UA Division of Genetics, Genomics & Precision Medicine co-chief Deb Meyers, PhD, and UA Arthritis Center/Division of Rheumatology’s Ernest Vina, MD, talked on “Genetics and Phenotypic Heterogeneity in Common Diseases (Asthma, COPD, etc.)” and “Gender and Race Disparities in Osteoarthritis.” See the archived video here.
Implications and Future Directions

- Smokers with SERPINA1 variants are at an increased risk for more severe disease. Findings need to be extended and replicated in TOPMed.
- Rare SERPINA1 variants (cumulative not so rare) will play a role in precision genetic profiling for disease risk and severity in COPD.

Role of CFTR variants in Asthma: Background

- Background: A recent meta-analysis found that risk of asthma was significantly higher in rare CFTR variant heterozygote carriers compared to non-carriers (Nielsen AO et al. J Cyst Fibros. 2016)
Non-Hispanic Whites: Results

- Multiple rare variants (frequency 0.0002-0.0005, CFTR2 database) were observed in 512 non-Hispanic whites and 243 African-Americans.

- Eight non-Hispanic whites were compound heterozygotes for non-disease causing variants (CFTR2 database). No differences were observed in FEV1 or FEV1/FVC.

Preliminary Results

- Seven African-Americans were compound heterozygotes for two CFTR potentially disease causing variants: Arg75His/ApaI, CFTR2 database 2 of 21 variants listed as "varying clinical consequences."
Photos courtesy of David Mogollón, Communications Coordinator, UA Department of Medicine, (520) 626-1137 or dmogollon@deptofmed.arizona.edu