BBiological Sexual Development

Gender Identity as a **Biological Process: Normal Prenatal Differentiation**

From the moment of conception, many biological factors contribute to the differentiation of male or female sex. In the following paragraphs, we will explore how biological sex differentiation occurs during prenatal development. Our discussion follows a chronological sequence: We begin at conception, looking at chromosomal differences between male and female, then continue with the development of gonads, production of hormones, the development of internal and external reproductive structures.

Genetic Sex

Our genetic sex is determined at conception by the chromosomal makeup of the sperm (male reproductive cell) that fertilizes an ovum or egg (female reproductive cell). Females have two similar chromosomes, labeled XX, whereas males have dissimilar chromosomes, labeled XY.

A normal female ovum (or egg) contains an X chromosome. A normal male sperm cell contains either an X or Y chromosome. If the ovum is fertilized by a sperm carrying a Y chromosome, the resulting XY combination will produce a male child. In contrast, if an X-bearing sperm fertilizes the ovum, the result will be an XX combination and a female child. Two X chromosomes are necessary for internal and external female structures to develop completely. But if one Y chromosome is present, male sexual and reproductive organs will develop (Harley et al., 1992; Page et al., 1987).

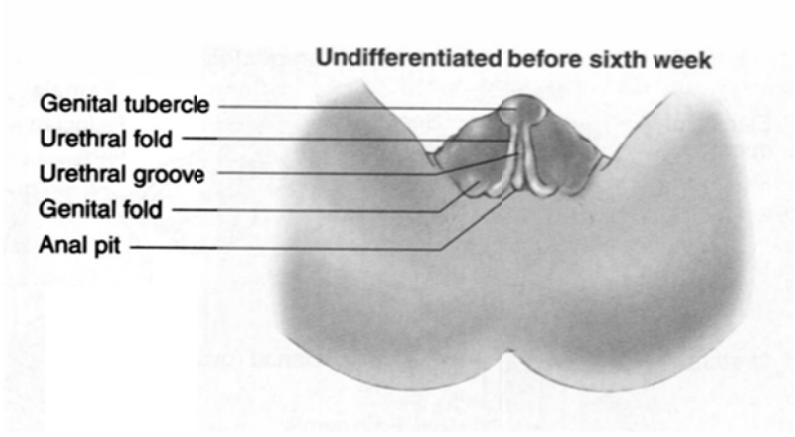
Why is the Y chromosome essential for male development?

In the first weeks after conception, male and female reproductive organs are the same. Differentiation begins about 6 weeks after conception. To put it simply, the presence of a Y chromosome triggers gonadal tissue to develop into testes. No Y chromosome and ovaries will develop and NO testes.

Once the testes or ovaries develop, these gonads begin releasing their sex hormones. As we will see next, these hormones become the critical factor in further sex differentiation, and genetic influence ceases.

The role of hormones

Like other glands in the *endocrine system* (a system of ductless glands that includes the pituitary, thyroid, parathyroids, adrenals, and pancreas), the gonads produce hormones and secrete them directly into the bloodstream. Ovaries produce **estrogens.** The primary hormone products of the testes are **androgens**. The most important androgen is *testosterone,* which influences both the development of male physical sex characteristics and sexual motivation. In both sexes the adrenal glands also secrete sex hormones, including small amounts of estrogen and greater quantities of androgen.



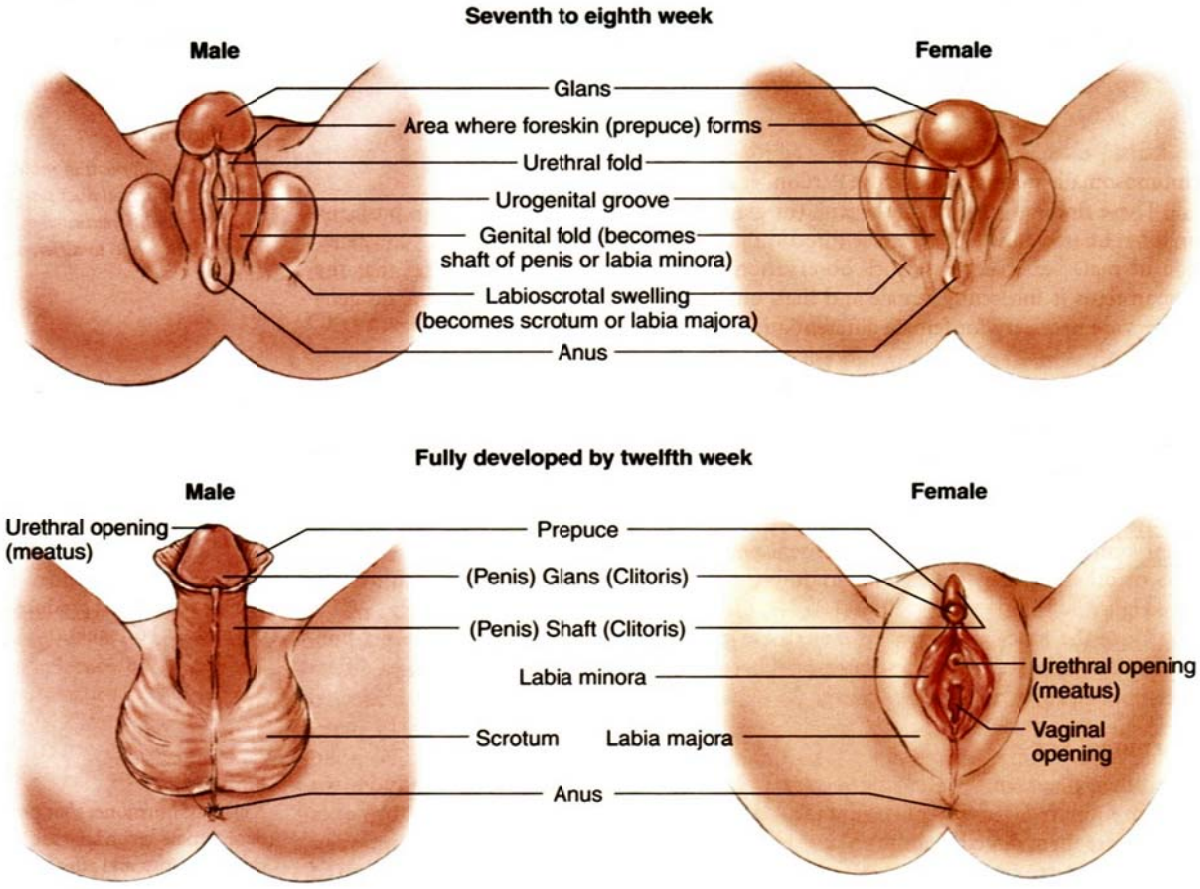
Sex of the External Genitals

Until the gonads begin releasing hormones during the sixth week, the external genital tissues of male and female fetuses are undifferentiated. (see picture at left)

These tissues will develop into either male or female external genitals depending on the presence or absence of a testosterone product released in males, known as *dihydrotestosterone,(DHT).* DHT stimulates the *labioscrotal swelling* to become the scrotum, and the *genital tubercle* and *genital folds* to differentiate into the glans and shaft of the penis, respectively. The genital folds fuse around the urethra to form the shaft of the penis, and the two sides of the labioscrotal swelling fuse to form the scrotum: these fusions do not occur in females. In the absence of testosterone (and possibly under the influence of a substance(s) triggered by the DSS or femaleness gene), the genital tubercle becomes the clitoris, the genital folds become the inner vaginal lips (labia minora), and the two sides of the labioscrotal swelling differentiate into the outer vaginal lips (labia majora). By the twelfth week, the differentiation process is complete: The penis and scrotum are recognizable in males; the clitoris and labia can be identified in females.

Because the external genitals, gonads, and some internal structures of males and females originate from the same embryonic tissues, it is not surprising that they have corresponding, or homologous, parts. Table 3.1 summarizes these female and male counterparts.

Table 3.1 Homologous Sex Organs	
Female	**Male**
Clitoris	**Glans of Penis**
Hood of clitoris	**Foreskin of penis**
Labia minora	**Shaft of penis**
Labia majora	**Scrotal sac**
Ovaries	**Testes**
Skene's ducts	**Prostate**
Bartholin's glands	**Cowper's glands**



Brief sum to this point

In sum, what will be male and female external genitals are the same in genetic males and females up to about age 6 weeks. If a Y chromosome is present, then testes form. If no Y, then ovaries form. If testes form, they do their thing – secreting testosterone. It is this hormone that causes the external genitals to develop as male. No testosterone and we develop looking like females.

We can have variations from normal developments at the level of chromosomes or hormones.

Variations in Chromosomes: Sex Chromosome Disorders

Errors occasionally occur at the first level of biological sex determination, and individuals are born with one or more extra sex chromosomes or missing one sex chromosome. Over 70 abnormalities of the sex chromosomes have been identified (Levitan & Montagu, 1977). These irregularities may be associated with various physical, health, and behavioral effects. We will consider two of the most widely researched of these abnormalities.

TURNER'S SYNDROME Turner's syndrome is a relatively rare condition characterized by the presence of only one sex chromosome, an X. This condition, estimated to occur in about one in every 2000 live female births (Gravholt et al,, 1998), results when an atypical ovum containing 22 autosomes and no sex chromosomes is fertilized by an X-bearing sperm. (The same ovum fertilized by a Y-bearing sperm does not survive.) The resulting chromosome number in the fertilized egg is 45 rather than the normal 46; the sex chromosome combination is designated XO. People with this combination develop normal external female genitals and consequently are classified as females. However, their internal reproductive structures do not develop fully—ovaries are absent or represented only by fibrous streaks of tissue. Turner's syndrome females do not develop breasts at puberty (unless given hormone treatment), do not menstruate, and, of course, are sterile. As adults, women with this condition tend to be unusually short (Gravholt et al,, 1998).

Because the gonads are absent or poorly developed and hormones are consequently deficient, Turner's syndrome permits gender identity to be formed in the absence of gonadal and hormonal influences (the second and third levels of biological sex determination). Turner's syndrome individuals identify themselves as female, and as a group they are not distinguishable from biologically normal females in their interests and behavior (Money & Ehrhardt, 1972). This characteristic strongly suggests that a feminine gender identity can be established in the absence of ovaries and their products.

KLINEFELTER'S SYNDROME A much more common sex chromosome error in humans is **Klinefelter's syndrome.** This condition, estimated to occur once in about every 500 live male births (Kruse et al,, 1998), results when an atypical ovum containing 22 autosomes and two X chromosomes is fertilized by a Y-bearing sperm, creating an XXY individual. Despite the presence of both the XY combination characteristic of normal males and the XX pattern of normal females, Klinefelter's syndrome individuals are anatomically male. This condition supports the view that the presence of a Y chromosome triggers the formation of male structures. However, the presence of an extra female sex chromosome impedes the continued development of these structures, and Klinefelter's syndrome males typically are sterile and have undersized penises and testicles. Their interest in sexual activity is often weak or absent (Money, 1968; Rabock et al., 1979). Presumably, this low sex drive is related, at least in part, to deficient production of hormones from the testes.

Klinefelter's syndrome males tend to be tall and are often somewhat feminized in their physical characteristics; they may exhibit breast development and rounded body contours. They also tend to be passive and to lack ambition, and they frequently show some intellectual impairment. Testosterone treatments during adolescence and adulthood can enhance the development of male secondary sexual characteristics and may increase sexual interest (Kolodny et al., 1979). These individuals usually identify themselves as male; however, they manifest a higher-than-expected degree of gender-identity confusion (Mandoki et al.,, 1991).

Variations in hormones that then influence development.

ANDROGEN INSENSITIVITY SYNDROME A rare genetic defect causes a condition known as **androgen insensitivity syndrome** (AIS), also called *testicular feminization syndrome*, in which the body cells of a chromosomally normal male fetus are insensitive to androgens (Clarnette et al,, 1997). The result is feminization of prenatal development, so that the baby is born with normal-looking female genitals and a shallow vagina. Not surprisingly, AIS babies are identified as female and reared accordingly. The anomaly is often discovered only in late adolescence, when a physician is consulted to find out why menstruation has not commenced. A study of 10 AIS individuals revealed that all but one—a child reared in a dysfunctional environment—had acquired a clear female gender identity and behaved accordingly (Money et al., 1968). These findings seem to support the importance of social learning in shaping gender-identity formation.

FETALLY ANDROGENIZED FEMALES In a second type of abnormal sex differentiation, chromosomally normal females are prenatally masculinized by exposure to excessive androgens, either from a genetically induced malfunctioning of their own adrenal glands *(adrenogenital syndrome)* or from androgenlike substances ingested by their mothers during pregnancy (Clarnette et al,, 1997). (In the 1950s, some pregnant women were given androgenlike drugs

to reduce the risk of miscarriage.) As a result, such babies are born with masculine-looking external genitals: An enlarged clitoris may look like a penis, and fused labia may resemble a scrotum. These babies are usually identified as female by medical tests, treated with minor surgery or hormone therapy to eliminate their genital ambiguity, and reared as girls. Nevertheless, one noteworthy study reported that 20 out of 25 fetally androgenized females identified themselves as "tomboys," engaged in traditionally male activities, and rejected behaviors and attitudes commonly associated with a female gender identity (Money & Ehrhardt, 1972). These findings, which appear to reflect the significant impact of biological factors in gender-identity formation, contrast markedly with the AIS study described earlier.

DHT-DEFICIENT MALES A third variety of abnormal prenatal differentiation is caused by a genetic defect that prevents conversion of testosterone into the hormone dihydrotestosterone (DHT), which is essential for normal development of external genitals in a male fetus. In males with this disorder, the testes do not descend before birth, the penis and scrotum remain undeveloped so that they resemble a clitoris and labia, and a shallow vagina is partially formed. Because their genitals look more female than male, these DHT-deficient males are typically identified as female and reared as girls. (Males who experience this abnormal sex-differentiation pattern are also sometimes described as exhibiting *5-alpha reductase syndrome. (**this is what your text call this on page 432**)* However, because their testes are still functional, an amazing change occurs at puberty as accelerated testosterone production reverses the DHT deficiency. This causes their testes to descend and their clitorislike organs to enlarge into penises. In short, these DHT-deficient males undergo rapid transformation, from apparently female to male. How do they respond?

In one study, a team of Cornell University researchers investigated 18 DHT-deficient males who had been reared as female in rural communities in the Dominican Republic (Imperato-McGinley et al., 1979). When their bodies changed at puberty, 16 responded to this transformation by embracing traditional male gender roles mandated by their culture. These findings challenge the widely held belief that once gender identity is formed in the first few years of life it may not be changed without severe emotional trauma. To the contrary, they suggest that gender identity may be malleable even as late as adolescence.

Some important questions have been raised about this study, however. First, the culture of this Caribbean nation is very male oriented, which may have influenced these youths to switch gender identity more readily. Indeed, some were exposed to extreme social pressure in the form of ridicule, as locals called them *quevote,* which means "penis at 12" or *machihembra*, "first woman, then man." And second, the research was conducted retrospectively, after the subjects were adults. Because people's recollections are not always reliable, it cannot be determined with certainty whether all these youths had experienced unambiguous female gender socialization as children.

Intersexuality

In sum, we know that chromosomes determine genetic sex but it is hormones that determine whether we will have a penis or clitoris. So a genetic female can get too much testosterone and genetic male could have conditions where his body can't recognize this testosterone. Children who are born with ambiguous genitals are called Intersexed. In other words, the doctor can't tell if what he sees is a big clitoris or a little penis.

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