Genetic basis of adaptation and speciation

Marina Panova
Adaptation and speciation: genes and alleles

Allele A: light phenotype

Mutation

Allele B: dark phenotype

Higher fitness

Ecological selection
Sexual selection

Probability of mating
Hybrid unfitness
Central Dogma of Molecular Biology: Eukaryotic Model

DNA

mRNA

Protein

Transcription and mRNA processing

Translation

Post-Translational Modification

Active Protein

Gene Control: Regulatory Regions

- There can be one or more transcription factors (also called DNA binding proteins) that can initiate (or stop) transcription.
- The transcription start site is where RNA polymerase transcribes mRNA from the DNA template.

**cis-regulatory mutations**

**structural mutations**

Source: NCBI-webpage
Milestones

• 1859: Charles Darwin "On the Origin of Species"
• 1942 Julian Huxley "Evolution: The Modern Synthesis"
• 1996: first complete sequence of eukaryote genome (yeast)
• 2001: publication of human genome
• 2009: 1193 complete genomes (123 eukaryotic); 5023 on-going (1257 eukaryotic)
Genomic era

Genome data for model organisms

Marker panels for nonmodel organisms

Example: Human genome

Nucleus containing DNA

DNA is organized into chromosomes: 22 pairs of autosomes (1-22) and 1 pair of sex chromosomes (X,Y).

Genes, the functional units of heredity, are carried on chromosomes.

Plus the mitochondrial DNA

Gene

Allele 1

Allele 2

(Genome)

(Transcriptome)
What we would like to know about adaptation/speciation genes

- Mechanistic explanation of function?
- Mutations and their effect on gene product?
- Effect on phenotype?
- Role in adaptation/speciation?
- One gene -> many traits (pleiotropy)
- Many genes -> one trait (polygenic)
- Interactions between genes (epistasis)
- For many genes function is unknown
"Speciation genes" in *Drosophila*

Haldane’s rule: hybrid unfitness in heterogametic sex

Dobzhansky-Muller incompatibility: dominant allele in autosomal gene - recessive allele in gene on X chromosome

Ex: *Odysseus* gene:
*D. simulans* x *D. mauritiana*
Location: X-chromosome
Function: *Hox* (transcription factor)
Process: development
"Speciation genes" in other organisms: *Xiphophorus* fishes and melanoma


Platyfish $\times$ Swordtail

<table>
<thead>
<tr>
<th>Genotype</th>
<th>A</th>
<th>B</th>
<th>C</th>
<th>D</th>
<th>E</th>
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Shift of research from lab cultures to organisms in their natural environments

• "It is embarrassing that, although many isofemale lines have been trapped in the wild since the pioneering work of Dobzhansky, the natural foods and larval habitats of Drosophila pseudoobscura and D. persimilis are virtually unknown" (M. Noor, pers. comm. in Mallet 2006)

• "The irony of studying "ecologically important traits" in Mus and Rattus is that neither of them have an ecology independent of human activities" (Storz & Hoekstra 2007).
Drosophila sechellia and Morinda citrifolia

Matsuo et al. 2007

1. Resistance to octanoic acid
2. Odor attraction to Morinda plants
3. Lack of escape behavior

Regulatory mutation in Odorant-binding protein57e gene
Phytophagous insects: promising systems

Goldenrod gall fly

Pea aphids

Leaf beetle

Larch bud moth

Leaf mining moth

Apple maggot fly

(from Matsubayashi et al. 2010)
"Speciation genes" in other organisms: Monkey flowers and pollinators

Bradshaw & Schemske Nature 2003

*Mimulus lewisii, YUP\textsuperscript{pink},*

*M. cardinalis, YUP\textsuperscript{red},*

hybrids
Flower plants and pollinators: *Aquilegia*

Kramer & Hodges 2010

"Petal and stamen" AP3-gene in *Arabidopsis*

Three genes, *AqAP3-1,2,3* in *Aquilegia*

Variation in corolla Staminodes
Pheromones in butterflies: fatty acids

Saturated: $\text{CH}_3(\text{CH}_2)_n\text{COOH}$

Desaturated: $\text{CH}_3(\text{CH}_2)_n\text{CH}=\text{CH}(\text{CH}_2)_m\text{COOH}$

*Desat1* gene in Diptera: $\Delta 9$ desaturase

Several *Desat* genes in Lepidoptera: desaturases $\Delta 9$ (16>18), $\Delta 9$ (18>16), $\Delta 9$ (14-26), $\Delta 11$... (Rooney 2009)
Genetics of coloration in mice

1. *Mc1r* gene: melanocortin-1 receptor; single mutation in coding sequence (Hoekstra et al. 2006)

2. *Agouti* gene: inhibitor of melanocortin-1 receptor; variation in regulatory sequences (Steiner et al. 2007)

3. Epistatic interactions between alleles in *Mc1r* and *Agouti*

4. Mutations in 60 genes affect pigmentation in laboratory mouse (Hoekstra et al. 2006)
Lactate dehydrogenase in the killifish

Dalziel et al. 2009

1. Mutations both in coding and regulatory regions
2. Epistasis with other genes

"Northern" and "southern" alleles of LDH
<table>
<thead>
<tr>
<th>Gene</th>
<th>Function of product</th>
<th>Phenotype</th>
<th>Taxon</th>
<th>References</th>
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<tbody>
<tr>
<td>AVPR1A</td>
<td>Vasopressin receptor</td>
<td>Creative dance performance</td>
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<td>Avpr1a</td>
<td>Vasopressin receptor</td>
<td>Paternal care</td>
<td>Rodents</td>
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<td>Cyp56G1</td>
<td>P450 enzyme</td>
<td>Pesticide resistance</td>
<td>Fruitflies</td>
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<td>DARC</td>
<td>Chemokine receptor</td>
<td>Resistance to infection with malaria</td>
<td>Humans</td>
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<td>e</td>
<td>Pigment synthesis</td>
<td>Colour pattern of abdomen</td>
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<td>hsp70</td>
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<td>Thermal tolerance</td>
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<td>Aggressive behaviour</td>
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<td>Memory, emotional status</td>
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</table>

*Table 1: Cis-regulatory mutations with interesting phenotypic consequences*
Single aminoacid substitution and toxin resistance

Garter snake

Rough-skinned newt

Geffeney et al. 2005
Conclusions

• We know too little to make generalizations
• But examples are inspiring!!!
• Complex epistasis and polygenic traits are probably very common but there may be single genes with major effect (ex: ”loss-of-function” mutation)
• Changes both in regulatory and coding parts of genes; relative importance may depend on the trait character
• Ecological studies of model genetic organisms and developing molecular tools for ecologically interesting systems