# Sex determination simplified essay

The biological meaning of sex has long been an issue of interest by society. During the early times when the scientific method has not yet been formulated, the types of sex were then arbitrarily described. Aristotle postulated the sex of a child is determined by the temperature of the semen due the act of copulation. In simpler terms, warm semen produced baby boys, while cold semen created baby girls. During another century, women have been ridiculed and put to shame when they are not able to bear male babies who were expected to inherit their parents' riches. King Henry VIII was upset with his wife for not bringing him a male baby that would inherit his kingdom.

Utterly upset, he commanded that his wife be put to death by beheading. We are indeed lucky at this day and age to be provided with scientific knowledge that would describe the process of sex determination in more reliable terms. It should be understood that sex in humans is determined at three different levels. The first level that will be discussed is the chromosomal level. A chromosome is the microscopic units of genetic material that are assorted and distributed during the regular cell division cycle.

In humans, each cell contains 46 chromosomes or 23 pairs of chromosomes. The twenty-two pairs of chromosomes are called autosomes or body chromosomes, which contain genetic information that is essential for the activities and functioning of the human body. The other more interesting pair of chromosomes are the sex chromosomes, which are of two specific types– X and Y. A pair of sex chromosomes defines the genetic sex of an individual (Affara, 1991). For example, an individual that has a pair of X chromosomes or XX is genetically programmed to develop into a female. On the other

hand, an individual with one sex chromosome of each type, XY, is genetically programmed to end up as a male.

Aside from microscopic chromosomes in a cell, there are also other factors that influence the development of a particular sex in a human being. Human chromosomes are studied through the process called karyotyping (Guo and Thompson, 1992). This involves the extraction of blood from the vein in the arm and the blood is then put into a high-velocity equipment called the centrifuge that separates the white from the red blood cells in the test tube based on the weight of the cells. The white blood cells or leucocytes are then collected after spinning and these cells are put in a culture medium are kept in a warm incubator for 72 hours.

The culture medium is a special formulation that contains glucose as the source of energy for the cultured leucocytes and important amino acids and minerals that would help the leucocytes lives in culture for the required amount of time. A special substance known as phytohaemagglutinin is also introduced into the blood cell culture. This substance induces the white blood cells to start dividing, tricking them to enter the cell cycle once again. The chromosomes that are viewed by scientists under the microscope are usually at the metaphase stage of the cell cycle. The metaphase stage is characterized by chromosomes aligning at the equatorial plate of the cell, wherein these are getting ready to separate into two chromatids and travel to the opposite ends or poles of the cell. This separate is a mechanism for chromosomes to get ready before the cell divides itself into two daughter cells.

By the time the dividing cells reach the metaphase stage of the cell cycle, the blood cultures are further processed at the 72nd hour of culture and spun down again to remove the culture medium. The cells drop down to the bottom of the test tube and this is resuspended in a hypotonic solution that will induce the cells to swell, a process that is important in preparing chromosomes for karyotyping. Once the cells are swollen, these are fixed with a preservative solution and the solution is dropped onto glass slides for viewing under the microscope. The cytogenetic technologist who has been trained in preparing chromosome slides and viewing them under the microscope analyzes all 46 chromosomes of each cell.

There are therefore only two normal sex chromosome combinations—XX for female and XY for males. There may be some cases wherein an individual may have more than 2 sex chromosomes. If such is the case, the individual is cytogenetically abnormal and he suffers from a numerical aberration or abnormality because he has more than the normal 2 chromosomes in each cell. When there are more than or less than 2 sex chromosomes in an individual, this condition is known as aneuploidy. In cases wherein there are 3 sex chromosomes in an individual, the condition is known as trisomy (Jurmain et al. , 2007).

One type of trisomy is Klinefelter syndrome, which typifies an individual with XXY sex chromosomes. These individuals have both male and female sex characteristics such as breasts and at the same time testes. These individuals are usually tall and lanky and are usually mentally retarded. They lose their ability to produce offspring.

Another type of trisomy is the so-called superfemale or trisomy X or XXX. The term superfemale is quite misleading because the individual is actually not a female to extremely presents female characteristics such as bigger breasts or prettier faces but actually the term superfemale only aims to denote the instead of just having two X chromosomes to denote femaleness, the individual has 3 X chromosomes which is more than enough or needed, hence the term superfemale. These individuals are often infertile and mentally retarded. They have normal female physical features.

In cases wherein there is an insufficient number of sex chromosomes such as that observed in patients diagnosed with Turner’s syndrome, the individual is determined to only have one X chromosome. Turner’s syndrome patients are usually short and stumpy and have webbed necks. They are generally mentally retarded and have difficulty engaging in social relationships. Another route for sex determination in human is through the development of primary sex characteristics which include the testes and the ovaries (Kalat, 2006). The development of these organs is dictated by the presence and concentration of sex-specific hormones. Hormones also play a critical role in the development of sex among humans.

Hormones are proteins that are secreted by specific organs of the body. Some hormones are produced in equal amounts by both males and females, while there are other hormones that are only produced by males, or by females. For example, the hormone estrogen is only produced by females and this hormone is responsible in regulating the monthly menstrual period. Another female-specific hormone is progesterone which is also known as the hormone of pregnancy because high levels of this hormone have been

observed during the course of pregnancy. Progesterone induces the uterus to thicken in order to provide protection and nourishment to the developing fetus.

During the initial stages of pregnancy, the fetus does not carry any specific sex because the fetus is then only composed on simple tissues that are common for both male and female fetuses (McLaren, 1988). Almost 50 years ago, a scientist postulated that all fetuses are initially programmed to develop into females, due to the presence of the female-specific hormones such as estrogen and progesterone. Eventually, during pregnancy, especially at the 6th week of gestation, the hormone testosterone is secreted and this masks the effect of the female-specific hormones. One the estrogen and progesterone are masked in terms of their effects, the testosterone hormone will induce the primitive sexual organs to develop into male-specific testes.

In case no testosterone is present in a developing fetus, the female-specific hormones of estrogen and progesterone will induce the primitive organs to develop into ovaries, which thus dictate that the developing fetus will be a girl (McLaren, 1990). The presence of testosterone which influences the Wolffian ducts to further develop into the epididymis, vas deferens, and seminal vesicles. Male-specific sexual characteristics such as the external genitalia will emerge when the cells of the urogenital tubercle convert the hormone testosterone into its analogue dihydrotestosterone which sways the development of the penis and scrotum. Another type of protein produced by Sertoli cells, is the Mullerian inhibiting substance (anti-Mullerian hormone), which influences the Mullerian ducts to self-destrut.

When these two proteins are not present, the Wolffian ducts degenerate and the Mullerian ducts develop into the oviducts, uterus, cervix, and upper vagina. As for the males, a gene identified as the sex-determining region in the Y chromosome or SRY has been linked to the formation of testes in a developing fetus. Research has shown that the protein generated by the SRY gene stops the secretion of other sex-specific hormones, thus letting testosterone alone to be produced in the developing fetus. This in turn, results in the development of male-specific organs in the fetus, including the testes and its related organs. The third route for sex determination in humans is through the development of secondary sexual characteristics which is also driven by specific hormones.

By linking deletions on the Y with the presence or absence of testes and by studying XX males which carry a tiny portion of the Y on one of their X chromosomes, investigators localized TDF to a 35-kb region of the Y short arm. Cloning of this region resulted in the identification of a gene designated sex-determining region Y (SRY) (Koopman et al. , 1991). Convincing evidence that SRY is the testis-determining gene was obtained when a 14. 6-kb genomic sequence of the mouse SRY locus was shown to be capable of inducing XX fetuses to develop into males in transgenic experiments.

The hypothesis is that SRY is a prime regulatory gene that triggers a battery of gene interactions that converts the fetal gonad into a testis. SRY codes for a member of the High Mobility Group-1/-2 (HMG) protein family whose components are characterized by an 80-amino acid DNA-binding sequence known as the HMG domain. Various HMG proteins such as the SRY, are proteins that control transcription that identifies and attaches a particular

DNA sequence and result in the attachment of DNA and reconfigure into an angle. The SRY target sequence has been located in the promoter segment of genes regulating differentiation such as Mullerian inhibiting substance and P450 aromatase, an enzyme that converts testosterone to estradiol. Furthermore it is present in the promoter region of SRY itself suggesting a positive feedback loop. It took over three decades from the recognization of the Y as testis-determining to the identification of SRY as TDF.

The isolation of the SRY gene is serves as a breakthrough in our understanding of human sex determination. The challenge is now to decode how SRY controls transcription and determining the genes existing adjacent to the SRY gene and linked to the sex determination pathway. Also interestingcontrasting mitosis and meiosis