Reduce Your Pelvis in 10,000 Years or Less

While the avid dieter obsesses about reduction of different body parts, permanent reduction of many structures seems to be achieved relatively effortlessly (though much more slowly) throughout evolution. In a recent article in Nature, Shapiro et al. examine one such example, the genetic basis of pelvic reduction in the threespine stickleback fish (2004). They conclude that a regulatory mutation in the Pitx1 gene is responsible for the pelvic reduction.

An amazing diversity of forms can exist within species or between closely related species, for example the Galapagos finches with different beak sizes and shapes, the lizard Hemiergis, with different numbers of digits, and the African cichlids with different jaw shapes, tooth shapes, and behaviors. Until recently, comparative studies of different species remained descriptive. With the advent of new molecular tools, it is now possible to examine evolution of diversity from the perspectives of modern developmental biology and genetics.

Not all systems are equally conducive to genetic and developmental analyses; studies can be severely hampered by complex inheritance systems, difficulty in raising an organism, large size of an organism, or inaccessibility of embryos. In particular, vertebrates have been difficult to examine. Fortunately, a few very interesting vertebrate species or species groups have properties that make them amenable to studies of a genetic and developmental nature. One such system, the threespine stickleback, contains thousands of different populations that occur in marine, brackish, and freshwater locations exhibiting differences in body size, number of spines, number of plates, pectoral fin size and shape, size of pelvis, size and shape of snout, number of gill rakers, number of vertebrae, and number of ribs (Bell and Foster, 1994). In recent years, a genetic map and cDNA libraries have been made of the threespine stickleback and quantitative trait analyses have been performed showing multiple quantitative trait loci (QTLs), chromosomal locations of genes, for a number of traits (Peichel et al., 2001; Colosimo et al., 2004). These genetic tools, and the amazing diversity within the group of sticklebacks, allow researchers to address previously inaccessible evolutionary questions.

The trait focused on by Shapiro et al. (2004) is pelvic reduction, which includes the length of the pelvic spines and the height and length of the pelvis. Pelvic reduction is hypothesized to evolve in locations characterized by low calcium and the presence of dragonfly naiad predators that grab the fish by their spines (Bell et al., 1999). The two parents in the current QTL analysis were a marine female with a large pelvis and long spines and a benthic, or freshwater, male with no spines and a reduced pelvis. An F2 population was made and progeny genotyped for many genetic markers. Five QTL were found for the traits of reduced pelvic structures—one accounting for the majority of the variance and four with minor inputs. Pitx1, a candidate gene known to be involved in hindlimb but not forelimb development, mapped to the location of the major QTL, increasing the LOD score to 80 (a LOD score of 3 is considered significant). In the F2 cross, all 70 individuals that showed complete lack of pelvic spines were homozygous for the benthic alleles in the Pitx1 marker showing no recombination between the phenotype of pelvic reduction and Pitx1.

Further support for the hypothesis that changes in Pitx1 are responsible for pelvic reduction comes from comparisons between the pelvic reduced sticklebacks and the Pitx1 knockout mouse. In this mouse, the forelimbs are normal and the hindlimbs are reduced (with the reduction more severe on the right side). Similarly, in pelvic reduced sticklebacks, the right side of the pelvis and the right pelvic spine are more reduced than the left side, and this difference also is linked to Pitx1.

If Pitx1 is indeed the causative gene of pelvic reduction in sticklebacks, in principle, there could be either a coding mutation or a regulatory mutation in this gene in the derived benthic form. No coding differences were found between the marine and benthic form, which is not surprising, because complete loss of this gene in sticklebacks would probably cause lethality. Addressing the possibility of a regulatory mutation by sequence comparison was not feasible because of the large area in which a regulatory mutation could be located. However, the experimenters did examine the expression of Pitx1 by in situ hybridization in the marine and benthic forms, assuming that a regulatory mutation would only affect certain domains of Pitx1 expression. Pitx1 was expressed in the marine form in many areas including the thymus, olfactory pits, neuromasts, caudal fin, and pelvic region. In the benthic form, there were two differences: the pelvic region expression was absent (Figure 1) and the caudal fin expression was reduced. Thus, it is likely that a Pitx1 regulatory mutation, perhaps in a pelvic fin/caudal fin enhancer, is responsible for the morphological difference in pelvises between the marine and benthic forms.
This study brings us closer to answering many questions about evolution of the pelvic reduction in the Paxton benthic population. A second interesting question is whether pelvic reduction evolved in the same way or different ways in other populations. Fortunately, because of the wealth of isolated populations of sticklebacks, Shapiro et al. were able to examine an Icelandic population with the reduced pelvic phenotype. When an Icelandic individual was crossed to a Paxton individual, the pelvic reduced phenotype was not complemented. Therefore, it is possible that mutations in the same gene are responsible for the pelvic reduction in both populations. This phenomenon of parallel evolution may even translate to the past; amazingly, a fossil stickleback with a reduced pelvis shows the Pitx1 knockout’s characteristic asymmetry.

This study shows the beauty of the stickleback system and its potential for answering many interesting questions in evolutionary biology. The developmental and genetic tools are quickly expanding for this system; it might soon be possible to find the mutations responsible for different phenotypes and to functionally test them by overexpression or transgenic analyses. Furthermore, by examining similar phenotypes in multiple populations, it will be possible to see what range of genetic changes cause the same phenotypic changes. The establishment of evolutionary model systems is allowing us to address questions about the evolution of body form that naturalists would never have thought possible and that dieters can only fantasize about.

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