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Adaptive evolution of pelvic reduction in sticklebacks by recurrent deletion of a *Pitx1* enhancer.

Chan YF, Marks ME, Jones FC, Villarreal G, Shapiro MD, Brady SD, Southwick AM, Absher DM, Grimwood J, Schmutz J, Myers RM, Petrov D, Jónsson B, Schluter D, Bell MA, Kingsley DM

Science 2010 Jan 15 **327**(5963):302-5 [[abstract on PubMed](#)]

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Selected by | Artyom Kopp / Sebastian Shimeld

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Faculty Member

Artyom Kopp

University of California,
United States of America
Developmental Biology

New Finding

Comments

This paper is a prime example of an integrated interdisciplinary approach leading to a deeper understanding of the genetic mechanisms of phenotypic evolution. The authors combine high-resolution genetic mapping, transgenic assays, gene expression analysis, and population-genetic evidence to show that cis-regulatory changes at the pituitary homeobox transcription factor 1 (*Pitx1*) locus contribute to the loss of pelvic structures in lake sticklebacks.

These changes are due to the deletion of essential sequences in the tissue-specific enhancer that controls *Pitx1* expression in the pelvis. Interestingly, different finless genotypes carry different deletions in this enhancer that produce similar expression changes but that almost certainly evolved independently. This result suggests that the evolutionary loss of morphological structures can have a simple genetic basis, and that the fixation of natural variation may be biased towards particular nexus points in developmental pathways. This bias can make genetic evolution somewhat predictable, in the sense that repeated morphological changes will be produced by changes in the same genes. Whether this predictability extends to the gain of new structures remains to be determined.

Competing interests: None declared

Evaluated 11 Jan 2010

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Sebastian Shimeld

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Developmental Biology

New Finding

This delightful paper shows that a major phenotypic change, the reduction of pelvic fins in some stickleback populations, can be traced to single mutations in the enhancer controlling pelvic expression of the *Pitx1* gene.

In a previous study, classical genetics and gene expression implicated the *Pitx1* locus in the pelvic reduction phenotype {1}. Now Chan and colleagues use a

variety of approaches, including more lab-based genetics, transgenic analysis of enhancers, population genetics and DNA flexibility modelling, to show that pelvic reduction in several independent stickleback populations is probably caused by the deletion of the enhancer responsible for pelvic Pitx1 expression. This shows that a surprisingly large, positively selected phenotypic change can be caused by a single mutation in a natural population. One note of caution in extrapolating from this finding though; the phenotypic change is essentially a loss of complexity. Whether such large changes linked to single mutations can occur in the other direction remains to be seen.

References: {1} Shapiro et al. Nature 2004, 428:717-23 [PMID:15085123].

Competing interests: None declared
Evaluated 21 Jan 2010

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