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)
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 if 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.
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

\[ 0.510\ 998\ 928 \pm 11\ \text{MeV} \]

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 possibilities.
- 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.

<table>
<thead>
<tr>
<th>Ball #1</th>
<th>Ball #2</th>
<th>Ball #3</th>
<th>Ball #4</th>
<th>Ball #6</th>
<th>Ball #7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tom's Lottery Picks</td>
<td>11</td>
<td>13</td>
<td>15</td>
<td>16</td>
<td>25</td>
</tr>
</tbody>
</table>

Playing the lottery (part 4)—kind of

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

Genealogy 101—DNA: Expectations and Realizations
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

```
<table>
<thead>
<tr>
<th></th>
<th>DYS391</th>
<th>DYS393</th>
<th>DYS385b</th>
<th>DYS19</th>
<th>DYS390</th>
<th>DYS426</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tom’s—yDNA</td>
<td>11</td>
<td>13</td>
<td>15</td>
<td>16</td>
<td>25</td>
<td>12</td>
</tr>
</tbody>
</table>
```
My yDNA results as reported in a project on the Family Tree DNA website.

<table>
<thead>
<tr>
<th>Row Number</th>
<th>Kit Number</th>
<th>Name</th>
<th>Paternal Ancestor Name</th>
<th>Country</th>
<th>Haplogroup</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>107637</td>
<td>England</td>
<td></td>
<td></td>
<td>E-M183</td>
</tr>
<tr>
<td>2</td>
<td>249825</td>
<td>Poland</td>
<td></td>
<td></td>
<td>I-554602</td>
</tr>
<tr>
<td>3</td>
<td>297473</td>
<td>England</td>
<td></td>
<td></td>
<td>H-M353</td>
</tr>
<tr>
<td>4</td>
<td>03579</td>
<td>Unknown Origin</td>
<td></td>
<td></td>
<td>J-Z3559</td>
</tr>
<tr>
<td>5</td>
<td>224491</td>
<td>Pedro Alfonso Sosa Gavirio</td>
<td>Indaliesco Sosa</td>
<td>Colombia</td>
<td>Q-L472</td>
</tr>
<tr>
<td>6</td>
<td>242864</td>
<td>Greece</td>
<td></td>
<td></td>
<td>R-M612</td>
</tr>
<tr>
<td>7</td>
<td>229576</td>
<td>Unknown Origin</td>
<td></td>
<td></td>
<td>R-CT511902</td>
</tr>
<tr>
<td>8</td>
<td>179780</td>
<td>Thomas John Ragaslin</td>
<td></td>
<td>Croatia</td>
<td>R-Z262</td>
</tr>
</tbody>
</table>
My two “closest” yDNA matches

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

<table>
<thead>
<tr>
<th>Genetic Distance</th>
<th>Name</th>
<th>Most Distant Ancestor</th>
<th>Y-DNA Haplogroup</th>
<th>Terminal SNP</th>
<th>Match Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>7</td>
<td>Mr. Daniel Durdov</td>
<td>[Icon] Y-DNA67</td>
<td>R-M512</td>
<td></td>
<td>5/19/2010</td>
</tr>
<tr>
<td>7</td>
<td>Mr. Miron Berezik</td>
<td>[Icon] Antonio Berezik, b.c.1785, Lesko, Poland</td>
<td>R-M512</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
The closeness of the match

- Generations do not exist!

- Think of years instead with 25 years to a generation

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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...

<table>
<thead>
<tr>
<th>Generations</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>4</td>
<td>0.18%</td>
</tr>
<tr>
<td>8</td>
<td>5.88%</td>
</tr>
<tr>
<td>12</td>
<td>26.45%</td>
</tr>
<tr>
<td>16</td>
<td>54.56%</td>
</tr>
<tr>
<td>20</td>
<td>77.16%</td>
</tr>
<tr>
<td>24</td>
<td>90.25%</td>
</tr>
</tbody>
</table>

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).
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 Dur dov and Thomas John Ragusi n shared a common ancestor within the last...

<table>
<thead>
<tr>
<th>Generations</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>8</td>
<td>2.73%</td>
</tr>
<tr>
<td>12</td>
<td>23.99%</td>
</tr>
<tr>
<td>16</td>
<td>53.04%</td>
</tr>
<tr>
<td>20</td>
<td>76.39%</td>
</tr>
<tr>
<td>24</td>
<td>89.92%</td>
</tr>
</tbody>
</table>

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 Dur dov and Thomas John Ragusi n did not share a common ancestor in the last 8 generation(s).
My mtDNA results

• Compared to a reference sequence, list of differences

• Formerly CRS, now RSRS
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
My Family Tree DNA homepage—the “dashboard”
Mark Whatford’s report page on 23andMe website.
Ancestry Reports page of the 23andMe website

- **Ancestry Composition**: The analysis considers DNA you received from all of your ancestors worldwide on both sides of your family. View Report
- **Haplogroups**: Haplogroups can tell you where a small portion of your ancestors originated thousands of years ago. Your haplogroups can shed light on... View Report
- **Neanderthal Ancestry**: Neanderthals were ancient humans who interbred with modern humans before becoming extinct 40,000 years ago. This report tells you how... View Report
DNA Relatives

Find and connect with genetic relatives to learn about relationships, shared ancestors and family history.

<table>
<thead>
<tr>
<th>Name</th>
<th>Strength of Relationship</th>
<th>Sharing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dean Brown</td>
<td>Third to Fifth Cousin</td>
<td></td>
</tr>
<tr>
<td>Ted Martin</td>
<td>Third to Fifth Cousin</td>
<td></td>
</tr>
<tr>
<td>Rebecca Hougher</td>
<td>Third to Sixth Cousin</td>
<td></td>
</tr>
<tr>
<td>Linda Dorei Jones</td>
<td>Third to Sixth Cousin</td>
<td></td>
</tr>
</tbody>
</table>

Showing 1728 out of 1728 relatives
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.
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
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
The punch line: My mother matches Rabbi Barbara
Genealogy 101—DNA: Expectations and Realizations

tjr 2/2016