Genetic Genealogy

DNA-Based Inference of Common Ancestry

Historical Record of Ancestors

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

great great grandparents

great grandparents

grandparent

parent

great great grandparents

great grandparent

grandparent

parent

2\textsuperscript{nd} cousin

1x removed

DNA Ancestors

DNA Siblings

DNA Connections
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.
Uni-Parental Inheritance

Y chromosome

mtDNA
Autosomal Inheritance

1:1 segregation each generation

Numbers of ancestors in each generation

Expected autosomal contribution of each ancestor

~ expectation ~
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
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.
### Matching of SNPs

<table>
<thead>
<tr>
<th>rsid</th>
<th>chromosome</th>
<th>position</th>
<th>Tester</th>
<th>Match</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs12564807</td>
<td>1</td>
<td>734462</td>
<td>AA</td>
<td>GG</td>
</tr>
<tr>
<td>rs3131972</td>
<td>1</td>
<td>752721</td>
<td>GG</td>
<td>AG</td>
</tr>
<tr>
<td>rs148828841</td>
<td>1</td>
<td>760998</td>
<td>CC</td>
<td>CC</td>
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<td>776546</td>
<td>AA</td>
<td>AA</td>
</tr>
<tr>
<td>rs115093905</td>
<td>1</td>
<td>787173</td>
<td>GG</td>
<td>GG</td>
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<td>rs11240777</td>
<td>1</td>
<td>798959</td>
<td>GG</td>
<td>AG</td>
</tr>
<tr>
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<td>1</td>
<td>824398</td>
<td>AA</td>
<td>AC</td>
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<tr>
<td>rs4475691</td>
<td>1</td>
<td>846808</td>
<td>TT</td>
<td>CT</td>
</tr>
<tr>
<td>rs7537756</td>
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<td>854250</td>
<td>GG</td>
<td>AG</td>
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<td>861808</td>
<td>GG</td>
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<td>TT</td>
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<td>rs147226614</td>
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<td>878697</td>
<td>GG</td>
<td>GG</td>
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<td>i6052728</td>
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<td>i6019302</td>
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<td>881843</td>
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<td>rs2272756</td>
<td>1</td>
<td>882033</td>
<td>AA</td>
<td>GG</td>
</tr>
</tbody>
</table>

**Half-Identical:**

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
Segment Matching Between Relatives
Identity by Descent 
half-1st Cousins

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

\[ r = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{16} \quad \text{or} \quad 6.25\% \text{ identical genome} \]
### Expectations for the Inheritance of Identical Matching Autosomal Segments Between Relatives

<table>
<thead>
<tr>
<th>Relationship</th>
<th>Genome Identical</th>
<th>Number of Identical Segment</th>
<th>Avg. Length of Shared Segment</th>
<th>(\rho(\text{none})^*) Donnelly</th>
</tr>
</thead>
<tbody>
<tr>
<td>1(^{st}) cousins</td>
<td>12.50%</td>
<td>41.4 segments</td>
<td>21.7 cM</td>
<td>0.00</td>
</tr>
<tr>
<td>2(^{nd}) cousins</td>
<td>3.13%</td>
<td>14.8 segments</td>
<td>15.1 cM</td>
<td>0.00</td>
</tr>
<tr>
<td>3(^{rd}) cousins</td>
<td>0.78%</td>
<td>4.8 segments</td>
<td>11.6 cM</td>
<td>0.02</td>
</tr>
<tr>
<td>4(^{th}) cousins</td>
<td>0.20%</td>
<td>1.5 segments</td>
<td>9.4 cM</td>
<td>0.31</td>
</tr>
<tr>
<td>5(^{th}) cousins</td>
<td>0.05%</td>
<td>0.4 segments</td>
<td>7.9 cM</td>
<td>0.70</td>
</tr>
</tbody>
</table>

*without detection errors
### Example Summary of DNA Matches

<table>
<thead>
<tr>
<th>Ancestors</th>
<th>DNA Match (relationship)</th>
<th>&quot;In Common With&quot; or &quot;Shared Match&quot;</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Grandparents</strong></td>
<td>Match-A (2C)</td>
<td>Match-A: self  Match-B: yes  Match-C: yes  Match-D: no</td>
</tr>
<tr>
<td><strong>GG Grandparents</strong></td>
<td>Match-B (3C)</td>
<td>Match-A: yes  Match-B: self  Match-C: no  Match-D: yes</td>
</tr>
<tr>
<td><strong>GG Grandparents</strong></td>
<td>Match-C (3C)</td>
<td>Match-A: yes  Match-B: no  Match-C: self  Match-D: no</td>
</tr>
<tr>
<td><strong>GGG Grandparents</strong></td>
<td>Match-D (4C)</td>
<td>Match-A: no  Match-B: yes  Match-C: no  Match-D: self</td>
</tr>
</tbody>
</table>
Example Summary of DNA Matches

<table>
<thead>
<tr>
<th>DNA Match (relationship)</th>
<th>Match-A</th>
<th>Match-B</th>
<th>Match-D</th>
<th>Match-C</th>
</tr>
</thead>
<tbody>
<tr>
<td>Match-A (2C)</td>
<td>self</td>
<td>yes</td>
<td>no</td>
<td>yes</td>
</tr>
<tr>
<td>Match-B (3C)</td>
<td>yes</td>
<td>self</td>
<td>yes</td>
<td>no</td>
</tr>
<tr>
<td>Match-D (4C)</td>
<td>no</td>
<td>yes</td>
<td>self</td>
<td>no</td>
</tr>
<tr>
<td>Match-C (3C)</td>
<td>yes</td>
<td>no</td>
<td>no</td>
<td>self</td>
</tr>
</tbody>
</table>
Genetic Structure of Human Populations

A global reference for human genetic variation

The 1000 Genomes Project Consortium

*Nature* 526, 68–74 (01 October 2015) | doi:10.1038/nature15393

- Each vertical line represents an individual
- Samples of individuals organized by continent
- Eight ancestry groups identified by color
- *Ancestry composition of each individual represented proportionally by each color*