Evolution of spontaneous mutation: patterns and consequences

Darwinian evolution

Heritable (genetic) variation

AND

Fitness differences (natural selection)
Structure of genetic variation

- Genetic epistasis (pleitropic effects of mutations)

- Biased production of variation
  - Differences in mutational targets
  - Mutational biases

Questions about mutational biases

- Do they exist?
Questions about mutational biases

- Do they exist?

- Do they vary in different organisms?

- Do they affect the course of evolution?
**Mutation** - first cause of evolution, ultimate cause of genetic variation

**Estimation problem**

- Direct measurement

  Problem: mutation is too infrequent
Mutation - first cause of evolution, ultimate cause of genetic variation

**Estimation problem**

- Direct measurement
  - Problem: mutation is too infrequent

- Inference from the observed variation
  - Problem: natural selection bias
Estimation solution

Pseudogenes → no functional constraints, ought to reflect mutational biases
(Li, Gojobori, and Nei 1981)

Problem → pseudogenes absent in many taxa

Transposition of a "dead-on-arrival" non-LTR retroelement

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Mutational Patterns and Evolution of Genome Size
Mutational Patterns and Evolution of Genome Size

Helena in the *D. melanogaster* subgroup

INTERNAL BRANCHES

TERMINAL BRANCHES

$P = 3.5 \times 10^{-15}$

$P = 0.76$

Deletion rate in *Drosophila* 0.13 deletions/substitution

(95% conf. interval 0.11 to 0.18 del/subst)
### HIGH RATE OF DNA LOSS IN DROSOPHILA

<table>
<thead>
<tr>
<th></th>
<th>Mammals</th>
<th>Drosophila</th>
</tr>
</thead>
<tbody>
<tr>
<td>Deletions per substitution</td>
<td>0.05</td>
<td>0.13</td>
</tr>
<tr>
<td>Average size of deletions (bp)</td>
<td>3.2</td>
<td>22.8</td>
</tr>
<tr>
<td>Half-life of a pseudogene (Myr)</td>
<td>884</td>
<td>14.3</td>
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</tbody>
</table>

(from D. Geau et al., JME 1989 28:279-285)

### Log10 genome size (bp)

- 6 7 8 9 10 11 12
- 3x10^9 bp (human genome)

- **Protists**
  - Euglenozoa
  - Ciliophora
  - SAR clade
- **Fungi**
  - Fungi
  - Sponges
  - Annelids
  - Mollusks
  - Crustaceans
  - Insects
  - Echinoderms
  - Agnathans
  - Sharks & Rays
  - Bony fishes
  - Amphibians
  - Reptiles
  - Birds
- **Animals**
  - Mammals
  - Algae
  - Phaeophytes
  - Gymnosperms
  - Angiosperms
- **Plants**
  - Log10 genome size (bp)
Factors in genome size evolution

- **Mutation**
  - $\uparrow \uparrow$ Transposable elements
  - $\downarrow \uparrow$ Deletions and insertions

- **Natural selection**
  - $\downarrow + \uparrow$ Strength
  - $\downarrow + \uparrow$ Direction

Hawaiian crickets, Genus Laupala

- Endemic to Hawaii
- 37 recognized species
- Not well studied molecularly
- Large genome size
  - (1800 MB vs 160 MB in Drosophila)
Mutational Patterns and Evolution of Genome Size

Deletion sizes in Drosophila and Laupala

- Drosophila: 0.13 (0.11-0.18) del/subst
- Laupala: 0.07 (0.05-0.10) del/subst

Significant at P=0.0006

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### Mutational Patterns and Evolution of Genome Size

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</tr>
<tr>
<td>Insertions per substitution</td>
<td>0.03</td>
<td>0.01</td>
</tr>
<tr>
<td>Deletion size</td>
<td>6.0 bp</td>
<td>22.8 bp</td>
</tr>
<tr>
<td>Insertion size</td>
<td>5.4 bp</td>
<td>2.9 bp</td>
</tr>
<tr>
<td>Pseudogene half-life (MYR)</td>
<td>560</td>
<td>14</td>
</tr>
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</table>

### DNA loss (per bp per point subst)

![Graph showing DNA loss and genome size](image-url)
Mutational Patterns and Evolution of Genome Size

[Graphs showing the relationship between DNA loss per bp per point substitution and genome size in Mbp for different species, including Podisma (grasshopper), Laupala (cricket), Mammals (humans and rodents), and Drosophila.]
# Acknowledgements

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