Heard PL, Carter EM, Crandall AC, Sebold CD, Hale DE, Cody JD. High Resolution Genomic Analysis of 18q- using oligo-microarray Comparative Genomic Hybridization (aCGH). In press.

You may have heard about a relatively new technology called microarray. Essentially, microarrays are a new way to look at a person's chromosomes for deletions and duplications. It allows scientists to identify chromosome abnormalities that are too small to see on a routine karyotype. However, this technology also helps us understand more about chromosome abnormalities that have already been diagnosed. We used microarray analysis to learn more about the nature of the deletions and duplications in our study population. We completed microarray analysis on 189 individuals with 18q-. As it turns out, deletions involving the long arm of chromosome 18 are frequently more complicated than they first appear on a karyotype. We learned that 17% of 18q deletions are interstitial. That means that they do not include the tip of the chromosome. 6% of 18q deletions included a duplication of the area next to the breakpoint. 8% of the deletions had more complex rearrangements that included the tips of other chromosomes. This shows that there is even more variability in chromosome abnormalities than we previously thought!