

Epigenetics

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Epigenetics

- Changes in gene expression that are not caused by a change in nucleotide sequence
  - Meaning is placed upon a gene without altering the nucleotide sequence
  - Often triggered by environment

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Examples of Epigenetics

1. Differentiation
2. X-inactivation
3. Genomic imprinting
4. A close-up look at specific disorders:  
*Down, Prader-Willi syndrome and Angelman syndrome*

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### Differentiation

- The process of cells specializing during development
  - Genes are silenced or “turned off”
  - The addition of methyl groups (CH<sub>3</sub>) to cytosine inactivates the gene
- All cells of the body have the same genome, but different genes are turned off in different cell types

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### X-inactivation

- An entire chromosome (with the exception of a few genes) is methylated in females
  - Creates barr body
- Random process
  - Maternally derived X is inactivated in some cells, paternally derived X is inactivated in other cells
  - Once it occurs, all descendants from that cell will have the same X inactivated
    - No difference in homozygotes, but X-inactivation may be visualized in heterozygotes

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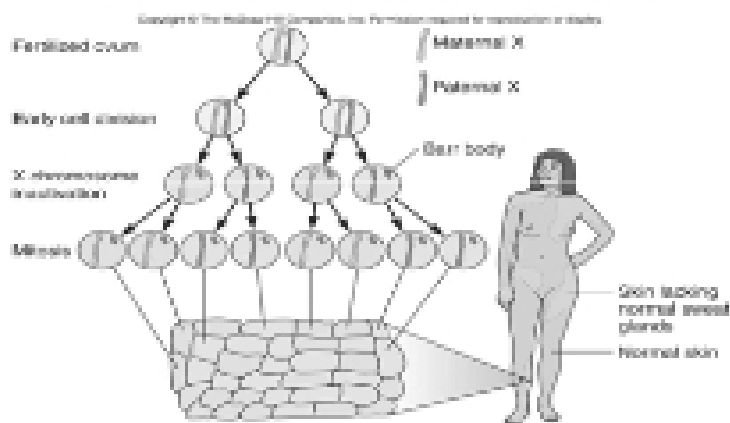
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### Observing X-inactivation: Anhidrotic Ectodermal Dysplasia




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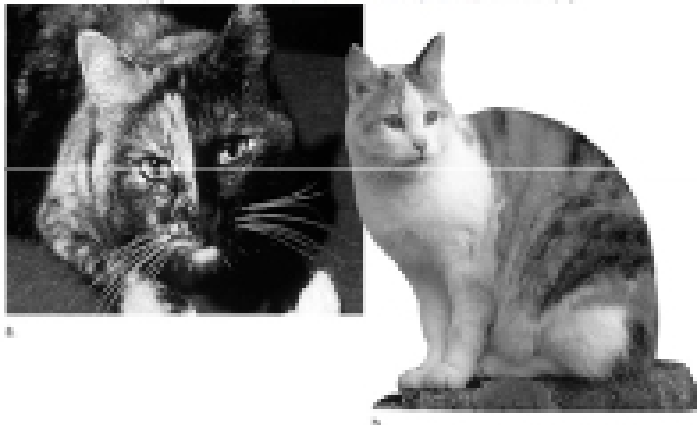
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### Observing X-inactivation: Calico Cat



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### Genomic Imprinting

- A gene is expressed when inherited from one parent and is silenced when inherited from the other parent
  - The gene that is silenced is said to be imprinted
- So, parental origin of a gene influences phenotype
- Function unknown, may play a role in development

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### Genomic Imprinting

- Imprints maintained in mitotic divisions but lost in meiosis
- Gene remains imprinted during mitosis of somatic cells
  - As if the gene remembers which parent it came from
- However, imprints are lifted and reset for the sex of the parent during meiosis

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