

Working To Understand Familial Lung Cancer And Its Genetic Underpinnings

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The primary focus of our research is to identify hereditary genetic variants that may predispose to lung cancer. In this study we processed blood and buccal swab samples collected from affected and unaffected members of families with statistically significant enrichment for lung cancer. DNA extracted will be sequenced and analyzed for variants associated with lung cancer risk. Blood or buccal swab samples were collected from individuals within lung cancer families. Samples were stored at -80 °C until sample processing. Specimen samples were processed using the Qiagen FlexiGene® DNA Kit or Gentra® Puregene® Buccal Cell kit. Purity was assessed through NanoDrop 2000 Spectrophotometry. A260/A280 spectrophotometry readings were obtained for DNA specimens from blood (N=59, mean=1.87, standard deviation 0.046) and buccal swabs (N=3, mean=1.94, standard deviation=0.08). Mean DNA yield for blood samples was 301 µg and for buccal swabs was 8.86 µg. The A260/A280 ratio assessed DNA purity, with a ratio of 1.8-2.0 generally accepted as optimal. Our average sample purity fell within this optimal quality range. The DNA yield required for genomic sequencing is 3 µg, so for blood specimens we have, on average, one hundred times more yield than is necessary, so excess DNA can be stored for future use. Meeting the quantity and quality standards, the DNA samples were shipped to the NIH Intramural Sequencing Center (NISC) for PCR-free whole genome sequencing. DNA variants identified through sequencing will be studied for association with hereditary lung cancer risk.