Organelle genome evolution
Plant of the day!

Rafflesia arnoldii

- largest individual flower (~ 1m)
- no true leafs, shoots or roots
- holoparasitic
- non-photosynthetic
Big questions

• What is the origin of organelle genomes?

• What were the major steps in organelle genome evolution?

• Why are organelle genomes maintained?

• Is organelle genome variation neutral or adaptive?
The Endosymbiotic Theory
The Endosymbiotic Theory

- MITOCHONDRIA: evolved from aerobic bacteria (α-proteobacteria) and a host

- CHLOROPLASTS: evolved from a heterotrophic eukaryote and a cyanobacteria
Evidence for the Endosymbiotic Theory

- Circular molecule
- No histones
- Protein synthesizing machinery (ribosomes, tRNA, rRNA)
- Some antibiotics block protein synthesis within the mitochondria and chloroplasts
- Structural similarity
- Reproduce through fission
- Strong phylogenetic evidence
The endosymbiotic theory

Futuyma (2009)
Organelle gene transfer
# Sizing-up mitochondrial genomes

<table>
<thead>
<tr>
<th>Plants and algae</th>
<th>kbp</th>
<th># protein coding genes</th>
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<tbody>
<tr>
<td>mt Pyraea litoralis</td>
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<td>52</td>
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<tr>
<td>mt Marchantia polymorpha</td>
<td>187</td>
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<tr>
<td>mt Laminaria digitata</td>
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<td>39</td>
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<td>mt Cyanidioschyzon merolae</td>
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<tr>
<td>mt Arabidopsis thaliana</td>
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<td>mt Chondrus crispus</td>
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<tr>
<td>mt Scenedesmus obliquus</td>
<td>43</td>
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<table>
<thead>
<tr>
<th>Various protists and fungi</th>
<th>kbp</th>
<th># protein coding genes</th>
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<tbody>
<tr>
<td>mt Reclinomonas americana</td>
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<td>67</td>
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<td>mt Malawimonas jakobiformis</td>
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<td>mt Naegleria gruberi</td>
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<td>mt Rhodomonas salina</td>
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<tr>
<td>mt Dictyostelium discoideum</td>
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<td>mt Phytophthora infestans</td>
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<tr>
<td>mt Acanthamoeba castellanii</td>
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<td>mt Cafeteria roenbergensis</td>
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<td>34</td>
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<tr>
<td>mt Monosiga brevicollis</td>
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<td>32</td>
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<tr>
<td>mt Physarum polycephalum</td>
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<td>20</td>
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<tr>
<td>mt Harpochrytrium sp</td>
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<td>14</td>
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<td>mt Candida albicans</td>
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<td>mt Cryptococcus neoformans</td>
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<td>mt Plasmodium falciparum</td>
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<table>
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<th>α-proteobacteria</th>
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<tr>
<td>Caulobacter crescentus</td>
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<td>Mesorhizobium loti</td>
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<td>Bradyrhizobium japonicum</td>
<td>~9100</td>
<td>~8300</td>
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References:
- NC_003055
- MP01MTCG
- AJ344328
- NC_000887
- MIATGENA
- MTCCGNMNE
- NC_002254
- NC_001823
- AF295546
- NC_002573
- NC_002572
- NC_000895
- NC_002387
- U12386
- NC_000946
- AF538053
- AB027295
- AY182006
- NC_002653
- NC_004336
- NC_001677
- AE006573
- BA000012
- BA000040
Sizing-up chloroplast genomes

Timmis et al. (2004)
Organelle gene transfer

modified from Kleine et al. (2009)
Recent organelle gene transfers

Comparisons of organelle genes and nuclear genes of the same species

- Gene transfer between cell compartments can occur
- This might be a continuous process
- The frequency of organelle-to-nucleus gene transfer
Mitochondrial Gene Transfer

- *rps10* gene phylogeny

inferred transfers to the nucleus

modified from Adams *et al.* (2000)
Chloroplast Gene Transfer

- *InfA* (translation initiation factor) gene phylogeny (Millen *et al.* 2001)
- ~24 chloroplast-to-nucleus gene transfers
- mutational decay/loss of chloroplast sequence
- *de novo* mechanism for chloroplast targeting?
Ancient organelle gene transfers

Comparisons of nuclear, organelle and candidate prokaryotic ancestor genomes

- The scale of organelle-to-nucleus gene transfer
- The fate of imported genes over time
Organelle gene transfers
(Arabidopsis)

Anabaena variabilis Nostoc sp.

Cyanobacterium-like endosymbiont

Plastid
Plastome size: 154 kb
Protein ORFs: 87

Nucleus
Nuclear genome size: ~ 125 Mb
Protein ORFs: 27,029

modified from Kleine et al. (2009)
The fate of transferred genes

- products routed back to the donor organelle
- products are targeted to other cellular compartments
- functional replacement of equivalent host genes
  (= endosymbiotic gene replacement)
Mitochondrial Gene Transfer Rates

• rate estimates:
  – 1 plasmid transfer to nucleus in 20,000 yeast cells (integration rare)
Chloroplast Gene Transfer Rates

Rate estimates from tobacco chloroplasts
- 1 transfer in 5 million leaf cells
- 1 transfer in 16,000 pollen grains

Higher rates of transfer in the pollen?
Degradation of the organelle genomes in pollen could make DNA fragments available for uptake
How do organelle genes get into the nucleus?

- **Bulk DNA**
  - Recombination between escaped organelle DNA and nuclear DNA
  - Experimental transfer in yeast
  - Non-coding sequence frequently transferred
  - Whole organelle sequences transferred

**MUST HAPPEN**
How do organelle genes get into the nucleus?

- **cDNA intermediates**
  - NUMTS and NUMPTS often lack organelle-specific introns and edited sites

MAY HAPPEN
modified from Henze & Martin (2001)
cDNA-hypothesis

nuDNA

Transfer

Mitochondria

Alternative

nuDNA

Recombination

Transfer

mtDNA

mtDNA

Reverse transcription

Splicing and editing

Transcription

modified from Henze & Martin (2001)
Why are organelle genomes maintained?

• **Hydrophobicity** - hydrophobic proteins are poorly imported

• **Redox-control** - fitness advantage if coding sequence and regulation are in same location

• Other constraints (RNA editing, genetic code)

• What about non-photosynthetic plants?
Why are organelle genomes maintained?

- **Hydrophobicity** - hydrophobic proteins are poorly imported

- **Redox-control** - fitness advantage if coding sequence and regulation are in same location

- Other constraints (RNA editing, genetic code)

- What about non-photosynthetic plants?
  - essential tRNAs (Barbrook *et al.* 2006)
Organelle genome evolution under changing environmental conditions
Structure of Plant cp Genomes

Noug cpDNA
151,762 bp

LSC – 84 kb
SSC – 18 kb
IR – 25 kb (each)

Dempewolf et al. 2010
Structure of Plant mt Genomes

Wheat mtDNA
452,528 bp

Ogihara et al. 2005
Neutral organelle DNA variation

- organelle-encoded proteins are highly conserved

- limited coding potential of organelle genomes (compared to the nuclear genome)

- reduced rates of sequence evolution (in plants)
## Rates of synonymous substitutions per million years

<table>
<thead>
<tr>
<th>Genome</th>
<th>Taxa compared</th>
<th>Rate</th>
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<tbody>
<tr>
<td>Plant</td>
<td></td>
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</tr>
<tr>
<td>mt</td>
<td>Maize/wheat Monocot/dicot</td>
<td>0.2 – 0.3</td>
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<tr>
<td></td>
<td></td>
<td>0.8 – 1.1</td>
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<tr>
<td>cp</td>
<td>Maize/wheat Monocot/dicot</td>
<td>1.1 – 1.6</td>
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<tr>
<td></td>
<td></td>
<td>2.1 – 2.9</td>
</tr>
<tr>
<td>nuc</td>
<td>Spinach/Silene Monocot/dicot</td>
<td>15.8 – 31.5</td>
</tr>
<tr>
<td></td>
<td></td>
<td>5.8 – 8.1</td>
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<tr>
<td>Animal</td>
<td></td>
<td></td>
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<tr>
<td>mt</td>
<td>Human/chimpanzee Mouse/rat</td>
<td>21.8 – 43.7</td>
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<tr>
<td></td>
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<td>18.2 – 54.5</td>
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<tr>
<td>nuc</td>
<td>Human/chimpanzee Mouse/rat</td>
<td>0.9 – 1.9</td>
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<tr>
<td></td>
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<td>3.9 – 11.8</td>
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### Evolution of three genomes

<table>
<thead>
<tr>
<th></th>
<th>Genome</th>
<th>Sequence evolution</th>
<th>Structural evolution</th>
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<tr>
<td><strong>Plant</strong></td>
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</tr>
<tr>
<td>mt</td>
<td>very slow</td>
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<tr>
<td>cp</td>
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<td>nuc</td>
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<td><strong>Animal</strong></td>
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<td>moderate</td>
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Adaptive organelle DNA variation?

Experimental evidence is accumulating:

- parallels between organelle capture and environmental variation
- experimental evolution of cytoplasm fitness effects
- direct tests of selection on organelle genes
Experimental evolution studies

Sambatti et al. (2008)

*Helianthus petiolaris*
- common in dry sandy soils

*Helianthus annuus*
- common in clay-based soils

Photos by J. Rick
Experimental evolution studies

Crossing design

Cytoplasm-by-habitat interaction

modified from Sambatti et al. (2008)
Tests of selection on organelle genes

- Kapralov & Filatov (2006)
- Schiedea genus (endemic to Hawaii)
- Species adapted to xeric conditions
- Positive selection detected at the cp \( rbcL \) gene
Co-adaptation of genotype and plasmotype

Woodson & Chory (2008)
Unanswered questions

• To what extent have organelle gene transfers shaped nuclear genomes?

• Is organelle gene transfer just a quirk of evolution?

• How often does organelle genetic variation contribute to local adaptation?

• What are the agents and traits under divergent selection?