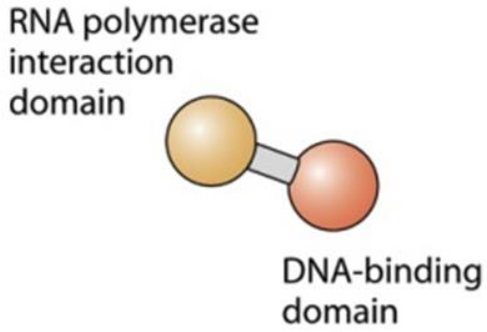
목 차

5. 유전자 발현 조절

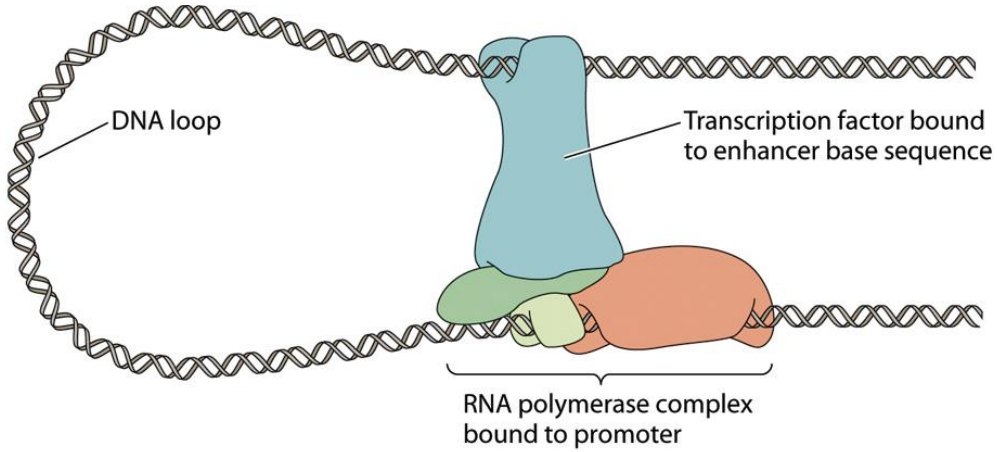
Differential Gene Expression

- Enhancer
  - Base sequence in DNA
  - Activate transcription
  - Far from the RNA polymerase binding site



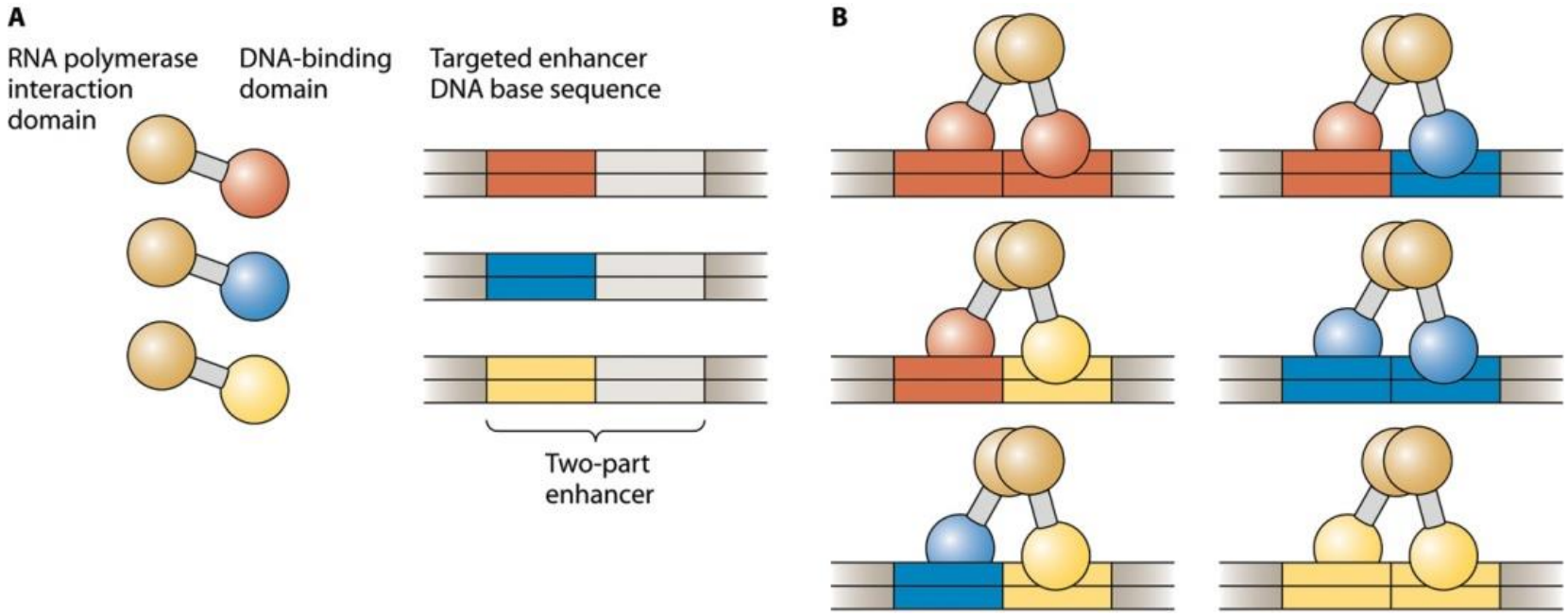
Transcription factor: consisting of two domains

- Silencer: turn off transcription



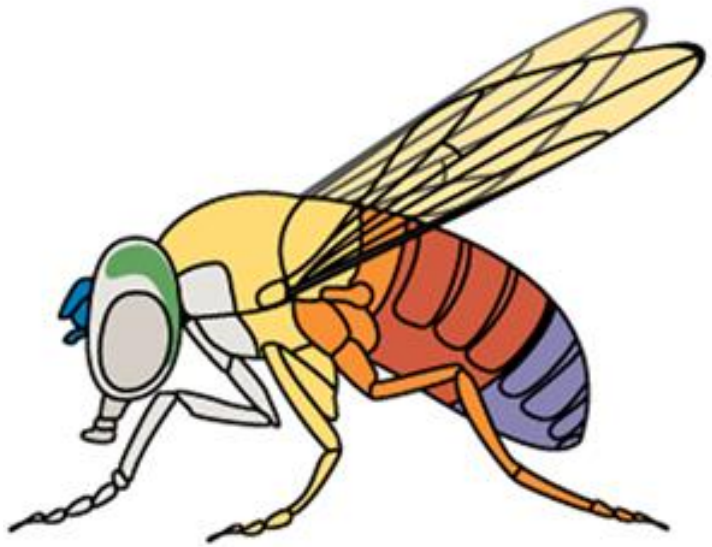
Transcription Factors

- Usually act as a complex with other proteins
- Regulation of gene expression with smaller number of transcription factors



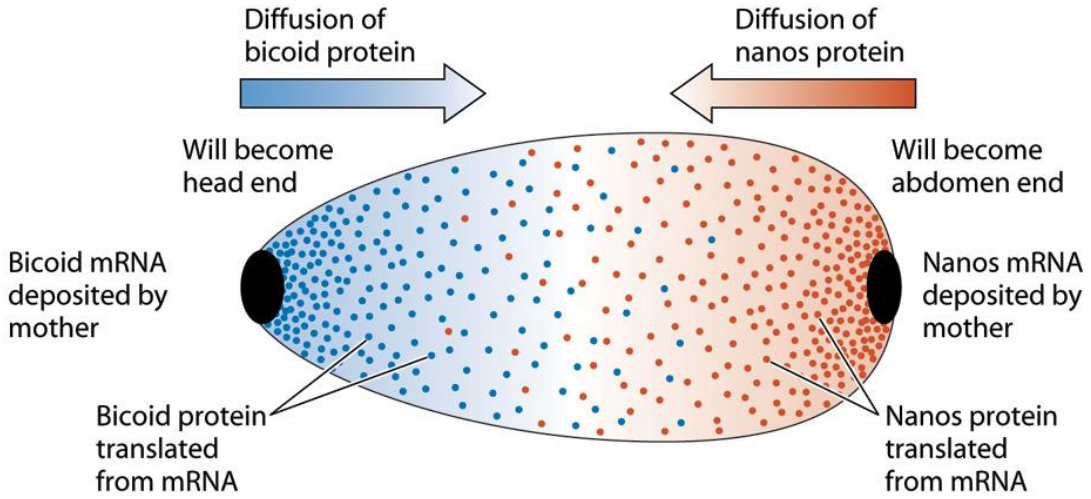
Determination of anterior-posterior body axis in fly

- Embryo produced by mutant fly
  - Mutant fly (mutation in bicoid) → embryo with two tails
  - Mutant fly (mutation in nanos) → embryo with two heads



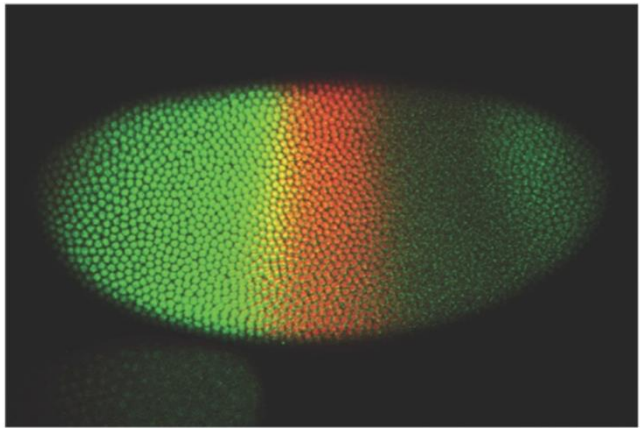
Determination of anterior-posterior body axis in fly

- Establishment of body plan by maternal genes
  - Maternal cells deposit bicoid and nanos mRNA at the opposite ends of embryo during embryo formation
  - Concentration gradient of bicoid and nanos upon fertilization
    - Bicoid end → head
    - Nanos end → tail



Segment formation

- Hunchback: gene required for development of thorax
  - Regulation of gene expression by bicoid and nanos proteins
    - Bicoid: activation of hunchback
    - Nanos: repression of hunchback



Green: hunchback protein

Red: Krupple protein

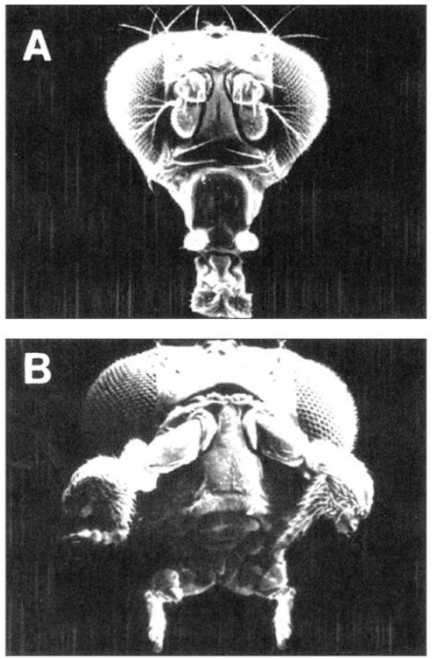
Yellow: both

- Genes turned on in the wake of the bicoid-nanos gradient divide the *Drosophila* embryo into segments
- Homologous genes in frog, chicken, zebrafish, mouse, and human

Homeotic Genes in Fly

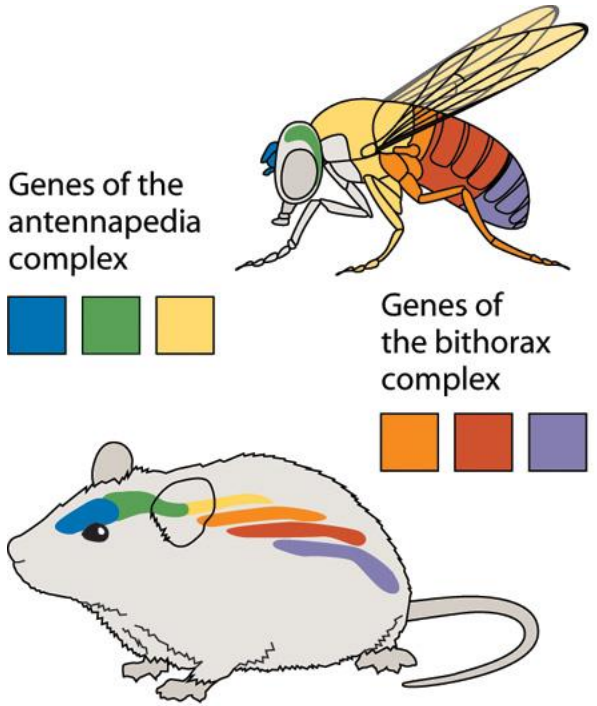
- Homeotic genes
  - The fates of the individual segments are controlled by an other family of genes: the homeotic genes
- Homeotic gene clusters
  - Bithorax complex
    - Controlling the development of the posterior half of the embryo
    - Gene arrangement on the chromosome is in the same order as the segments of the fly body they controls
  - Antennapedia complex
    - Controlling the development of the anterior part

A. Normal
B. Antennapedia



Homeotic Genes in Vertebrates

- Similar to *Drosophila* homeotic genes
- Instead of one bithorax cluster and one antennapedia cluster, mouse and human have 4 copies of each.
- The proteins encoded by homeotic genes have similar DNA binding domains, called the homeodomain.
- Homeotic genes would specify segment fate by turning different sets of genes on and off.



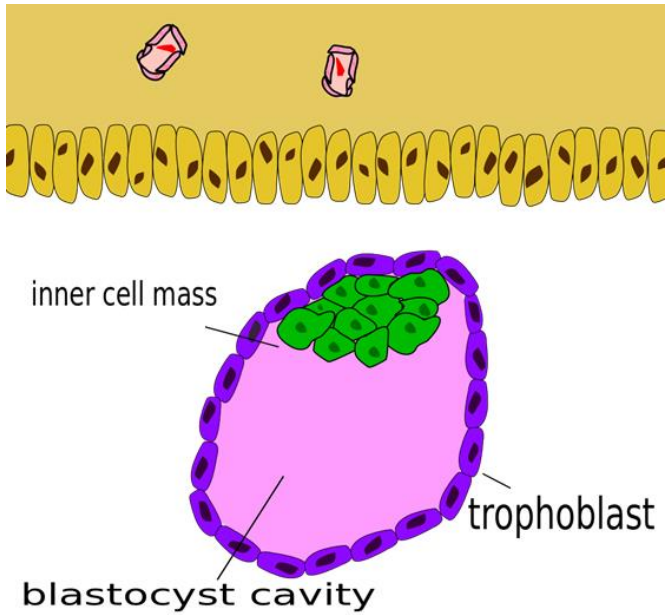
6. 포유동물의 초기 발생

Early Cell Division and Implantation

- Blastocyst
  - Inner cell mass
  - Trophoblast (outer cell layer)
    - The trophoblast cells form a fluid-filled ball with the inner cell mass.
    - The trophoblast cells will form the embryo's portion of placenta.
    - Implantation into the uterus
- Extraembryonic tissues
  - Fetal side of placenta

    +
  - Membranes surrounding the fetus
- After implantation, the cells of the inner cell mass undergo gastrulation.

Blastocyst

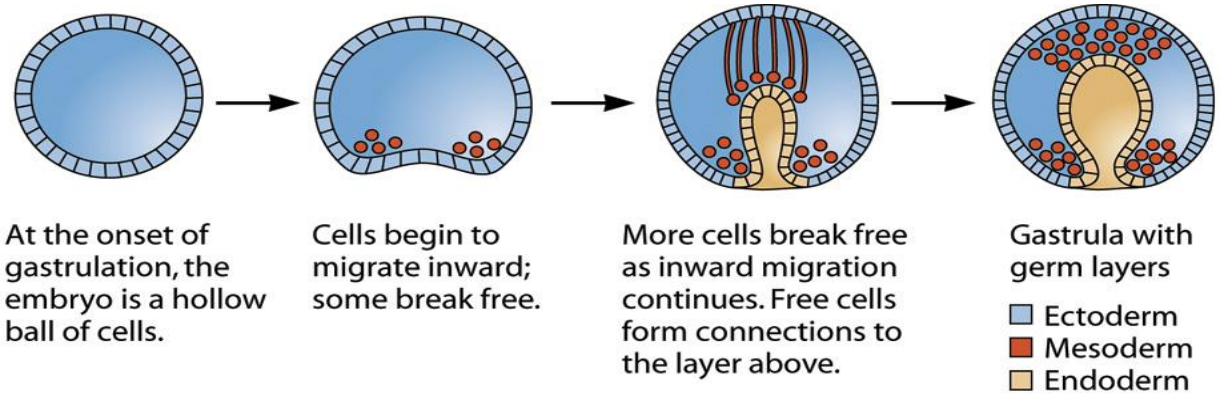


※ 출처 : commons.wikimedia.org

Early Development in Mammals

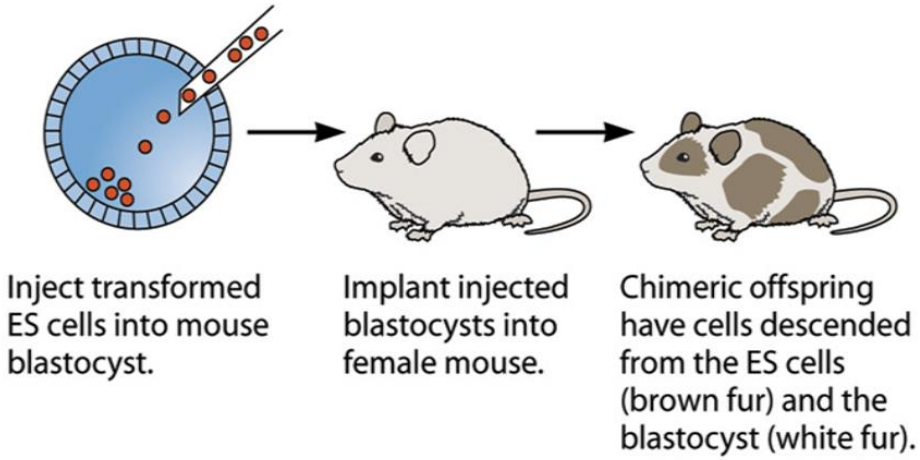
- Gastrulation (The blastula undergoes a dramatic rearrangement.)
  - Formation of three germ layers
    - Ectoderm → outer layer of the skin and the nervous tissue
    - Endoderm → inner linings of the digestive organs and circulatory system
    - Mesoderm → muscle, bone, blood, and other internal organs and tissues

- Differentiation into specific tissues and organs
    - Homeotic genes



Twin and Chimera

- Identical twin
  - They develop from the same inner cell mass, which splits and forms two embryos
- Chimera
  - a single organism composed of two different embryo's inner cell mass



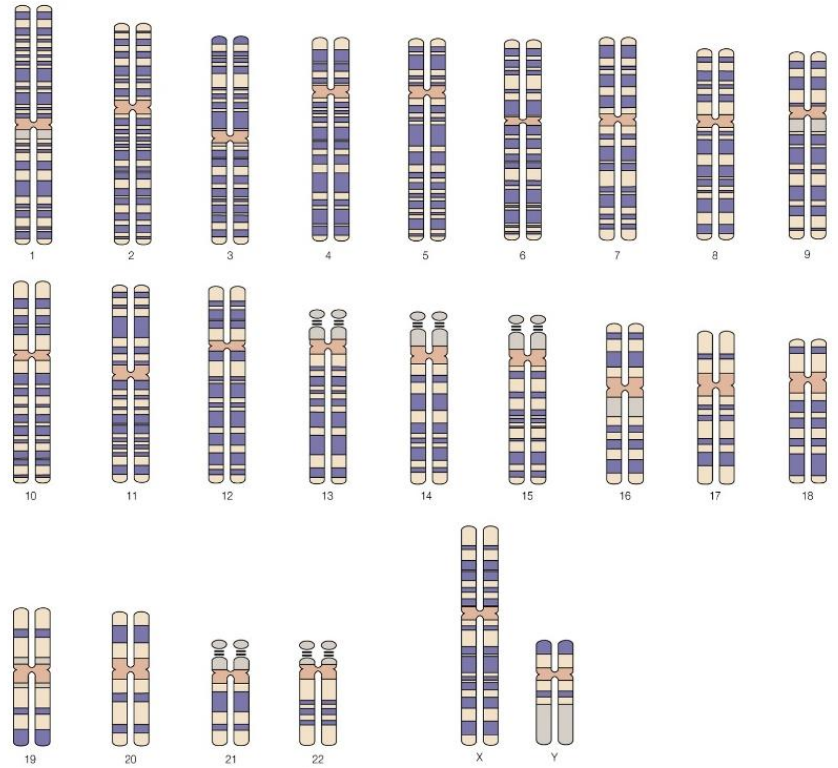
7. 성의 분화 (Sex Differentiation)

Sex Differentiation

- Primary sex determination
  - Determination of the gonads: ovaries or testes
    - Genetic
    - Environmental
      - Reptile ; depending on the temperature
- Secondary sex determination
  - Sexual phenotype outside the gonads
    - Male mammals: penis, seminal vesicles, prostate gland
    - Female mammals: vagina, cervix, uterus, oviducts, mammary glands
- Different from species to species

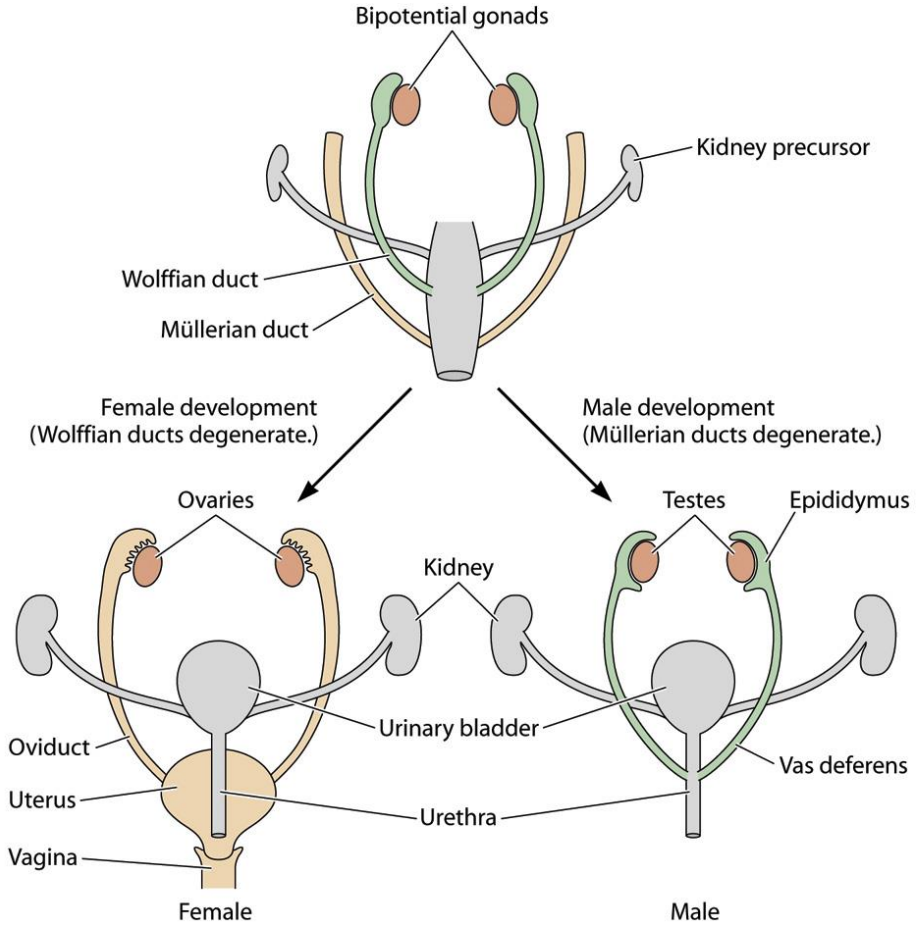
Primary Sex Determination

- 23 pairs of human chromosomes
  - Autosomes: 22 pairs (homologous chromosomes)
  - Sex chromosomes
    - Male: XY, Female: XX
    - X chromosome :1500 genes not related to gender development, essential for survival
    - Y chromosome: small, 100 genes
      - SRY: sex-determining region of the Y chromosome
      - Regulation of early gene expression



Sex Differentiation

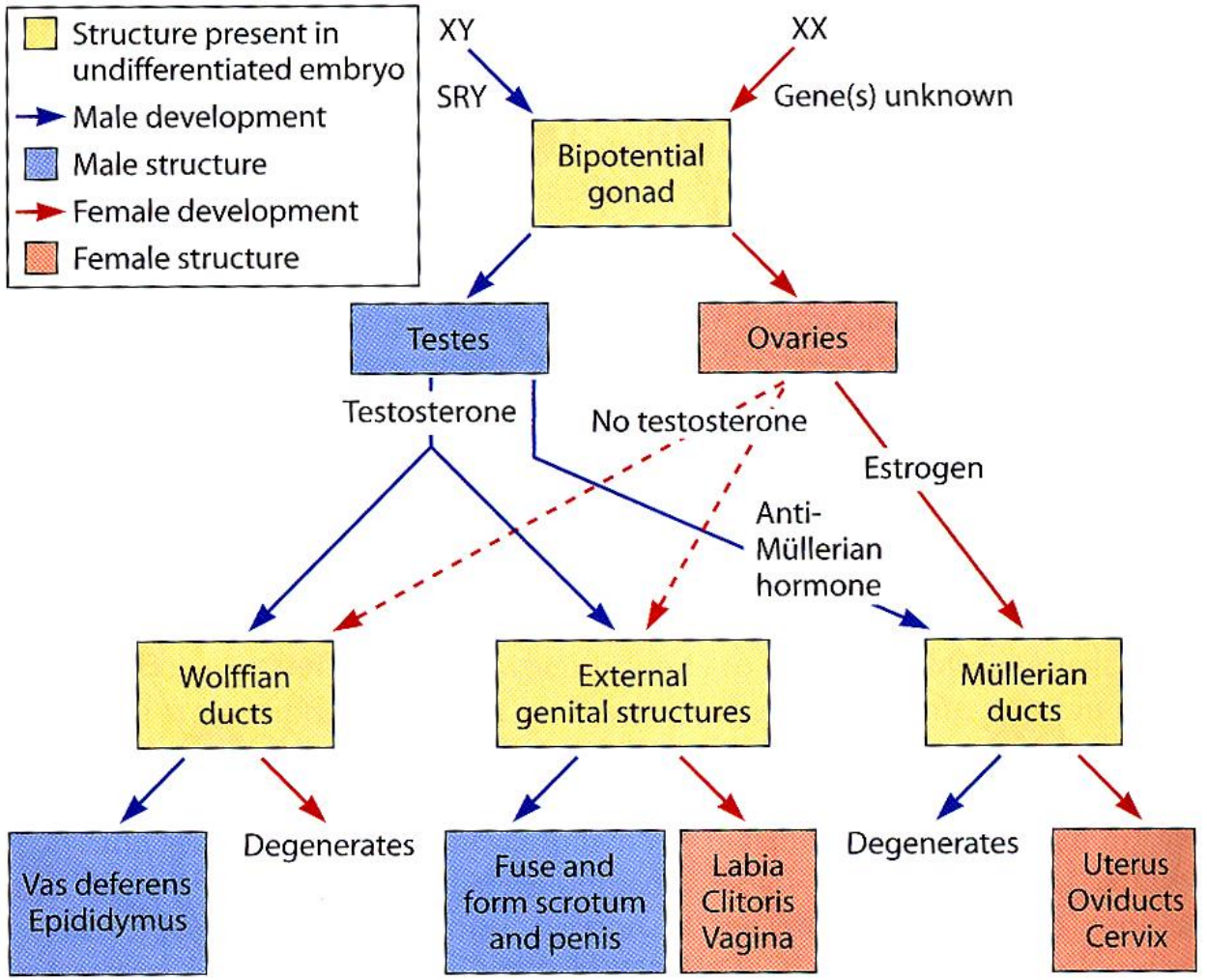
- Primordial gonadal structure : bipotential gonads
  - Mullerian and Wolffian ducts
- Female development
  - Degeneration of Wolffian ducts
  - Generation of ovaries and eggs
- Male development
  - Degeneration of Mullerian ducts
  - Generation of testes and sperms



Sex Differentiation

- With SRY
  - Expressed around week 7 of development
    - Stimulation of testes formation
  - Hormones secreted from testes
    - Anti Mullerian hormone (AMH)
    - Testosterone
      - Stimulate development of male sex organs
- Without SRY
  - Development of ovaries
  - Hormones secreted from ovaries
    - Estrogen
      - Generation of female sex organs
- Two X chromosomes are necessary for complete female sexual development
  - Turner's syndrome: one X, no Y chromosome

Sex Differentiation



Sex Hormones

- No strict female and male hormone
- Estradiol
  - Responsible for growth spurts of boys and girls at puberty
  - Conversion of testosterone to estradiol in the bone of boys
- Testosterone
  - Generated in the adrenal glands of the kidney and in the ovaries
  - Stimulation of the growth of mammary glands, uterus, and clitoris in rats
- Estrogen
  - Produced from the adrenal glands in both males and females
  - Necessary for complete development of the Wolffian ducts
  - Fertility in adult males
    - Water resorption during semen formation

8. 성 발생의 비정상적 변형

Variations in Sex Development

- Androgen (male hormone secreted from testes) insensitivity: XY female
  - Androgen: male hormone
  - Mutation of the testosterone receptor in X chromosome
    - Testes formation because of SRY gene
    - Female external genital structures

Variations in Sex Development

- DHT deficiency
  - Testosterone converted into 5α-dihydrotestosterone (DHT) in the fetal external genitalia

    Testosterone → DHT

  - Mutation of the converting gene on chromosome 2
  - High concentration of testosterone at puberty
    → development of external genitalia at puberty
  - Common in a certain population in the Caribbean

Variations in Sex Development

- CAH (Congenital adrenal hyperplasia)
  - No cortisol-synthesizing enzyme
  - Cortisol precursor is same as androgen precursor.
  - Overproduction of testosterone and other androgens from adrenal gland
  - Female fetus → Male-like genital structure

Gender Identity

- Testosterone
  - Key factor in the development of sexual identity