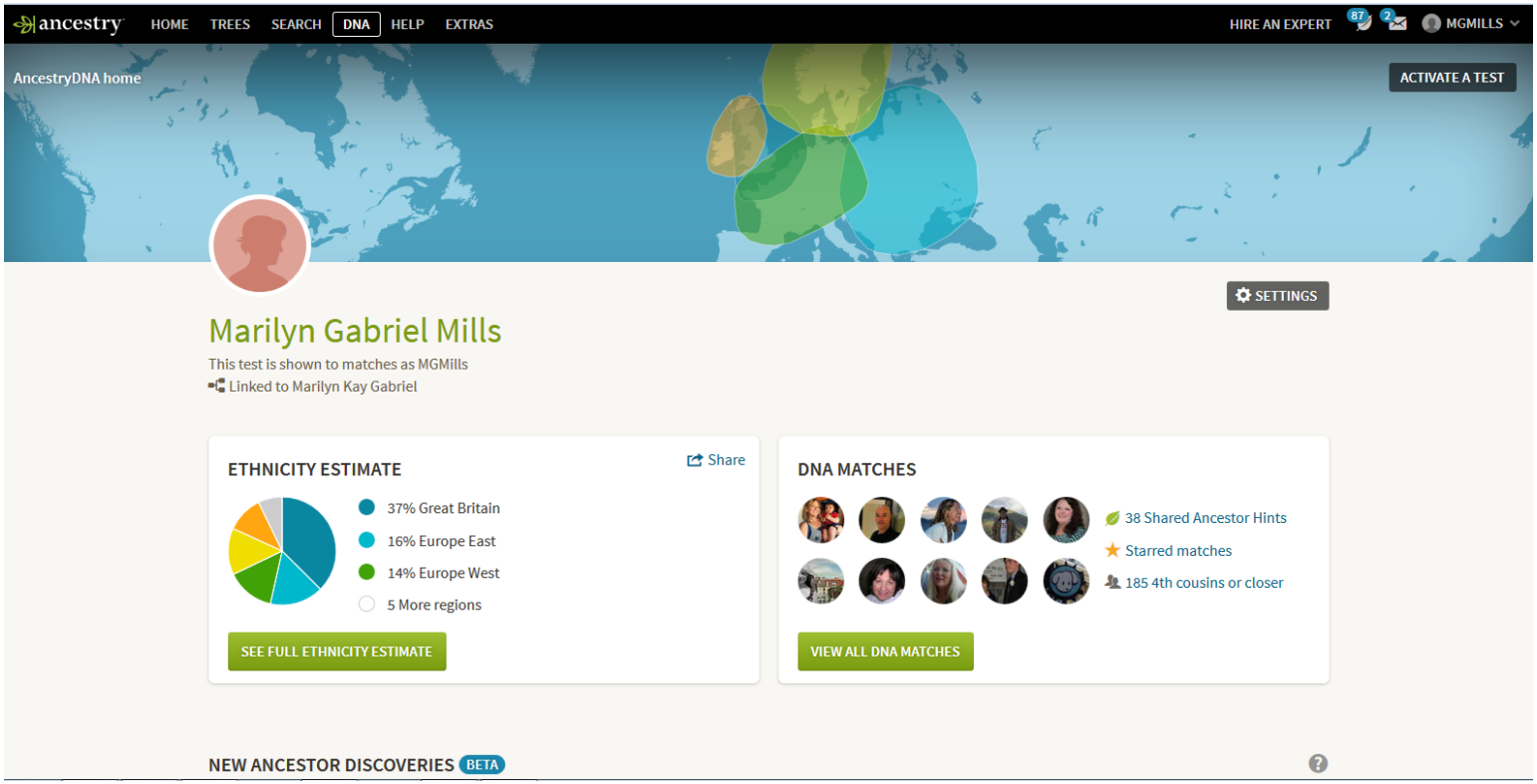
Test results page from 23andMe website for Mark Whatford

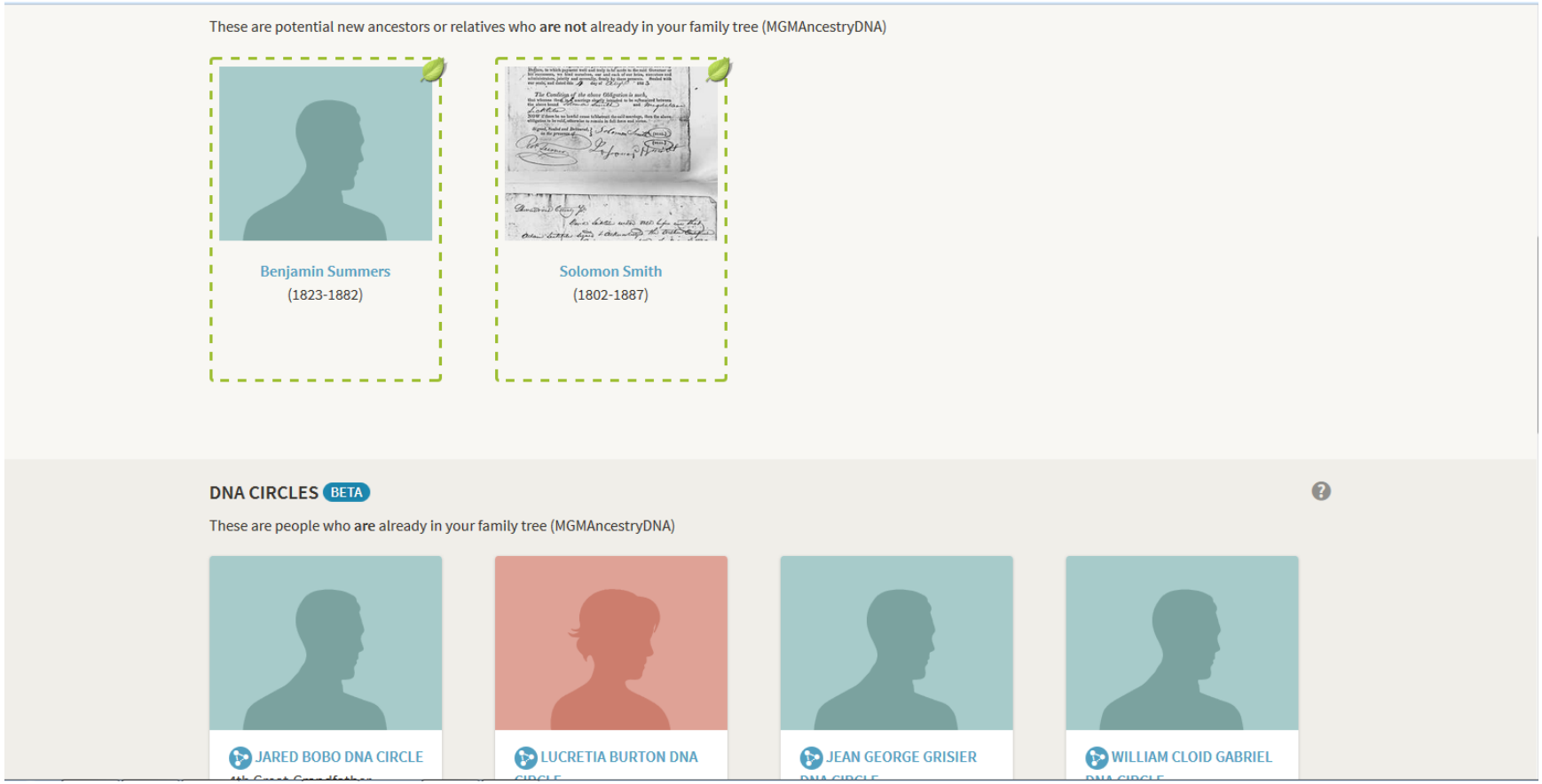


Genealogy 101—DNA: Expectations and Realizations

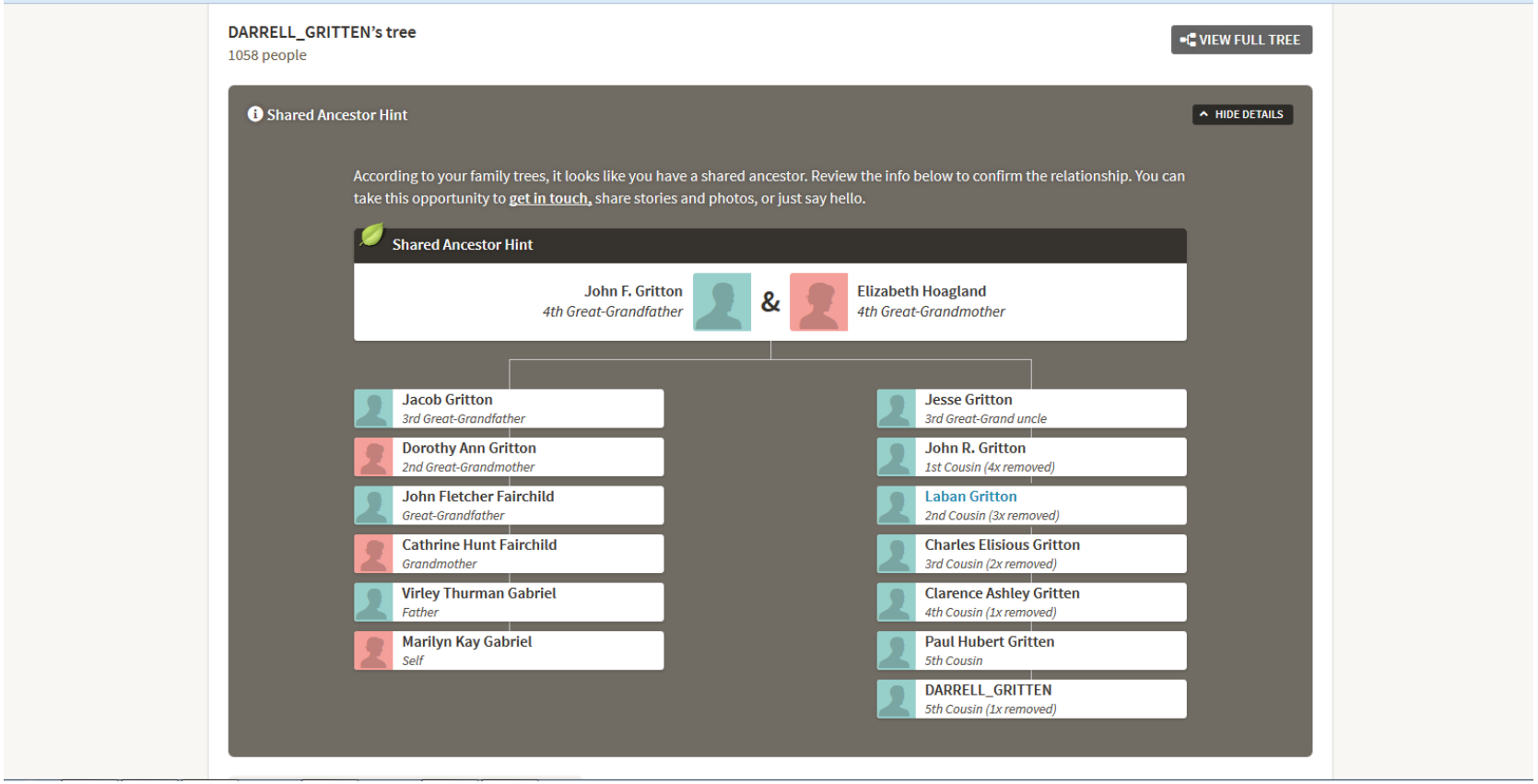
Marilyn Mill's homepage on Ancestry.com website



Marilyn Mill's homepage on Ancestry.com website. This information is found at the bottom of the previous page



Marilyn Mill's comparison with Darrell Gritten on Ancestry.com website



The people (We are all human)

- If you are searching for Great-grandpa Buck's parents the so are others.
- Adoptees.
- Immigrants—non American, non English.
- You may not correspond with the person who took the test. Some don't respond. Others respond nastily.
- Are the family genealogies sufficient?

The people (part 2) My “rules of thumbs”

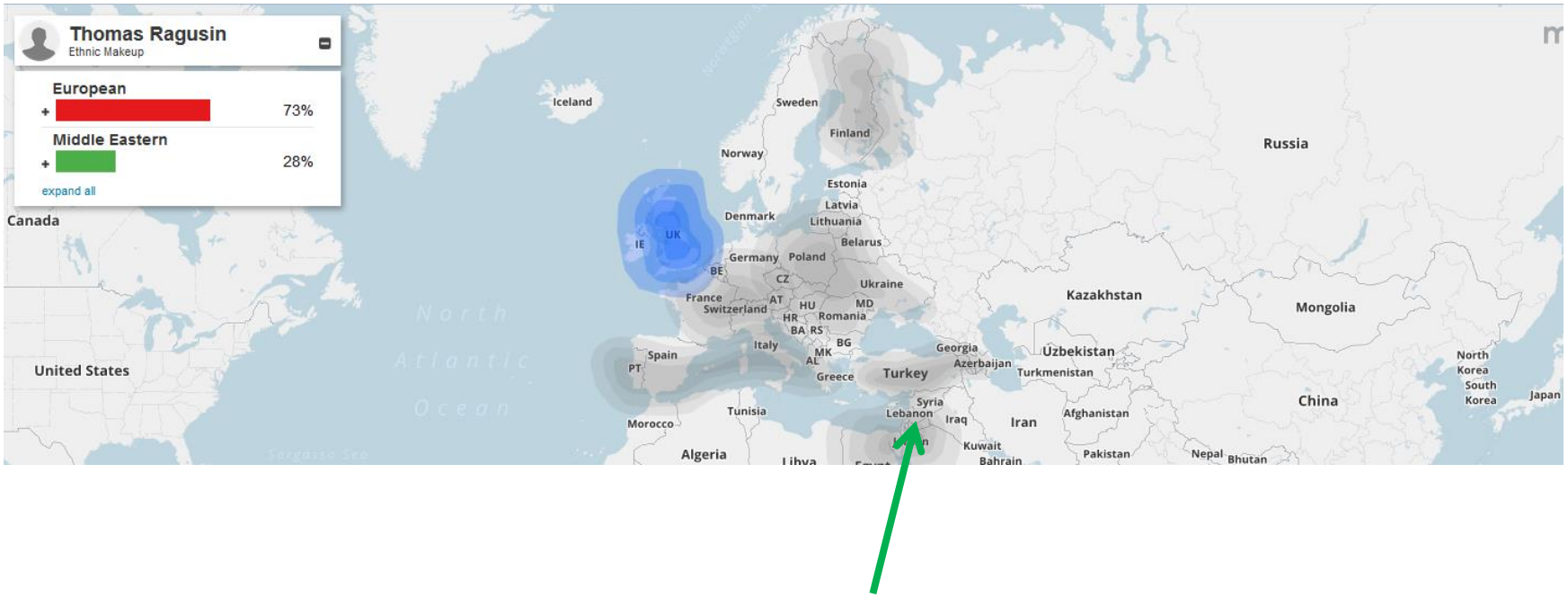
- I do not address them by the given name, but use Mr. or Mrs. Then I see how they sign the response.
- I sign my email as Tom.
- I introduce myself. I give my kit number, and what DNA test I talking about.
- I give them something. “Based on your surname list/family tree the enclosure is my lineage back to XX who married YY. Are these the same people from your lineage? If so, then this would make us nth cousins.”

The Final Story

- Three of my four grandparents were immigrants
- There lineages in Europe are based on Roman Catholic Church records.
- My sole American born grandparent is descended from every form of Protestantism that immigrated to the American colonies.
- Unspoken expectation—my ancestors were Christians.

Genealogy 101—DNA: Expectations and Realizations

My ethnic makeup: I am 28 % Middle Eastern originally labelled as Jewish



What do I do now?

- Got a yamulke.
- Merchants of Venice were Jewish
- Rumors about the Steinmans
- The Republic of Ragusa was a haven for Jews in the Middle Ages
- Spanish Jews were driven into southern Italy and Ragusa during the Inquisition
- Research of Rabbi Barbara Aiello

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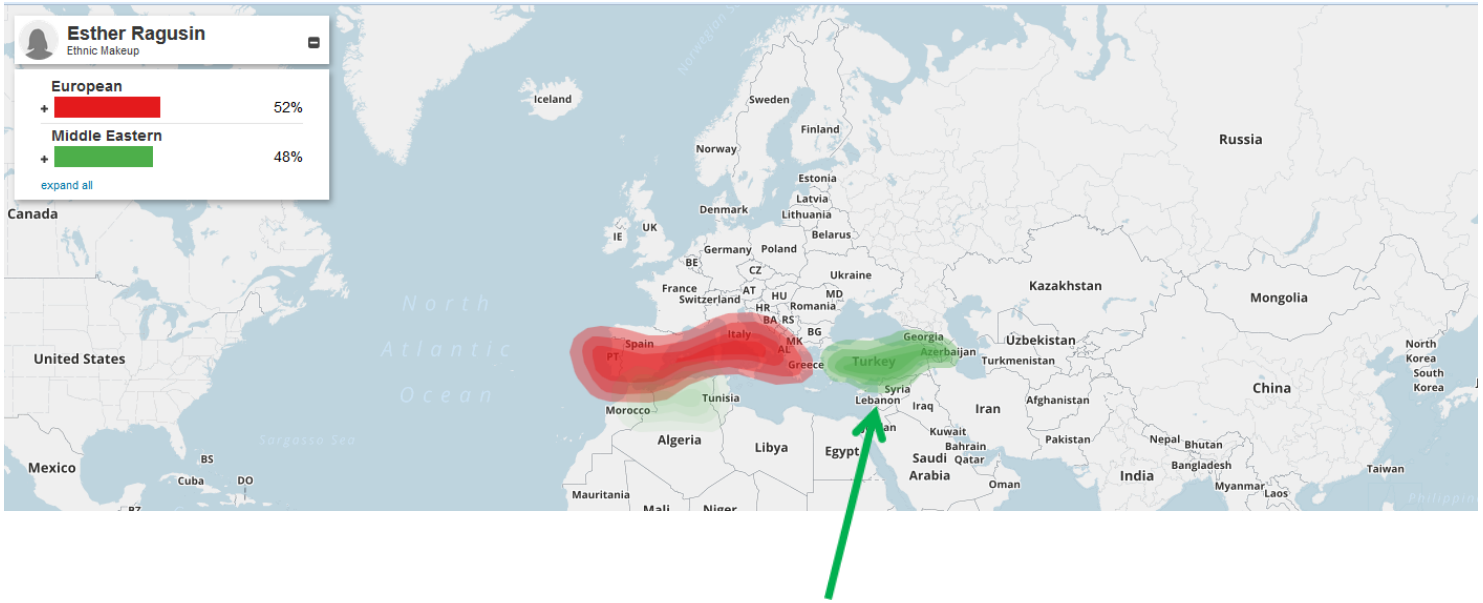
My mother's ethnic makeup: Her approximately 50 % fully explains the approximately 25 % "Jewishness" of myself and my siblings



Rabbi Barbara's website describing records from the Inquisition identifying Calabrian Jews who were forced to convert

rabbi barbara
Rabbi Barbara Aiello

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ITALIAN JEWISH ROOTS AND SURNAME RESEARCH

The Italian Jewish Cultural Center of Calabria (IjCCC) is an international organization based in Calabria, the southernmost region of Italy. Founded by Rabbi Barbara Aiello, whose Jewish ancestry includes B'nai Anusim (Italians whose ancestors were forced into adult baptism and Christian conversion during Inquisition times), is the founding director of the IjCCC, an organization dedicated to help those with Italian heritage determine if their family surnames indicate Italian Jewish roots. Although the IjCCC does not establish a blood line, our staff initiates a search of Italian family surnames to determine a Jewish connection.

Staff combs through ancient Inquisition records, searching for matches between family surnames and Jewish religious persecution. For example, in Rabbi Barbara's family's the surname "GRANDE" is prominent. Research determined that families named GRANDE, coming from the same towns and villages as Rabbi Aiello's family members, were arrested and tortured for "judaizing," practicing Judaism in secret.

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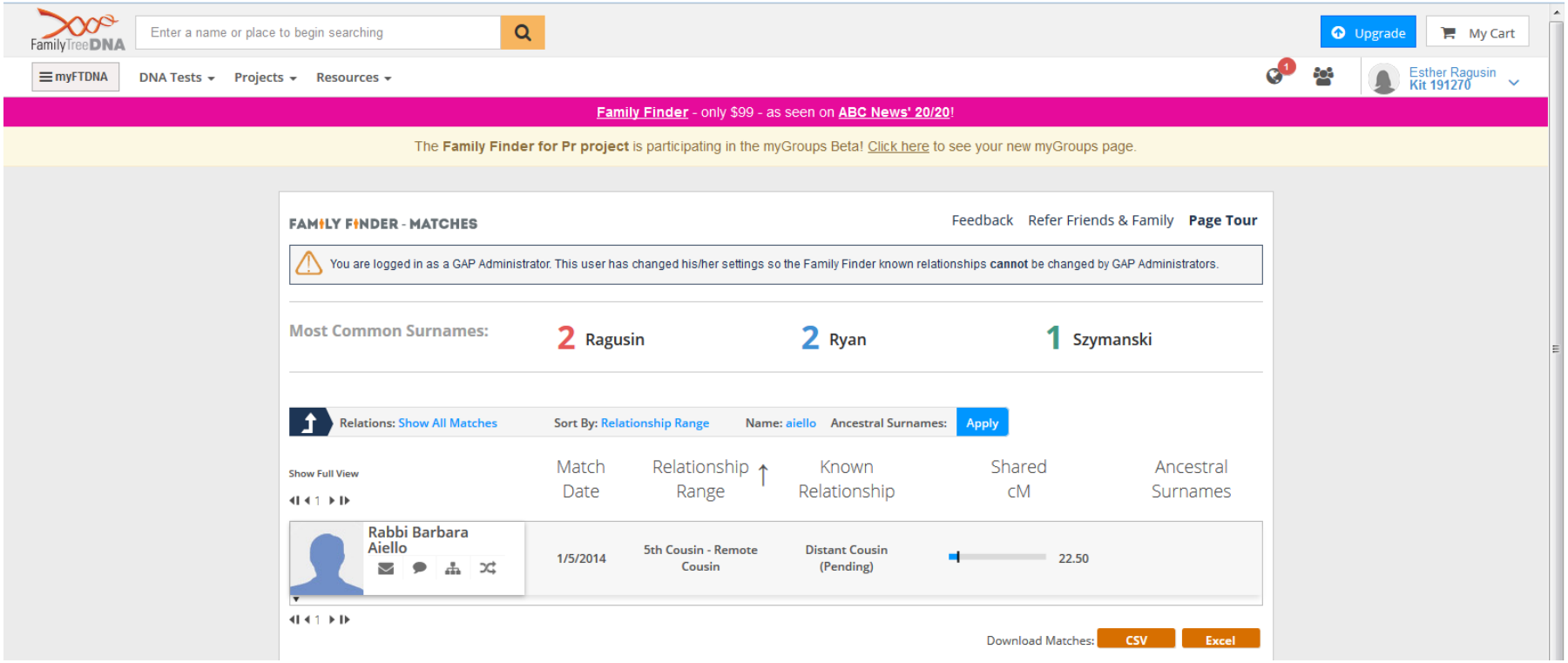
The punch line: My mother matches Rabbi Barbara



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