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NEWS ANALYSIS

A Path for Precision Medicine

By Gina Kolata

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For three years, Dr. Robert Green, a researcher at Brigham and Women's Hospital and Harvard Medical School, has been painstakingly gathering genetic data on thousands of Alzheimer's patients, trying to figure out whether genetic differences explain why the disease progresses over a quarter century in some people and kills others within five years. He has called in favors from colleagues and filled out the reams of paperwork required to share data across institutions, but still has only half of what he needs to do the analysis.

The hope of scientists working on diseases like Alzheimer's, diabetes and cancer is that the so-called precision medicine plan that President Obama proposed last week would speed such efforts to understand genetic variations within diseases and to develop treatments for them. The plan — part of the budget the administration sent to Congress on Monday — would establish a coordinated way for researchers to get genetic and clinical data on a million people. It would cost $215 million in the next fiscal year, including $70 million for the National Cancer Institute.

"Everyone with every disease wants to do this," Dr. Green said. "Is the Type 2 diabetes that results in the loss of a limb the same disease as the one that is easily controlled with diet? Right now, we lump them together."

The plan was shaped by a 2011 report from a National Academy of Sciences expert committee that urged the federal government to pour money into a new taxonomy of diseases that would define them by their molecular and environmental causes rather than their physical signs and symptoms.

To illustrate the potential of the approach, the committee contrasted two hypothetical patients.

The first, with breast cancer, would today have her tumor analyzed to determine which drugs would probably work against it. She might also have genetic tests to reveal whether she had a risky gene mutation.

The second patient, not so lucky, would have Type 2 diabetes, “an imprecise category,” the report said. “No concrete molecular information is available to customize Patient 2’s therapy to reduce his risk for kidney failure, blindness or other diabetes-related complications. No tests are available to measure risk of diabetes for his siblings and children.”

The report also noted that the diabetes diagnosis “gives little insight into the specific molecular pathophysiology of the disease and its complications; similarly, there is little basis for tailoring treatment to a patient’s pathophysiology.”

The goal of precision medicine is to give the diabetes patient — and many others — the same sort of molecular diagnosis and targeted treatment as today’s breast cancer patient.

Cancer has shown the enormous potential of precision medicine, researchers say. Dr. Charles Sawyers, who was a co-chairman of the National Academy of Sciences committee and directs the human oncology and pathogenesis program at Memorial

Sloan Kettering Cancer Center, said further progress on cancer — “the low-hanging fruit” — could be greatly accelerated with the proposed support.

Some patients have already benefited as doctors discover genes driving their tumor’s growth and prescribe drugs aimed at those genes.

“No matter what tumor type you have, a certain percentage of patients, often a small percentage, have mutations that would likely result in a treatment that would work and that we never would have thought of,” Dr. Sawyers said.

Researchers, he said, are now doing what they call basket trials. Instead of studying, say, lung cancer patients, they are studying all patients who have a mutation in their tumor that might be blocked with a particular drug.

“Lots of dramatic successes have come out in the last year or so,” Dr. Sawyers said.

He envisions not only more drug discoveries but also more widespread sequencing of patients’ tumors. Today, Dr. Sawyers said, tumor gene sequencing is mostly done at major cancer centers using money from philanthropies. Even at Sloan Kettering, he said, only subsets of patients have their tumor genes sequenced because there is only so much money.

The hope is that every disease has molecular underpinnings that, once understood, will lead to new treatments or even cures. And the proposed accumulation of genetic and clinical data from as many as a million people may also help scientists figure out genetic features that predispose people to disease and why. That sort of work has been a focus at the Broad Institute in Cambridge, Mass., where Dr. Sekar Kathiresan has had the frustration of trying to pull together scattered genetic data.

In one of his recent studies, he asked if HDL cholesterol, the so-called good cholesterol, really protects against heart disease or whether it is more like gray hair and aging. People get gray hair when they grow old, but gray hair does not cause aging. People with a lower risk of heart disease often have higher levels of HDL, but does HDL actually reduce their risk?

To get an answer, Dr. Kathiresan and his colleagues needed data from more than 116,000 people in 20 studies. In each instance, his team had to contact investigators on the studies to ask for help analyzing their data.

In the end, Dr. Kathiresan's team found that HDL did not directly protect against heart disease. If the precision medicine initiative supplies genetic and clinical data in a form that is easy to use, it would speed such studies, scientists say.

"This is an experiment we are poised to do," Dr. Sawyers said. "It is not a giant 'put a man on the moon' type project. It is a very tractable question."

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