Genetics and Evolution 101

adapted from the University of Cal-Berkeley's webpage

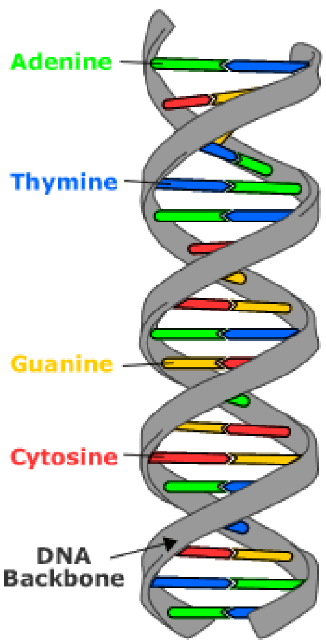
"The thing that a lot of people cannot comprehend is that Mother Nature doesn't have a bullet with your name on it, she has millions of bullets inscribed with 'to whom it may concern".

Breeders need to understand the complexity of Mother Nature's bullets - or alleles - if we are to make wise breeding decisions, no matter the strategy chosen for our breeding programs. An understanding of development is similarly important. This brief primer, adapted from the University of Cal-Berkeley's website, is presented to help understand the basics of DNA, heredity and development. To follow the lessons in a logical fashion, simply pass the links and scroll through the lessons. Otherwise, hit the link of interest. It will take you to that lesson.

"Talk to Your DNA"
by Marlis Jermutus
http://www.starseedgallery.net

Understanding DNA

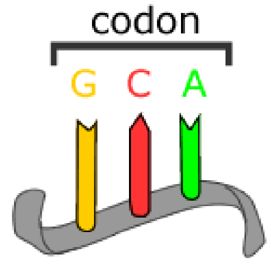
DNA contains a set of instructions for building a dog. These instructions are inscribed in the structure of the DNA molecule through a genetic code. It works like this:



DNA is made of a long sequence of smaller units strung together. There are four basic types of unit: A, T, G, and C. These letters represents the type of base each unit carries: adenine, thymine, guanine, and cytosine.

The sequence of these bases encodes instructions. Some parts of your DNA are control centers for turning genes on and off, some parts have no function, and some parts have a function that we don't understand yet. Other parts of your DNA are genes that carry the instructions for making proteins–which are long chains of amino acids. These proteins help build an organism.

Protein-coding DNA can be divided into codons–sets of three bases that specify an amino acid or signal the end of the protein. Codons are identified by the bases that make them up–in the example at right, GCA, for guanine, cytosine, and adenine. The cellular machinery uses these instructions to assemble a string of corresponding amino acids (one amino acid for each three bases). The amino acid that corresponds to "GCA" is called alanine; there are twenty different amino acids synthesized this way in humans. "Stop" codons signify the end of the newly built protein.



The completed protein is then released to do its job in the cell.

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Genetic Variation

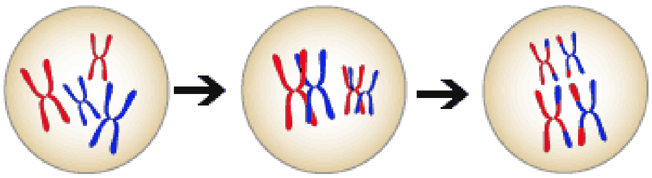
Without genetic variation, some of the basic mechanisms of evolutionary change cannot operate.

There are three primary sources of genetic variation:

1. Mutations are changes in the DNA. A single mutation can have a large effect, but in many cases, evolutionary change is based on the accumulation of many mutations.

2. Gene flow is any movement of genes from one population to another and is an important source of genetic variation.

3. Sex can introduce new gene combinations into a population. This genetic shuffling is another important source of genetic variation.



Genetic shuffling is a source of variation.

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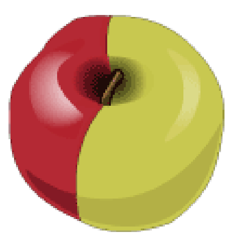
Mutations

Mutation is a change in DNA, the hereditary material of life. An organism's DNA affects how it looks, how it behaves, and its physiology–all aspects of its life. So a change in an organism's DNA can cause changes in all aspects of its life.

Mutations are random.
Mutations can be beneficial, neutral, or harmful for the organism, but mutations do not "try" to supply what the organism "needs." In this respect, mutations are random–whether a particular mutation happens or not is unrelated to how useful that mutation would be.

Not all mutations matter to evolution.
Since all cells in our body contain DNA, there are lots of places for mutations to occur; however, not all mutations matter for evolution. Somatic mutations occur in non-reproductive cells and won't be passed onto offspring.



For example, the golden color on half of this Red Delicious apple was caused by a somatic mutation. The seeds of this apple do not carry the mutation.

The only mutations that matter to large-scale evolution are those that can be passed on to offspring. These occur in reproductive cells like eggs and sperm and are called germ line mutations.

A single germ line mutation can have a range of effects:

No change occurs in phenotype.
You've probably heard of "junk DNA," DNA that doesn't seem to do anything. Mutations in junk DNA get passed on to offspring, but as far as we know, have no obvious effect on the phenotype.

Small change occurs in phenotype.

A single mutation caused this cat's ears to curl backwards slightly.

Big change occurs in phenotype.

Some really important phenotypic changes, like DDT resistance in insects are sometimes caused by single mutations[1]. A single mutation can also have strong negative effects for the organism. Mutations that cause the death of an organism are called lethals–and it doesn't get more negative than that.

There are some sorts of changes that a single mutation, or even a lot of mutations, could not cause. Neither mutations nor wishful thinking will make pigs have wings; only pop culture could have created Teenage Mutant Ninja Turtles–mutations could not have done it.

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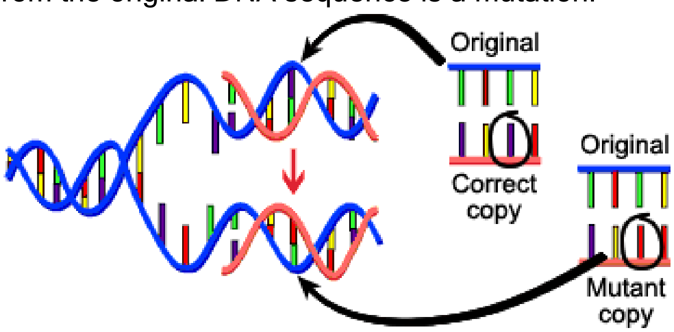
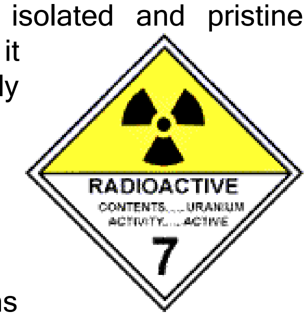
The Causes of Mutations

Mutations happen for several reasons.

1. DNA fails to copy accurately.
   Most of the mutations that we think matter to evolution are "naturally-occurring." For example, when a cell divides, it makes a copy of its DNA–and sometimes the copy is not quite perfect. That small difference from the original DNA sequence is a mutation.



2. External influences can create mutations.
   Mutations can also be caused by exposure to specific chemicals or radiation. These agents cause the DNA to break down. This is not necessarily unnatural–even in the most isolated and pristine environments, DNA breaks down. Nevertheless, when the cell repairs the DNA, it might not do a perfect job of the repair. So the cell would end up with DNA slightly different than the original DNA and hence, a mutation.



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Types of Mutations

Knowing a few basic types of mutations can help you understand why some mutations have major effects and some may have no effect at all.

Substitution

A substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G). Such a substitution could:

1. change a codon to one that encodes a different amino acid and cause a small change in the protein produced. For example, sickle cell anemia is caused by a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein produced.

2. change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations.

3. change an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein. This can have serious effects since the incomplete protein probably won't function.

Insertion

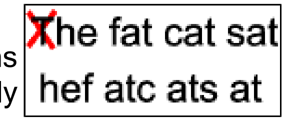
Insertions are mutations in which extra base pairs are inserted into a new place in the DNA.

Deletion

Deletions are mutations in which a section of DNA is lost, or deleted.

Frameshift

Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly parsed. These changes are called frameshifts. For example, consider the sentence, "The fat cat sat." Each word represents a codon. If we delete the first letter and parse the sentence in the same way, it doesn't make sense. In frameshifts, a similar error occurs at the DNA level, causing the codons to be parsed incorrectly. This usually generates proteins that are as useless as "hef atc ats at" is uninformative.



There are other types of mutations as well, but this short list should give you an idea of the possibilities.

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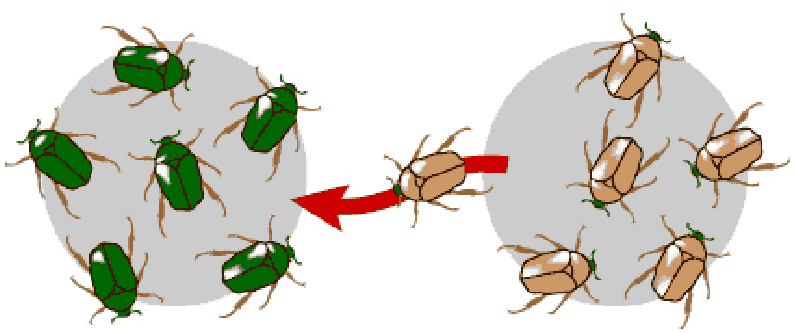
Gene Flow

Gene flow—also called migration—is any movement of genes from one population to another. Gene flow includes lots of different kinds of events, such as pollen being blown to a new destination or people moving to new cities or countries. If genes are carried to a population where those genes previously did not exist, gene flow can be a very important source of genetic variation. In the graphic below, the gene for brown coloration moves from one population to another.



Gene flow has several important effects on evolution:

Within a population:

It can introduce or reintroduce genes to a population, increasing the genetic variation of that population.

Across populations:

By moving genes around, it can make distant populations genetically similar to one another, hence reducing the chance of speciation. The less gene flow between two populations, the more likely that two populations will evolve into two species.

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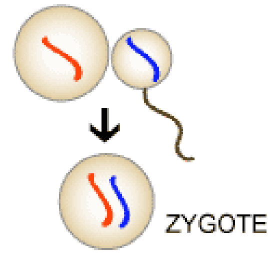
Sex and Genetic Shuffling

Recombination introduces new gene combinations into populations.

Here is a quick and dirty review of the genetics of sexual canine reproduction.

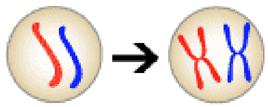
Genes are located on long chains of DNA called chromosomes.



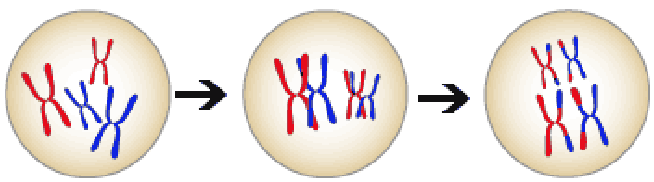
Canines have 39 pairs of chromosomes: one member of each pair was inherited from the dam and the other from the sire. Correspondingly, dogs have two versions of every gene, one from the mother and one from the father.

If dogs reproduced by taking 39 pairs of chromosomes from the dam and 39 pairs of chromosomes from the sire, the puppy would have too many chromosomes (78 pairs). So eggs and sperm carry only half the usual number of chromosomes–just 39 unpaired chromosomes, carrying one version of each gene. When the egg and sperm get together, the puppy receives the normal 39 matched pairs.

When eggs and sperm are produced, the parent cell first copies each chromosome, leaving the duplicate pairs attached to one another.

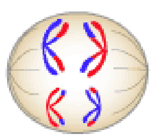


Producing eggs and sperm is the first opportunity for mixing and matching genes. When the dam makes an egg, her chromosomes first find their matched partners and exchange some DNA with each other. That's called recombination. Because of this shuffling, genes from the dam's dam and genes from the dam's sire can wind up next to one another on the same stretch of DNA. (The same thing happens in the sire's sperm.)

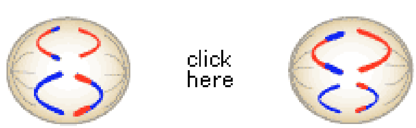


Only after chromosomes recombine do they segregate into different egg cells, so that each egg cell ends up with one version of each chromosome.

Meiosis, step one (click the image for animation):



Meiosis, step two (click the image for animation):



When egg and sperm meet, the puppy inherits a combination of genes that is totally unique: it carries versions of genes from all 4 grandparents plus any mutations that occurred when the dam and sire were making the egg and sperm.

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Genetic Drift

Genetic drift–along with natural selection, mutation, and migration–is one of the basic mechanisms of evolution.

In each generation, some individuals may, just by chance, leave behind a few more descendents (and genes, of course!) than other individuals. The genes of the next generation will be the genes of the "lucky" individuals, not necessarily the healthier or "better" individuals. That, in a nutshell, is genetic drift. It happens to ALL populations–there's no avoiding the vagaries of chance.

Look at this cartoon. Genetic drift affects the genetic makeup of the population but, unlike natural selection, through an entirely random process. So although genetic drift is a mechanism of evolution, it doesn't work to produce adaptations.

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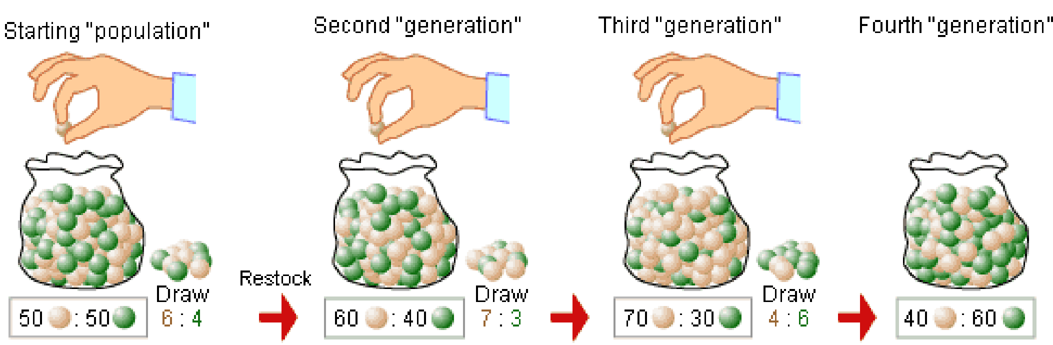
Sampling Error and Evolution

Genetic drift–one of the basic mechanisms of evolution–is simply the evolutionary equivalent of a sampling error.

Imagine a game in which you have a bag holding 100 marbles, 50 of which are brown and 50 green. You are allowed to draw 10 marbles out of the bag. Now imagine that the bag is restocked with 100 marbles, with the same proportion of brown and green marbles as you have just drawn out. The game might play out like this:



It's clear that the ratio of brown to green marbles "drifts" around (5:5, 6:4, 7:3, 4:6 . . .)

This drifting happens in populations of organisms. Due to many random factors, the genes in one generation do not wind up in identical ratios in the next generation, and this is evolution. It is possible for the frequency of genes for brown coloration to increase in a population of beetles without the help of natural selection. While this is evolution, it is evolution due to chance, not selection.

Genetic drift has several important effects on evolution:

1. Drift reduces genetic variation in populations, potentially reducing a population's ability to evolve in response to new selective pressures.
2. Genetic drift acts faster and has more drastic results in smaller populations. This effect is particularly

important in rare and endangered species.

3. Genetic drift can contribute to speciation. For example, a small isolated population may diverge from the larger population through genetic drift.

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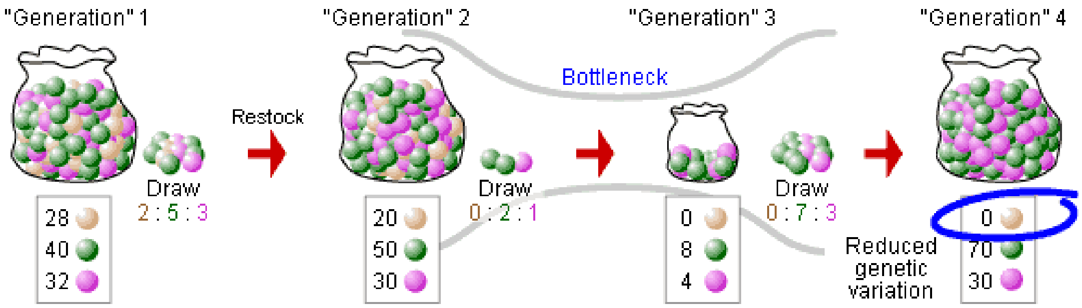
Bottlenecks and Founder Effects

Genetic drift can cause big losses of genetic variation for small populations.

Population bottlenecks occur when a population's size is reduced for at least one generation. Because genetic drift acts more quickly to reduce genetic variation in small populations, undergoing a bottleneck can reduce a population's genetic variation by a lot, even if the bottleneck doesn't last for very many generations. This is illustrated by the bags of marbles shown below, where, in generation 2, an unusually small draw creates a bottleneck.



Reduced genetic variation means that the population may not be able to adapt to new selection pressures, such as climatic change or a shift in available resources, because the genetic variation that selection would act on may have already drifted out of the population.

An example of a bottleneck:
Northern elephant seals have reduced genetic variation probably because of a population bottleneck humans inflicted on them in the 1890s. Hunting reduced their population size to as few as 20 individuals at the end of the 19th century. Their population has since rebounded to over 30,000—but their genes still carry the marks of this bottleneck: they have much less genetic variation than a population of southern elephant seals that was not so intensely hunted.

Founder effects

A founder effect occurs when a new colony is started by a few members of the original population. This small population size means that the colony may have:

- reduced genetic variation from the original population.
- a non-random sample of the genes in the original population.

For example, the Afrikaner population of Dutch settlers in South Africa is descended mainly from a few colonists. Today, the Afrikaner population has an unusually high frequency of the gene that causes Huntington's disease, because those original Dutch colonists just happened to carry that gene with unusually high frequency. This effect is easy to recognize in genetic diseases, but of course, the frequencies of all sorts of genes are affected by founder events.

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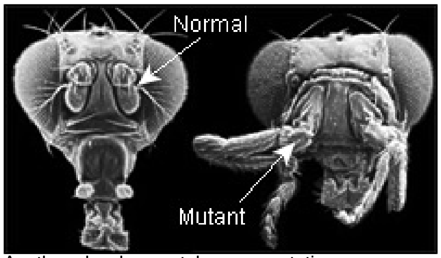
Development

Development is the process through which an embryo becomes an adult organism and eventually dies. Through development, an organism's genotype is expressed as a phenotype, exposing genes to the action of natural selection.

Studies of development are important to evolutionary biology for several reasons:

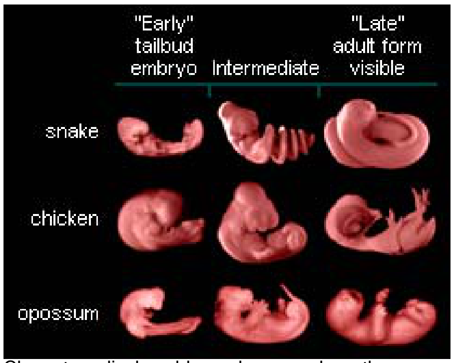
Explaining major evolutionary change
Changes in the genes controlling development can have major effects on the morphology of the adult organism. Because these effects are so significant, scientists suspect that changes in developmental genes have helped bring about large-scale evolutionary transformations. Developmental changes may help explain, for example, how some hoofed mammals evolved into ocean-dwellers, how water plants invaded the land, and how small, armored invertebrates evolved wings.

Mutations in the genes that control fruit fly development can cause major morphology changes, such as two pairs of wings instead of one.



Another developmental gene mutation can cause fruit flies to have legs where the antennae normally are, as shown in the fly on the right.

- Learning about evolutionary history
  An organism's development may contain clues about its history that biologists can use to build evolutionary trees.



Characters displayed by embryos such as these may help untangle patterns of relationship among the lineages.

- Limiting evolutionary change
  Developmental processes may constrain evolution, preventing certain characters from evolving in certain lineages. For example, development may help explain why there are no truly six-fingered tetrapods.

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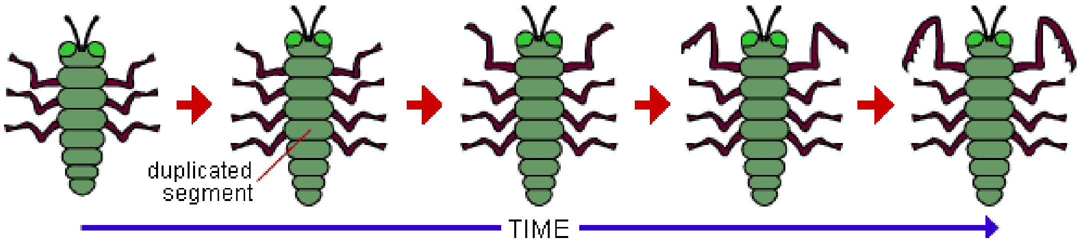
Types of Developmental Change

Developmental changes have likely been involved in important morphological changes—like the evolution of novel and complex structures.

Here are a few of the sorts of developmental changes that may have been important:

Module duplication and adaptation
A module refers to a unit that can be duplicated and further adapted. For example, arthropods have various numbers of body segments. Segment duplication and loss is a developmental change that probably occurred many times in the evolution of this clade. The graphic below shows a hypothetical example of module duplication and adaptation.



Individualization
This is the modification of a particular module, usually when there is selection for a specialized function.

Scorpion image courtesy of the California Academy of Sciences.
Spider image courtesy of Michael Hedin, San Diego State University.

Heterochrony
Heterochrony is a change in the timing of developmental events. For example, a change in timing might slow down the development of the body, but not alter the maturation of the reproductive system. This change yields an adult organism with a form similar to the ancestral juvenile form.

Larval salamander image courtesy of Jeff LeClere.
Tiger salamander image courtesy of Greg Sievert.
Axolotl image courtesy of Barbara Shardy.

Salamanders go through a larval stage in which they have feathery, external gills (left). Most salamanders lose these gills when they metamorphose into adults (center). Because of heterochrony, axolotls now retain the juvenile external gills as fully reproductive adults (right).

Allometric growth
Allometric growth is a change in the rate of growth of a dimension or feature relative to other features. For example, we can describe some of the evolutionary changes that produced bats in terms of allometry. Bat wings are basically paws with really long fingers and skin stretched between them. In order for these wings to evolve, the rate of growth of finger bones must have increased relative to the growth of the rest of the bat's body—or

perhaps the rate of growth of the rest of the body decreased relative to the fingers. Either way, it is allometry.
Bat image courtesy of Ben Waggoner.

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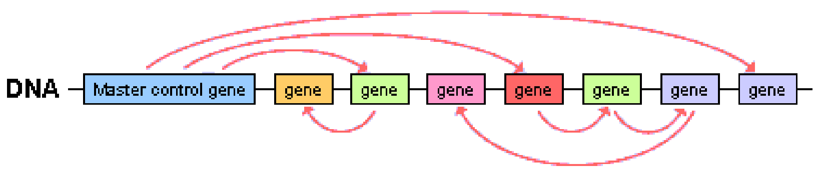
Understanding Complexity

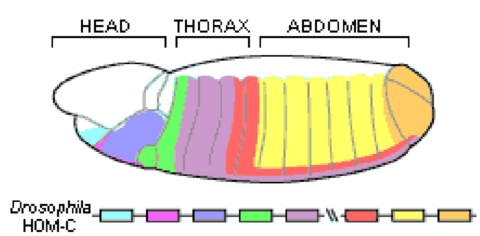
Going from a simple fertilized egg cell to a fly over the course of a few weeks is an amazing transformation. A lot of things have to happen: the body form must be laid out (head/tail, back/belly), different tissues must be built, and organs must be grown. The adult fly is composed of a lot of very different parts made of different cells –yet every cell carries the same genetic instructions. So how does it "know" what to do?

- Certain genes control where and when other genes are expressed.
  Not all genes code for "building material" proteins (such as keratin that makes up part of your skin, or rhodopsin that makes eyes sensitive to light). Regulatory genes control when and where other genes get turned on. For example, these sorts of genes tell the cells of the fly when and where to start building wings. This occurs during the larval stage on the second and third segments of the thorax. Regulatory genes can start a "chain reaction" of effects, turning on and off other genes, whose products affect other genes, whose products in their turn affect other genes, and so on. A single regulatory gene can thus control the construction of a body part as complex as a leg or eye.



The diagram above illustrates how the master control gene regulates other genes, which in turn, regulate other genes.

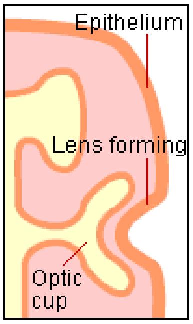
- Different cells have different genes expressed.
  For example, eye cells turn on the genes that make proteins necessary for vision–but the cells lining the digestive tract don't turn on these genes. Instead, they turn on genes that create digestive enzymes.



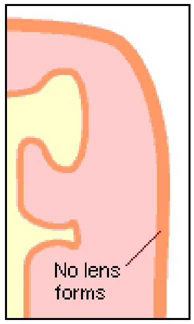
Different segments of the developing fly embryo express different genes.

- Chemical signals also influence the fate of cells.

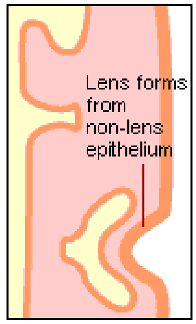
Chemical signals from the environment and from other cells can affect which genes are turned on in a particular cell. For example, in the developing vertebrate eye, chemical signals from the retina probably cause adjacent cells to become lens cells instead of some other type of cells. Here we see a diagram of the optic cup, of which the retina is part, developing normally. As a result, it sends signals to nearby cells, causing them to form a lens from the epithelium. The pictures below illustrate what happens to lens development if the optic cup is removed, transplanted, or replaced by other tissue.



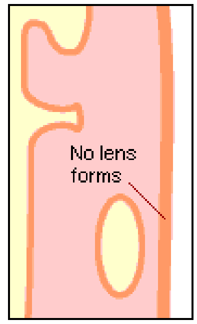
Optic cup removed:



Optic cup transplanted:



Other tissue transplanted:



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