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## Karen Miga

### The dark side of the human genome: introducing a new era of satellite DNA genomics

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Our understanding of the human genome remains incomplete, with an estimated 10% of bases, largely representing highly repetitive sequences from centromeres and the acrocentric short arms, that are omitted from genome-wide studies of cellular function and human health. As a result, sequence structure and epigenetic regulation in these uncharacterized regions remain largely detached from contemporary human genomic studies. To address this challenge, I have implemented a graph-based approach to model sequence content and organization within each centromeric region. Centromeric DNAs are expected to vary in repeat composition and copy number between individuals in the population. Therefore as an extension of this foundational work, I have performed an initial assessment of centromeric sequence variation in the human population, and will discuss efforts to include these regions in association studies of human disease. Further, in an effort to explore the role of centromeric sequences in nuclear biology and satellite-DNA based mechanisms of gene expression, I have used existing ENCODE data to characterize transcription factors, non-coding expression, and long-range spatial interactions involving centromeric DNAs. These studies are expected to establish a new niche in the genome sciences that integrates the use of both computational and experimental approaches to better understand these specialized regions of the genome.

**14:30 h, Lecture hall B 01.027**

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