

## LECTURE 8

### Sex determination

Seven different *genetic* mechanisms will be discussed:

- a) dominant “Y” (humans)
- b) gene interaction/epistasis (but with few genes)
- c) environment
- d) haplo-diploidy
- e) X/A balance
- f) Polygenes
- g) X/A epistasis

#### a) dominant Y

- (i) “discovered” by ascertaining sex of sex-chromosome abnormal compared to “normals”

XX and XO are female  
XY and XXY are male

led to the conclusion that presence or absence  
of a Y chromosome was the determining factor

- (ii) a testis-determining-factor (TDF) is produced by a sex-determining-region (SRY) located on the short arm of the Y chromosome

- (iii) existence of SRY was deduced by examining chromosomes of XX males and XY females, where a small piece of the Y (and which contained SRY) was deleted from a Y and inserted into an X

XX males -- had the small Y insert with SRY, and  
XY females -- had the Y with the SRY deleted

- (iv) the testis determining factor (TDF) directs the primordial (undifferentiated) gonads to develop into testes,; the testes then produce (secrete) testosterone which stimulates development of secondary sex characteristics of males

- (v) birds are ZZ:ZW -- may be dominant Z but not known for sure

#### b) gene interaction/epistasis (with few genes)

- (i) occurs in plants (which almost never contain sex chromosomes) yet have “sexual” phases in the sense of an external gamete producer (“males”, pollen) and an external gamete receiver that produces its own gametes (“females”, ovary)

- (ii) good example is corn: monoecious -- both male and female (hermaphroditic, selfing)  
diecious -- either male or female



d) haplo-diploidy [occurs in honeybees and many other, but not all, Hymenoptera]

- (i) three sexes: drones → males, genetically haploid (1N)  
workers → females, genetically diploid (2N), typically sterile  
queens → females, genetically diploid (2N), fertile
- (ii) *queens* are diploid and produce gametes by normal meiosis
- (iii) *males* are haploid and produce gametes (essentially) by mitosis (first meiotic division aborted)
- (iv) system is an effective screen of recessive lethals (not surprisingly)
- (v) males have a grandfather, but no father

e) X/A balance [*Drosophila*]

- (i) where the ratio of the number of X chromosomes to sets of autosomes (ploidy level) “determines” the sex of an individual  
  
2X/2A -- 1.0, female  
1X/2A -- 0.5, male
- (ii) under this mechanism, XX and XXY (diploid) individuals are female, whereas XY and XO (diploid) individuals are male
- (iii) The Y chromosome in *Drosophila* is necessary for sperm maturation and tails; XO individuals are sterile
- (iv) at the “molecular” level, a sex (X) linked gene called Sex-lethal (*Sxl*) essentially “counts” the number of X chromosomes and sets of autosomes and “responds” to the X/A ratio  
  
if X/A ratio is 1.0 or greater, *Sxl* is turned on  
if X/A ratio is 0.5 or less, *Sxl* is not turned on

f) polygenes [swordtail fish, *Xiphophorus helleri*]

- (i) where sex is a function of the action of numerous, additive genes and there are no sex chromosomes at all; in this case, the sex of an individual is a function of the sum of small male- or female-contributing genes in any given individual  
  
 $\Sigma$  male genes >>  $\Sigma$  female genes → male  
 $\Sigma$  female genes >>  $\Sigma$  male genes → female
- (ii) inferred from normal distribution of the distribution of sex ratios of individual broods, i.e., sex ratios vary but the average (mean) is roughly 1:1

h) X/A epistasis

- (i) sex is determined by epistatic interaction(s) among genes on defined sex chromosomes and genes on autosomes; normally, genes on sex chromosomes are epistatic to those on autosomes and the sex of an individual is that expected from its sex chromosome constitution
- (ii) the epistatic interaction(s) can break down resulting in “spontaneous sex reversals” where the sex of an individual is opposite that of its sex chromosomes
  - (a) not uncommon in lower vertebrates (e.g., several fish species) where “spontaneous sex reversals are found in nature

Note general homology of sex chromosomes (e.g., X and Y)

XX males: fertile with “normal” X chromosomes (gametes all X bearing)

XY females: fertile with “normal” X and Y chromosomes (gametes are  $\frac{1}{2}$  X and  $\frac{1}{2}$  Y)

- (iii) can experimentally induce “sex reversal” in appropriate species by judicious application of opposite, sex-specific hormones at the appropriate developmental window (after which sex appears to be irreversibly determined)
  - (a) monosex culture in many aquacultured fish species has several advantages