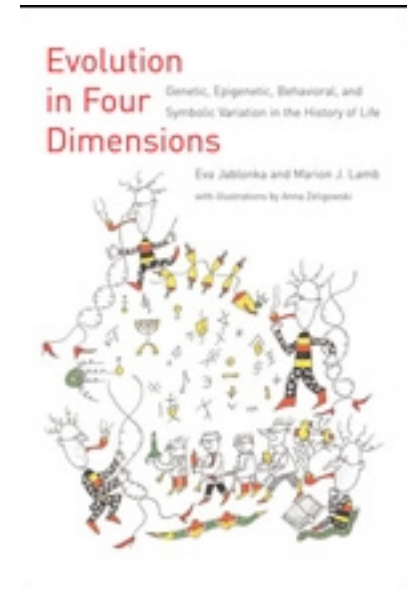
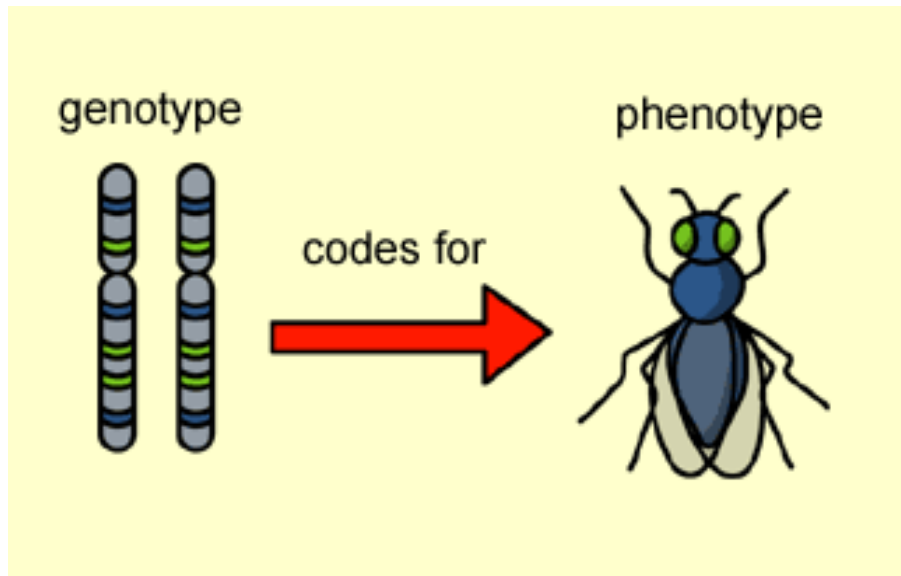


# Evolve fenotypu II



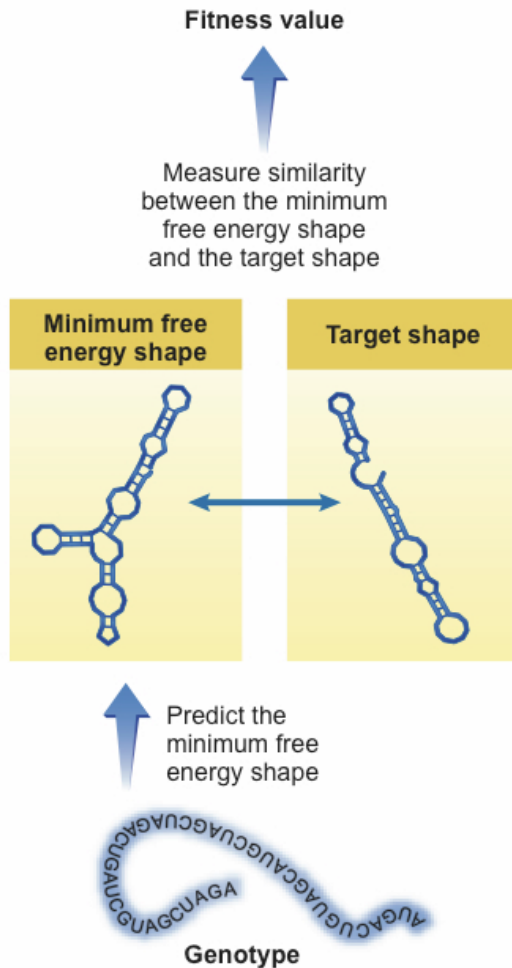
## Vztah genotyp → fenotyp

- plán? počítačový program? knihovna?
- genotypová astrologie (Jablonka a Lamb)

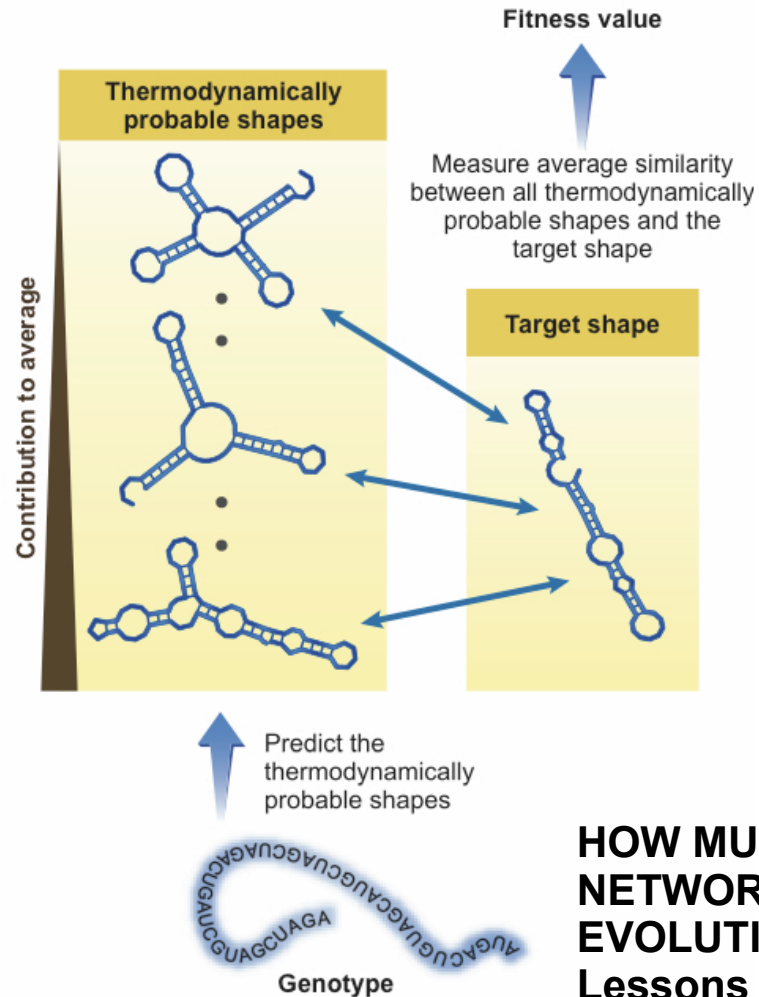


# Modely „RNA“ - různé vážení: A-U, G-C, G-U interakcí, penalizace za neodpovídající si báze

**a** “Simple” model



**b** “Plastic” model

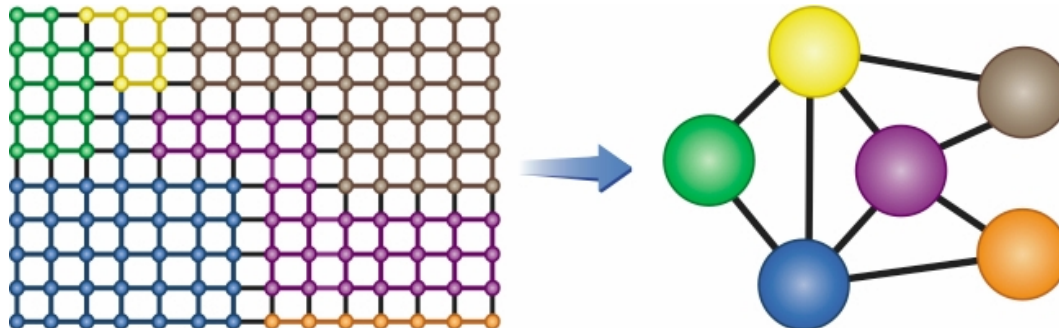


## HOW MUTATIONAL NETWORKS SHAPE EVOLUTION: Lessons from RNA Models

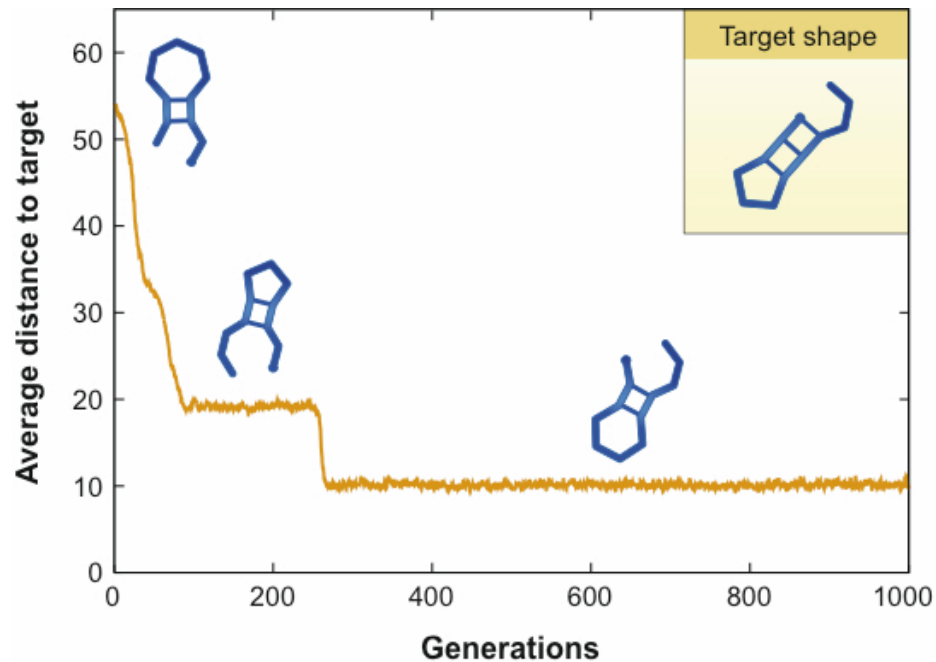
Annu. Rev. Ecol. Evol. Syst. 2007

## Modely „RNA“

- spousta genotypů odpovídá jednomu fenotypu (oblasti neutrality)
- pravděpodobný směr změny fenotypu záleží na genotypu (historické náhodě)



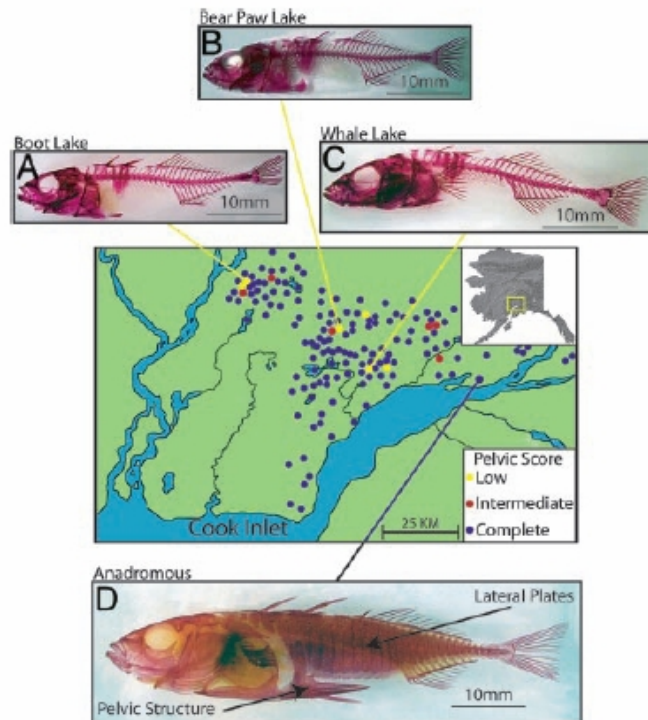
- zpravidla punktualistická evoluce



## Cvičení z genetiky pro zahřátí

### Parallel genetic basis for repeated evolution of armor loss in Alaskan threespine stickleback populations

William A. Cresko<sup>1\*</sup>, Angel Amores<sup>2</sup>, Catharina V. <sup>PNAS</sup> | April 20, 2004 | vol. 181 | no. 16 | Patrick Phillips<sup>1</sup>, Michael A. Bell<sup>1</sup>, Charles B. Kimmel<sup>3</sup>, and John H. Postlethwait<sup>4,5</sup>



**Fig. 1.** Map of the Matanuska-Susitna (Mat-Su) Valley of Alaska with trypsin cleared and Alizarin red stained stickleback showing Boot Lake (A), Bear Paw Lake (B) and Whale Lake (C) freshwater low-armor populations, and anadromous Rabbit Slough complete-armor ancestor (D). The complete-armor anadromous form has a full set of lateral plates and fully formed pelvic structure (arrows), whereas each freshwater population lacks most of the lateral plates and has either highly reduced (Boot, Bear Paw) or completely absent (Whale) pelvic structures. Blue dots indicate additional populations with mostly complete pelvic structure (mean score of 5.0), red are mostly intermediate (2.0–4.0), and yellow are low (0.0–2.0).

- Jsou rozdíly v brnění a přítomnosti pánevních trnů důsledkem fenotypové plasticity?

- Liší se populace koljušek v genetické kontrole brnění a pánevních trnů? Jsou kontrolovány dominantními alelami? Liší se jednotlivé populace v těchto lokusech?

- Kolika lokusy jsou kontrolovány rozdíly v brnění a přítomnosti pánevních trnů u koljušek?

- Jsou „pánev“ a brnění kontrolovány jinými lokusy?

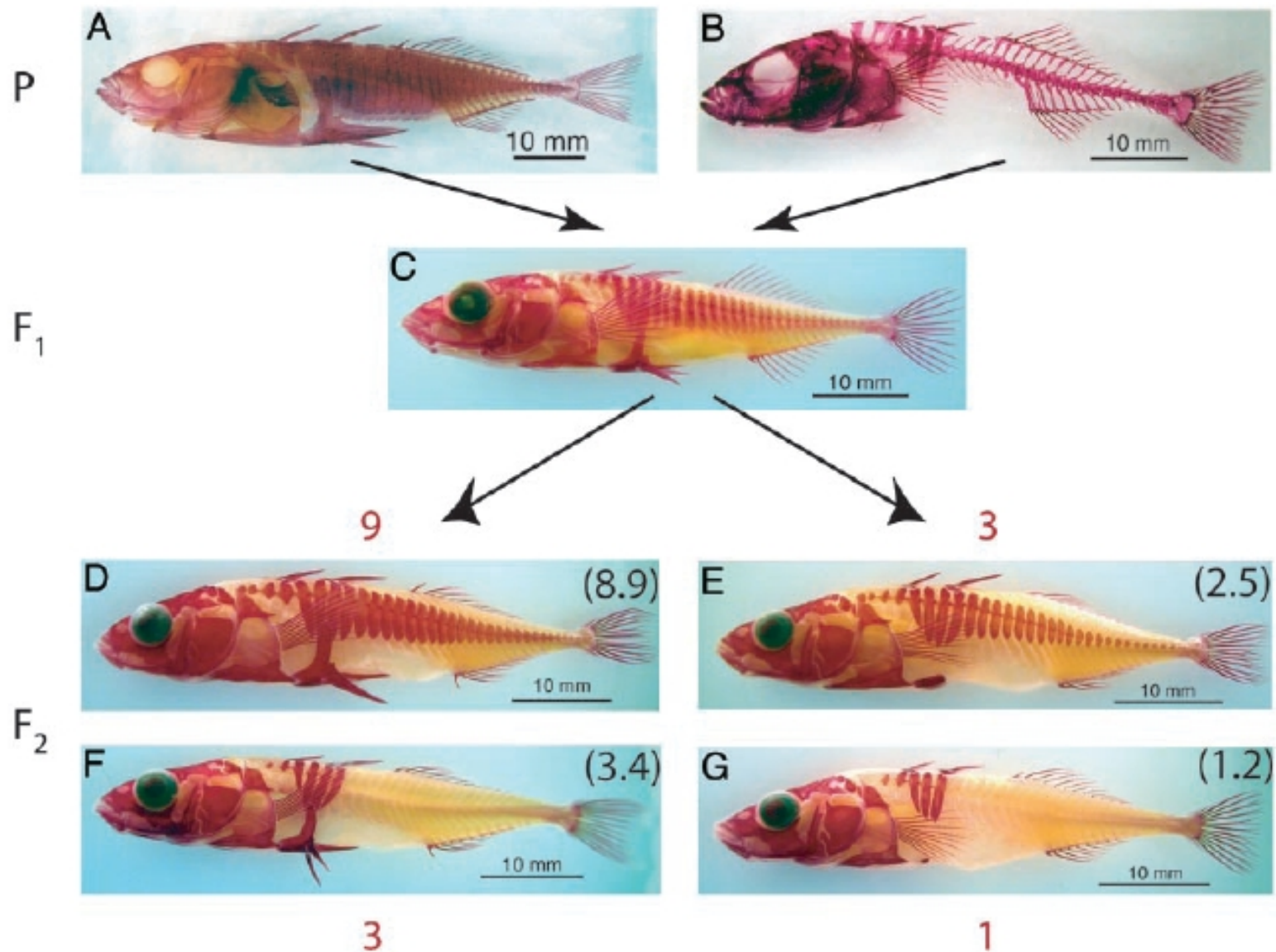
**Table 1. Variation in lateral plate number and pelvic score across populations and cross types**

Type of collection or cross		n	Lateral plates		n	Pelvic structure	
			Left	Right		Left	Right
Parental Anadromous	Wild	116	<b>33.2 (0.06)</b>	<b>33.2 (0.07)</b>	116	<b>5.0 (0.00)</b>	<b>5.0 (0.00)</b>
	Laboratory	115	<b>33.1 (0.06)</b>	<b>33.1 (0.06)</b>	115	<b>5.0 (0.00)</b>	<b>5.0 (0.00)</b>
Parental Bear Paw Lake	Wild	102	3.9 (0.08)	3.9 (0.08)	102	1.0 (0.08)	0.7 (0.07)
	Laboratory	131	3.8 (0.08)	3.8 (0.08)	131	1.0 (0.09)	0.5 (0.05)
Parental Boot Lake	Wild	106	5.2 (0.08)	5.2 (0.08)	106	0.9 (0.10)	0.9 (0.09)
	Laboratory	135	4.8 (0.08)	4.7 (0.07)	135	1.0 (0.08)	0.8 (0.06)
Parental Whale Lake	Wild	105	2.3 (0.12)	2.2 (0.12)	105	0.5 (0.05)	0.2 (0.04)
	Laboratory	38	4.6 (0.09)	4.6 (0.12)	38	0.1 (0.03)	0.0 (0.00)
F <sub>1</sub> complementation	Bear Paw-by-Boot	225	5.4 (0.06)	5.5 (0.06)	225	2.0 (0.12)	1.1 (0.07)
	Boot-by-Whale	57	4.9 (0.15)	4.7 (0.15)	57	0.8 (0.15)	0.5 (0.11)
F <sub>1</sub> mapping	Bear Paw-by-Anadromous	50	<b>32.1 (0.13)</b>	<b>32.3 (0.15)</b>	50	<b>5.0 (0.00)</b>	<b>5.0 (0.04)</b>
	Boot-by-Anadromous	33	<b>31.4 (0.17)</b>	<b>31.4 (0.23)</b>	33	<b>5.0 (0.00)</b>	<b>5.0 (0.03)</b>
	Whale-by-Anadromous	42	<b>32.2 (0.13)</b>	<b>32.3 (0.16)</b>	42	<b>5.0 (0.00)</b>	<b>5.0 (0.02)</b>
F <sub>2</sub> mapping	Bear Paw complete armor	281	<b>32.6 (0.07)</b>	<b>32.6 (0.08)</b>	324	<b>5.0 (0.00)</b>	<b>5.0 (0.00)</b>
	Bear Paw low armor	123	6.7 (0.17)	6.6 (0.19)	88	2.6 (0.18)	2.2 (0.19)
	Boot complete armor	264	<b>32.5 (0.08)</b>	<b>32.5 (0.07)</b>	296	<b>5.0 (0.00)</b>	<b>5.0 (0.00)</b>
	Boot low armor	99	7.1 (0.23)	7.2 (0.36)	70	2.7 (0.19)	2.4 (0.20)
	Whale complete armor	191	<b>32.6 (0.10)</b>	<b>32.5 (0.19)</b>	207	<b>5.0 (0.00)</b>	<b>5.0 (0.00)</b>
	Whale low armor	81	5.7 (0.13)	5.7 (0.15)	75	2.0 (0.16)	1.6 (0.15)

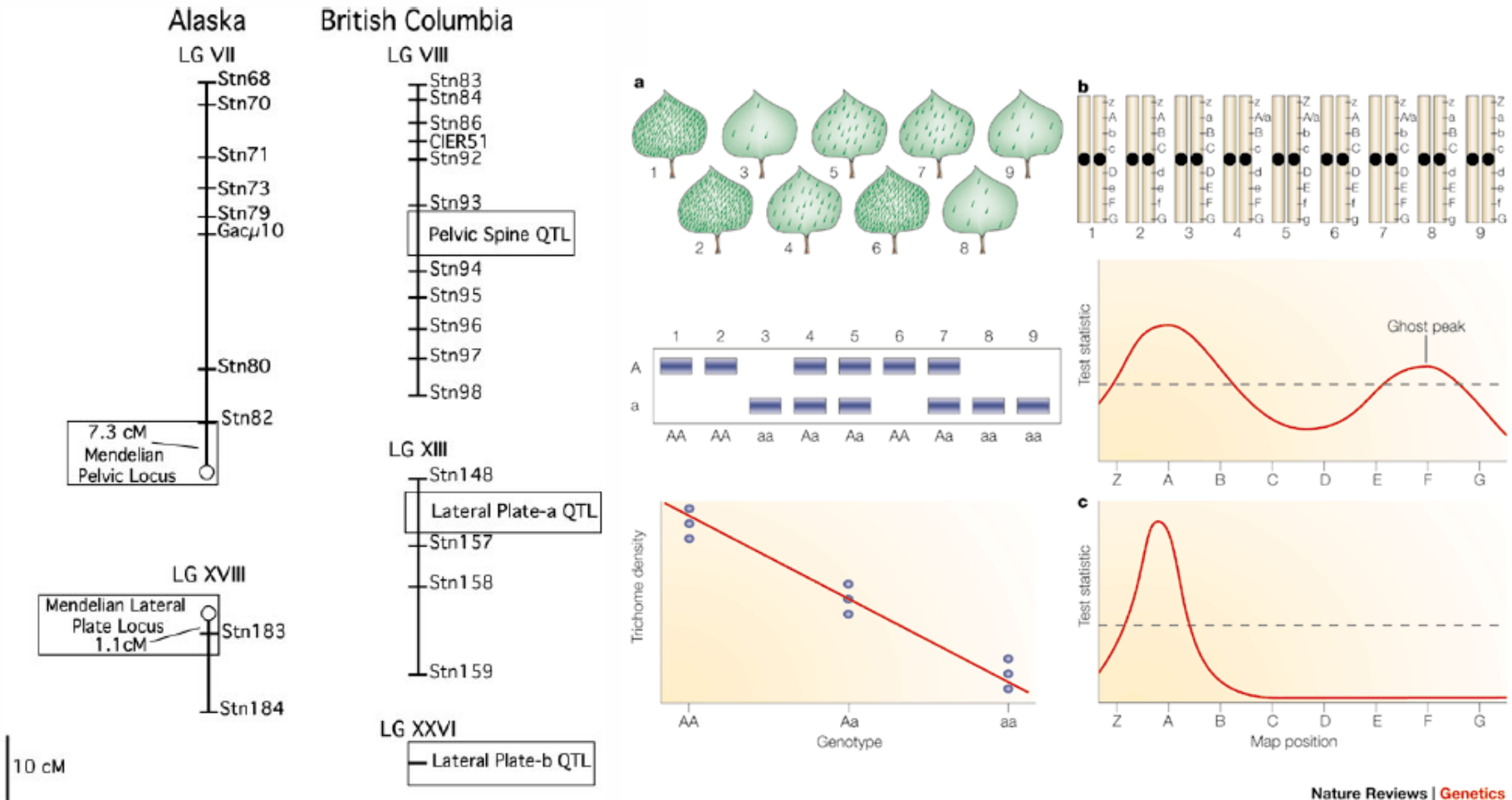
Mean (SE) numbers of lateral plates and pelvic scores on the left and right side, along with number of individuals sampled (*n*) for wild-caught and laboratory-bred and reared individuals, crosses between different low-armor populations (F<sub>1</sub> complementation), hybrid F<sub>1</sub> individuals produced by crossing low-to complete-armor fish (F<sub>1</sub> mapping), and the offspring of full-sib crosses between F<sub>1</sub> mapping individuals divided between complete and low lateral plate and pelvic classes (F<sub>2</sub> mapping). Bold indicates scores within the complete-armor class.

- Jsou rozdíly v brnění a přítomnosti pánevních trnů důsledkem fenotypové plasticity? ■
- Liší se populace koljušek v genetické kontrole brnění a pánevních trnů? ■
- Jsou kontrolovány dominantními alelami? Liší se jednotlivé populace v těchto lokusech? ■
- Kolika lokusy jsou kontrolovány rozdíly v brnění a přítomnosti pánevních trnů u koljušek? ■
- Jsou „pánev“ a brnění kontrolovány jinými lokusy? ➔

- Jsou „pánev“ a brnění kontrolovány jinými lokusy?



# - A jakými lokusy? QTL mapping



**Fig. 5.** Positions of Alaskan Mendelian lateral plate (Mendelian Lateral Plate Locus) and pelvic (Mendelian Pelvic Locus) loci on the stickleback linkage map (22). Alaskan Mendelian plate and pelvic loci localized to linkage group (LG) VII and LGXVIII, respectively. The Alaskan lateral plate and pelvic locus maps 1.1 centiMorgans (cM) away from Stn183, and the pelvic phenotype locus maps 7.3 cM from marker Stn82. Open boxes on the right side of the figure indicate the position of QTLs important for lateral plate and pelvic spine variation in a cross between low-plate, complete-pelvic stickleback species from British Columbia (22).



# A jakými lokusy? Kandidátské geny

MC1R

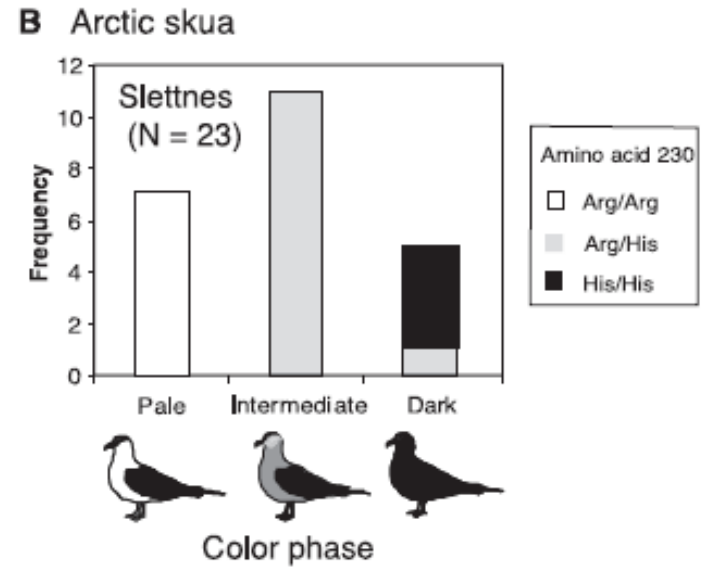
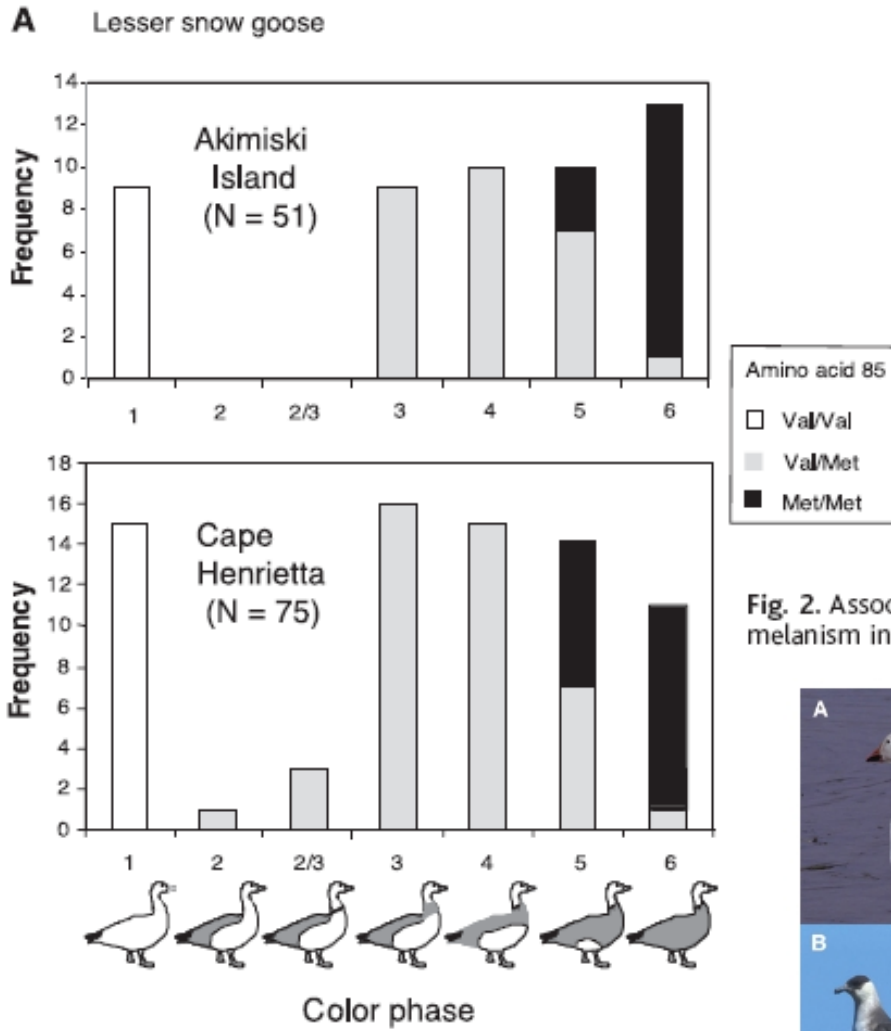
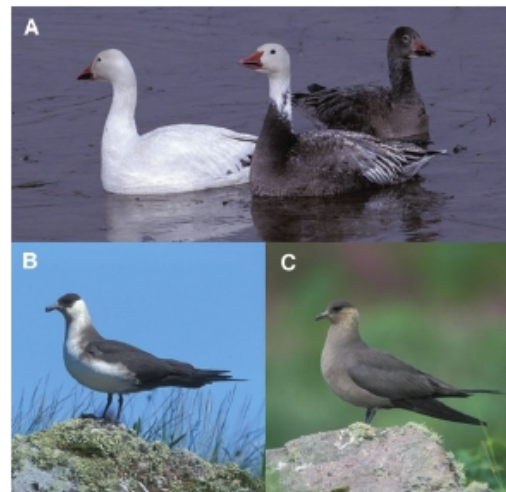


Fig. 2. Association between copy number of variant *MC1R* alleles and degree of melanism in lesser snow geese (A) and arctic skuas (B).



Conserved Genetic Basis of a Quantitative Plumage Trait Involved in Mate Choice

Nicholas I. Mundy,<sup>1\*</sup> Nichola S. Badcock,<sup>2</sup> Tom Hart,<sup>2</sup> Kim Scribner,<sup>3</sup> Kirstin Janssen,<sup>4</sup> Nicola J. Nadeau<sup>1</sup>

19 MARCH 2004 VOL 303 SCIENCE

# Je náhoda, že došlo k mutacím v tomto jednom genu (receptoru pro MSH - melanocyt stimulující hormon)?

*Evolution*, 58(8), 2004, pp. 1794–1808

## ADAPTIVE REPTILE COLOR VARIATION AND THE EVOLUTION OF THE *MC1R* GENE

ERICA BREE ROSENBLUM,<sup>1,2</sup> HOPI E. HOEKSTRA,<sup>3,4</sup> AND MICHAEL W. NACHMAN<sup>3,5</sup>

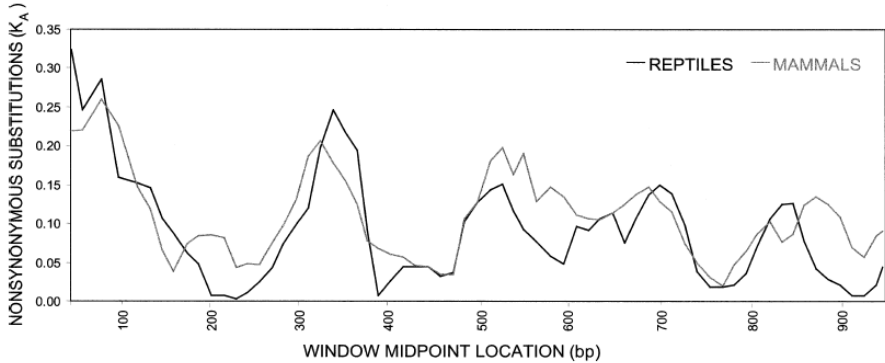
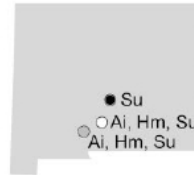


FIG. 3. Evolutionary constraint across *Mc1r* in reptiles and mammals. Sliding window comparison of nonsynonymous substitution rate ( $K_A$ ) for mammals and reptiles across all regions of the *Mc1r* gene.

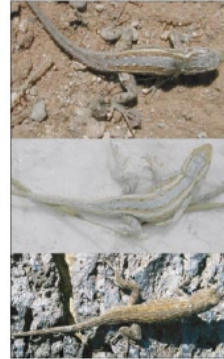
### SOUTHERN NEW MEXICO



*Aspidoscelis inornata*



*Holbrookia maculata*



*Sceloporus undulatus*

### SOUTHERN CALIFORNIA



*Anniella pulchra*



*Phrynosoma platyrhinos*



*Uta stansburiana*

### GREAT LAKES REGION



*Thamnophis sirtalis*

100 mi

Co jsou funkčně synonymní a nesynonymní mutace?

Změny v lokusech ano, ale jaké?

- změna v kódující sekvenci

# Evoluční novinky

*Jak pracuje evoluce*

**LUKÁŠ  
KRATOCHVÍL**

Vesmír 88, září 2009



## Evolution at Two Levels: On Genes and Form

Sean B. Carroll



PLoS Biology | [www.plosbiology.org](http://www.plosbiology.org)

July 2005 | Volume 3 | Issue 7

# Změna v kódující sekvenci

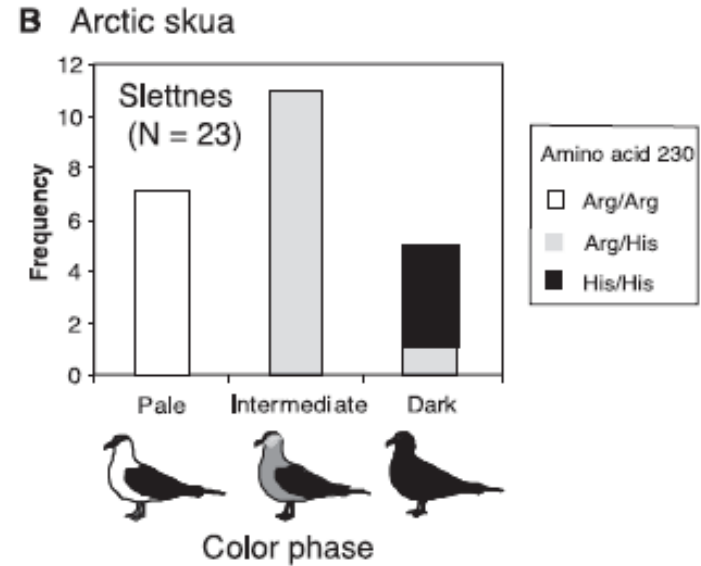
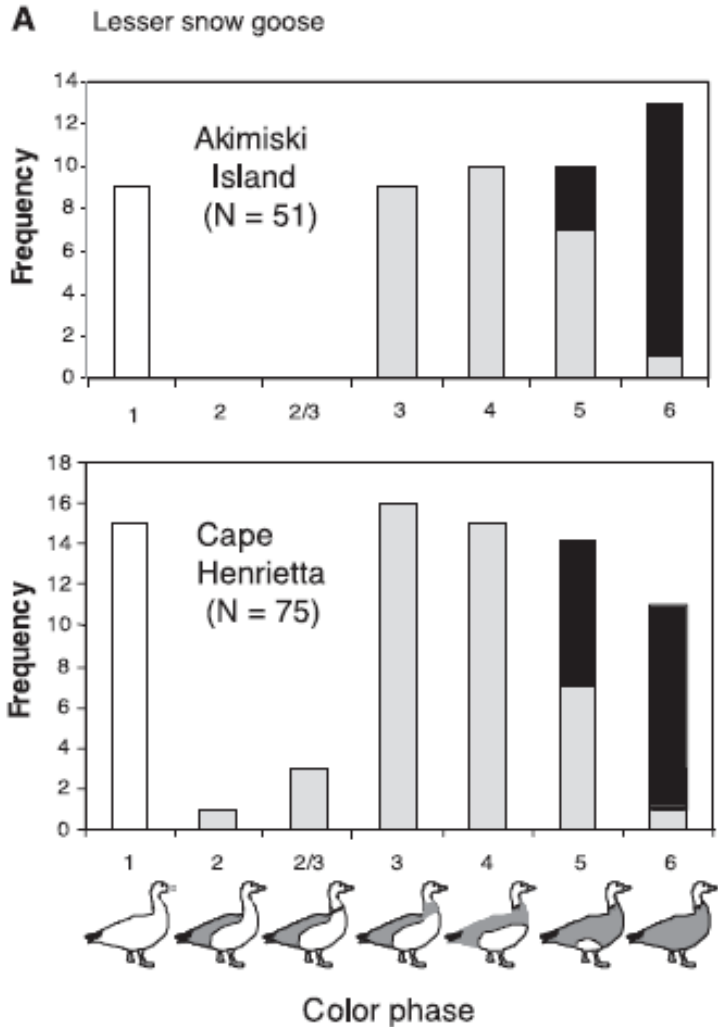
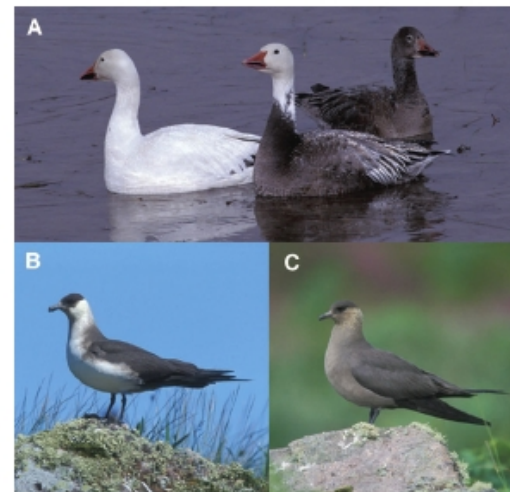


Fig. 2. Association between copy number of variant *MC1R* alleles and degree of melanism in lesser snow geese (A) and arctic skuas (B).



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19 MARCH 2004 VOL 303 SCIENCE

## THE LOCUS OF EVOLUTION: EVO DEVO AND THE GENETICS OF ADAPTATION

# Molecular origins of rapid and continuous morphological evolution

John W. Fondon III\* and Harold R. Garner

„Repeat expansions or contractions vary in a locus-specific manner and occur at rates up to 100,000 times higher than point mutations, because of the distinct mutational mode of slipped-strand mispairing rather than an incorporation error“

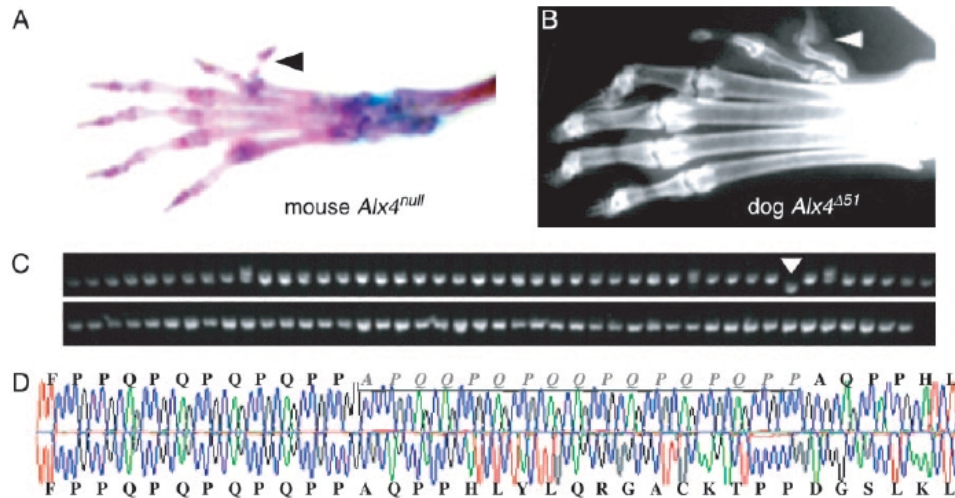


Fig. 1. Large magnitude repeat length mutations can result in gross morphological change. (A) *Alx4*<sup>-/-</sup> mice exhibit a duplication of the first digit (arrowhead). [Adapted with permission from ref. 18 (Copyright 1998, The Company of Biologists).] (B) A radiograph of the rear paw of a Great Pyrenees shows the typical double dewclaw phenotype specified in the breed standard (arrowhead). (C) Polydactylous Great Pyrenees are homozygous for a 51-nucleotide repeat contraction in the *Alx4* gene. PCR amplification of the repeat-containing regions of *Alx4* from 89 dog breeds reveals that this deletion is unique to the Great Pyrenees breed (arrow). Phenotypically normal basset hounds, flat-coated retrievers, and harriers were heterozygous for distinct two amino acid insertions (doublets). (D) DNA sequencing reveals that the deletion is caused by a contraction of the PQ<sub>n</sub> repeat that results in the removal of 17 aa within the repeat.



Table 2. Alleles with modest alterations in developmental regulator gene coding repeat length are common in natural populations of placental mammals

Species	Panel size	No. of <i>Runx-2</i> alleles	No. of <i>Dlx-2</i> alleles	No. of <i>Twist-1</i> alleles
<i>Canis lupus</i> (gray wolf)	70	5	5	16
<i>Canis latrans</i> (coyote)	98	7	5	17
<i>Vulpes vulpes</i> (red fox)	10	4	1	1
<i>Vulpes velox</i> (swift fox)	1	1	2	2
<i>Alopex lagopus</i> (Arctic fox)	2	2	3	3
<i>Enhydra lutris</i> (sea otter)	3	2	2	3
<i>Lontra canadensis</i> (river otter)	1	2	1	2
<i>Ursus maritimus</i> (polar bear)	2	1	1	2
<i>Ursus arctos</i> (brown bear)	2	1	1	1
<i>Odobenus rosmarus</i> (walrus)	2	2	2	1
<i>Sylvilagus floridanus</i> (Eastern cottontail rabbit)	2	1	2	1
<i>Lasiorycteris noctivagans</i> (gray-eared bat)	1	2	2	1

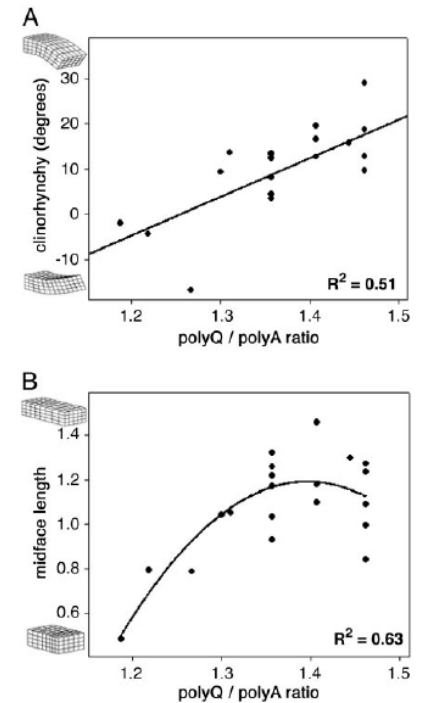


Fig. 2. Tandem repeat length in a developmental gene is quantitatively correlated with continuous morphological features. (A and B) Reported (9, 11) effects on transcription of polyglutamine and polyalanine repeats suggested that these two domains may be involved in competitive activities and that the relative lengths of these domains may be more instructive than their aggregate length. A Pearson correlation test of this hypothesis revealed a significant correlation between *Runx-2* polyglutamine to polyalanine ratio and clinorhynchus (D/V nose bend,  $P = 0.0001$ , Pearson one-sided significance,  $n = 27$ , A) and midface length ( $P = 0.0002$ ,  $n = 27$ , B) (24). The nature and direction of these correlations is indicative of longer relative *Runx-2* glutamine repeats resulting in increased midface growth, consistent with observations from human cleidocranial dysplasia patients (25). Published studies (9–16) indicate that amino acid repeat length-function relationships are typically nonlinear; however, fitting a quadratic or exponential to the clinorhynchus data (A) does not provide sufficient improvement in residuals to support the use of a nonlinear function over a simple line.

# Změny v lokusech ano, ale jaké?

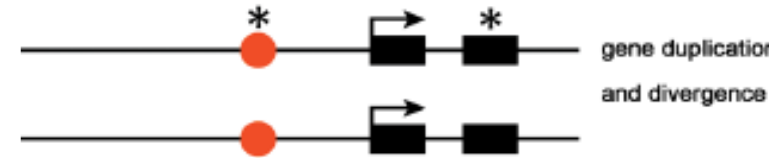
## Duplikace genů

Susumu Ohno: *Evolution by Gene Duplication*

- málo pleiotropních následků (srv. *MC1R*)

- frekvence genové duplikace je asi 1/100 000 000 let na gen  
(Lynch a Conery *Science* 2000)

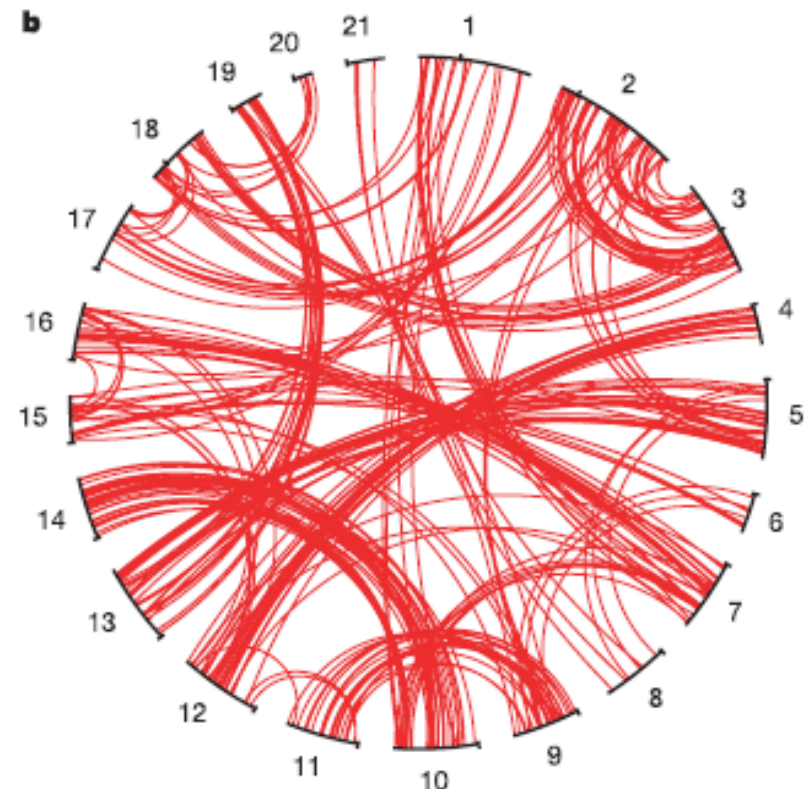
- genomová duplikace



## Genome duplication in the teleost fish *Tetraodon nigroviridis* reveals the early vertebrate proto-karyotype

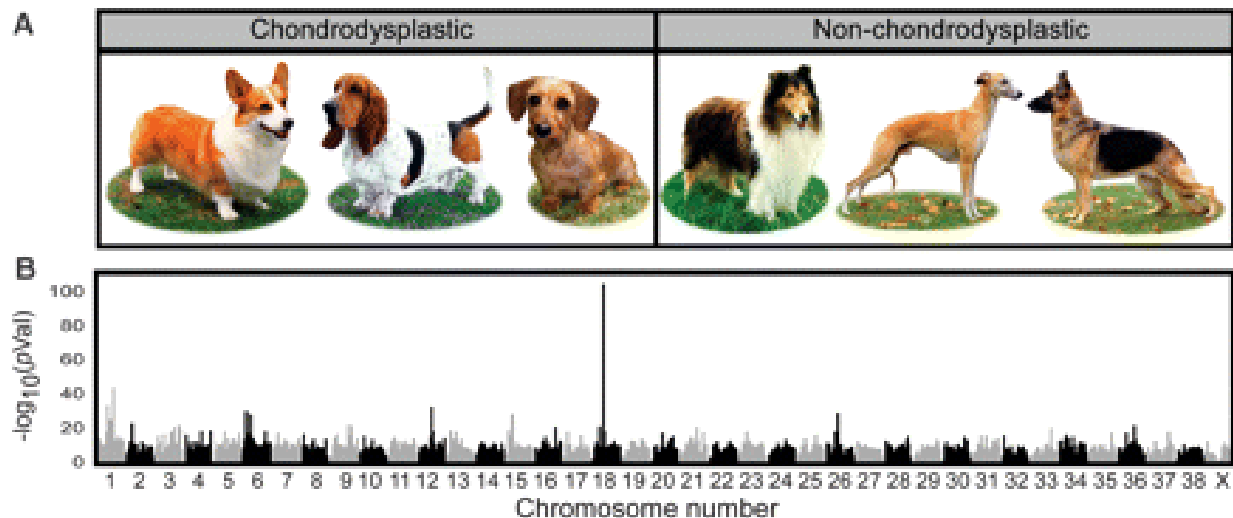
Olivier Jaillon<sup>1</sup>, Jean-Marc Aury<sup>1</sup>, Frédéric Brunet<sup>2</sup>, Jean-Louis Petit<sup>1</sup>, Nicole Stange-Thomann<sup>3</sup>, Evan Mauceli<sup>3</sup>, Laurence Bouneau<sup>1</sup>, Cécile Fischer<sup>1</sup>, Catherine Ozouf-Costaz<sup>4</sup>, Alain Bernot<sup>1</sup>, Sophie Nicaud<sup>1</sup>, David Jaffe<sup>3</sup>, Sheila Fisher<sup>3</sup>, Georges Lutfalla<sup>5</sup>, Carole Dossat<sup>1</sup>, Béatrice Segurens<sup>1</sup>, Corinne Dasilva<sup>1</sup>, Marcel Salanoubat<sup>1</sup>, Michael Levy<sup>1</sup>, Nathalie Boudet<sup>1</sup>, Sergi Castellano<sup>6</sup>, Véronique Anthonard<sup>1</sup>, Claire Jubin<sup>1</sup>, Vanina Castelli<sup>1</sup>, Michael Katinka<sup>1</sup>, Benoît Vacherie<sup>1</sup>, Christian Biéumont<sup>7</sup>, Zineb Skalli<sup>1</sup>, Laurence Cattolico<sup>1</sup>, Julie Poulain<sup>1</sup>, Véronique de Berardinis<sup>1</sup>, Corinne Cruaud<sup>1</sup>, Simone Duprat<sup>1</sup>, Philippe Brottier<sup>1</sup>, Jean-Pierre Coutanceau<sup>4</sup>, Jérôme Gouzy<sup>8</sup>, Genis Parra<sup>6</sup>, Guillaume Lardier<sup>1</sup>, Charles Chapple<sup>6</sup>, Kevin J. McKernan<sup>9</sup>, Paul McEwan<sup>9</sup>, Stephanie Bosak<sup>9</sup>, Manolis Kellis<sup>3</sup>, Jean-Nicolas Volff<sup>10</sup>, Roderic Guigó<sup>6</sup>, Michael C. Zody<sup>3</sup>, Jill Mesirov<sup>3</sup>, Kerstin Lindblad-Toh<sup>3</sup>, Bruce Birren<sup>3</sup>, Chad Nusbaum<sup>3</sup>, Daniel Kahn<sup>8</sup>, Marc Robinson-Rechavi<sup>2</sup>, Vincent Laudet<sup>2</sup>, Vincent Schachter<sup>1</sup>, Francis Quétier<sup>1</sup>, William Saurin<sup>1</sup>, Claude Scarpelli<sup>1</sup>, Patrick Wincker<sup>1</sup>, Eric S. Lander<sup>3,11</sup>, Jean Weissenbach<sup>1</sup> & Hugues Roest Crolius<sup>1\*</sup>

NATURE | VOL 431 | 21 OCTOBER 2004



# An Expressed *Fgf4* Retrogene Is Associated with Breed-Defining Chondrodysplasia in Domestic Dogs

Heidi G. Parker,<sup>1</sup> Bridgett M. VonHoldt,<sup>2</sup> Pascale Quignon,<sup>1</sup> Elliott H. Margulies,<sup>3</sup> Stephanie Shao,<sup>1</sup> Dana S. Mosher,<sup>1</sup> Tyrone C. Spady,<sup>1</sup> Abdel Elkahloun,<sup>1</sup> Michele Cargill,<sup>4\*</sup> Paul G. Jones,<sup>5</sup> Cheryl L. Maslen,<sup>6</sup> Gregory M. Acland,<sup>7,8</sup> Nathan B. Sutter,<sup>8</sup> Keiichi Kuroki,<sup>9</sup> Carlos D. Bustamante,<sup>10</sup> Robert K. Wayne,<sup>2</sup> Elaine A. Ostrander<sup>1†</sup>

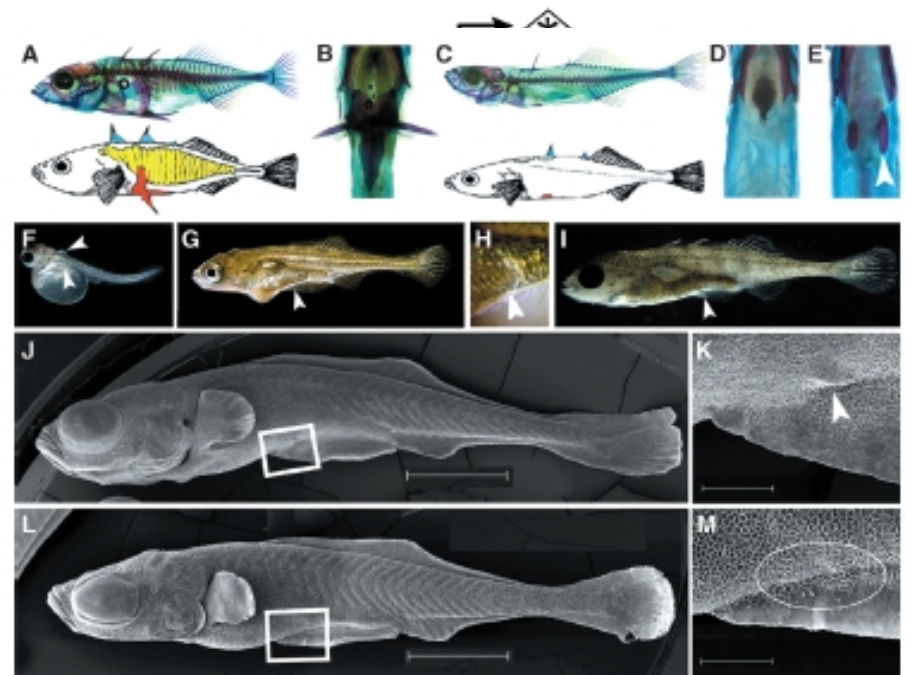
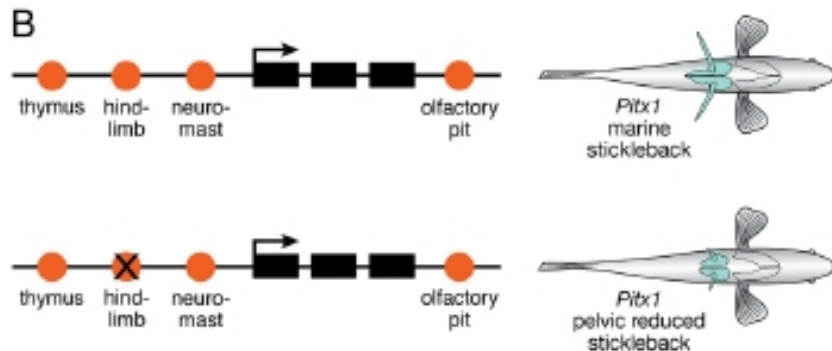


# Změny v lokusech ano, ale jaké? cis-regulace (změny v promotoru)

## Expression of limb initiation genes and clues to the morphological diversification of threespine stickleback

Nicholas J. Cole<sup>1</sup>, Mikiko Tanaka<sup>1,2</sup>, Alan Prescott<sup>1</sup> and Cheryl Tickle<sup>1</sup>

Current Biology Vol 13 No 24  
R952



Current Biology

Figure 1. Skeletal anatomy (bone in red) and embryology of paired fin development in spined and spine-deficient stickleback.

(A) Lateral view of spined adult and diagram highlighting skeletal elements; blue, dorsal spines; yellow, lateral plates; red, pelvic girdle and spine. Modified from [1]. (B) Ventral view of pelvic region of spined adult showing girdle and spines. (C) Lateral view of spine-deficient adult and diagram. Modified from [1]. (D and E) Ventral view of pelvic region of two spine-deficient fish showing complete absence of pelvic structures (D) and tear shaped bones (E). Note asymmetry in (E) — left bone (arrowhead) larger than right. (F) Hatched spined fish (lateral view). Pectoral fins present (arrowheads) but no pelvic fins. Pectoral fin buds also develop in spine-deficient fish at same age. (G and H) Spined 5 mm TL larva. (G) Lateral view. (H) Detail of pelvic region with transparent fin bud (arrowhead). (I) Lateral view of 10 mm TL spined juvenile; arrowhead indicates pelvic spine. (J–M) Scanning EMs. (J) Spined larva, scale bar 750  $\mu$ m. (K) Detail of pelvic region in (J), arrowhead indicates developing pelvic spine, scale bar 200  $\mu$ m. (L) Spine-deficient larva, scale bar 750  $\mu$ m. (M) Detail of pelvic region in (L), showing no evidence of pelvic spine (circled), scale bar 200  $\mu$ m.



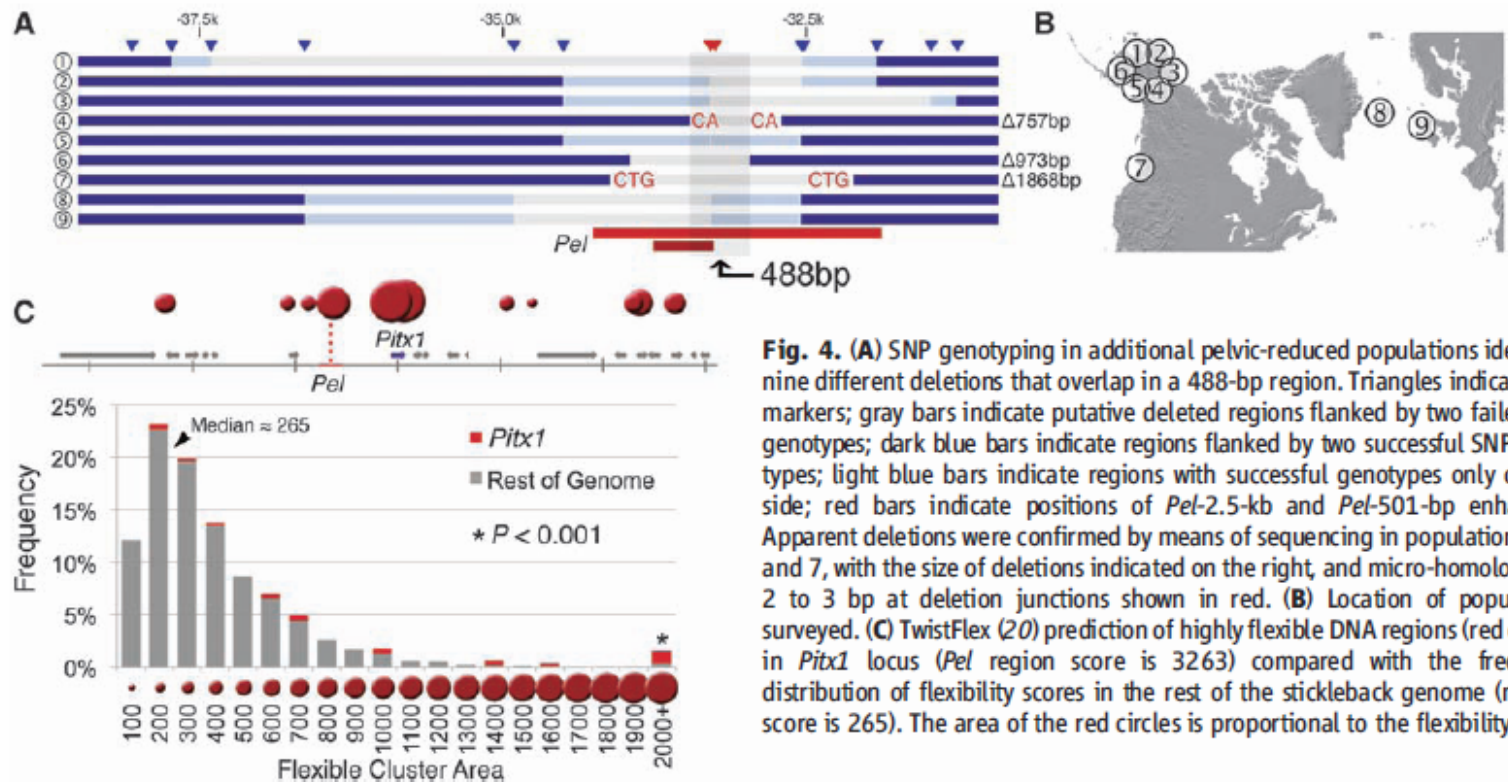
Změny v lokusech ano, ale jaké?  
*cis*-regulace (změny v promotoru)



# Adaptive Evolution of Pelvic Reduction in Sticklebacks by Recurrent Deletion of a *Pitx1* Enhancer

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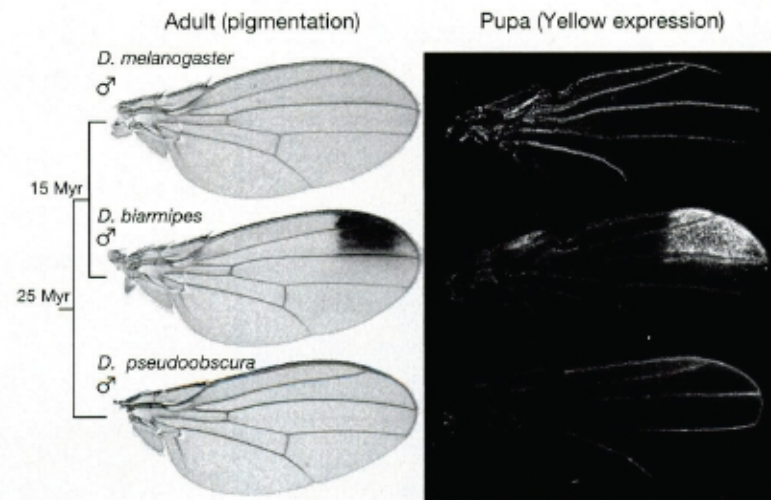
**Fig. 4.** (A) SNP genotyping in additional pelvic-reduced populations identifies nine different deletions that overlap in a 488-bp region. Triangles indicate SNP markers; gray bars indicate putative deleted regions flanked by two failed SNP genotypes; dark blue bars indicate regions flanked by two successful SNP genotypes; light blue bars indicate regions with successful genotypes only on one side; red bars indicate positions of *Pel*-2.5-kb and *Pel*-501-bp enhancers. Apparent deletions were confirmed by means of sequencing in populations 4, 6, and 7, with the size of deletions indicated on the right, and micro-homologies of 2 to 3 bp at deletion junctions shown in red. (B) Location of populations surveyed. (C) TwistFlex (20) prediction of highly flexible DNA regions (red circles) in *Pitx1* locus (*Pel* region score is 3263) compared with the frequency distribution of flexibility scores in the rest of the stickleback genome (median score is 265). The area of the red circles is proportional to the flexibility score.

## Evolve *cis*-regulace – často koopce už existujících signálů

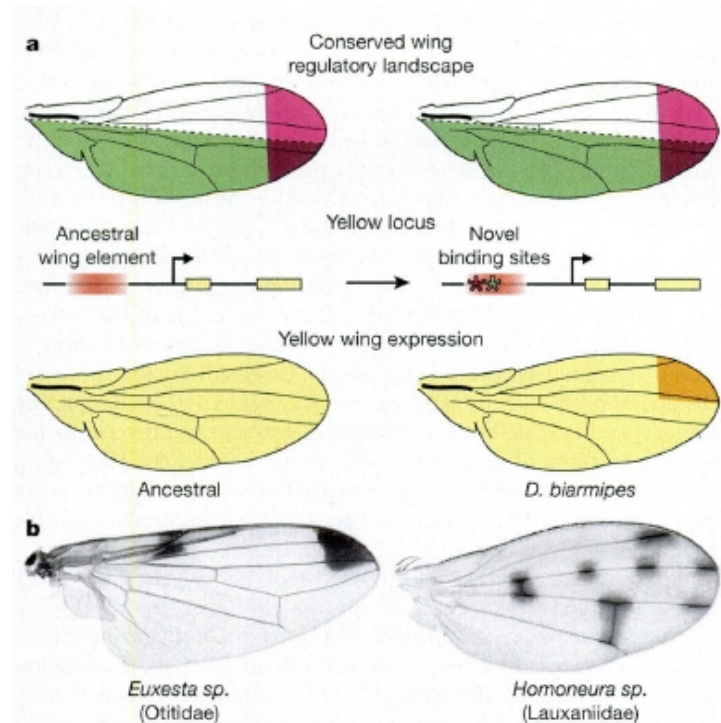
# Chance caught on the wing: *cis*-regulatory evolution and the origin of pigment patterns in *Drosophila*

Nicolas Gompel<sup>†</sup>, Benjamin Prud'homme<sup>\*</sup>, Patricia J. Wittkopp<sup>†</sup>, Victoria A. Kassner & Sean B. Carroll

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**Figure 1** Expression of the Yellow protein prefigures adult wing pigmentation. The conspicuous spot of dark pigmentation present at the tip of the male wing of *Drosophila biarmipes* (left) is a new trait evolved among species of the *Drosophila melanogaster* group<sup>14,46</sup> (about 15 Myr of divergence; divergence time is 60–80 Myr for the family Drosophilidae<sup>30</sup>), superimposed on the ancestral pattern of uniform grey shading and darker veins found both in *D. melanogaster* and in *D. pseudoobscura*, a species from the sister *D. obscura* group (25 Myr of divergence<sup>29,30</sup>). In all three species the male pupal distribution of Yellow in the wing, revealed by a specific antibody (right), foreshadows the adult pigmentation.



**Figure 6** Cryptic prepatterns and the evolution of novel gene expression patterns through the evolution of *cis*-regulatory sequences. **a**, The upper panel shows a model of the conserved landscape of transcriptional regulators that pattern and shape the *Drosophila* wing (green and pink represent repressor and activator, respectively). The evolution of binding sites for a subset of these regulators in the *yellow* wing *cis*-regulatory element (coloured stars) co-opts them to modify *yellow* expression (lower panel). Combined with other regulatory changes at other loci, the changes at the *y* locus result in a novel pigmentation spot. **b**, Wing pigmentation patterns similar to *D. biarmipes* (left) or *D. guttifera* (right) evolved independently in other fly families (here Otitidae and Lauxaniidae).

# Spor o důležitost jednotlivých mechanismů v evoluci

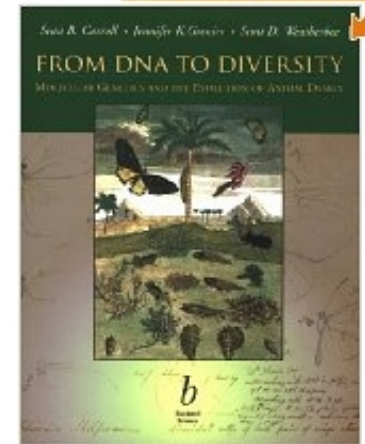
- Sean Carroll
  - Jerry Coyne, Hopi Hoekstra
- ?transkripční faktory? promotory? strukturní geny?

## THE LOCUS OF EVOLUTION: EVO DEVO AND THE GENETICS OF ADAPTATION

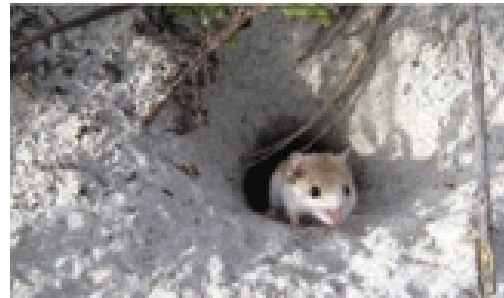
Hopi E. Hoekstra<sup>1,2</sup> and Jerry A. Coyne<sup>3,4</sup>

*Evolution* 61-5: 995–1016

Click to **LOOK INSIDE!**



Greg Wray and Jerry Coyne





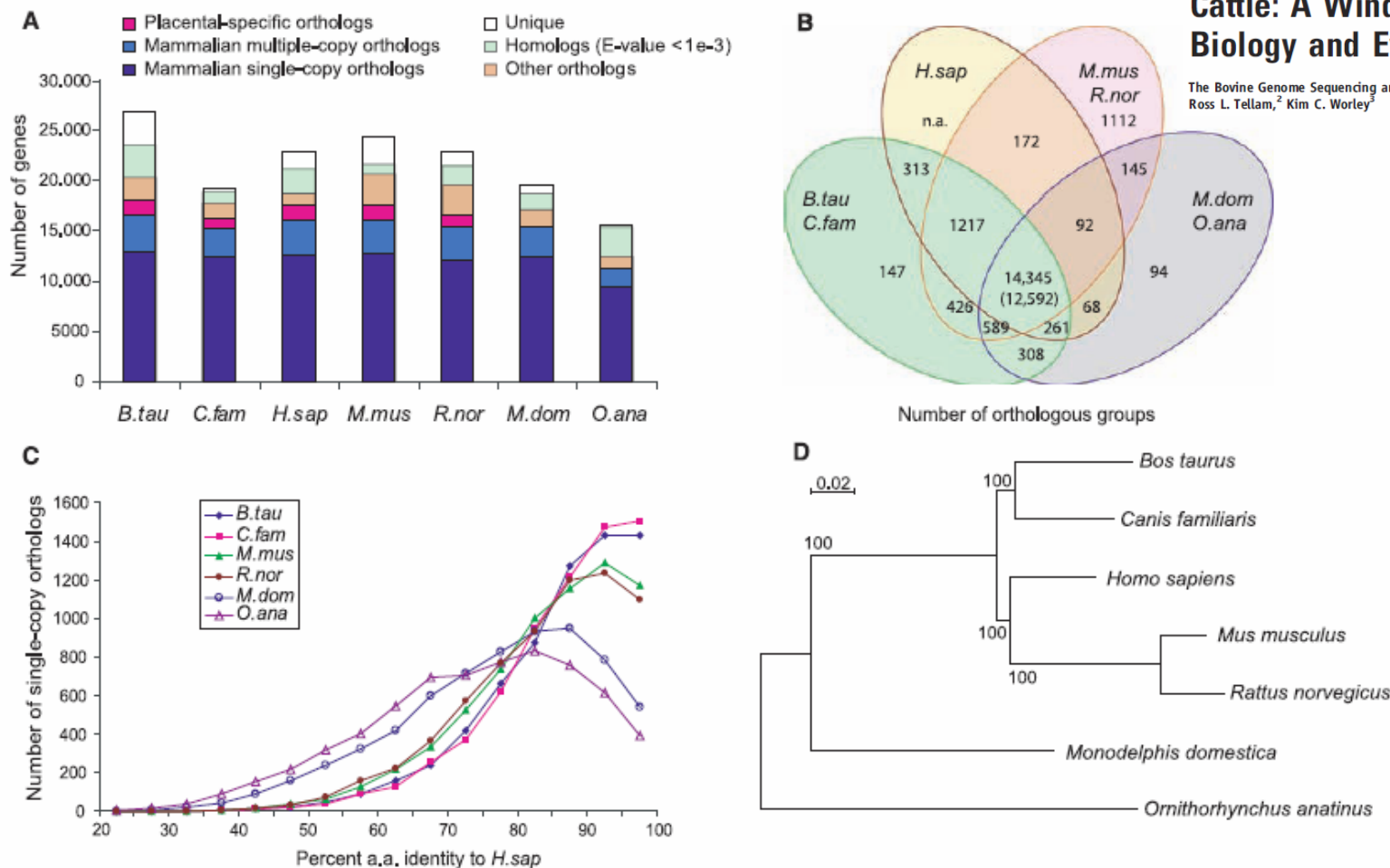
## „Sean Carroll vs. Jerry Coyne“



- Jacob a Monod objevili zásadní mechanismus, ten lze použít i na vysvětlení vzniku evolučních novinek
- všichni máme stejné strukturní geny (99-% šimpanzi: King a Wilson 1975) - pleiotropie
- duplikace příliš vzácná
- změny v transkripčních faktorech příliš mnoho pleiotropních efektů
- *cis*-regulace je řešením
- Carroll mluví jen o morfologii, kde se toho moc neví – proč by měla být jiná než fyziologie a chování?
- Je složité říct, co je strukturní gen a co regulační (histony)
- duplikace zas tak vzácná není
- pravděpodobně 21% lidských protein-kódujících genů nemá homologa u myši (a 6 % u šimpanze)
- 80% proteinů se mezi člověkem a šimpanzem liší aspoň v jedné aminokyselině
- nevíme, co je v evoluci především důležité, ale spíš změny ve strukturních genech

# The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution

The Bovine Genome Sequencing and Analysis Consortium,<sup>1</sup> Christine G. Elsik,<sup>1</sup> Ross L. Tellam,<sup>2</sup> Kim C. Worley<sup>3</sup>



**Fig. 1.** Protein orthology comparison among genomes of cattle, dog, human, mouse, and rat (*Bos taurus*, *Canis familiaris*, *Homo sapiens*, *Mus musculus*, *Rattus norvegicus*, representing placental mammals), opossum (*Monodelphis domestica*, marsupial), and platypus (*Ornithorhynchus anatinus*, monotreme). (A) The majority of mammalian genes are orthologous, with more than half preserved as single copies (dark blue); a few thousand have species-specific duplications (blue); another few thousand have been lost in specific lineages (orange). We also show those lacking confident orthology assignment (green), and those that are apparently lineage specific [unique (white)]. Placental-specific orthologs are shown in pink. Single- or multiple-copy genes were

defined on the basis of representatives in human, bovine, or dog; mouse or rat; and opossum or platypus. (B) Venn diagram showing shared orthologous groups (duplicated genes were counted as one) between laurasiatherians (cattle and dog), human, rodents (mouse and rat), and nonplacental mammals (opossum and platypus) on the basis of the presence of a representative gene in at least one of the grouped species [as in (A)]. (C) Distribution of ortholog protein identities between human and the other species for a subset of strictly conserved single-copy orthologs. (D) A maximum likelihood phylogenetic tree using all single-copy orthologs supports the accepted phylogeny and quantifies the relative rates of molecular evolution expressed as the branch lengths.

## Základní pojmy a teze

- odhalit způsob genetické kontroly je dosažitelné u znaků podmíněných několika málo lokusy
- i jen znaky s jednoduchou dědičností (kontrola několika lokusy) mohou produkovat rozdílné fenotypy a vést k adaptivním radiacím
- nejdůležitějšími mechanismy jsou patrně změny v protein-kódující sekvenci, v promotoru a genové (genomové) duplikace
- důležitost jednotlivých mechanismů je sporná (navíc existují ještě mnohé další mechanismy)