Nemours

Dr. Erin Crowgey Appointed Associate Director of Bioinformatics

Repertoire to include genome sequencing in study of cancer, sickle cell and kidney disease

The sequencing of the human genome is leading to advancements in personalized medicine, but the process of associating genetic variation to a specific human disease and treatment is still complex. Recent progress in DNA sequencing technologies, known as next generation sequencing (NGS), is enabling the detection of many genomic alterations. Experts with training and skill in bioinformatics and the ability to analyze massive volumes of genomic data are fostering novel approaches to the study and treatment of diseases like cancer and sickle cell disease.

Erin Crowgey, PhD, who recently earned her doctorate in bioinformatics at the University of Delaware, has joined Nemours as Research Associate Director of Bioinformatics. Dr. Crowgey has unique expertise in identifying genetic variation captured from regions of interest in DNA samples. To do this, she develops software scripts (sometimes referred to as pipelines) that sift through enormous amounts of data with the goal of identifying genetic alterations that are linked to a specific disease.

- Dr. Crowgey's doctoral studies have included a stint as a teaching assistant at King's College in London where she learned to use precise algorithms. In Dr. Cathy Wu's lab at the University of Delaware (UD), Dr. Crowgey has been able to process big data thanks to extensive computational resources — 'BioHen' supercomputer cluster — not widely available at most research institutions.
- At UD, one of the projects Dr. Crowgey tackled involved acute myeloid leukemia (AML) in children, leading her to E. Anders Kolb, MD, Director, Nemours Center for Cancer and Blood Disorders and to Denni Ferrara, President, Leukemia Research Foundation of Delaware (LRFDE). Funding from the LRFDE allowed Dr. Crowgey to perform complex analytical work and helped recruit her to the full time Nemours faculty by covering part of her salary as a doctoral student.
- Through Dr. Kolb's participation in the Children's Oncology Group (COG), Dr. Crowgey obtained a huge data set generated from children with AML located across the U.S. She looked at the genomic DNA of the children from three distinct points in the course of their disease: diagnosis, remission, and relapse. The remission samples (when the disease is in abatement) helped Dr. Crowgey to identify somatic genomic variants in the diagnosis and relapse stages. Relapse is dangerous in AML. While survival is high (60-80%) after the first round of treatment, many patients who relapse do not survive.
- As a result of her findings, Dr. Crowgey and collaborators are close to making a standardized laboratory test kit available for anyone newly diagnosed with AML. The kit will allow clinicians to prospectively guide AML treatment based on the patient's genomic sequencing.
- Another example of how NGS can be applied in a preventive way is the sickle cell project next on the docket for Dr. Crowgey. A small subset of sickle cell patients are at high risk of stroke before age 5. Since the disease is very much regulated by genetics, Crowgey will look for the genomic alteration responsible for this risk. The goal is that eventually, on the day a baby with sickle cell is born, standard testing will show which children are at risk for early stroke and thus guide an ongoing course of preventive treatment.
- Dr. Crowgey will also work with the Nemours Research Molecular Diagnostic Laboratory, Nephrology Division Chief Joshua Zaritsky, MD, and Piper's Kidney Beans Foundation to create a comprehensive next generation sequencing panel for steroid-resistant nephrotic syndrome.

Tim Bunnell, PhD, Nemours Director of Bioinformatics, said that Dr. Crowgey's solid undergraduate grounding in biology enables her to apply bioinformatics capabilities in a meaningful way. Moreover, he added, "Dr. Crowgey is extremely adept at teasing out genomic variations by cleverly designing her analyses. In other words, she knows what to look for and she has a knack for finding it." As Dr. Kolb put it, "Erin can find a needle in a needle stack."

NGS, bioinformatics, and Dr. Crowgey herself all represent the next generation of opportunities in the evolution of science and medicine.

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