

Painted chromosomes and sex probes: limitations and prospects of using genetic markers to identify gender of California Chinook salmon

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There are a wide variety of pollutants that are capable of altering the endocrine system of animals, or interfering with the genetic information that directs cellular processes and the physiological functions that they control. Pollutants that directly interfere with the integrity or repair of genetic information by binding or damaging DNA are genotoxic. DNA damage is not necessarily limited to the individual in which the damage originally occurs. DNA damage that occurs in the progenitor cells of gametes may be inherited by subsequent generations if that damage is not corrected by cellular repair mechanisms. Chronic exposure to genotoxic substances can, over time, have a progressively more negative effect on exposed populations. Genetic damage or disruption of the endocrine system during early development may be the precursors of some of the numerous effects reported at higher levels of biological organization such as the feminization of males, developmental abnormalities, and infertility. Problems such as these would contribute to the loss of genetic variation within and decreased reproductive potential of affected populations.

In 2001, fall-run Chinook salmon with incongruent genetic and phenotypic sex ('apparent' XY-females) were detected in naturally spawning and hatchery populations throughout the Central Valley. Controlled breeding experiments were performed to test the validity of available male-specific genetic markers to identify sex. These breeding experiments inferred that a genetic rearrangement or mutation rather than altered sexual development caused by endocrine disruption is responsible for observed incongruence between sexual genotype and phenotype.

'Apparent' XY-female Chinook salmon may be symptomatic of exposure of fish to genotoxic environmental contaminants. Mutational degradation of the Y-chromosome and subsequent loss of the sex determining region, or incorrect repair of damage to the Y-chromosome leading to a translocation between the sex chromosomes may be responsible for the 'apparent' XY-female Chinook. Frequent, true sex-reversal, as would occur by the presence of a Y-chromosome that lacks a functional sex-determining locus, could have long-term serious implications for genetic diversity of a species. Conversely, if male-specific genetic markers have merely moved to the X chromosome or an autosome, and are thus no longer exclusively associated with the Y chromosome, apparent sex-reversal through such a mechanism may pose no serious threat to Chinook salmon populations. In order to discriminate between alternate models used to explain the incongruence between sexual genotype and phenotype, breeding experiments were performed using genetically normal and 'apparent' XY-females. Offspring were evaluated cytogenetically via Fluorescent In Situ Hybridization (FISH) to examine how the male-specific probes localized on chromosomes. In addition, Mendelian inheritance of rainbow trout derived microsatellite loci were tested in families used for cytogenetic analysis. 'Apparent' XY-female offspring appear to have inherited a truncated Y-chromosome (Y*). Microsatellite linkage and FISH analyses suggest that apparent XY-female fall-run Chinook salmon in California are not the result of a Y chromosome to autosome whole-arm translocation. The Y* chromosome transmitted by 'apparent' XY-females may be a Y that no longer confers genetic information necessary for development as a phenotypic male. The Y* chromosome could be the result of a mutation or recombination between sex chromosomes.