Silverman - Chronic Obstructive Pulmonary Disease (COPD)

Updated 10/24/2018

Introductory slides (Phase 1) from the June 4, 2015 Steering Committee/EAP meeting (requires log-in). [1]

Introductory slides (Phase 3) from the April 24, 2017 All Hands call (requires log-in) [2]

Boston Early-Onset COPD Study: This study has enrolled subjects with severe COPD (forced expiratory volume in one second (FEV1) < 40% predicted) at an early age (< 53 years) without alpha-1 antitrypsin deficiency (a known Mendelian risk factor for COPD). Extended pedigrees are enrolled, primarily in New England, although some more geographically distant subjects have been included. This study has been used for epidemiological studies, familial aggregation analysis, linkage analysis, and candidate gene association analysis. Approximately 100 of the severe, early-onset COPD probands will undergo whole genome sequencing in this project. Sequencing data from the subjects with IRB approval for data sharing will be submitted to dbGaP. The Boston Early-Onset COPD Study web site is: http://bostoncopd.org/ [3]

COPDGene: COPDGene (also known as the Genetic Epidemiology of COPD Study) is an NIH-funded, multicenter study. A study population of more than 10,000 smokers (1/3 African American and 2/3 non-Hispanic White) has been characterized with a study protocol including pulmonary function tests, chest CT scans, six minute walk testing, and multiple questionnaires. Five years after this initial visit, all available study participants are being brought back for a follow-up visit with a similar study protocol. This study has been used for epidemiologic and genetic studies. Previous genetic analysis in this study has been based on genome-wide SNP genotyping data.Approximately 1900 subjects will undergo whole genome sequencing in this NHLBI WGS project, including severe COPD subjects and resistant smoking controls. The COPDGene Study web site is: http://www.copdgene.org/ [4]