

## ACMG CLINICAL GENETICS MEETING

### It's not too late to register for the 2012 ACMG Annual Clinical Genetics Meeting

Advance Registration deadline is February 15. After that date, registrations will be accepted online and on-site but will be charged the Late Registration fee. Visit [www.acmgmeeting.net](http://www.acmgmeeting.net) for registration fees and the link to the online registration site.

#### Session Highlights

The detailed program schedule and speakers can be found at [www.acmgmeeting.net](http://www.acmgmeeting.net) in the Program Sessions and Events Schedule section of the website. Attendees can view the entire schedule or plan their session itinerary online.

**PLENARY SESSIONS INCLUDE:** 2012 Presidential Plenary; 2012 ACMG Awards and “The Coming Revolution in Medical Genetics: From Double Helix to Genomics and Back Again”

**43<sup>RD</sup> ANNUAL MARCH OF DIMES CLINICAL GENETICS CONFERENCE:** Genetics of Aging — From Progeroid Syndromes to Centenarians

**HIGHLIGHTS PLENARY SESSION:** Developmental Epigenetics

**JOINT ACMG/SIMD PLENARY SESSION:** “Making Sense of Mitochondrial Disease”, Charlotte Convention Center

This year the ACMG Meeting will begin one day early—on Tuesday—and end on Saturday afternoon. On Tuesday, in addition to Special Interest Group Forums, two CME Short Courses will be held on the following topics:

- Clinical Cancer Genetics: New Paradigms and Concepts for Understanding Cancer Susceptibility



Bechtler Museum of Modern Art — Charlotte, NC

- Next Generation Sequencing: Clinical Utility, Laboratory Implementation and Bioinformatics Analysis

Invited sessions throughout the meeting will focus on topics such as Current Concepts in Reproductive Genetics, Informatics in Medical Genetics, Developmental Eye Disorders, Assessing the Value of a Genetic Diagnosis, Single Gene Causes of Cancers, Laboratory Implementation of Prenatal aCGH, Cardinal Signs of Selected Syndromes and Cardinal Signs and Symptoms of Common and Rare Important Inborn Errors of Metabolism, Genetics of Sports Performance, Challenges in Newborn Screening Diagnosis and Follow-up, Practical Implementation of Next Generation Sequencing, and the Genetic Basis and Diagnosis of Overgrowth Syndromes.

Additional highlights of the meeting include Oral Abstract Platform Presentations on Thursday and Friday, Poster Presentations in the exhibit hall, an Industry Supported Symposia on Friday morning and an exhibition of close to 100 companies showcasing the newest treatment advances and methodologies, laboratory and genetic testing, and educational resources.

### Laboratory Quality Assurance Committee Announces Major Change to Cytogenetics Guidelines for Detection of Sex Chromosome Mosaicism

Based on new evidence in the scientific literature reporting that inclusion of 30 cells, instead of the standard 20 cells, does not result in a significant increase in the detection of sex chromosome mosaicism. Section E5.1.2.2 of the *Standards and Guidelines for Clinical Genetics Laboratories* has been changed to read, “Cases being studied for possible sex chromosome abnormalities, in which mosaicism is common, should include the standard 20-cell assessment. If mosaicism is confirmed,

the analysis is complete. A minimum of 10 additional metaphase cells should be evaluated when one cell with a sex chromosome loss, gain or rearrangement is observed within the first 20 cells analyzed.” This change will be reflected in the CAP Cytogenetic Checklist, as well. The entire *Standards and Guidelines for Clinical Genetics Laboratories* can be accessed from the ACMG website ([www.acmg.net](http://www.acmg.net)), under the Publications tab.