

# **BCH 7412 “Epigenetics of Human Disease and Development”**

1 module, 1 credit

Fall semester, Module 3

Meets Wednesday and Friday, 2:00-4:30, begins 11/2/05, ARB Rm. R3-265 (Room and time may vary, please check current official course listing)

Prereq: GMS 6001 or consent of instructor, BCH 6415 recommended

**Course director:** Keith Robertson, Ph.D.

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## **Description**

In depth lecture and literature-based assessment of epigenetic mechanisms of mammalian gene regulation, including: DNA methylation, histone modifications, genomic imprinting, inherited genetic diseases, viral gene regulation, and epigenetic reprogramming in embryonic stem cells and cloning. Particular emphasis will be placed on how loss of proper epigenetic control leads to human disease and how novel therapies to correct epigenetic defects may be possible. Basic mechanisms and mouse models of human diseases are also emphasized.

## **Course Format and Grading**

Epigenetics of Human Disease and Development will be offered in the Fall semester. One hour of lecture by the instructor followed by one hour of discussion of recent research papers by the students will be conducted twice per week. Grading scale will be letter grades.

## **Evaluation will include the following:**

**70% Oral presentation.** A selected published paper will be presented in the class. The presenter will introduce the background and rationale for the study, explain the supporting data, and summarize the major findings of the paper. The presenter is also encouraged to critique the paper, pointing out its strengths and weaknesses.

**30% Group discussion.** Students will be expected to contribute to the discussion and critique of the paper being presented in the class.

## Tentative Course Outline

- 1) **Epigenetic Mechanisms of Gene Regulation I** – Basic mechanisms, methylation and the genome, DNA methyltransferases, and their connections to chromatin
- 2) **Epigenetic Mechanisms of Gene Regulation II** – Basic mechanisms, methyl binding proteins and their connections to chromatin, methylation as a genome defense system
- 3) **DNA Methylation and Cancer** – Global and region-specific changes in DNA methylation, effects on transcription & chromatin structure, therapeutic opportunities
- 4) **Genomic Imprinting** – CTCF and BORIS as imprint regulators, parent-of-origin effects, loss of imprinting in cancer (LOI)
- 5) **Diseases Associated with Abnormal Imprinting** – Beckwith-Wiedemann syndrome (BWS), Prader-Willi/Angelman syndromes
- 6) **Methods in Epigenetics** – Methods for the analysis of DNA methylation patterns (global, gene-specific, & high-throughput), chromatin structure, and histone modification patterns
- 7) **DNA Methylation, Viral Gene Expression, and Carcinogenesis** – Herpesviruses as a model systems, viral gene regulation by DNA methylation and chromatin structure
- 8) **Epigenetic Reprogramming, Cloning, and the Role of DNA Methylation in Mammalian Development** – Roles for methylation in embryogenesis, aberrations in this process during cloning in animals and *in vitro* fertilization procedures in humans
- 9) **Epigenetics, Embryonic Stem Cells, Pluripotency, and Differentiation** – Epigenetic mechanisms regulating ES cell differentiation down certain lineages and in maintaining pluripotency

Faculty lecturers include Drs. K. Robertson, K. Brown, T. Yang, D. Bloom, J. Resnick, M. Kladde, and N. Terada. Topics and lecturers may vary from year to year.