Different Selective Pressures Shape the Molecular Evolution of Color Vision in Chimpanzee and Human Populations

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Introduction

- Genetic analysis reveal how human and chimpanzee lineages have diverged
- X-linked color vision variation result in L-opsin (red) and M-opsin (green)
- Correlation between opsin wavelength absorption maxima and single amino acid variants
- How color vision gene variation are different between humans and chimpanzees

Materials and Methods

- Primate samples
  - Pan troglodytes verus and other subspecies: DNA of 40 species from western Africa, 3 from central, 1 from eastern, and 1 Nigerian
  - Human: 163 males from Africa and 73 males from “non-Africa”
  - Long-wavelength opsin color vision gene
- Gene fragment amplification and sequencing

Materials and Methods

- Data analysis and statistical tests
  - D test: assess SNP frequency spectrum, D values consistent with population structure or expansion
  - LD and recombination parameter: examine haplotype structure at the gene
  - McDonald and Kreitman test of neutrality to examine older lineage-specific changes in selective pressures

Results

- Chimpanzee gene diversity
  - 42 variants among 56 chimpanzee chromosomes

Results

- Chimpanzees are far less variable than humans in silent site diversity
- Replacement SNPs are found across human populations and some show significant differences

<table>
<thead>
<tr>
<th>Table 1</th>
<th>Population Diversity Estimates and Tests of Neutrality at OPL/M-LH</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample</td>
<td>Theta (θ)</td>
</tr>
<tr>
<td>Pan-opsin</td>
<td>0.060</td>
</tr>
<tr>
<td>Pan-opsin non-Pan-opsin</td>
<td>0.060</td>
</tr>
<tr>
<td>African</td>
<td>0.060</td>
</tr>
<tr>
<td>non-African</td>
<td>0.060</td>
</tr>
</tbody>
</table>
Results
- Subspecies of chimpanzees show nucleotide diversity higher than humans
- Silent diversity of *P. t. troglodytes* similar to African while that of *P. t. verus* similar to non-African
- Much different in amino acid replacement sites variation between chimpanzees and humans

<table>
<thead>
<tr>
<th>Sample</th>
<th>Few nucleotide polymorphism</th>
<th>Few nucleotide amino acids</th>
<th>Few nucleotide rarer</th>
<th>Few nucleotide rarest</th>
</tr>
</thead>
<tbody>
<tr>
<td>F. t. subspecies</td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>F. t. troglodytes</td>
<td>0.052</td>
<td>0.062</td>
<td>0.075</td>
<td>0.080</td>
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<tr>
<td>F. t. verus</td>
<td>0.044</td>
<td>0.065</td>
<td>0.067</td>
<td>0.070</td>
</tr>
</tbody>
</table>

Discussion
- Abundance of amino acid polymorphism at human gene might be result of gene conversion
- Similar patterns in silent and amino acid polymorphism, and fixation
- Unusual pattern of haplotype diversity only in humans

Discussion
- Human gene replacement SNPs due to balancing selection for spectral tuning
- Only one X-linked opsin gene exist in chimpanzees, no variation?
- Hunter-gatherer societies, distinguish color of fruits and other food on shaded background
- Sexual selection and mate recognition
- Color vision is unique in human and primate evolution