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Making genomic medicine a reality in the community hospital setting

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The Inova Translational Medicine Institute (ITMI) is a not-for-profit research institute that spent the past four years enrolling, consenting and banking samples (blood, saliva, urine and placenta) from families who gave birth at the Inova Fairfax Medical Center (IFMC). To date, over 3500 mother/father/baby family trios have enrolled in two major studies: A preterm Birth Study and an 18 year long childhood longitudinal study. Whole genome sequencing data has been generated on approximately 7000 individuals. Expression, methylation and miRNA data have been generated on the mother's samples. We have utilized these data to generate novel ancestral genomic references to enhance the identification of rare disease causing variants in WGS data; Perform an *in silico* newborn screen that is compared to results returned by the Commonwealth of Virginia; Parse cancer causing genes by ancestry; Generate prediction models for preterm birth; Develop a private hybrid cloud database infrastructure and tools to support research and Launch a pharmacogenomics panel test that is offered free to any baby born at our hospital. ITMI has developed a novel WGS based test, referred to as a 'genomic physical' that evaluates approximately 3000 pediatric and adult onset disease causing genes as well as inherited familial cancer genes in a clinical laboratory environment to provide patients with an accurate assessment for the risk of developing genetic disease and a clinical interpretation of the WGS results. In combination, these projects and tests are helping the IFMC to integrate genomics into the healthcare of patients in the community hospital.

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Understanding the Fanconi anemia pathway: A novel avenue in cancer genomics

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During the past decade, study of the rare inherited chromosome instability disorder, Fanconi Anemia (FA), has uncovered a novel DNA damage response pathway. Studies into the molecular basis of this disease have unmasked a network of DNA repair mechanisms that contributes to safeguarding the stability of the genome. To date, 19 genes have been identified that encode FA complementation group proteins, many of those have been linked to breast and ovarian cancer, acute myeloid leukemia, solid tumors and skin cancer. Previous studies from our group and others have identified a critical role of FANCM in repairing DNA interstrand crosslinks (ICLs) caused due to cisplatin, a chemotherapy drug used to treat different types of cancer. ICLs are among the most deleterious DNA lesions, since the covalent linkage formed between the Watson and Crick strands prevent DNA unwinding, posing formidable blocks to vital cellular processes such as genome duplication, cell cycle regulation, growth and division. In the presentation, I will summarize the recent progress in understanding the molecular pathogenesis of FA and discuss roles of FANCM as a breast cancer susceptibility gene.

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