Evolution & Mendelian Genetics

Background

Evolutionary theories require that there be a mechanism (or mechanisms) for transferring characteristic trait information from parents to offspring - and from ancestors to descendants.

- This transfer of traits is observed in nature.

Speciation requires that the information carried by this mechanism be subject to modification by blending, copying errors and/or external influences - Speciation is observed in nature.

The properties of this transfer mechanism were determined from experiments conducted by Gregor Mendel (1822-1884). Mendel's conclusions are contained in two empirical laws :

• The Law of Segregation,

The Law of Independent Assortment

Mendel's Laws are, essentially, the "rules" governing the transfer of traits from ancestors to their descendants. Mendel's Laws enable quantitative predictions of how traits are made manifest (or not) in successive generations.

Mendelian Genetics

Some History

Mendel originally intended to illustrate and verify Lamarck's views on evolution, but found instead that offspring possessed the basic traits of their parents, independent of the external environmental conditions.

Mendel was aware of the works of Darwin when he published the results of his studies in 1865. However, this work was not "discovered" until 1900, well after the 1859 publication of Darwin's "The Origin of Species ..."

The information-carrying units are called genes. The corresponding physical bodies are the chromosomes (observed by Hoffmeister in 1848) which bear the information-carrying molecules of DNA (identified by Meischer in 1869). Note that these discoveries antedated the works of both Darwin and Mendel.

The double-helix structure of the DNA molecule, and the mechanism of its replication was finally described and explained by Crick & Watson in 1953.

The basic mechanism of transfer of traits from ancestor to offspring, and the processes giving rise to Mendel's Laws are now understood at the levels of molecular chemistry - biochemistry.

<u>Mendel's Laws of Genetics</u> Gregor Mendel (1822 - 1884) *Versuche über Pflanzen-Hybriden* (1865) ("Experiments in Plant Hybridization")

Foundations of Genetics: Mendel's Experiments

Experiments involved breeding and cross-breeding 28,000 pea plants. (Note that these pea plants are capable of self-fertilization.) Exceedingly careful experimental techniques, environmental controls, etc.

Peas

Recorded Traits (and Characteristics)#1: Seed form ("round" & "wrinkled")#2: Seed albumen color ("yellow" & "green")#3: Flower color ("violet" & "white")= Seed coat color ("gray-brown" & "white")#4: Seed pod form ("inflated" & "constricted")#5: Seed pod color ("green" & "yellow")= Stalk-leaf-calyx color ("green" & yellow")#6: Flower position on stem ("axial" & "terminal")#7: Stem length ("long" & "short")

...basically seven independent traits, with only two characteristics each





Mendelian Genetics

Mendel's Experiments: Methodology • Create pure strains true-breeding for some trait characteristic (*e.g.* round seeds) or group of trait characteristics (*e.g.* round, yellow seeds white flowers,...)

Cross-breed pure (but different) strains to produce hybrids

• Observe trait characteristics of these hybrids ... establishing which trait characteristics are dominant (*e.g.* green dominates yellow in seed color)

Observe characteristics of the offspring of self-fertilizing hybrids

Experimental Result: Mendel's Laws of Genetics Law of Segregation

Allele <u>pairs</u> separate (segregate) during gamete formation and randomly unite at fertilization. Offspring receive one from each parent. The two alleles determine the character of the trait.

Law of Independent Assortment

Allele pairs separate independently during gamete formation so that traits are transmitted independently of one another.

Mendel's First Set of Experiments

For clarity, consider a single Trait with two Characters, G and g (*e.g.*, Seed color: G = "Green", g = "not green") [We assign the capital letter to the trait characteristic which turns out to be dominant.]

Procedure

Generate two pure (true-breeding) strains of green (G) and yellow (g) character.

Cross-breed the green and yellow strains. These are the parental (P) generation.

<u>Result</u>: The offspring, the <u>first filial</u> (F1) generation are always 100% green.

Self-pollinate the F1 generation to produce the <u>second filial (F2)</u> generation.

<u>Result</u>: On the Average 75% of the F2 generation are green (G), 25% yellow (g).

<u>Hypothesis</u>: Offspring acquire half of their traits from each parent. If of different character, one is always dominant. The <u>gene</u> of a plant consists of an <u>allele</u> pair.

Tests of Hypothesis: Repeat experiment using true breeding stains displaying differing characteristics of some other trait (*e.g.*, stem length). Confirm that hybridizing with characters (Y) and (y) <u>always</u> gives an F1 generation displaying (Y) only - and the F2 generation, *on the average*, gives (Y) and (y) in a 3:1 ratio.

Mendel's Law of Segregation

Allele pairs separate (segregate) during gamete (sperm or ovum) formation and unite at fertilization. Offspring therefore receive one allele from each parent. The two alleles (one of which may be dominant) determine the character of the trait.

> **Explaining the First Experiment** Parents (P) are GG and gg

F1 Generation: $GG + gg \rightarrow Gg \& gG$ Hence <u>all</u> offspring display the dominant "G" character

F2 Generation: Gg \rightarrow GG, Gg, gG & gg 3/4 of offspring display the "G" character, 1/4 the recessive "g" character

Digressions: Problem: Producing true breeding ("pure) strains (GG and gg) Caution: Random combinations and statistical results.

> A New Question: Are traits transmitted independently?

Mendel's Second Set of Experiments

Procedure

Begin with two strains true-breeding for a pair of traits say (G +Y) and (g + y)

Cross-breed the (G + Y) and (g + y) strains - the parental (P) generation.

<u>Result</u>: The first filial (F1) generation are always all (100%) of character (G + Y).

Self-pollinate the F1 generation to produce the second filial (F2) generation.

<u>Result:</u> On the Average

56.25% of the F2 generation are (G + Y) 18.75% of the F2 generation are (G + y) 18.75% of the F2 generation are (g + Y) 6.25% of the F2 generation are (g + y)

<u>Hypothesis</u>: Offspring acquire half of all traits from each parent. The allele for each trait is assigned randomly during gamete formation: Traits are transferred independently.

Test of Hypothesis: Repeat these experiments using true breeding stains displaying different pairs of characteristics of other traits. Confirm that this always gives an F1 generation displaying only the dominant pair and that the F2 generation has the above proportions of dominant/recessive combinations.

Mendel's Law of Independent Assortment

Allele pairs separate (segregate) independently during gamete (sperm or ovum) formation so that traits are transmitted independently of one another.

Explaining the Second Experiment Parents (P) are GGYY and ggyy

F1 Generation: $GGYY + ggyy \rightarrow GgYy$ Hence all offspring always display the dominant "G +Y" characters

> F2 Generation: $GgYy + GgYy \rightarrow$ GG, Gg, gG & gg in combination with YY, Yy, yY, & yy There are 4 x 4 = 16 combinations of traits There are 3 x 3 = 9 combinations giving (G + Y) There are 1 x 3 = 3 combinations giving (g + Y) There are 3 x 1 = 3 combinations giving (G + y) but only 1 combination (*i.e.*, ggyy) giving (g + y)

The proportions are 9/16 = 56.25%, 3/16 = 18.75%, 3/16 = 18.75%, and 1/16 = 6.25% ... as is observed!

One can further test these two Mendelian Laws by hybridizing strains breeding true for three (or more) traits (GYX with gyx, for example). Both laws hold under these more complex situations so no additional laws of genetics are required.

Notes on Mendelian Genetics

Mendel's Laws are empirical - based upon experiments and observation.

Mendel's experiments only examined traits determined by a single gene.

Mendelian genetics are probabilistic: Predictions are for statistical averages.

Some traits are <u>perfectly</u> correlated, suggesting a single gene determines both. (*e.g.*, pod color and stem color among Mendel's peas.)

Other traits are correlated, suggesting some genes contribute to several traits. (*e.g.*, blonde hair and blue eyes)

Mendel found that offspring possessed the basic traits of their parents, independent of the (experimentally controlled) environmental conditions. The Lamarckian hypothesis is not supported!

Mendelian genetics <u>requires</u> a genetic change ("mutation") if evolution is to occur. The unmodified genetic lineage of G & g alleles can only be GG, Gg, and gg - nothing else.

To this point the concept of "genes" is still an abstraction in the sense that the physical mechanism of information storage (*e.g.*, DNA) is not specified. It is sufficient to recognize that some such mechanism must exist. (*cf.* the status of Kepler's Laws)

Mutation and Mendelian Genetics

A lineage (ancestors and descendants) based upon the (G,g) allele pair can consist only of individuals with GG, Gg, gG, and gg allele pairs. (The character is either G or g.)

An allele might be altered prior to fertilization (*e.g.,* during gamete formation) and the fertilized ovum would then have a Gx or gx gene. The subsequent lineage might include Gx, gx, and xx pairs in addition to GG, Gg, and gg (The characters could now be G, g, or x.)

Possible Causes of Mutation

Environmental Influences (radiation, cosmic rays, poisoning, heat,) Deliberately acquired changes coupled to environment? (*cf*., Lamarckism) Copying Errors & Genetic Drift

(Note: There are some provisions for "self repair" of damaged genes.)

Possible Consequences

(from most probable to least likely)

Fertilization does not occur.

• The mutation is lethal: The offspring do not live to reproduce.

• The altered lineage terminates: The offspring are sterile.

• The offspring are fertile but "flawed".

The offspring are fertile and "improved"

The last two outcomes are subject to the pressures of <u>Natural Selection</u>!

Mutation and Evolution

Diversity and Diversification

A propagated mutation (*i.e.*, fertile offspring) can lead to diversity in traits. GG, Gg, gg \rightarrow GG, Gg, gg, Gx, gx, xx

The Rôle of Natural Selection

General vigor of the trait - bearing members of the species

Improved adaptation to the ambient environment

Ability to respond to environmental changes ("hidden talents")

Digression: Convergent Evolution

Inventions and reinventions: Limbs, wings, and fins Ears, eyes, and other senses

... and atrophy

Comment: Evolution by descent with modification together with the workings of natural selection will be neither unidirectional nor optimal. *I.e.,* there is no intelligence in the design ...