

## What is Genetics?

It is the field that studies the “language” that dictates the nature of all living things!

This is how families retain similarity

The study of **heredity** (the transmission of characters from parents to offspring).

The study gene expression and of **variation** among individuals in populations.

This is why individuals are different!!

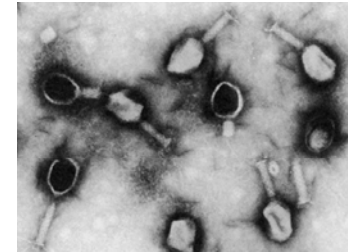
## Why is Genetics Important?

1) Field that studies the common “language” or source of biological information used by all living things (and some not so living things).

**Vertebrates**

to

**Viruses**



DNA creates the “blueprint” for all living things, but RNA is used in some

2) It's the study of Inheritance or Heredity: Why offspring look like their parents.

His Mom  
Snuppy

His Clone



3) It's the study of Variation: Why no two individuals are exactly the same.



Mary-Kate and Ashley use to be identical.

Now that they are older, they are not!!!

Genes and Environment influence the patterns of variation that we see!  
And finance influence how we label these differences.



The “**Variation**” we do see is created by mutations and recombination.

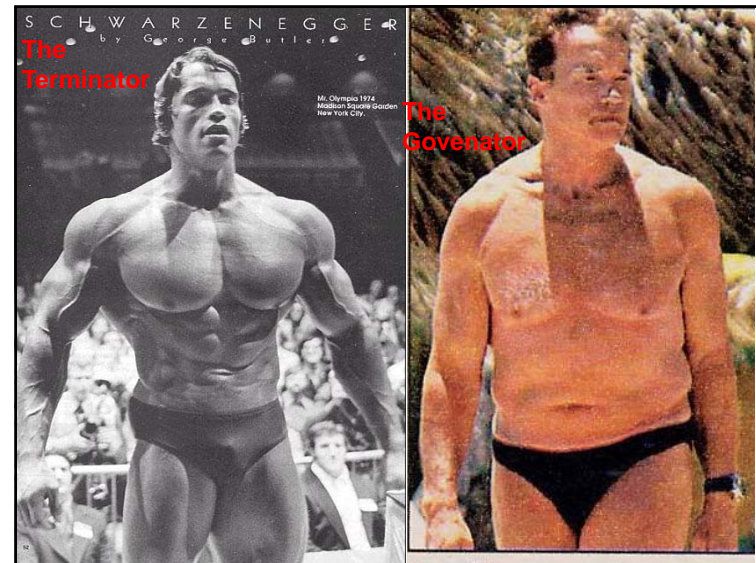
The raw material used by evolutionary processes (**Natural Selection**) to shape the biological diversity (biodiversity) that we see in the world.

The raw material that has allowed humans to use **Artificial Selection** to shape the domesticated species that we



5 genes here

Is all the variation that we see caused by changes in our ge



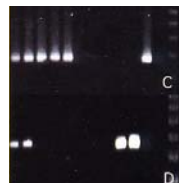
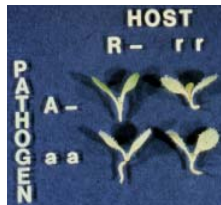
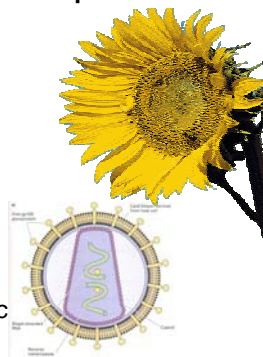
Important practical implications:

#### Agriculture

Breeding  
GMOs

#### Human Health

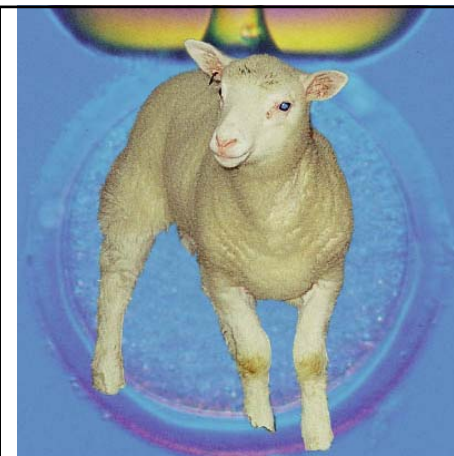
HIV  
BRCA1  
Antibiotic  
Resistant  
H1N1



Many other areas: Forensics (DNA fingerprinting),  
Environmental protection/bio-remediation....  
Cloning (Dolly, horse, Raelians)

#### Genetics in the News

Who is Dolly and what happened to her?



(a) Dolly

Credit: I. Willmut, A.E. Schnleke, J. McWhir, A.J. Kind & K.H.S. Campbell. With permission from Nature, from cover of Nature, Feb. 27, 385: Issue 6619, 1997.

Technical Sidelight – Dolly the cloned sheep

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## N Korea orders UN team to leave

- Doubt and disgust over birth of 'Eve'
- Four more on way, claims scientist

By TOBY HARDEN in WASHINGTON  
AND BOBBI HIGGINS, SCIENCE EDITOR

A SCIENTIST belonging to a cult called the Raelians that believes mankind was created by aliens claimed yesterday that one of its followers had given birth to the world's first human clone, a girl called Eve.

In the face of intense skepticism and revulsion from scientists worldwide, the acceptance of the use of human eggs by Rosseter, a French chemist, had to wait. She set her company, Clinard, but it was not until 1964 that the tests would confirm she was an exact peer-to-peer of her mother.

By 1967, the French doctor, Jean Bado, who was not a specialist in reproductive medicine, said a price of \$100,000 was being offered for the assurance the alleged birth. The oldest child, she said, had been born by cesarean section, and the mother was a French doctor.

Independent expert would say the child was the daughter of the mother.

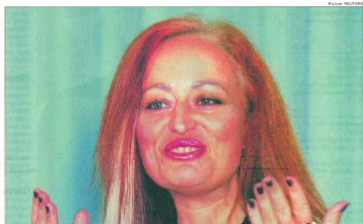
Rosseter had suffered no reaction to her lack of claim.

She said she had a "very persistent genetic test," said Dr. Robert Leach, of the National Institute of Health, who had been a member of the scientific committee.

The scientists also expressed grave concerns about the ethical and medical aspects of the use of human eggs, and the fact that the child was born by cesarean section, and the mother was a French doctor.

In 1967, by Claude Vincent, a French French doctor, and a French doctor, who was a member of the scientific committee.

former motor racing journalist who says he was once abducted by extra-



By TOBY HARRISON

NORTH KOREA ordered United Nations inspectors monitoring its nuclear reactors to leave the country yesterday, intensifying the crisis over the communist dictatorship's weapons programme.

The United States was accused of making North Korea "the target for a nuclear pre-emptive strike".

North Korea's Yongbyon nuclear complex, which is capable of producing weapons-grade plutonium, was being reactivated, the letter said, because President George W. Bush had cancelled shipments of fuel oil promised in 1994 and said the country was part of an "axis of evil" with Iran and Iraq.

The White House condemned

The White House condemned

Who is Eve and what happened to her?

The Boston Globe

## A deadly disease is said to lack cure

The spread of a new, highly resistant form of tuberculosis that is "virtually untreatable" is causing alarm among international health officials who say that it has now been identified in "all regions of the world," according to the World Health Organization.



**Use Genetics to screen for diseases.**

Colleen and Jeff Convery hold their daughter, Erika, who was conceived after fertility specialists at Brigham and Women Hospital screened the parents' fertilized eggs to avoid Duchenne muscular dystrophy, a disease that strikes mainly boys.

## Muscular Dystrophy

## A very early checkup

## Genetic screening of embryos helps ease parents' fears, but is it a step toward 'designer babies'?

By M.E. Malone

Colleen Convery did not want her baby to suffer the same fate as her three



**Use Genetics to produce resistant crops and reduce pesticide use.**

**Fig 1.10b Side-by-side comparison of corn stalks from plants that are resistant (top) and susceptible (bottom) to the corn borer.**

## Use genetics to look at human history!!

### Women have spread genes farther, study says

ASSOCIATED PRESS

NEW YORK – Women have done better than men have at spreading their genes around the globe, a new study suggests.

The explanation, according to researchers: Through history, women moved to their mates' communities to start families.

Over thousands of years, these short migrations apparently enabled

#### Explanation: Migration to start families

women's genes to get around more than men's, said researcher Mark Seielstad.

Seielstad, a fellow in the program for population genetics at the Harvard School of Public Health, did the work with L. Luca Cavalli-Sforza and Eric Minch at the Stanford University School of Medicine. Their

findings were published in the November issue of the journal *Nature Genetics*.

Another genetics specialist said he suspects there is more to the story.

The study used data on modern-day genes. It looked for the effects of men and women moving away

from their native communities over time and producing children somewhere else. It relied on the fact that the Y chromosome is passed only by fathers. In contrast, ge-

net material called mitochondrial DNA, or mtDNA, is passed along on mothers.

Both kinds of genetic material contain tiny variations that are tracked in different populations. More a specific variation is common to a particular population, the

**Mitochondrial Genes are maternally** inherited and can be used to track female lineage;  
Genes on **Y chromosome** are **paternally** inherited and can be used to track male lineage.

Atlas of the Human Journey:

<https://www3.nationalgeographic.com/genographic/atlas.html>

## Use of genetics to fight crime!! Forensics...CSI

### Dog's DNA may be the smoking gun in Wash. murder trial

AP/WIDEWORLD

SEATTLE – DNA from a dog may be used as evidence in a murder trial in Washington, perhaps the first such use in the United States.

King County Superior Court Judge Richard Jones has given preliminary approval for prosecutors to submit DNA blood tests from a pit bull that was shot and killed – along with its two owners – in a South Se-

attle house in 1996.

Prosecutors say the dog's DNA is crucial to their case.

Except for identical twins, no two people have the same genetic fingerprint found in the DNA in every cell of their bodies. Forensic courts are using DNA tests in criminal trials as evidence to link defendants to crime scenes or victims through semen, blood, hair, saliva or other biological traces.

In the case involving the pit bull, prosecutors say two men charged in the slayings had the dog's blood on their jackets – evidence that places both men at the scene of the killings.

A California company – PE Ag-Gen – extracted bloodstains from its spots on clothing and matched them with blood from the dog. The test showed there was one chance in 100 million that the blood was not the dog's, prosecutors said.

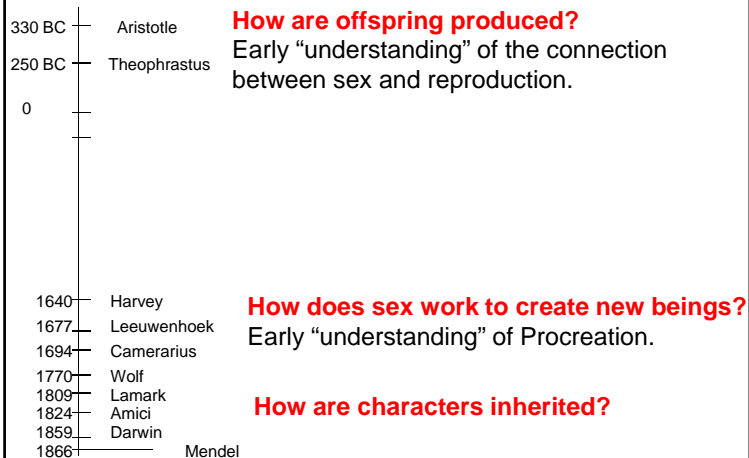
"I think it [the dog DNA] is extremely relevant to our case," Timothy Brinkshaw, senior deputy prosecutor, said. "It's the most probative evidence and the most objective evidence that the defendants were present while the crime was committed."

The dog, a pit-bull mix named Chief, died 20 hours after being shot, reportedly in a house in South Park on Dec. 9, 1996. Chief's owners, Jay Johnson, 25, and Rafael Rivera, 20,

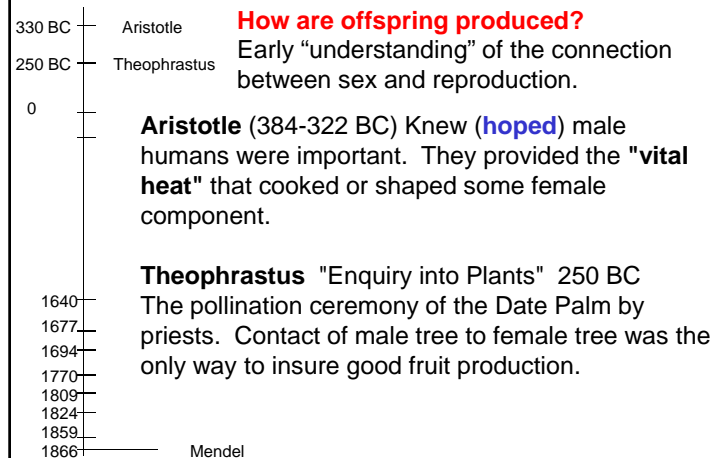
prosecutors say the men accused of the killings – Trifunovic and Kennedy Leitch – killed down the street of the house after Johnson refused to sell them marijuana, then shot the dog, Johnson and John Rivera.

Jay Halpern, assistant at PE AgGen, said that DNA was used in a case in Canada, but she thinks that will be the first time dog DNA has been used in the United States.

## 2500 Year History of Genetics



## 2500 Year History of Genetics





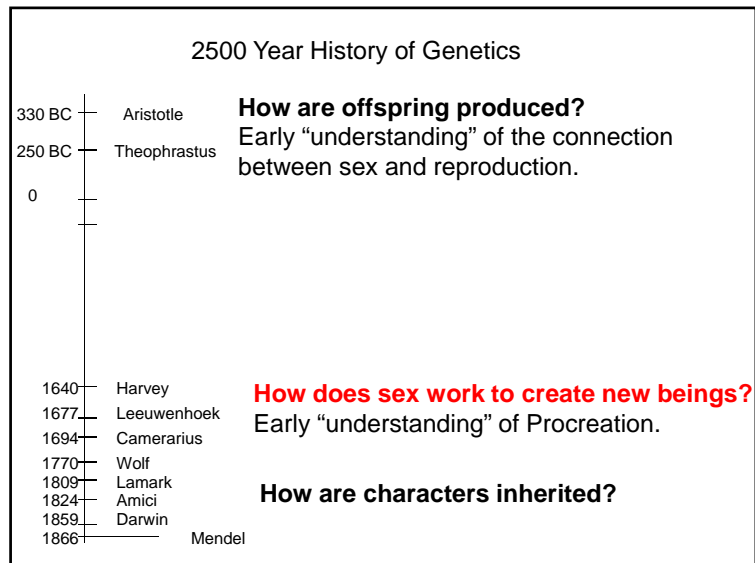
**Leeuwenhoek** Dutch (1677) discovered (with primitive microscope) sperm in animals "wild animalcules arising in seminal fluids by putrefaction."

**Camerarius (1694)** at Tübingen Bot Gardens (Germany)  
Observed that Female mulberry without Male yields no seed. Worked with many plants (corn) and showed the removal of stamens prevented seed formation.

**"In the plant kingdom the production of seed which is the most perfect gift of nature and the general means of maintenance of a species, does not take place unless anthers have previously prepared the young plant contained in the ovary".**



At this stage the Sex-Reproduction Connection was made in the "scientific community". Again, for at least animals this connection was clear earlier.



### How does sex work to create new beings?--Early Understanding of Procreation.

Theories:

**Preformation** and homunculus (ovists vs. spermists!!??).

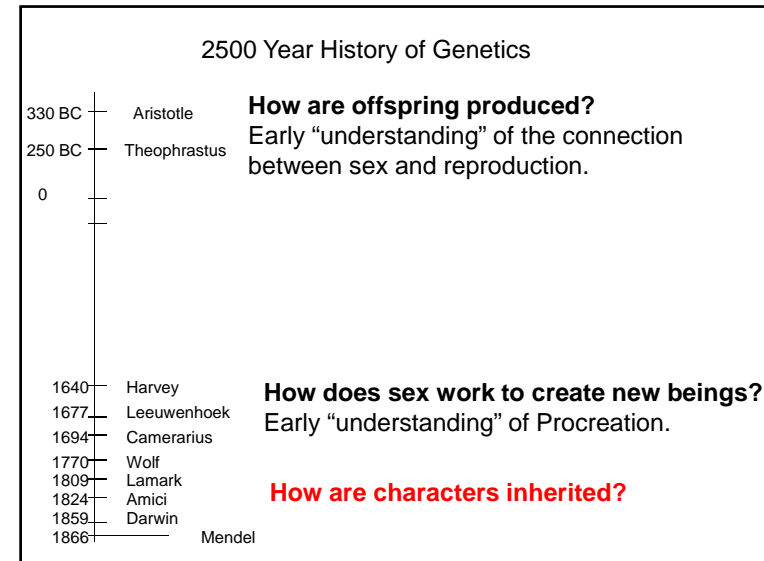
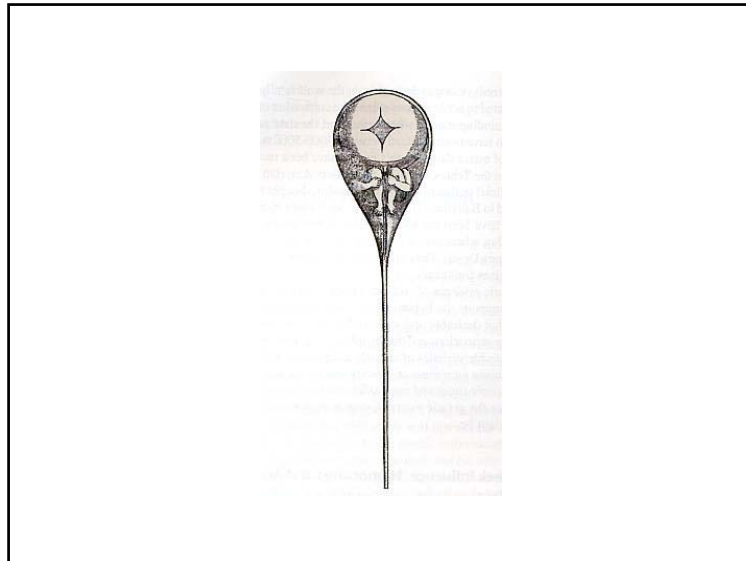
**Epigenesis:** structures not present in early embryos but newly formed (Harvey 1640s)

**Casper Wolf** (1733-1794) an embryologist showed adult structures absent in embryo.

**Amici (1824)** an astronomer. Watched for 3 hrs and saw pollen grain germinate, grow and enter embryo sac. **"tubes elongate bit by bit and finally come in contact with ovules, one tube for each ovule."**

These observations killed Preformation Theory and showed fusion of male and female cells was required for reproduction





Lack of understanding about how sex leads to reproduction (union of male and female cells) does not mean there was no understanding of inheritance.

**People certainly have had a sense that characters are inherited. Simple understanding that "like begets like"**

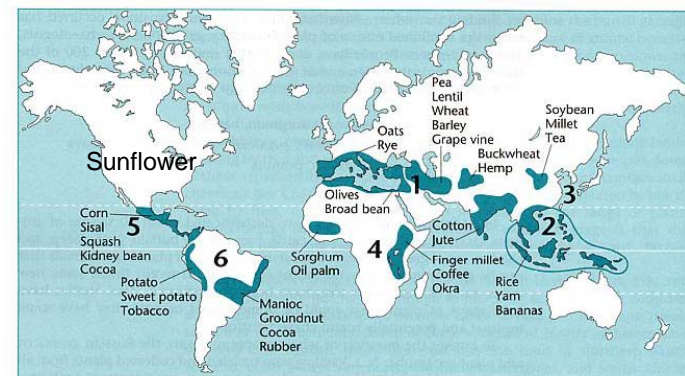
Hindu saying: "a man of base descent never escapes his origin"

disease and occupation are inherited??!!

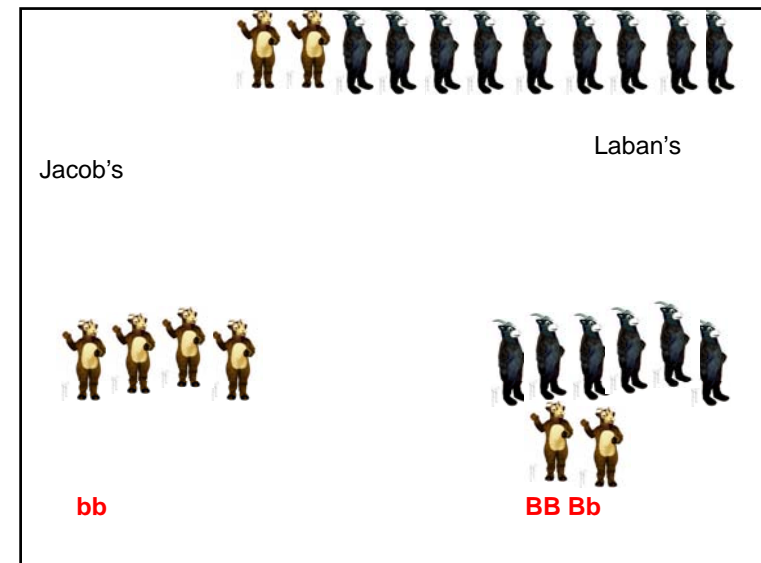
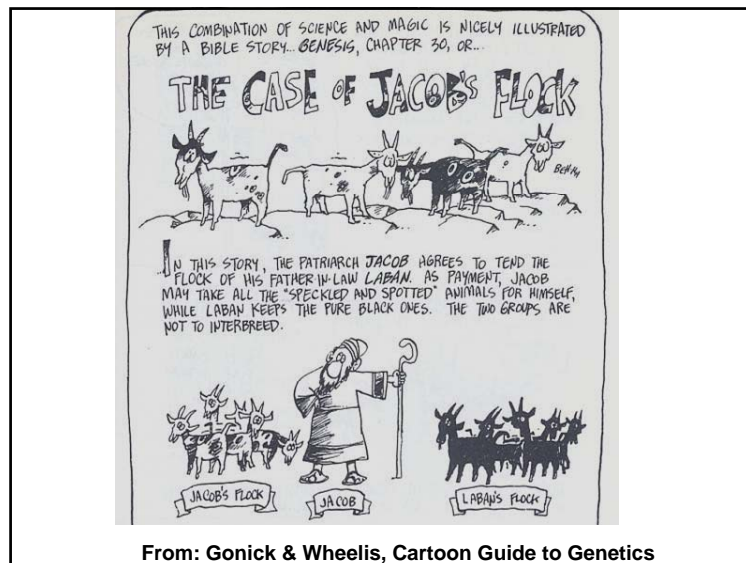
Agriculture: (Breeding...controlled evolution)

Biblical story:

How are characters inherited?



Primitive methods creating Genetically Modified Organisms (GMOs)



Once the understanding of sex and reproduction was clarified (union of male and female cells) in the 1700s, the direction of **discussion** changed from problem of **procreation** to **inheritance and variation**.

**How are characters inherited? Why do they vary? Why are individuals and species different?**

**Blending inheritance** (intermediacy of hybrids) was the prevailing theory in 1700-1800s.

### Blending Inheritance: the problem:

**Darwin** realized that blending inheritance however would diminish variation.

**Evolution by Natural Selection** (nature does what humans have been doing by artificial selection) depends on heritable variation.

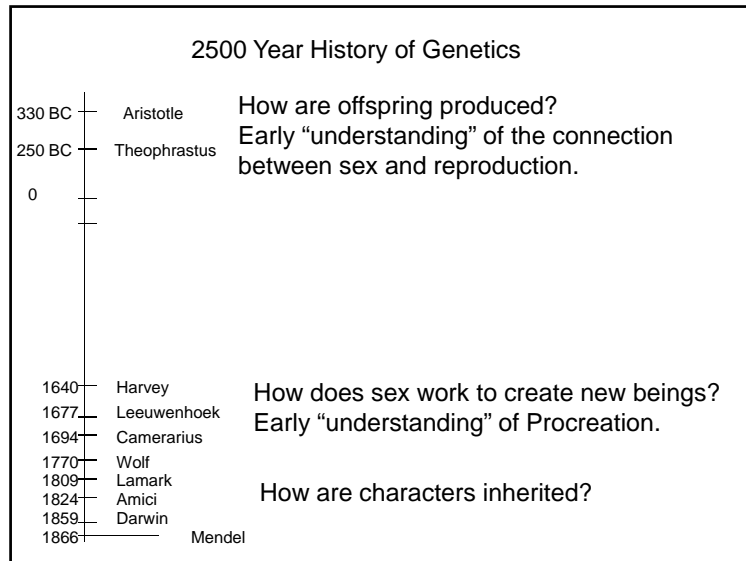
Darwin adopted Lamarck's (1809) ideas to solve blending inheritance theory deficiency.

Doctrine of **use and disuse** and **inheritance of acquired characteristics** would generate variation lost by Blending.

Theory of **pangenesis** that heritable materials (gemmules) were floating in the body and come to together at conception. The gemmules can be affected by the environment.

Frances Galton's test.





**By late 1800's two merging approaches:**

- A. **Transmission studies:** (hybridizers) **Kolreuter**, and culminating with **Mendel's** (1866) work. Ch. 3-5
- B. **Cytology**; starting with primitive microscopes used by Leeuwenhoek through Amici and culminating with **T. Boveri** and **Walter Sutton**. Sutton proposed the **chromosomal theory of heredity (1903)**. They proposed that the hereditary factors of Mendel were located on chromosomes. Ch. 2, 7, 8

**Beginning in the 1950s a third approach:**

- C. **Molecular Genetics:** With the discovery of the structure of DNA this approach rapidly advanced Ch. 10-