



PRESS RELEASE

Stepping Closer to Personalized Medicine

UNM Cancer Center Scientist wins 3-year, \$1.35 million National Institutes of Health grant for genomics research

FOR IMMEDIATE RELEASE:

September 10, 2013 — Albuquerque, NM (UNM Cancer Center) — Why do some people develop disease while others with the same genetic mutation don't? For example, the National Cancer Institute estimates that women who inherit a harmful BRCA2 mutation have a 45 percent chance of developing breast cancer by age 70. What protects the other 55 percent with the same BRCA2 mutation? Jeremy Edwards, PhD, thinks the answer lies in our genes. Dr. Edwards is a University of New Mexico Professor in the Department of Molecular Genetics and Microbiology and in the Department of Chemical Engineering. He is a full member of the UNM Cancer Center. Dr. Edwards recently won a 3-year, \$1.35 million grant from the National Institutes of Health to develop technology to complete the human genome and hopefully answer these puzzling questions.

What we think of today as a “complete” genome is not truly complete. But, with the new technology Dr. Edwards is developing, along with the Proton Genome Sequencer available at the UNM Cancer Center, Dr. Edwards and his team can finally sequence an entire “complete” genome from an individual. The new technology gives unprecedented access to the genome and uncovers many genetic variations that may cause disease. His research will allow scientists to study the genetics of complex diseases like cancer. It could also lead to better tools for personalizing medicine for many complex diseases, not just for cancer.

In his first subset of studies, Dr. Edwards will develop and apply new DNA sequencing technologies to search for long-range genome interactions. The 23 pairs of chromosomes inside each of our cells makes up our genome. Each chromosome is a long molecule of “DNA” that, when stretched out, looks like a twisting ladder. Each rung consists of two smaller molecules, called “bases.” The four bases can be ordered in almost infinite ways. It is the sequence of these base pairs—subsets of this sequence form the “genes”—that encode the protein-making instructions for the cell. A cell copies this sequence every time it divides into daughter cells. If the cell makes a mistake in the copying process, however, it could create diseased daughter cells. Finding these mistakes has been the focus of genetic research until now, but it hasn't fully explained complex diseases like cancer.

“Right now, when we sequence a genome, we get a list of variants,” says Dr. Edwards. A variant is a gene that differs from the reference human genome sequence. “But, there's no way to tell which one of the chromosome pair they're on.” Dr. Edwards thinks this information could shed light on how complex diseases arise and why some people develop symptoms while others don't. Additionally, he says, “The

genome inside the cell gets folded and twisted, so there could be long range interaction.” Genes that are far apart in the sequence could thus become physically close and influence each other. Using the new technology along with the Ion Proton Genome Sequencer, Dr. Edwards will methodically study the genomes of many individuals to find such interactions.

In the second subset of studies, Dr. Edwards will contribute to the next version of the reference human genome sequence. The Human Genome Project publicly announced the first human genome sequence in June, 2000, and the Genome Reference Consortium is currently on revision 37. Dr. Edwards explains, “There are certain regions that have never been sequenced, most likely because they are identical to the genome in another place.” The Human Genome Project and subsequent revisions relied on sequencing technology that read parts of the genome and then fitted them together like puzzle pieces. So, identifying the specific chromosomes from which these snippets came was not always possible. These unknown parts of the genetic sequence could be important in explaining complex diseases. Dr. Edwards will be able to identify many missing pieces in the reference human genome using the newly developed innovative technology.

The third subset of studies will focus on using full-genome sequencing in the clinic. In genetic testing today, clinicians identify only the differences between the individual and the reference human genome. Rather than using the reference genome, Dr. Edwards would like to assemble a brand new reference for each person. He says, “Assembling a *de novo* genome for each and every individual will identify a lot of complex differences you can’t uncover by just looking for differences with the reference.” He goes on to explain that a *de novo* assembly “will be much more accurate in finding structural changes, rearrangements in the genome and changes where chunks are deleted, inserted and flipped around. It will make the genetic information people get now much more valuable and informative.” Providing timely genetic information in a clinical setting will require more than just running the genome sequencer. Dr. Edwards and his team will need to create a whole process to protect privacy and convey information accurately, among other considerations.

All three subsets of studies focus on the molecular nature of genetics, not on a specific technology. Says Dr. Edwards, “We’re machine or platform agnostic. Our work can be applied to any sequencing machine out there.”

About the UNM Cancer Center

The UNM Cancer Center is the Official Cancer Center of New Mexico and the only National Cancer Institute-designated Cancer Center in the state. One of just 68 premier NCI-Designated Cancer Centers nationwide, the UNM Cancer Center is recognized for its scientific excellence, contributions to cancer research, the delivery of high quality, state of the art cancer diagnosis and treatment to all New Mexicans, and its community outreach programs statewide. Annual federal and private funding of over \$72 million supports the UNM Cancer Center’s research programs. The UNM Cancer Center treats

more than 60 percent of the adults and virtually all of the children in New Mexico affected by cancer, from every county in the state. It is home to New Mexico's largest team of board-certified oncology physicians and research scientists, representing every cancer specialty and hailing from prestigious institutions such as M.D. Anderson Cancer Center, Johns Hopkins University, and the Mayo Clinic. Through its partnership with Memorial Medical Center in Las Cruces, the UNM Cancer Center brings world-class cancer care to the southern part of the state; its collaborative clinical programs in Santa Fe and Farmington serve northern New Mexico and it is developing new collaborative programs in Alamogordo and in Roswell/Carlsbad. The UNM Cancer Center also supports several community outreach programs to make cancer screening, diagnosis and treatment available to every New Mexican. Learn more at www.cancer.unm.edu.

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