



## **Mountain States Genetics 2008 Annual Education Conference Syllabus**

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### **Session I: Current Issues in Genetics and Forensics**

**Instructors/Presenters:** Maria Liu, JD, partner, Collins, Liu, Lyons, Greeley, CO  
Matt Kaplan, Ph.D., U OF A

#### **Course Description:**

1. Discuss crime scene reconstruction and the use of DNA in criminal cases
2. Review why it was believed that epithelial cells would be on the victim's clothing
3. Explain the legal issues encountered and surpassed.
4. Review of the Shoah Project for genotyping Holocaust survivors.
5. Describe the emotional and social issues involved in working with the families.
6. Describe the concerns and challenges of high-throughput genotyping.

#### **Learning Objectives:**

1. Review of the history and current use of DNA analysis in CSI labs.
  2. Review of the Tim Masters murder case.
  3. Discuss the extended time lapse and circumstantial evidence leading to the first conviction in 1998, and the DNA evidence that exonerated Masters in 2008.
  4. Review the use of DNA analysis in determining familial relationships
  5. Discuss the issues involved in performing a forensic reconstruction using descendants of the survivors that have dispersed across the globe.
  6. Identify the technological advances required to perform this project
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### **Session II: UPDATE ON DOWN SYNDROME: Prenatal Screening Modalities**

**Instructors/Presenters:** Sarah Hartway, MS, RN, Mile High Down Syndrome, Denver, CO

#### **Course Description:**

1. Discuss the impact of the diagnosis of Down syndrome on new or expectant parents and the range of parental responses.
2. List 3 guidelines for presenting a diagnosis of Down syndrome to parents in a supportive manner.
3. Identify two trends in prenatal testing and Down syndrome birth rates in Colorado.

#### **Learning Objectives:**

1. Outline patient and family response and effects of prenatal diagnosis of Down Syndrome
  2. Give examples of genetic counseling options for patient guidance during gestation and post delivery periods.
  3. Review population incidence and testing modalities for Down Syndrome in Colorado
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**Session III: Emerging Technology Forum – Results of Phase II clinical trials of Genz 112638 for patients with type 1 Gaucher Disease**

**Instructor/Presenter:** Timothy Miller, MD, Genzyme Therapeutics

**Course Description:**

1. Describe clinical trial protocols for investigational oral therapy for Gaucher Disease with Genz-112638.
2. Analyze data from an ongoing open-label Phase 2 clinical trial of the investigational oral therapy Genz-112638.
3. Report of spleen volumes, hemoglobin level, platelet counts and Chitotriosidase levels after six months of oral therapy

**Learning Objectives:**

1. Review of the phenotypic and physiological characteristics of Gaucher Disease
  2. List of existing and emerging therapies for Gaucher Disease
  3. Examination of Phase II clinical trial data and outcomes, showing pre-and –post therapy spleen volumes, hemoglobin level, platelet counts and Chitotriosidase levels after six months.
  4. Review of protocols for 12-24 month trial
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**Session IV: Keynote Presentation: Preimplantation Genetic Diagnosis & Changing Patterns of Screening for Fetal Chromosomal Disease**

**Instructor/Presenter:** Mark Hughes, MD Ph.D., Genesis Genetics  
Phil Wyatt, MD, PhD, Genzyme Genetics

**Course Description:**

1. Identify the principles behind DNA screening and diagnostic testing in single-cells.
2. Explain the current status of Preimplantation Genetics in 2008 and list indications for single-gene and chromosome testing of embryos.
3. Describe the role of new genetic technologies in reproductive diagnostics and the ethical implications of these procedures.
4. Summarize an historical overview of screening for fetal chromosomal disease.
5. Discuss current screening methods.
6. Discuss the minimal standards for screening and which methods represent current standard of care.
7. Future trends in serum/invasive screening.

**Learning Objectives:**

1. Review of the science of single-cell chromosome analysis
  2. Review preimplantation diagnosis and current usage.
  3. Discuss genetics and reproductive diagnostics, and the associated ethical issues.
  4. Review the history of prenatal diagnostic screening.
  5. Discuss current standard practice for prenatal screening.
  6. Review current methodology and developing technology for future screening options
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**Session V: Cardiac Genetics**

**Instructors/Presenters:** Colleen Brown, ScM, UC San Francisco  
Matt Taylor, MD, UC School of Medicine

**Course Description:**

1. Identify genetic conditions commonly encountered in the adult cardiac genetics clinic.
2. Discuss the factors that are driving growth and interest in adult cardiac genetic conditions.
3. Discuss which diseases can be currently evaluated using DNA-based testing.
4. Explain the challenges of current genetic testing methods in terms of: genetic heterogeneity, sensitivity, costs, and quality of 'mutation-causes-disease' databases.
5. Recognize potential of unique aspects of cardiac genetic diseases along with ethical situations that arise in this population cohort.

**Learning Objectives:**

1. Review of adult cardiac disorders associated with chromosome abnormalities.
  2. Discuss social and population factors affecting interest and causes of cardiac disease.
  3. Review specific testing modalities for cardiac genetic disorders.
  4. Review of the costs, limitations and value of cardiac genetic testing.
  5. Discuss therapeutic and counseling options for patient cohort.
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**Session VI: Bob McCurdy Lecture: Presymptomatic Testing for Neurological Disorders**

**Instructor/Presenter:** Robert C. Green, MD, PhD, Boston University SOM

**Course Description:**

1. Review and understand the genetic and environmental risk factors for Alzheimer's disease.
2. Discuss APOE genotypes and the influence of these genotypes on risk of AD.
3. Discuss disclosure issues surrounding presymptomatic testing for AD.

**Learning Objectives:**

1. Identify of current environmental factors associated with Alzheimer's disease and population incidence.
  2. Review of current population data for AD
  3. Review of current genetic legal issues and counseling models for presymptomatic testing
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**Session VII: Cancer Genetics**

**Instructor/Presenters:** Carol Clericuzio, MD, University of New Mexico SOM  
Lori Ballinger, MS, University of New Mexico SOM

**Course Description:**

1. Describe 3 congenital anomalies associated with childhood cancers.
2. Describe a tumor surveillance program for children with sporadic aniridia.
3. Explain the type of genetic test available for multiple endocrine neoplasia 2B and know how to access [www.genetests.org](http://www.genetests.org)
4. Instruction about common inherited cancer syndromes.
5. Discussion of how risk determination is made.
6. Examples of how patients who may be at increased risk for inherited cancer syndromes are identified.

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7. Discussion of the impact of these syndromes on the management of patients and family.

**Learning Objectives:**

1. Review of slides from three affected patients
  2. Explanation of the surveillance and database system currently available to track affected children
  3. Description of the testing modalities and online resources for patients/families affected with multiple endocrine neoplasia 2B
  4. Live instruction of how to access information at [www.genetests.org](http://www.genetests.org)
  5. Review of currently identified inherited cancers
  6. Description of risk analysis calculation
  7. Review of counseling strategies for families and patients
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**Session VIII: New Frontiers in Prenatal & Neonatal Diagnosis: Applications of Genomic Microarray Analysis**

**Instructor/Presenter:** Sau Wai Cheung, PhD, Baylor School of Medicine

**Course Description:**

1. Review of cytogenetic and molecular testing modalities for prenatal and neonatal diagnosis.
2. Discuss the use of targeted BAC clone array as a platform for comparative genomic hybridization (aCGH) to enable detection of a wide range of pathologic copy number changes in DNA.
3. Review of developing database of more than 300 cases using new microarray techniques.

**Learning Objectives:**

1. Historical review of the application of cytogenetic and molecular (FISH) techniques in prenatal and neonatal diagnosis of chromosomal abnormalities
  2. Comparative review of how aCGH may detect somatic chromosomal mosaicism that would be missed by conventional cytogenetics.
  3. Review of Baylor's developing database of prenatal/neonatal patients and parents to determine efficacy and accuracy of microarray screening technology.
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**Session IX: Laboratory Case Workshop**

**Instructor/Presenter:** Mary Haag, PhD, Moderator

**Course Description:**

1. Review of molecular diagnostic modalities.
2. Use of PCR and FISH technologies for cancer diagnosis.
3. Review of slides of PCR and FISH results to confirm prenatal cytogenetic diagnosis various genetic disorders.

**Learning Objectives:**

1. Examination of cytogenetic results compared to molecular testing.
  2. Review of expanded options in the use of FISH probes for cancer diagnosis and therapy monitoring
  3. Comparative review of PCR and FISH slides results against cytogenetic reports.
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