GENETICS FOR ANCESTRY,
GENEALOGY,
AND MORE

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222/224 Biology Bldg (BB)
bryant-mcallister@uiowa.edu
**Course Schedule**

- **Week 1, Feb. 4**  DNA and the Human Genome
- **Week 2, Feb. 11**  Ancestry Analysis from DNA
- **Week 3, Feb. 18**  Connecting Cousins for Genetic Genealogy
- **Week 4, Feb. 25**  Recreational Genetics

https://wiki.uiowa.edu/display/2360159/G4G

Darwin Day Speakers:  Feb. 18-20
http://iowacitydarwinday.org/events.html
OBJECTIVES

• Navigate lists of DNA relatives and individual DNA matches
• Recognize the utility of clustering DNA relatives into family groups
• Apply genetic principles to predict patterns of DNA identity between biological relatives
Genetics for Genealogy: Major Testing Options

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

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

• **Family Finder Test** at Family Tree DNA ($99) – Broad set of features to examine DNA matches; website is difficult to navigate and database is small ($37 transfer of AncestryDNA data to Family Finder database)
Placement within Human Family Tree

✓ Shared sequence in DNA of maternally-inherited mitochondrion or male-limited Y chromosome
  • Placement on a branch of the human family tree
  • Revelation of closest relatives by fewest mismatches

✓ Genetic similarity with reference populations sampled throughout the globe
  • Prevalence of mtDNA and Ychr haplogroups
  • Estimate of compositional mixture from different ancestry groups

➢ Segments of base identity along the DNA of autosomes and X chromosome
  • Number and length of identical segments decreases with relationship distance
Genetic Genealogy

DNA-Based Inference of Common Ancestry

2\textsuperscript{nd} - 3\textsuperscript{rd} cousins

Historical Record of Ancestors

great great grandparents
great grandparent
great grandparent
parent
parent

DNA Ancestors
DNA Siblings
DNA Connections

2\textsuperscript{nd} cousin
1x removed
GENOME-WIDE SNP MATCHING
DNA matching using segments of identity in SNP data
DNA is the Cellular Recipe for Life

- A **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
My mum’s hair colour is grey.
My mom’s hair color is gray.
Each company uses a common testing technology that assesses about 700,000 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

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Matching of SNPs

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<td>1</td>
<td>882033</td>
<td>AA</td>
<td>GG</td>
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Half-Identical: at least one common variant at all SNPs in this segment.
DNA REALATIVES

Clustering DNA relatives through common relationships
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<th>Strength of Relationship</th>
<th>Sharing</th>
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<td>Second Cousin Once Removed</td>
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<td>Male</td>
<td>1.16% shared, 7 segments</td>
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<td>Male</td>
<td>0.49% shared, 2 segments</td>
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<td>3</td>
<td>name</td>
<td>Third to Fourth Cousin</td>
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<tr>
<td></td>
<td>Male</td>
<td>0.99% shared, 2 segments</td>
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</tr>
<tr>
<td>4</td>
<td>Anonymous</td>
<td>Third to Fourth Cousin</td>
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<tr>
<td></td>
<td>Male</td>
<td>0.82% shared, 3 segments</td>
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<tr>
<td>5</td>
<td>Anonymous</td>
<td>Third to Fourth Cousin</td>
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<td>Female</td>
<td>0.66% shared, 3 segments</td>
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<td>6</td>
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<td>Third to Fourth Cousin</td>
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<tr>
<td></td>
<td>Male</td>
<td>0.56% shared, 3 segments</td>
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### Most Common Surnames:

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<td>Williams</td>
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<td>Brown</td>
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### Relations:

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<th>Shared cM</th>
<th>Ancestral Surnames</th>
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<td>1/7/2016</td>
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<tr>
<td>2/13/2015</td>
<td>2nd Cousin - 4th Cousin</td>
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<td>117.29</td>
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<td>2nd Cousin 1R (Pending)</td>
<td>70.49</td>
<td>Lane / Neill / Alkire / Hunter / Ott / Stone</td>
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<tr>
<td>10/28/2014</td>
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<td></td>
<td>56.31</td>
<td>Browne / Parsons / Anderson / Barlow / Carlton...</td>
</tr>
</tbody>
</table>
name

@att.net

N/A
Y-DNA HAPLOGROUP

U5b2a1a1
mtDNA HAPLOGROUP

Most Distant Ancestors

Paternal:

Maternal:
Margaret Evaline Williamson

About Me
No information entered.

Ancestral Surnames
Alkire, Hunter, Lane, Neill, Ott, Stone
DNA Matches in AncestryDNA

**Filters**

**Search matches**

McAllister  

Include similar surnames

**2ND COUSIN**

Possible range: 2nd - 3rd cousins
Confidence: Extremely High
Last logged in Oct 24, 2015

1153 people

**2ND COUSIN**

Possible range: 2nd - 3rd cousins
Confidence: Extremely High
Last logged in Oct 7, 2015

107 people
Without an active Ancestry.com subscription

Discover how you’re connected.

Maximize your DNA results with the power of Ancestry.

Make meaningful connections to your past.

Expand your family history experience.

View other members' family trees and you could find out how you’re related.

Discover the fascinating facts of how your family story led to you.

Get hints along the way to find shared ancestors, grow your family tree, and start telling your family story.
With an active Ancestry.com subscription

**Predicted relationship: 3rd Cousins**

Possible range: 3rd - 4th cousins (What does this mean?)

Confidence: Extremely High

**Surnames (10 generation pedigree)**

- Acuff
- Brock
- Burnett
- Gatewood
- McAllister
- McDaniel
- McReynolds
- Wolfenbarger

**Ethnicity**

- Regions: Great Britain, Ireland, Europe East
- Trace Regions: Italy/Greece, Caucasus, Iberian Peninsula, Scandinavia, Asia South, European Jewish
DNA Matches in AncestryDNA

**2ND COUSIN**

Possible range: 2nd - 3rd cousins
Confidence: Extremely High
Last logged in Oct 24, 2015

Possible range: 2nd - 3rd cousins
Confidence: Extremely High
Last logged in Oct 7, 2015
Ancestor(s) Shared with DNA Match

- Thomas Francis McAllister
  - Grandfather
- Mary Alice Diggins
  - Grandmother

- George Peter (Sully) McAllister
  - Father
- Self

- Lottie Ellen McAllister
  - Aunt
  - 1st Cousin
  - 1st Cousin (1x removed)
  - 1st Cousin (2x removed)
DNA Matches in AncestryDNA

Thomas F McAllister
b. 1859 Ohio
d. 1907 Louisiana

Mary A Diggins
b. 1861 Kentucky
d. 1926 Louisiana

Filters
Search matches
McAllister
Search by birth location
SEARCH Cancel

Include similar surnames

2ND COUSIN
Possible range: 2nd - 3rd cousins
Confidence: Extremely High
Last logged in Oct 24, 2015

1153 people
VIEW MATCH

107 people
VIEW MATCH

Last logged in Oct 7, 2015
Thomas F McAllister  
- b. 1859 Ohio  
- d. 1907 Louisiana

Mary A Diggins  
- b. 1861 Kentucky  
- d. 1926 Louisiana

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

Shared matches with A

CLOSE FAMILY
- Possible range: Close family - 1st cousins  
  Confidence: Extremely High

1ST COUSIN
- (administered by bmcallis86)  
  Possible range: 1st - 2nd cousins  
  Confidence: Extremely High

2ND COUSIN
- Possible range: 2nd - 3rd cousins  
  Confidence: Extremely High

Last logged in Feb 13, 2016
DNA Circles

Mary Alice Diggins DNA Circle
Grandmother
(1861-1926)
5 Members

Thomas Francis McAllister DNA Circle
Grandfather
(1859-1907)
5 Members

McAllister Family Group
3 members
<table>
<thead>
<tr>
<th>Name</th>
<th>Match Date</th>
<th>Relationship Range</th>
<th>Known Relationship</th>
<th>Shared cM</th>
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<td>Relationship</td>
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<td>Shared cM</td>
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<td>Combs / Lane / Neill / Perkins / Berd / Biakely...</td>
</tr>
</tbody>
</table>
Sara Combs  
b. 1824 TN  
d. 1914 MO

William Lane  
b. 1820 TN  
d. 1852 AR

Matches D & E are likely also connected to this same family.
Mechanisms for Clustering Relatives

• “Shared Matches” tab in AncestryDNA
  • Shared matches limited to 4th cousin and closer to tester
  • Shared matches limited to 4th cousin and closer of 1° match
  • Sharing results of a DNA test bypasses this limit
• “DNA Circles” and “New Ancestor” Discoveries in AncestryDNA
  • DNA kit linked with shared family tree
  • Dependent on both DNA and Ancestor Information matches
• “In Common With” sorting in Family Finder (FTDNA)
  • Use “Family Finder Matrix” to compare multiple DNA relatives
• No mechanism for clustering match lists in 23andMe
  • Greatest limitation of 23andMe platform for genealogical research
Eric Mc
This test is shown to matches as E. M.

ETHNICITY ESTIMATE
- 43% Europe West
- 33% Ireland
- 15% Great Britain
- 3 Other regions

DNA MATCHES
- 0 Shared Ancestor Hints
- 0 Starred matches
- 391 4th cousins or closer

Sharing DNA results
These people have been invited to view the results of the DNA Test. To change the role assigned to each person, click the Edit button. You can also take away a person’s permission to view the test by clicking the “Remove” link.

<table>
<thead>
<tr>
<th>Invited Person</th>
<th>Role</th>
<th>Tasks</th>
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<tr>
<td>Eric Mc</td>
<td>Guest</td>
<td>Remove</td>
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</table>

EDIT ROLES INVITE OTHERS TO ACCESS DNA RESULTS
AUTOSOMAL INHERITANCE

Three fundamental principles govern autosomal transmission
Mendelian Principles

Segregation
Chromosome pairs separate and are transmitted individually and equally to gametes
Chromosomal Inheritance
Autosomal Inheritance
1:1 segregation each generation

Numbers of ancestors in each generation
32 16 8 4 2

Expected autosomal contribution of each ancestor
50% 25% 12.5% 6.25% 3.125%

~ expectation ~
Mendelian Principles

Segregation
Chromosome pairs separate and are transmitted individually and equally to gametes
Mendelian Principles

Segregation
Chromosome pairs separate and are transmitted individually and equally to gametes

Independent Assortment
Different pairs of chromosomes sort independently during gamete formation
Autosomal Inheritance

Recombination
Independent assortment
and exchange between
chromosome pairs forms
new genetic combinations
Patterns of Inheritance

Compare the genome of:
Margo Fisher (Grandma)

To the genome of:
Erin Mendel (Daughter)

Genome-Wide Comparison
Comparison across all of the genome data

Estimates
- Half-identical (1.3 Gb)
- Completely identical (0 Gb)
- Not identical
- Not enough information
- Genes/regions associated with the selected trait.
### SNP Matching

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**45.8 Mb half-Identical segment:** at least one common variant at all SNPs in this segment.
The Autosomal Genome

Chromosome Measurements

Physical Size – basepairs
3 Gbp ~ half the genome
6 Gbp ~ whole genome

Genetic Size – centimorgans
3,590 cM = all autosomes
Avg. 160 cM for each pair
The Autosomal Genome

Chromosome Measurements

Physical Size – basepairs
3 Gbp ~ half the genome
6 Gbp ~ whole genome

Genetic Size – centimorgans
3,590 cM = all autosomes
Avg. 160 cM for each pair
Predicted relationship: 4th Cousins

Possible range: 4th - 6th cousins (What does this mean?)

Confidence: Very High

Amount of Shared DNA
21.1 centimorgans shared across
3 DNA segments

What does this mean?

Name: RW
Strength of Relationship: Fourth Cousin
Sharing:

Female
0.33% shared, 2 segments

McAllister / Bason / Downey / Dyke / Houlihan...
AUTOSOMAL RELATEDNESS

Predicting segment identity based on the relationship
DNA Support of Common Ancestry

Patrick McAllister  
- b. 1830 Ireland  
  - d. 1880 Tennessee

Mary  
- b. abt 1789 Ireland  
  - d. after 1860 Ohio

Thomas McAllister  
- b. 1818 Ireland  
  - d. 1875 Ohio

Me!
Identity by Descent

Observation:
6.04% DNA shared across 10 segments
Predicted Relationship 2nd Cousin

\[ r = \frac{1}{16} \]
Expectation: 6.25% shared identity
Degree of Autosomal Relatedness

Probability of inheriting same segment from grandfather

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{16} \] or 6.25% shared identity by descent

\[ r = \frac{1}{2} \]

\[ \frac{1}{2} = r \]

\[ r = \frac{1}{2} \]

\[ \frac{1}{2} = r \]
Degree of Autosomal Relatedness

25% shared identity by descent

\[ r = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4} \]

[Diagram showing the calculation of relatedness]
Degree of Autosomal Relatedness

\[ r = \frac{1}{2} \times \frac{1}{2} = 1/4 \]

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = 1/8 \]
Degree of Autosomal Relatedness

\[ r = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4} \]

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8} \]
Degree of Autosomal Relatedness

25% shared identity by descent

\[ r = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4} \]

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8} \]

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8} \]

\[ r = \frac{1}{2} \times \frac{1}{2} + \frac{1}{4} = \frac{1}{4} \]
Expectations for the Inheritance of Identical Matching Autosomal Segments Between Relatives

<table>
<thead>
<tr>
<th>Relationship</th>
<th>Number</th>
<th>Avg. Length</th>
<th>Total Length</th>
<th>Genome Identical</th>
<th>p(none)*</th>
</tr>
</thead>
<tbody>
<tr>
<td>1st cousins</td>
<td>41.4</td>
<td>21.7 cM</td>
<td>897.5 cM</td>
<td>12.50%</td>
<td>0.00</td>
</tr>
<tr>
<td>2nd cousins</td>
<td>14.8</td>
<td>15.1 cM</td>
<td>224.4 cM</td>
<td>3.13%</td>
<td>0.00</td>
</tr>
<tr>
<td>3rd cousins</td>
<td>4.8</td>
<td>11.6 cM</td>
<td>56.1 cM</td>
<td>0.78%</td>
<td>0.01</td>
</tr>
<tr>
<td>4th cousins</td>
<td>1.5</td>
<td>9.4 cM</td>
<td>14.0 cM</td>
<td>0.20%</td>
<td>0.23</td>
</tr>
<tr>
<td>5th cousins</td>
<td>0.4</td>
<td>7.9 cM</td>
<td>3.5 cM</td>
<td>0.05%</td>
<td>0.64</td>
</tr>
</tbody>
</table>

**total autosomal length 3590 cM**

*without detection errors
AUTOSOMAL MATCHING

Common ancestry revealed by identical DNA segments
DNA Support of Common Ancestry

Patrick McAllister
b. 1830 Ireland
d. 1880 Tennessee

Thomas McAllister
b. 1818 Ireland
d. 1875 Ohio

Mary?
b. abt 1789 Ireland
d. after 1860 Ohio

Me!
Expectation
0.20% shared
Fourth Cousin
0.33% shared, 2 segments

Expectation
0.20% shared
<table>
<thead>
<tr>
<th>Matrix Matches</th>
<th>A</th>
<th>B</th>
<th>C</th>
<th>D</th>
<th>E</th>
</tr>
</thead>
<tbody>
<tr>
<td>Match A</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Match B</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Match C</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Match D</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Match E</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
</tbody>
</table>

- ✓ - This person is identified as a match.

Sara Combs  
b. 1824 TN  
d. 1914 MO

William Lane  
b. 1820 TN  
d. 1852 AR
GENETICS FOR GENEALOGY
Considerations
Genetics for Genealogy

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

**DNA MATCHES**

- American Colonial Ancestry
  - 194 Shared Ancestor Hints
  - Starred matches
  - 529 4th cousins or closer

- 19th Century German Migrants to Texas
  - 2 Shared Ancestor Hints
  - Starred matches
  - 9 4th cousins or closer
Genetics for Genealogy

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

![DNA Relatives Chart](image)

- American Colonial Ancestry & 1/8 19th Century Irish Migrants
- 19th Century German Migrants
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.
Genetics for Genealogy: Major Testing Options

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

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

- **Family Finder Test** at Family Tree DNA ($99) – Broad set of features to examine DNA matches; website is difficult to navigate and database is small ($37 transfer of AncestryDNA data to Family Finder database)