**Chromosome Abnormalities**
Moderator: Angela E. Lin, Harvard Medical School, and MassGeneral Hospital for Children, Boston, MA

**Syndrome Review II: Common Deletion/Microdeletion Syndromes**
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Chromosome deletion and microdeletion syndromes account for an increasing number of clinically recognizable genetic conditions. New deletion syndromes continue to be characterized, and a number of previously-described syndromes of unknown etiology have been found to be due to chromosomal deletions or microdeletions. Chromosomal microarray technologies are in wide clinical use, and are diagnosing an ever expanding number of microdeletions and microduplications, which, when recurrent, are being characterized as new syndromes. As new deletions (and duplications) are being identified, the genes associated with the physical characteristics of the patients are being determined. Future studies in this area will focus on identifying candidate genes for the phenotypic features of deletion and microdeletion syndromes toward the goal of understanding the pathophysiology of the abnormal developmental and physiologic processes involved in each syndrome.

This presentation will focus on several classic and some newly-described deletion and microdeletion syndromes, and illustrate how studies are identifying genes involved in birth defects and developmental problems.