

BIOL 153: INTRODUCTORY GENETICS (MEDICAL GENETICS)

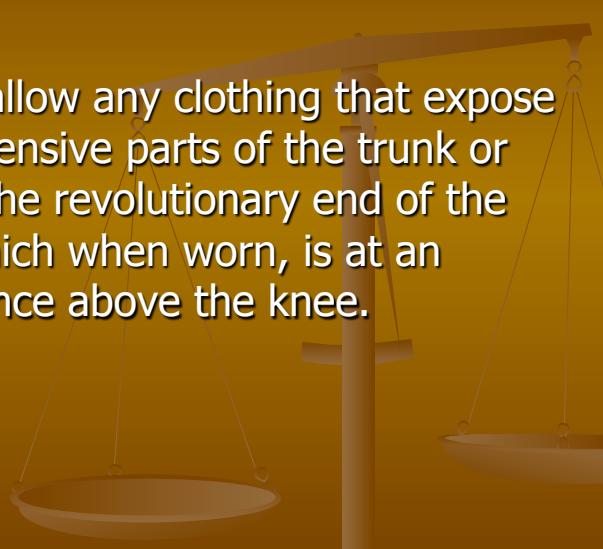
Class: OD1

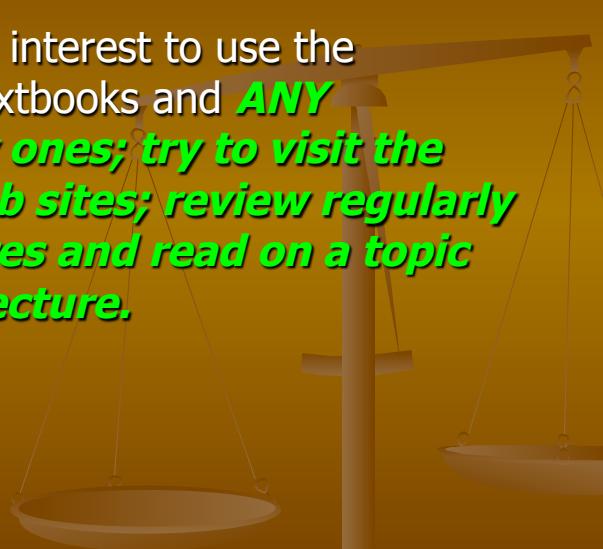
Lecturer: Dr. Alexander K. Anning

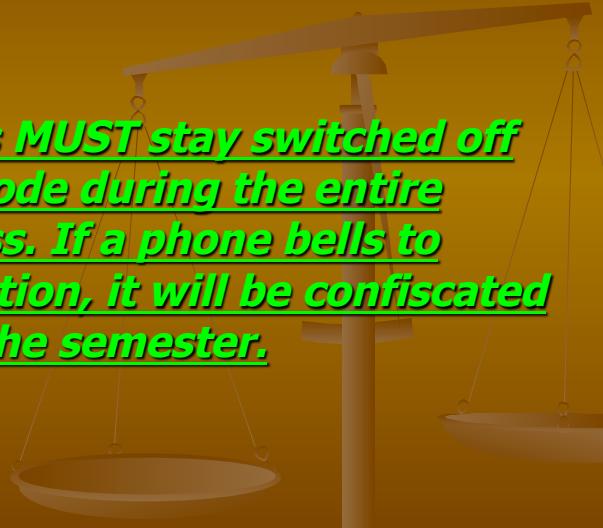
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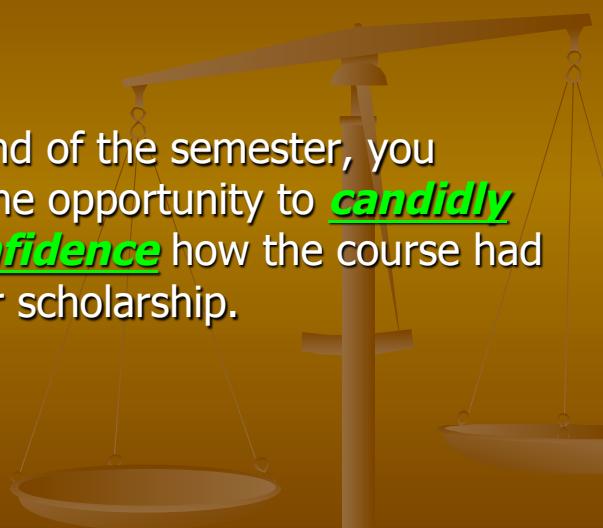
General Comments

1. Lateness to class is not entertained under any circumstances. You may be turned away once the lectures get under way.
2. Lectures and labs are formal sessions and students are advised to dress properly. Would not allow baseball caps, hats, track suits, sleeveless T-shirts etc.

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3. Would also not allow any clothing that expose the midriff or extensive parts of the trunk or short clothes at the revolutionary end of the fashion scale; which when worn, is at an outrageous distance above the knee.

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4. It will be in your interest to use the recommended textbooks and **ANY supplementary ones; try to visit the publishers' web sites; review regularly previous lectures and read on a topic before a new lecture.**

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5. **Mobile phones MUST stay switched off or in silence mode during the entire period of a class. If a phone bells to cause a distraction, it will be confiscated till the end of the semester.**

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6. Getting to the end of the semester, you would be given the opportunity to **candidly evaluate in confidence** how the course had impacted on your scholarship.

7. Plagiarism and cheating in any form by either copying from a fellow student or copying verbatim from internet sites and textbooks would ATTRACT THE SEVEREST FORM OF SANCTIONS. THE CANDIDATE SHALL LOSE ALL CONTINUOUS ASSESSMENT MARKS.

MIDSEMESTER EXAMS AND CONTINUOUS ASSESSMENT

Both the mid-semester and the continuous assessment will make up 30% semester mark.

The final paper (end of semester) will also make up 70%.

Mid-semester exams will be organized most likely in the first week of November.

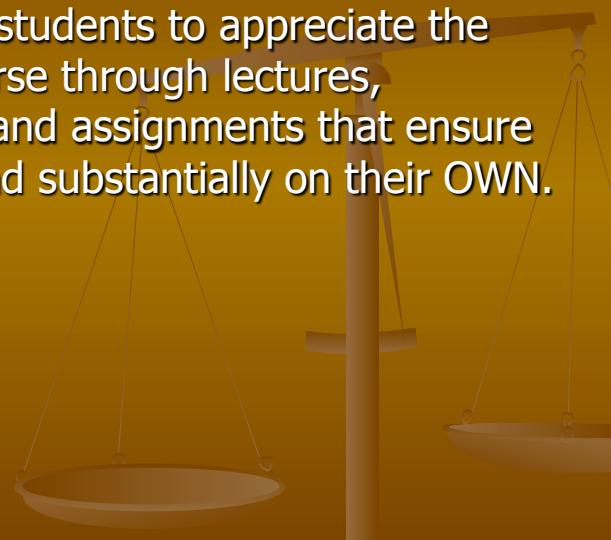
Recommended Textbooks:

- ❖ Principles of Genetics; 8th Edition (1991)
Gardner, E. J.; Simmons, M. J. & Snustad D. P.
- ❖ Principles of Genetics; 4th Edition (1993)
Tamarin, R. H.
- ❖ Modern Genetics; 2nd Edition (1984)
Ayala, F. J. & Kiger Jr, J. A.
- ❖ Concepts of Genetics; (1983)
Klug, W. S. & Cummings, M. R.

INTRODUCTION & COURSE OBJECTIVE

- ❖ This introductory course in Genetics is run over two semesters as BIOL 153 (Introductory Genetics) and BIOL 154 (Population Genetics and Evolution)
- ❖ The two will introduce students to the science of genetics and provide a broad overview of the entire scope of genetics.
- ❖ However, the course provides sufficient detail to give a basic prologue to higher genetics such as BIOL 355 (Molecular Genetics) BIOL 356 (Gene Expression & Modification) and BIOL 451 (Genetic Engineering)

- ❖ It will also allow students to appreciate the scope of the course through lectures, laboratory work and assignments that ensure that students read substantially on their OWN.



COURSE OUTLINE

- ❖ The Course will focus on the introduction to heredity:
 - ❖ Preformation
 - ❖ Epigenesis
 - ❖ Pangenesis and Lamarckism
 - ❖ Mendelism: Independent assortment, factors which influence Mendel ratios



Course Outline

- ❖ Cellular basis of inheritance
 - ❖ Mitosis and meiosis
 - ❖ Meiosis and gene segregation with examples of human genetic disorders
- ❖ Variations in chromosome number
- ❖ Chromosomal abnormalities and human diseases

Course outline cont'd

- ⌘ Sex determination, sex linkage and pedigree analysis
- ⌘ Gene interaction
- ⌘ Multiple alleles
- ⌘ DNA structure and function

WHAT IS GENETICS?

- ❖ It is the branch of biology that deals with heredity and variation.
- ❖ The hereditary units that are transmitted from one generation to the next (i.e. inherited) are called genes.

What is Genetics?

- ❖ Genetics is concerned with the transmission, expression, and evolution of genes (i.e. the molecules that control the function, development and the ultimate appearance of individuals).

Early Ideas About Hereditary

- ❖ The existence of biological heredity is obvious in the resemblance of children to their parents.
- ❖ It was long known that in humans and animals, the sexual act was involved in procreation.
- ❖ It was therefore natural to assume that semen was the carrier of heredity, but how this was accomplished proved difficult to establish.

THEORIES

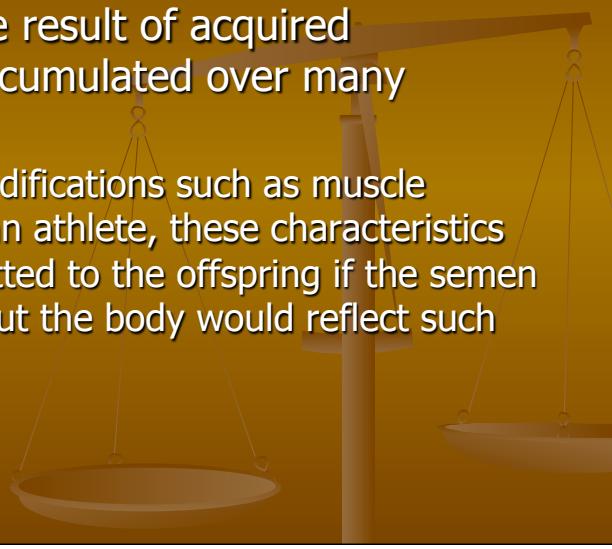
- ❖ Pangenesis, Lamarckism, Preformation and Epigenesis attempted to explain the development of plants and animals.
- ❖ The theory of pangenesis states that every structure which is inherited will pass on its characteristics by contributing a small amount to the semen.
- ❖ According to this theory, particles (pangenes or gemmules) form in each body part are transported through the blood vessels to sperms/eggs and then inherited by offspring.

Pangenesis

- ❖ The similarity between parents and offspring was accounted for by postulating that the pangenes or gemmules formed in each part of the body reflected the characteristics of that part.
- ❖ This theory was supported by Aristotle and other ancient Greeks. It prevailed for many centuries into the 19th Century.

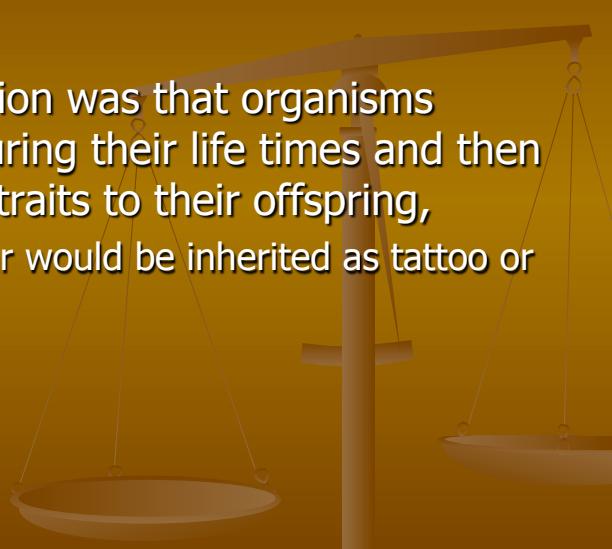
Lamarckism

- ❖ Evolution was the result of acquired characteristics accumulated over many generations:
 - ❖ e.g., in body modifications such as muscle development in an athlete, these characteristics could be transmitted to the offspring if the semen formed throughout the body would reflect such modifications.



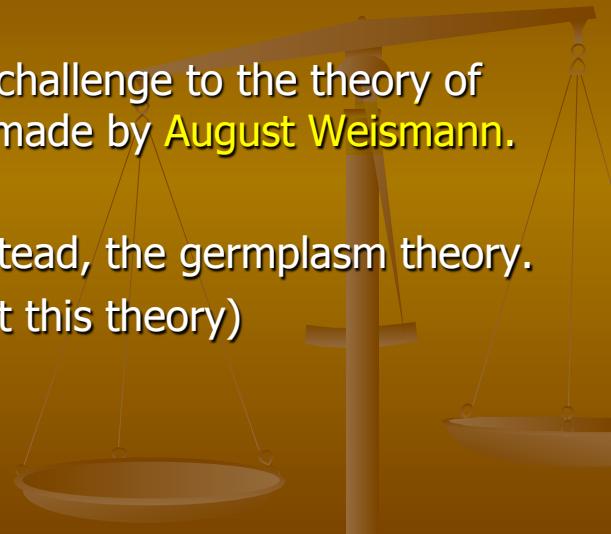
Lamarckism

- ❖ Another explanation was that organisms acquired traits during their life times and then passed on those traits to their offspring,
 - ❖ e.g. tattoo or scar would be inherited as tattoo or scar pangenies.



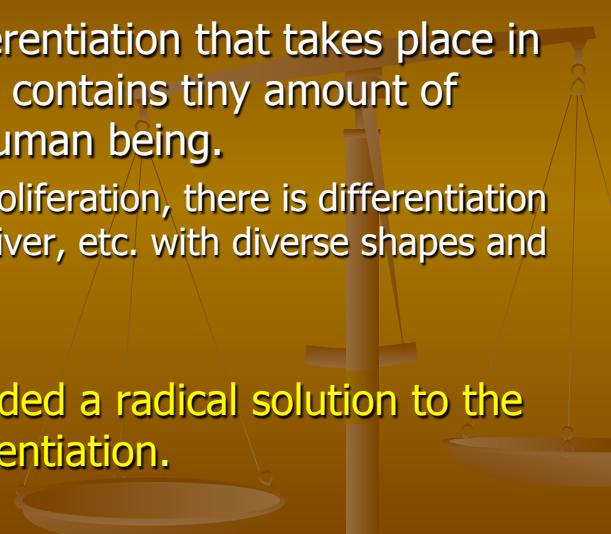
Germplasm Theory

- ❖ The first serious challenge to the theory of pangenesis was made by August Weismann.
- ❖ He proposed, instead, the germplasm theory.
(Read more about this theory)



Preformation

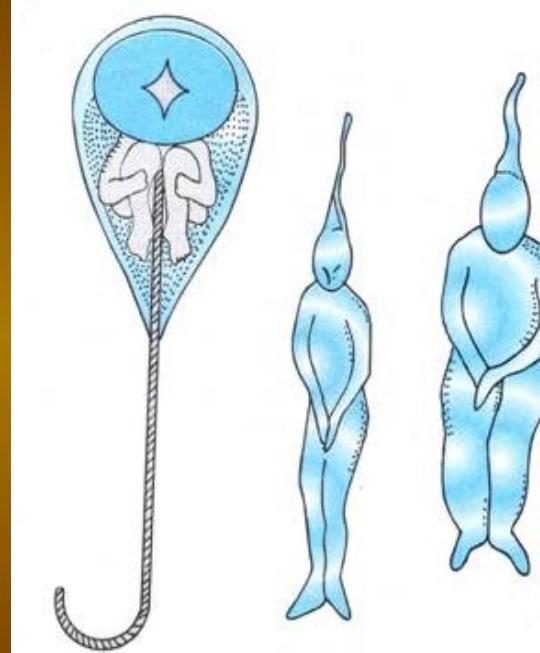
- ❖ Explains the differentiation that takes place in the zygote which contains tiny amount of material into a human being.
 - ❖ E.g., after cell proliferation, there is differentiation into arms, legs, liver, etc. with diverse shapes and functions.
- ❖ This theory provided a radical solution to the problem of differentiation.



Preformation

- ❖ In the late 17th century, an observer using a primitive microscope and a lively imagination claimed to have seen a miniature figure called (*homunculus*) of a man inside the human spermatozoon.
- ❖ Jan Swammerdam and Charles Bonnet discovered this theory and was supported by other wishful thinkers.

Homunculus:
**little man in a
sperm cell**



Preformation

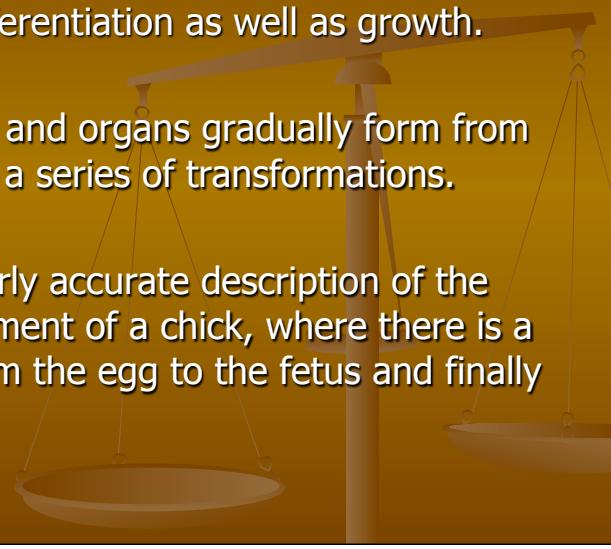
- ❖ The human body, according to some preformationists, was already preformed in the spermatozoon.
- ❖ Development was simply a matter of growth of the tiny *homunculus*.
- ❖ There were the “**spermatists**” and the “**ovists**”.

Epigenesis

- Kasper Wolff and Karl Ernst von Baer proposed this theory in the 18th and 19th centuries.
- This theory discredited or disproved Pangenesis, Lamarckism and Preformation.
- According to this theory, the sex cells are largely homogenous bits of organic matter and contains nothing resembling the body that will develop from them.

Epigenesis

- ❖ Development is differentiation as well as growth.
- ❖ The various tissues and organs gradually form from the zygote through a series of transformations.
- ❖ They provided a fairly accurate description of the embryonic development of a chick, where there is a gradual change from the egg to the fetus and finally to the adult body.



Conclusion:

- ❖ Theory of epigenesis was more accurate than that of preformation.
- ❖ The organism is not preformed in the zygote.
- ❖ What is transmitted from parents to offspring is a set of “instructions”, i.e., the genetic information in the DNA, which interacting with the environment, direct the development of the organism.

