

Evolution of spontaneous mutation:  
patterns and consequences

Darwinian evolution

Heritable (genetic) variation

AND

Fitness differences (natural selection)

### Structure of genetic variation

- Genetic epistasis (pleiotropic effects of mutations )
- Biased production of variation
  - Differences in mutational targets
  - Mutational biases

### Questions about mutational biases

- Do they exist?

## Questions about mutational bias s

- Do they exist?
- Do they vary in different organisms?

## Questions about mutational bias s

- 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**

**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**

**Mutation - first cause of evolution,  
ultimate cause of genetic variation**

**Estimation problem**

- **Direct measurement**

**Problem: mutation is too infrequent**

- **Inference from the observed variation**

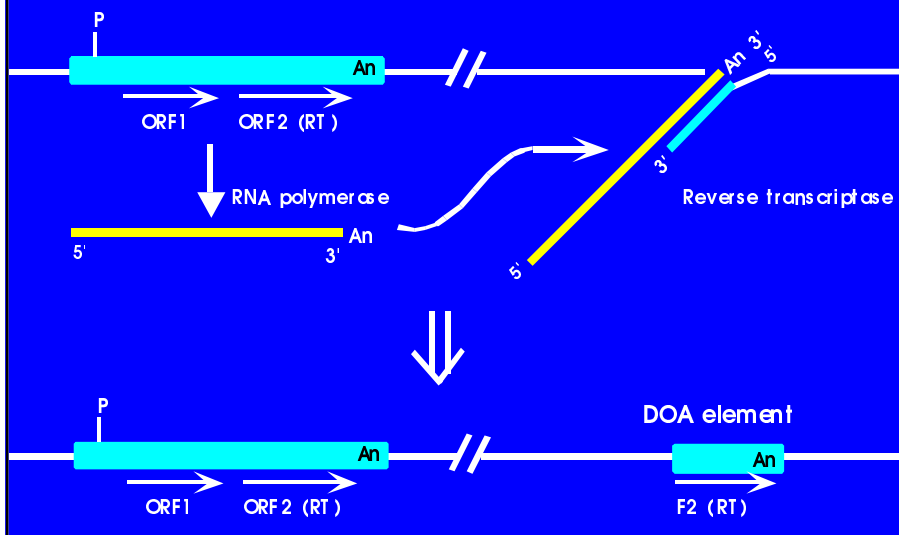
**Problem: natural selection bias**

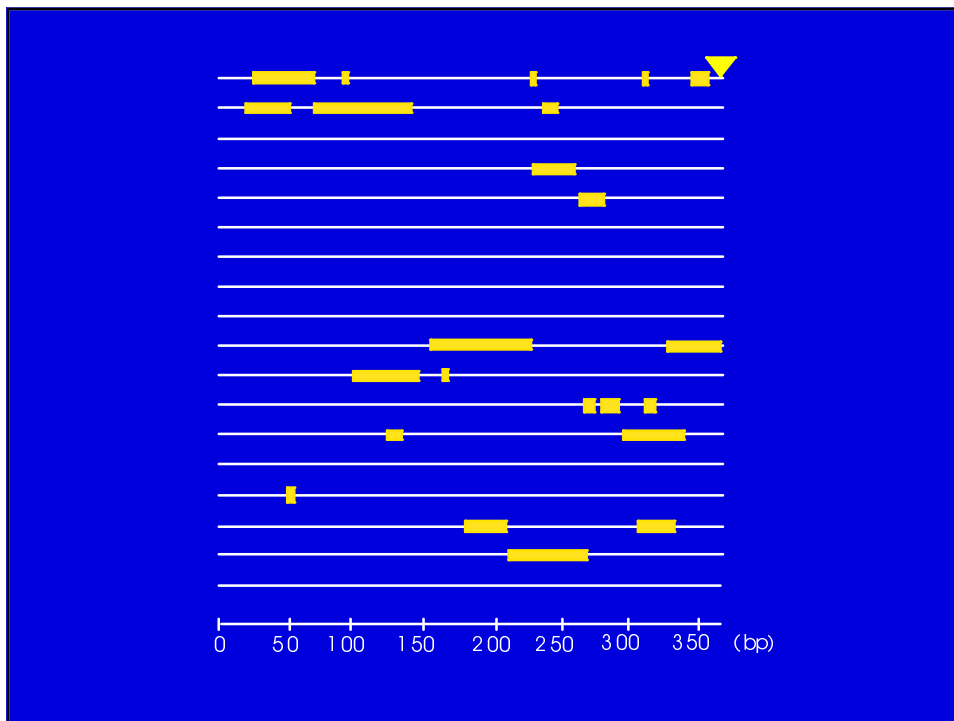
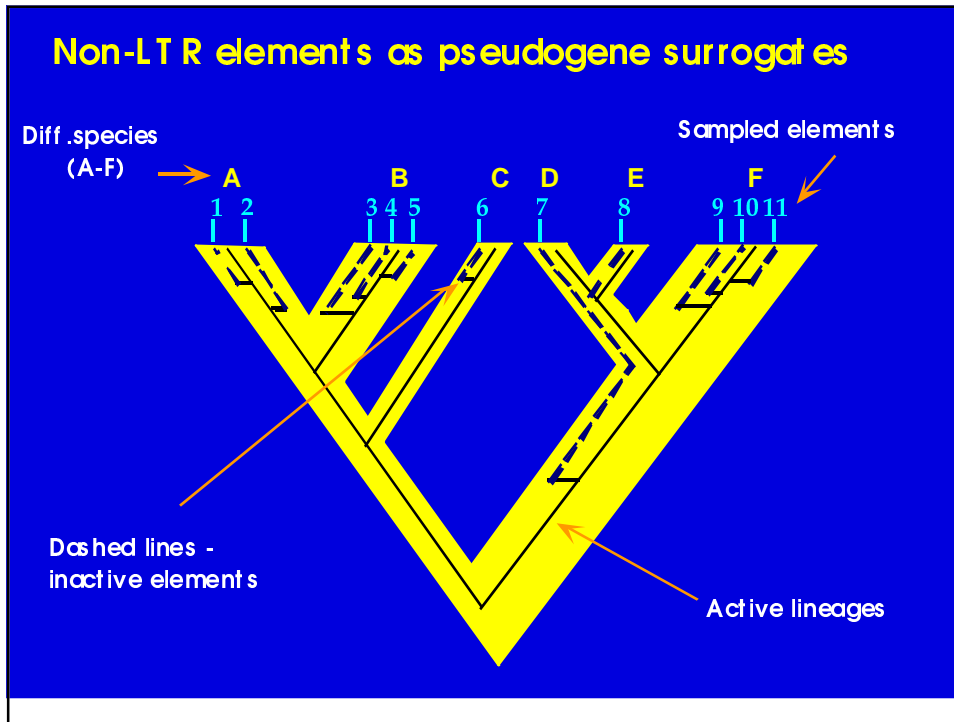
## Estimation solution

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

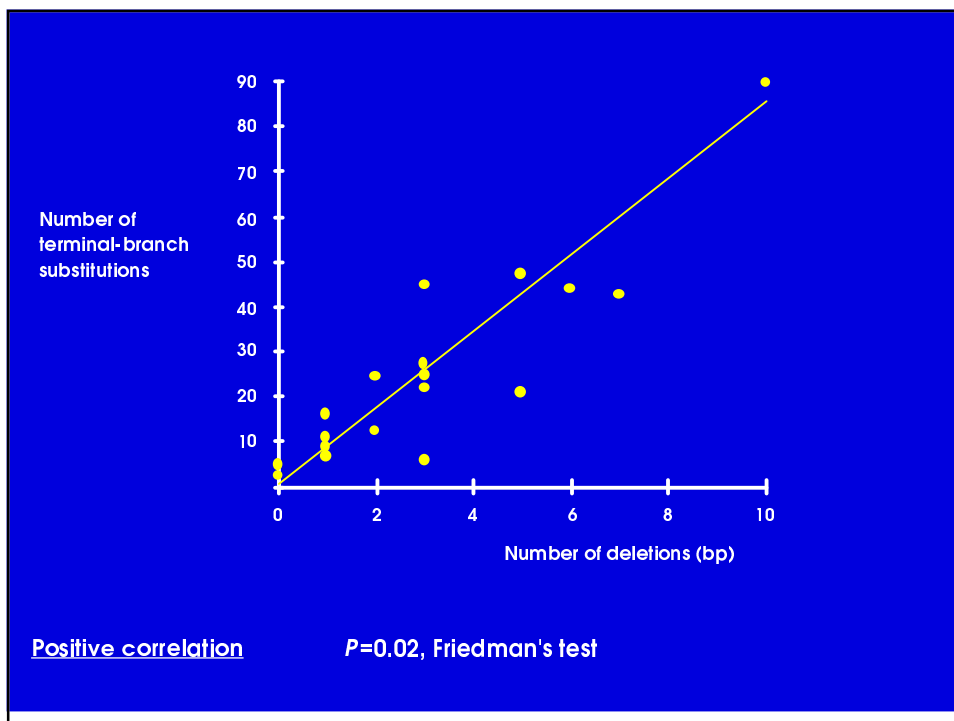
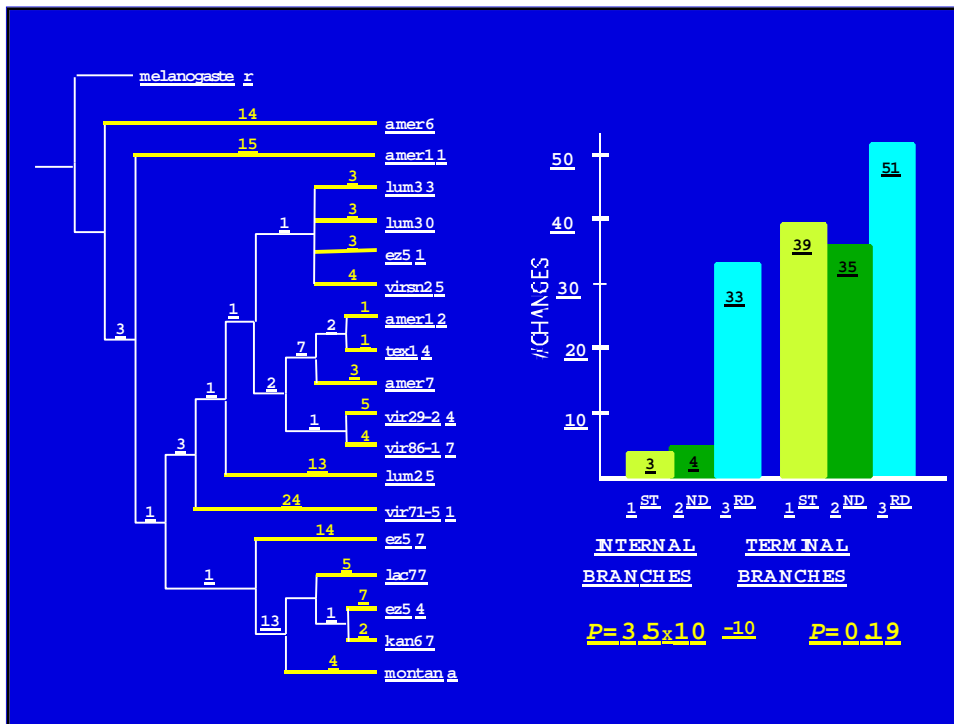
**Problem** → pseudogenes absent in many taxa

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

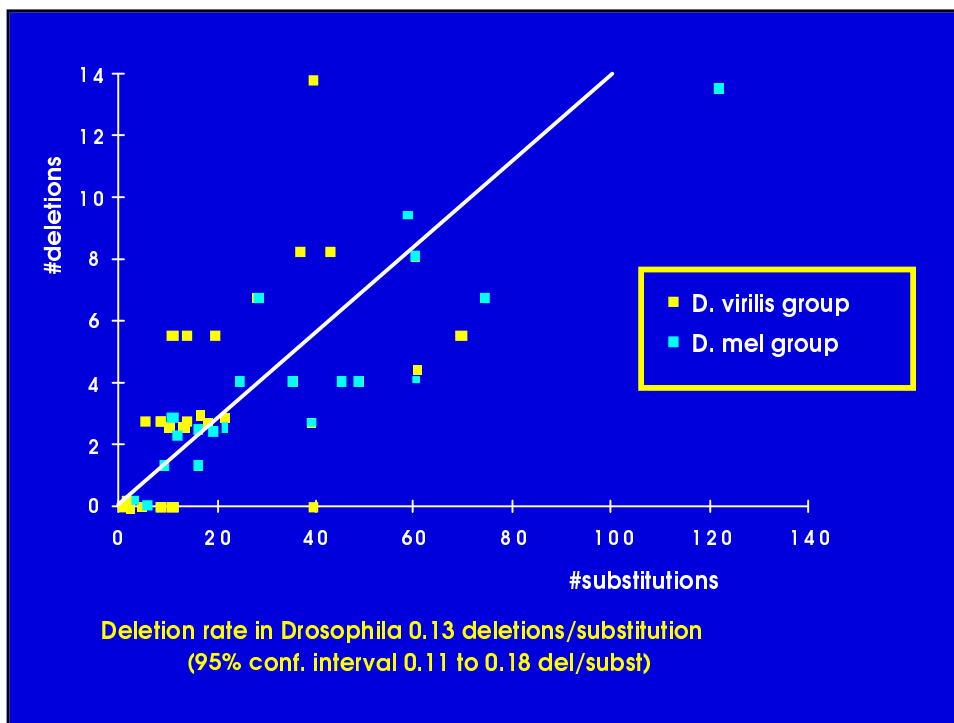
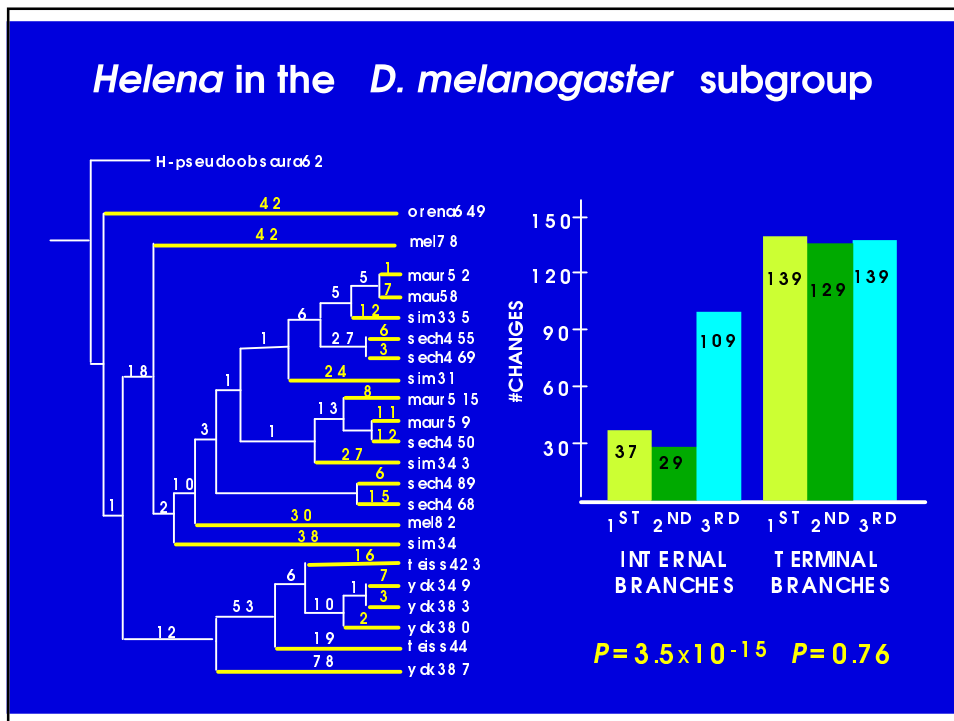




# Mutational Patterns and Evolution of Genome Size



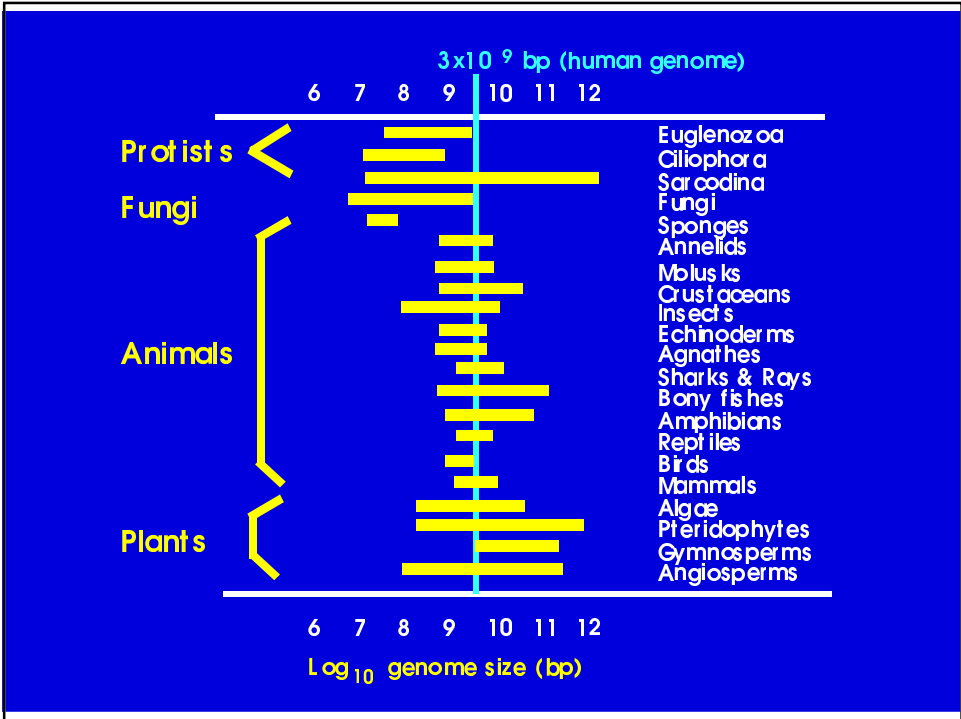




### HIGH RATE OF DNA LOSS IN DROSOPHILA

	Mammals	Drosophila
Deletions per substitution	0.05	0.13
Average size of deletions (bp)	3.2	22.8
Half-life of a pseudogene (Myr)	884	14.3

from D. Grauretal.  
JME, 1989 28:279-285



## Factors in genome size evolution

- Mutation

+ ↑ Transposable elements

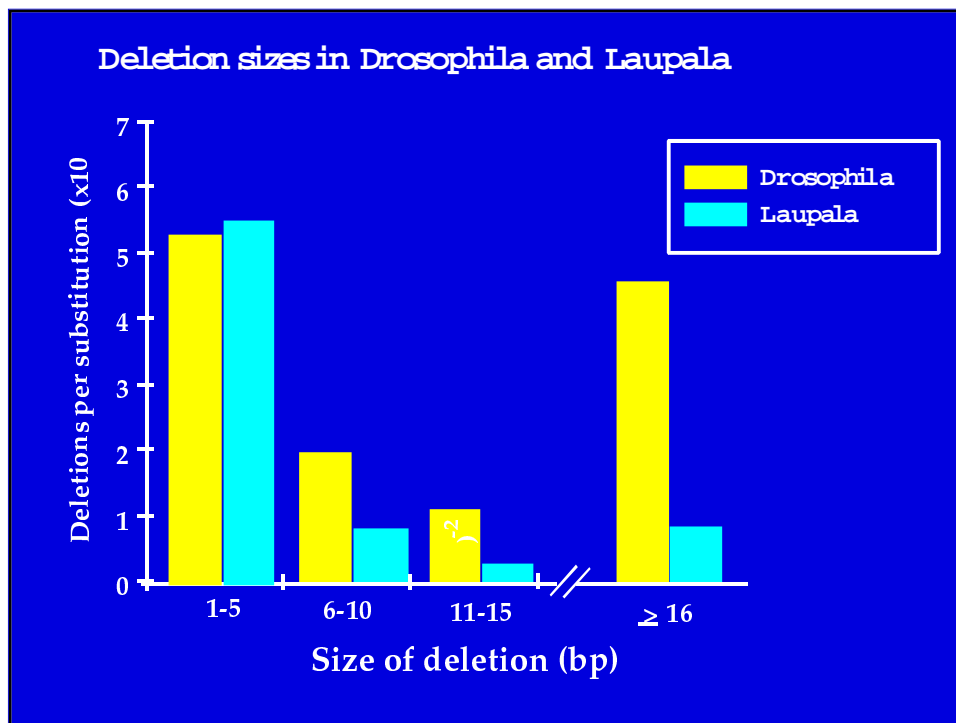
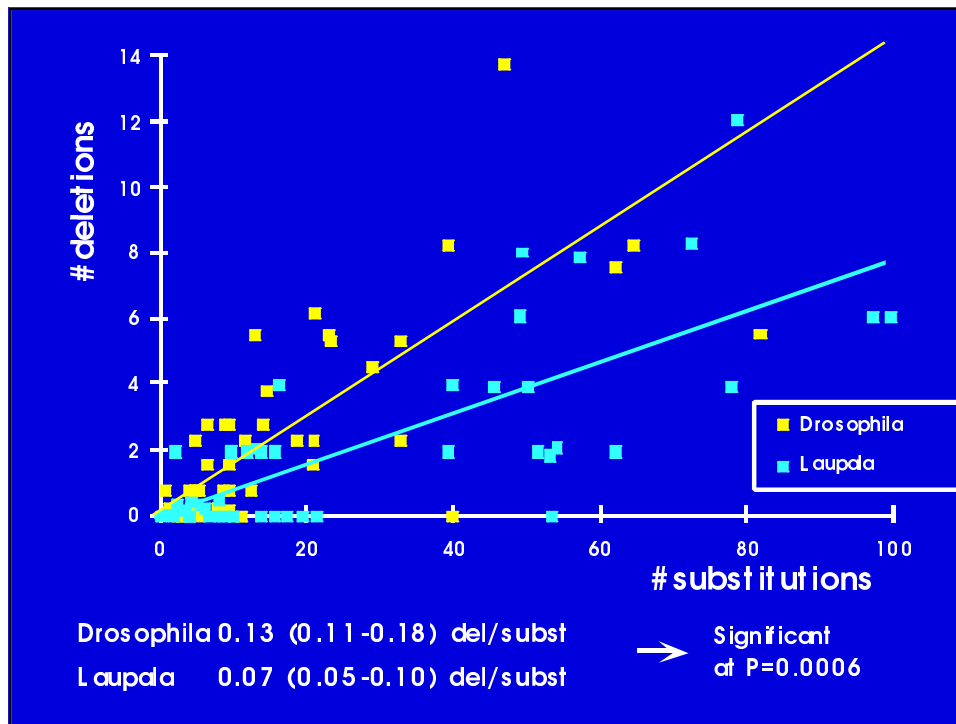
- ↓ + ↑ Deletions and insertions

- Natural selection

- ↓ + ↑ Strength  
Direction

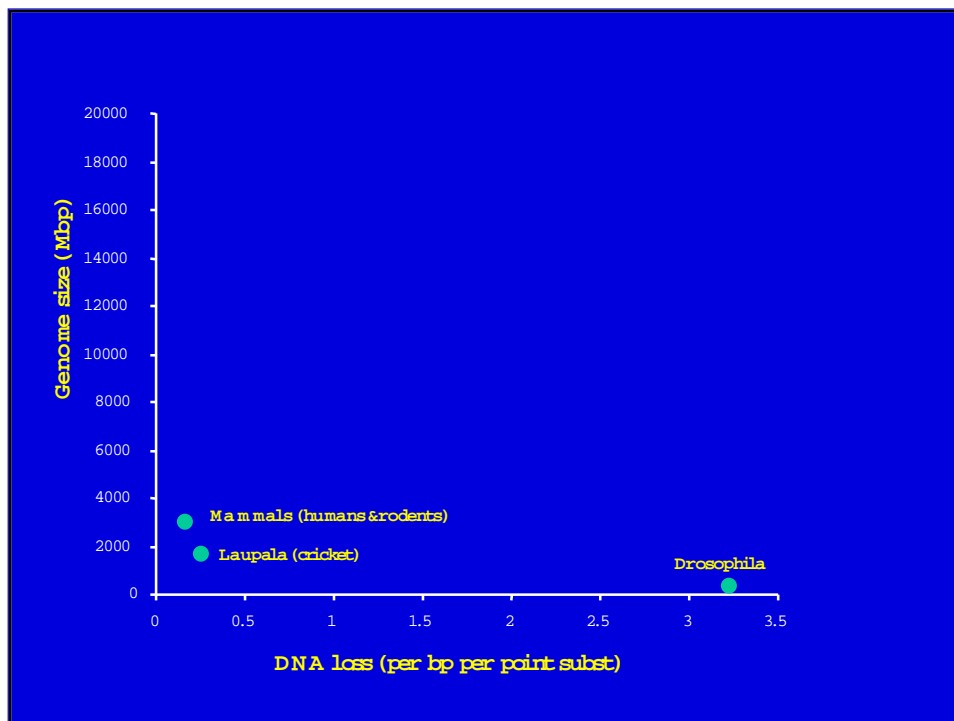
## Hawaiian crickets, Genus *Laupala*

- Endemic to Hawaii
- 37 recognized species
- Not well studied molecularly
- Large genome size  
(1800 MB vs 160 MB in *Drosophila*)

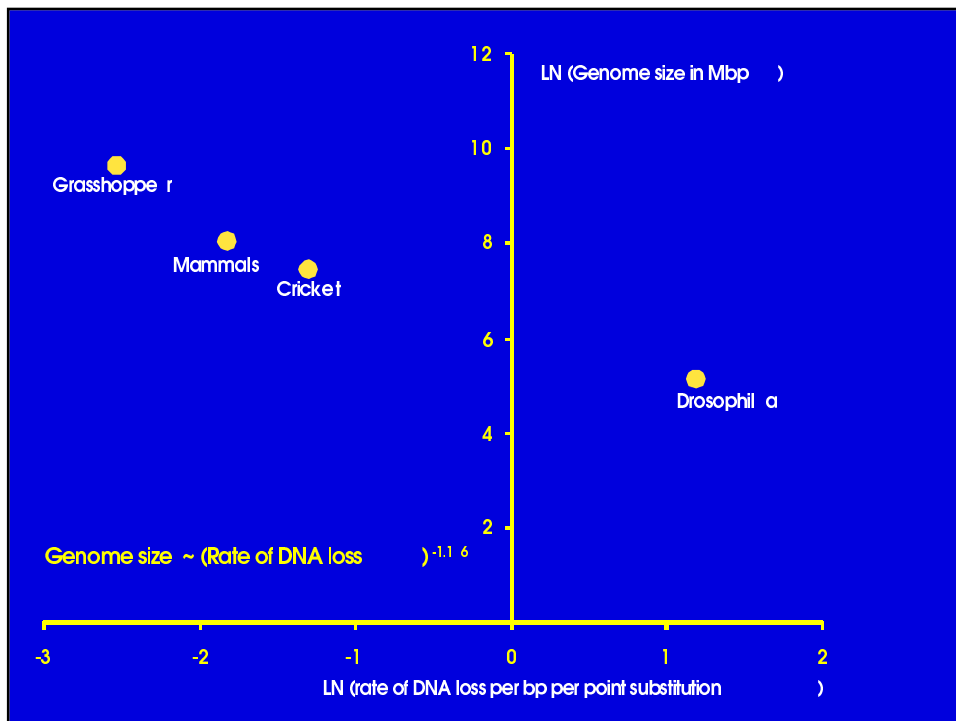
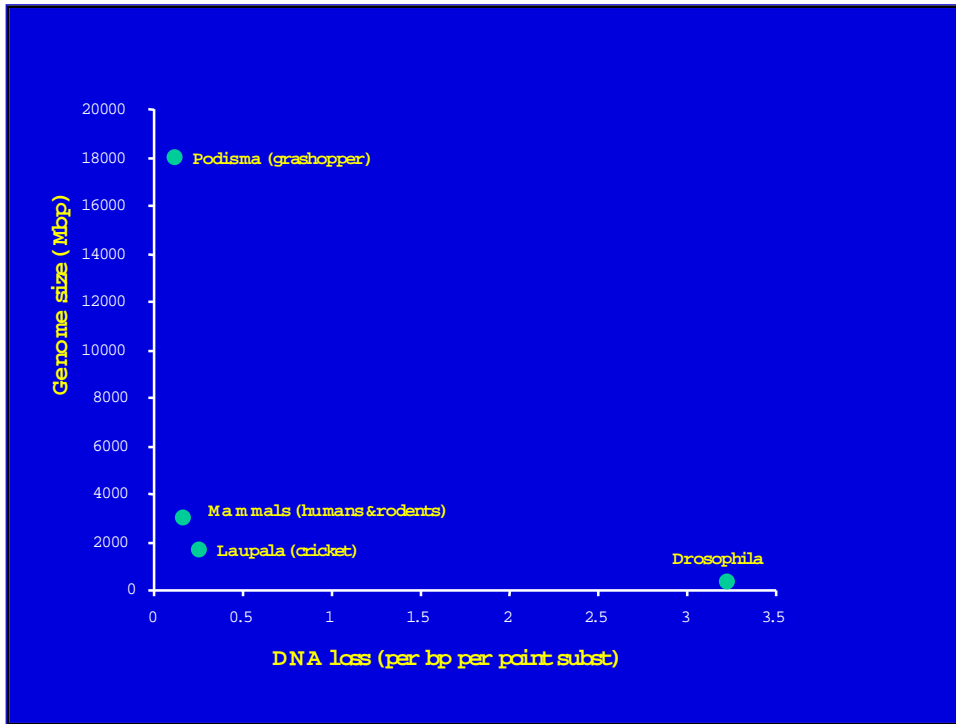


## Mutational Patterns and Evolution of Genome Size

	Laupala	Drosophila
Deletions per substitution	0.07	0.13
Insertions per substitution	0.03	0.01
Deletion size	6.0 bp	22.8 bp
Insertion size	5.4 bp	2.9 bp
Pseudogene half-life (MYR)	560	14



# Mutational Patterns and Evolution of Genome Size



## Acknowledgements

Collaborators:

**Daniel Hartl**  
**Douda Bensasson**  
**Etsuko Moriyama**  
**Kerry Shaw**  
**Todd Sangster**  
**Spencer Johnston**  
**(Texas A&M)**

Comments  
& support:

**Lewontin & Hartl**  
**labs**  
**Dick Lewontin**  
**Mark Siegal**