Navigating the DNA Marketplace

Bryant McAllister, PhD
Associate Professor of Biology
University of Iowa
bryant-mcallister@uiowa.edu

The Triangle Club
February 18, 2018
The Personal Genome

- **Recreational Genetics** – analyses of individual DNA sequence data to inform geographic ancestry, biological relationships and physical/behavioral traits through direct-to-consumer (DTC) genetic tests.

- **Personalized Medicine** – analyses of individual DNA sequence data to diagnose genetic causation of a medical condition and/or tailor medical treatment under guidance of a physician and supported by other medical services.
AncestryDNA can reveal the source of your greatness. Discover your ethnic origins—and get inspired by the places in your past.

ONLY $69* $99

Comparison based on prevailing price on 7/12/2017

BUY NOW

The AncestryDNA Team Pack
The more you order, the more you can discover. Buy now
Growth in DTC Genetic Testing

Cumulative Customers

Calendar Years

Jan-07   Jan-08   Jan-09   Jan-10   Jan-11   Jan-12   Jan-13   Jan-14   Jan-15   Jan-16   Jan-17   Jan-18   Jan-19

23andMe Press

AncestryDNA Press
DNA Interest Group

Meetings: 4th Tuesday of each month, 6-7 pm
Iowa City Public Library, Room A

• February 27 – Race & Ancestry
• March 27 – Why the Y Chromosome
• April 24 – National DNA Day
• May 23 – Pharmacogenetics
Personal Genome Learning Center

Mission: foster knowledge of genetic and evolutionary principles through engagement with personal genomics

- Engage students in the application of genetic and evolutionary principles through the exploration of personal genome data.
- Involve students in public outreach events and workshops that support understanding of personal genome results and applications.
- Develop resources that support learning genetic and evolutionary principles relevant to understanding personal genome data.
The Family Finder Experience
World’s most comprehensive database

- Autosomal test that finds your DNA matches within five generations.
- Includes myOrigins, a mapping tool that provides a detailed ethnic and geographic breakdown of where your ancestors came from.
- New parental matching feature enables users to sort results by parental and maternal matches.

Order Now  ONLY $79 USD

Father’s Line DNA  FAMILY FINDER
SHOP NOW  
Starting at $248

Family Finder  FAMILY FINDER
SHOP NOW  
Only $79

mtFull Sequence DNA  FAMILY FINDER
SHOP NOW  
Only $278
Genome-wide SNP Analysis: Major Testing Options

- **23andMe** – Report revised in 2015. Includes Ancestry Estimates & DNA Matching ($99 list); Carrier Status & Trait Prediction reports also available ($199 list).

- **AncestryDNA** – Includes Ancestry Estimate and DNA Matching ($99 list). Integrated with Ancestry.com family tree for an extra subscription fee.

- **MyHeritageDNA** – Includes Ancestry Estimate and DNA Matching ($99 list). DNA matches on same platform as tree-building infrastructure.

- **Family Tree DNA** – Various tests available including Family Finder SNP test ($79 list).
I AM
1 OF 705,343
PARTICIPANTS

<table>
<thead>
<tr>
<th>Northern European</th>
<th>Mediterranean</th>
<th>Southwest Asian</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>British (United)</strong></td>
<td>49%</td>
<td>33%</td>
</tr>
<tr>
<td><strong>You</strong></td>
<td>43%</td>
<td>37%</td>
</tr>
<tr>
<td><strong>Native American</strong></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
GENOME-WIDE SNP ANALYSIS

Chromosomes, DNA, and your unique genetic profile
DNA is the Cellular Recipe for Life

- **Genome** is the entire set of DNA molecules present in a cell.
- Each DNA molecule contains a string of A=T pairs and G≡C pairs as a DNA sequence.
- Cells of an individual contain the same genome sequence, but different from any other individual.
Chromosomes:
23 pairs of linear DNA molecules inherited from both parents; 22 pairs of autosomes, and a single pair of sex chromosomes

Gametes (egg or sperm) contain only 1 member of each pair
The Human Genome

Chromosomes:
23 pairs of linear DNA molecules inherited from both parents; 22 pairs of autosomes, and a single pair of sex chromosomes

Gametes (egg or sperm) contain only 1 member of each pair
SNP Analysis
Single Nucleotide Polymorphism

My mum’s hair colour is grey.
My mom’s hair color is gray.
SNP Analysis
Single Nucleotide Polymorphism

My mom’s hair color is gray.
My mom’s hair color was gray.
Genome-Wide SNP Analysis

Each company uses a common testing technology that assesses about 600,000 different known variable sites (SNPs) in the DNA of your genome (including mtDNA in some cases). Most of the same sites are used in the tests of different companies.
# Genome-Wide SNP Analysis

<table>
<thead>
<tr>
<th>rsid</th>
<th>chromosome</th>
<th>position</th>
<th>genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs12564807</td>
<td>1</td>
<td>734462</td>
<td>AA</td>
</tr>
<tr>
<td>rs3131972</td>
<td>1</td>
<td>752721</td>
<td>AG</td>
</tr>
<tr>
<td>rs148828841</td>
<td>1</td>
<td>760998</td>
<td>CC</td>
</tr>
<tr>
<td>rs12124819</td>
<td>1</td>
<td>776546</td>
<td>AA</td>
</tr>
<tr>
<td>rs115093905</td>
<td>1</td>
<td>787173</td>
<td>GG</td>
</tr>
<tr>
<td>rs11240777</td>
<td>1</td>
<td>798959</td>
<td>AG</td>
</tr>
<tr>
<td>rs7538305</td>
<td>1</td>
<td>824398</td>
<td>AC</td>
</tr>
<tr>
<td>rs4475691</td>
<td>1</td>
<td>846808</td>
<td>CT</td>
</tr>
<tr>
<td>rs7537756</td>
<td>1</td>
<td>854250</td>
<td>AG</td>
</tr>
<tr>
<td>rs13302982</td>
<td>1</td>
<td>861808</td>
<td>GG</td>
</tr>
<tr>
<td>rs55678698</td>
<td>1</td>
<td>864490</td>
<td>CC</td>
</tr>
<tr>
<td>rs1110052</td>
<td>1</td>
<td>873558</td>
<td>TT</td>
</tr>
<tr>
<td>rs147226614</td>
<td>1</td>
<td>878697</td>
<td>GG</td>
</tr>
<tr>
<td>i6052728</td>
<td>1</td>
<td>878697</td>
<td>GG</td>
</tr>
<tr>
<td>i6019302</td>
<td>1</td>
<td>881843</td>
<td>GG</td>
</tr>
<tr>
<td>rs2272756</td>
<td>1</td>
<td>882033</td>
<td>GG</td>
</tr>
</tbody>
</table>
ANCESTRY COMPOSITION

Predicting the geographic origins of your ancestors
Prediction of Ancestry Composition

Given an individual with this genotype, what geographic areas is the person likely from?

chr1:752721 A/G

http://popgen.uchicago.edu/ggv/
Bryant McAllister 100.0%

Europe 98.3%
- North and West Europe 98.3%
  - English 85.6%
  - Irish, Scottish, and Welsh 10.9%
  - Finnish 1.8%

America 1.7%
- Central America 1.7%

Europe 99.2%
- Sub-Saharan African 0.7%
- Middle Eastern & North African < 0.1%
- East Asian & Native American < 0.1%

Northern European 43%
Mediterranean 37%
Southwest Asian 17%
Native American 2%
DNA Story for Bryant McAllister

Ethnicity Estimate

- Great Britain: 49%
- Ireland/Scotland/Wales: 34%
- Europe West: 8%
- Scandinavia: 5%

Low Confidence Regions

Migrations

- **Lower Midwest & Virginia Settlers**
  From your regions: Great Britain, Ireland/Scotland/Wales...

- **Mississippi & Louisiana Settlers**
  From your regions: Great Britain, Ireland/Scotland/Wales...

- **Southern States Settlers**
  From your regions: Great Britain, Ireland/Scotland/Wales...

See all 150+ regions
Uni-Parental Inheritance

Maternal Transmission of mtDNA

Paternal Transmission of the Y chromosome
Uni-Parental Haplogroups

Haplogroup H Migration

**Origin:** H originated in the Near East, but expanded into isolated pockets of Europe. After the Ice Age, this haplogroup spread across Europe where it is the most prevalent haplogroup today. It is present in about half of the Scandinavian population and is also common along the continent's Atlantic coast.

**Highlight:** Mitochondrial DNA extracted from the remains of St. Luke belong to haplogroup H.

**Example Populations:** Basques, Scandinavians

Haplogroup R-M269 Migration

**Origin:** R-M269 is the most common haplogroup in western Europe, where its branches are clustered in various national populations, including in the Basque, in Ireland, and on the fringes of the North Sea.

**Highlight:** R-M269 is found in more than 50% of men in western Europe.

**Example Populations:** Irish, Basques, British, French
Haplogroup R (also known as R-M207) is the most recent common ancestor of all R lineages, including your haplogroup.
Y-DNA Ancestry Tests
Discover your heritage on your father's line

Connect on your direct father's line
Uncover your paternal heritage

only $169
Add to cart
Males only

Trace the history of your surname.
Join FREE surname research projects.
Find your genetic matches in the world's largest Y-DNA database.
DNA RELATIVES
Revealing common ancestry through DNA matching
Segment Matching Between Relatives

identical segment
### Matching of SNPs

<table>
<thead>
<tr>
<th>rsid</th>
<th>chromosome</th>
<th>position</th>
<th>Tester</th>
<th>Match</th>
<th>Half-Identical: at least one common variant at all SNPs in this segment</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs12564807</td>
<td>1</td>
<td>734462</td>
<td>AA</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>rs3131972</td>
<td>1</td>
<td>752721</td>
<td>GG</td>
<td>AG</td>
<td></td>
</tr>
<tr>
<td>rs148828841</td>
<td>1</td>
<td>760998</td>
<td>CC</td>
<td>CC</td>
<td></td>
</tr>
<tr>
<td>rs12124819</td>
<td>1</td>
<td>776546</td>
<td>AA</td>
<td>AA</td>
<td></td>
</tr>
<tr>
<td>rs115093905</td>
<td>1</td>
<td>787173</td>
<td>GG</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>rs11240777</td>
<td>1</td>
<td>798959</td>
<td>GG</td>
<td>AG</td>
<td></td>
</tr>
<tr>
<td>rs7538305</td>
<td>1</td>
<td>824398</td>
<td>AA</td>
<td>AC</td>
<td></td>
</tr>
<tr>
<td>rs4475691</td>
<td>1</td>
<td>846808</td>
<td>TT</td>
<td>CT</td>
<td></td>
</tr>
<tr>
<td>rs7537756</td>
<td>1</td>
<td>854250</td>
<td>GG</td>
<td>AG</td>
<td></td>
</tr>
<tr>
<td>rs13302982</td>
<td>1</td>
<td>861808</td>
<td>GG</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>rs55678698</td>
<td>1</td>
<td>864490</td>
<td>CC</td>
<td>CT</td>
<td></td>
</tr>
<tr>
<td>rs1110052</td>
<td>1</td>
<td>873558</td>
<td>GG</td>
<td>TT</td>
<td></td>
</tr>
<tr>
<td>rs147226614</td>
<td>1</td>
<td>878697</td>
<td>GG</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>i6052728</td>
<td>1</td>
<td>878697</td>
<td>GG</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>i6019302</td>
<td>1</td>
<td>881843</td>
<td>GG</td>
<td>GG</td>
<td></td>
</tr>
<tr>
<td>rs2272756</td>
<td>1</td>
<td>882033</td>
<td>AA</td>
<td>GG</td>
<td></td>
</tr>
</tbody>
</table>
## AncestryDNA Results for Bryant McAllister

### Filters
- HINTS
- NEW
- STARRED
- MOTHER
- FATHER

### Sort by:
- Relationship
- Date

### PARENT/CHILD

<table>
<thead>
<tr>
<th>Name</th>
<th>Possible range</th>
<th>Confidence</th>
<th>Relationship</th>
</tr>
</thead>
<tbody>
<tr>
<td>Judith Anne Lane</td>
<td>Parent, Child - immediate family member</td>
<td>Extremely High</td>
<td>Judith Anne Lane is your Mother</td>
</tr>
<tr>
<td>Thomas Allen McAllister</td>
<td>Parent, Child - immediate family member</td>
<td>Extremely High</td>
<td>Thomas Allen McAllister is your Father</td>
</tr>
</tbody>
</table>

- Judith Anne Lane: 3557 people
- Thomas Allen McAllister: 1636 people
2nd Cousin

Possible range: 1st - 2nd cousins
Confidence: Extremely High

3rd Cousin

Possible range: 3rd - 4th cousins
Confidence: Extremely High

4th Cousin

Possible range: 4th - 6th cousins
Confidence: High

Distant Cousin

Possible range: 5th - 8th cousins
Confidence: Good
Predicted relationship: Distant Cousins
Possible range: 5th - 8th cousins (What does this mean?)
Confidence: Good

3,174 people

SHARED SURNAMES
Direct ancestor surnames that appear in both tree and Bryant McAllister’s tree
Johnston McAllister Smith
DNA Support of Common Ancestry

Mary?

b. abt 1789 Ireland

d. after 1860 Ohio

Thomas McAllister

b. 1818 Ireland
d. 1875 OH

Patrick McAllister

b. 1830 Ireland
d. 1880 Tennessee

Thomas F. McAllister

b. 1859 OH
d. 1907 LA

Peter McAllister

b. 1857 PA
d. 1932 OH

Mary Ann McAllister

b. 1849 PA
d. 1905 OH

Me!
Predicted relationship: Distant Cousins
Possible range: 5th - 8th cousins

Amount of Shared DNA:
19.7 centimorgans shared across 2 DNA segments

Ethnicity:
Regions: Ireland, Great Britain, Scandinavia

<table>
<thead>
<tr>
<th>Name</th>
<th>Strength of Relationship</th>
<th>Sharing</th>
</tr>
</thead>
<tbody>
<tr>
<td>RW</td>
<td>Fourth Cousin</td>
<td>Half IBD 25 cM</td>
</tr>
<tr>
<td>Female</td>
<td>0.33% shared, 2 segments</td>
<td>23andMe</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Name</th>
<th>Match Date</th>
<th>Relationship Range</th>
<th>Shared Centimorgans</th>
<th>Longest Block</th>
<th>Ancestral Surnames</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>03/08/2016</td>
<td>2nd Cousin - 4th Cousin</td>
<td>53</td>
<td>17</td>
<td>McAllister / Smith / Bason / Dyke / Downey / Houlihan / Hunter / Kane /</td>
</tr>
</tbody>
</table>
Expectation (4th cousins):
14 cM or 0.20% shared
Genetics for Genealogy

- Results vary dramatically based on individual ancestry and database composition – *test the oldest generation.*

### DNA Matches

- **426 Shared Ancestor Hints**
- **184 Starred matches**
- **1000+ 4th cousins or closer**

### American Colonial Ancestry

- **2 Shared Ancestor Hints**
- **2 Starred matches**
- **60 4th cousins or closer**

### 19th Century German Migrants to Texas
Genetics for Genealogy

- Results vary dramatically based on individual ancestry and database composition – *test the oldest generation.*
Genetics for Genealogy

- Results vary dramatically based on individual ancestry and database composition – *test the oldest generation*.
- Endogamy increases the background level of relatedness among individuals.
- Lists of DNA matches contain two types of errors:
  - The genetic identity shared with the relative was not inherited from your most common ancestor(s), but rather a more distant ancestor.
  - Relatives descended from a known common ancestor(s) do not appear in the list of DNA relatives.
- DNA relatives may be connected through multiple pathways of common ancestry.
Family Testing Strategy

~50% identical
Genetics for Genealogy: Major Testing Options

- **AncestryDNA** ($99 list) – Fully integrated with active Ancestry.com tree with powerful features; no ability to directly examine matching DNA segment(s)

- **MyHeritageDNA** ($99 list) – Detailed information on genetic basis of DNA matches; DNA matches exist on site for building family trees (accepts raw data from 23andMe or AncestryDNA)

- **Family Finder Test** at Family Tree DNA ($79 list) – Broad set of features to examine DNA matches; website navigation challenging and database relatively small (accepts raw data from 23andMe or AncestryDNA)

- **23andMe** ($199 or $99) – Good for broad set of interpretations, entertainment, & validating relationships; most users are private and not motivated by genealogy
TRAITS

Inference of personal characteristics influenced by your genetic profile
Chromosome pair 10
Carriers for a rare condition called Usher 1F have a variant in the PCDH15 gene.
View your Usher 1F report.
Traits Reports
Discover how certain genetic variants can influence your personal characteristics and attributes.

- Taste and Smell (3 Reports)
- Facial Features (6 Reports)
- Hair (6 Reports)
- Physical Characteristics (4 Reports)
- Physical Responses (1 Report)
- Skin (2 Reports)
Lactose Intolerance

Almost everyone is born with the ability to digest dairy products. But most people lose that ability as they age, becoming lactose intolerant.

Bryant, based on your genetics, you are not likely to be lactose intolerant.

Your genetic result suggests that you are able to eat or drink dairy without experiencing digestive problems.
Digestion and indigestion

People can blame most symptoms of lactose intolerance on the bacteria that live in our intestines. A healthy digestive system is home to trillions of bacteria, which help to digest the food we eat. But when a person who doesn’t produce lactase eats or drinks dairy products, intestinal bacteria will start digesting the lactose instead, producing gases that lead to bloating, abdominal pain, and flatulence. Undigested lactose can also lead to diarrhea.

Evolution in action

Your DNA determines whether you can produce lactase after childhood, a trait known as "lactase persistence." Research suggests that ancient humans were lactose intolerant, and different genetic variants associated with lactase persistence evolved at different times in different parts of the world. This report is based on a genetic variant associated with lactase persistence that evolved in Europe within the last 20,000 years.

Genetic result

<table>
<thead>
<tr>
<th>GG</th>
<th>Likely lactose intolerant</th>
</tr>
</thead>
<tbody>
<tr>
<td>AG</td>
<td>Likely not lactose intolerant</td>
</tr>
<tr>
<td><strong>AA</strong></td>
<td><strong>Likely not lactose intolerant</strong></td>
</tr>
</tbody>
</table>

See the percentage of customers with these results

This report does not diagnose any health conditions or provide medical advice. Consult with a healthcare professional before making any major lifestyle changes or if you have any other concerns about your results.
Bryant, you likely produce the lactase enzyme.

**Likely not lactose intolerant**

**Variants Detected**

0

**Marker Tested**

C/T-13910

**Gene:** Near LCT

**Marker:** rs4988235

**Your Genotype***

A

Typical copy from one of your parents

A

Typical copy from your other parent

**View All Tested Markers**

1

**Additional Information**

Ancestry_BFM.txt

rs4988235

1

<table>
<thead>
<tr>
<th>Marker</th>
<th>Genotype 1</th>
<th>Genotype 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs4988262</td>
<td>136604210</td>
<td>C</td>
</tr>
<tr>
<td>rs4988243</td>
<td>136607703</td>
<td>T</td>
</tr>
<tr>
<td>rs3213871</td>
<td>136615222</td>
<td>C</td>
</tr>
<tr>
<td>rs1827549</td>
<td>136608646</td>
<td>A</td>
</tr>
<tr>
<td>rs4988199</td>
<td>136618520</td>
<td>A</td>
</tr>
</tbody>
</table>
CONSIDERATIONS
“There is also a continuing need for educational initiatives that will allow consumers to understand what test results will mean for them in order to make informed decisions about whether to use such services.”
Questions About DNA Tests

- What outcome(s) do you want in the test results?
- What does the DNA test assay?
- What interpretations are delivered in the results?
- How would you use these results?
- What is the cost, and how does it compare with competitors?
- Are there additional fees associated with access or use of test results?
- Are other customers satisfied with the service?
- Are the terms of service acceptable to you?
Genetic testing brings families together
And sometimes tears them apart

by Julia Belluz on September 9, 2014
Potential Consequences of the Results from a Personal Genetic Test

• Surprising findings about family relationships
  • New relatives discovered by genetic matching
  • Discovery of genetic relationship between your parents
  • Different relationship than expected with other tested family members

• Surprising prediction of ancestry composition
  • Individuals contain a genetic mixture representative of their ancestry through human history

• Surprising discovery of genetic susceptibility to disease
  • You and/or your children may have an increased chance of developing an incurable genetic disease