Sex Determination and Sex Linakge
Genetics Course

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Sex linkage

- Heredity is equal from both parents-Mendels studies of garden pea.
- Inheritance is equal from both parents –sex linkage
- It occurs when the gene controlling a trait is located on a chromosome a so called **sex chromosome,** that is not present in morphologically identical pairs (two homologous copies) in both sexes.
- Such Traits exhibit unique **sex-linked patterns** of inheritance recognized in pedigree studies.
- Roles of sex chromosomes in controlling sex phenotypes is important and mechanisms of sex determination will also be reviewed here.

Mechanisms of Sex Determination

- Nature encompasses a vast array of diverse mechanisms of sex determination in different species.
- Sex determination focus on two species
- *Drosophila* melanogaster
- Humans
- Important in development of genetic concepts.

Explanation

- In fruit fly and human, in many other species, especially higher animals individuals normally exhibit one of the two sex phenotypes
- Female (eggs, ovules, or macrospores)
- Male (sperm, pollen, or microspores)
- Species with seperation of sexes in different individuals ----dioecious.e.g all higher animals and some higher plants
- Species in which both male and female gametes produced by each individual ---monoecious.
- Hermaphroditism—production of both eggs and sperms by the same organisms e.g lower animals, hermaphrodites producing both types of gametes by the same organism.
- The two sex phenotypes are usually quite easily distinguished in humans and fruit flies , not universally the case.
- In Isogamy two genetically distinct types of gametes are sometimes morphologically indistinguishable e.g lower eukaryotes.
- In humans, fruit flies and most higher eukaryotes , the gametes produced are of two quite different types, namely, eggs and sperms.

Identification of sex chromosomes

- First investigation relating chromosomes to sex determination at turn of century
- H.Henking a German biologist discovered in 1891 , a particular nuclear structure traced throughout spermatogenesis of certain insects. Half sperm received this structure and half didn't .
- Henking didn't speculate on the significance of this body, but identified it as " X" body and showed that sperm differed by its presence or absence.
- In 1902 C.E. McClung verified and extended such observations and made cytological observations on different species of grasshoppers and demonstrated that
- Somatic cells in female grasshoppers carry different chromosome numbers than do corresponding cells in male. So,
- He followed X body in spermatogenesis but didn't succeed in tracing oogenesis of female grasshopper. McClung associated X body with sex determination but erroneously considered it to be peculiar to males.

Contributions of E.B. Wilson and associates to basic knowledge of sex determination

- Wilson reported extensive cytological investigations on several different insects. He succeeded in following oogenesis as well as spermatogenesis.
- The X body of Henking thus found to be a chromosome that determined sex. It was identified in several insects and became known as sex or X chromosome.
- All eggs of these insects carried an X chromosome, but it was included in only half of the cells forming sperm. All sperm had usual complement of other chromosomes (autosomes).
- Eggs fertilized by sperm containing X chromosome produced zygotes with two X chromosomes, which became females.
- Eggs receiving sperm without an X chromosome produced zygotes with one X, which became males.
- Males are referred to as hemizygous (half that present in a zygot) for X chromosomes or for genes located on X chromosomes. This hemizygosity of males for genes located on X chromosomes is responsible for altered patterns of inheritance observed for X-linked traits.

xx- x y Mechanism of sex Determination

- Wilson observed another chromosome arrangement in the milkweed bug. Same number of chromosomes were present in the cells of both sexes observed in this bug. The homolog of X was distinctly smaller and was called Y chromosome.
- XX zygotes became females, and XY zygotes became males. This was called XX-XY mechanism of sex determination. Examples most higher animals and some plants. It is also found in fruit fly and humans. Both exhibit same pattern of transmission of X and Y chromosomes in normal individuals . In humans X chromosome is considerably longer than the Y chromosome.
- The total complement of human chromosomes includes 44 autosomes
- XX in female
- XY in male
- Eggs produced by the female in oogenesis have usual complement of autosomes (22) plus an X chromosome. Sperm from the male have the same autosomal number and either an X or a Y. Eggs fertilized with sperm containing a Y chromosome result in zygotes that develop into males: those fertilized with sperm containing an X chromosome develop into females.
- Control of sex determination by a particular chromosome provided a tangible evidence that genes are in chromosomes. Females are called homogametic sex and males are called heterogametic .

Species with Heterogametic Females

- Many species, including most birds, moths, and some fish, a chromosomal mechanism of sex determination occurs, basically identical to XX-XY mechanism but with females being heterogametic(designated as ZW) and males being homogametic (usually designated as ZZ).
- This mechanism is sometimes called ZZ-ZW. However, mechanistically this is identical to XX-XY system, but with relationship between sex chromosomes and sex phenotypes reversed. Stated differently, in birds, the chromosome composition of the egg determines the sex of the offspring, whereas in humans and fruitflies, the chromosome composition of the sperm determines the sex of the offspring.

The Y chromosome and Sex Determination in Mammals

- In both *Drosophila* and human, normal males have an XX sex chromosome composition and normal males have an XY sex chromosome composition. Sex determination is quite different in both.
- **In mammals, the presence of a Y chromosome is required for the development of a male sex phenotype. In contrast, the Y chromosome plays no significant role in sex determination in *Drosophila*.**
- Irregular sex chromosomes constitutions are fairly common in humans. Surprisingly, X chromosomes present in any number (e.g., XXX or XXXX) **in the absence of a Y chromosome give rise to a female sex phenotype.** A Y chromosome is required for maleness; moreover the presence of a single Y chromosome is sufficient even in the presence of several X chromosomes (e.g., XXXXY).

Explanation

- The Y chromosome induces development of the undifferenciated gonadal medulla into a testis, whereas an XX chromosomal complement induces the undifferenciated gonadal cortex to develop ovaries. The gene on the Y chromosome in humans that is responsible for the development of testis is called *TDF* (for *T*estis *D*etermining *F*actor).
- The *TDF* gene has been isolated (cloned), characterized structurally (sequenced), and found to encode a protein with features that suggest that it acts by regulating the expression of other genes. So *TDF* gene may be the master regulator that triggers the expression of a large number of genes that produce the male phenotype. In the absence of *TDF* gene , the genes that produce the female sex phenotype would be expressed. *TDF* gene (and possibly other male determining genes on the Y chromosome) exhibits a very dominat effect on the development of the sex phenotype. Even in the presence of three or more X chromosomes, a single Y chromosome is usually sufficient to produce testis and male characteristics.