

Audio Report Transcript 08-12-15

California's Precision Medicine Initiative
Targets Hard-to-Diagnose Diseases

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Publication: *California Healthline*
Date Posted: August 12, 2015

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The field of precision medicine is gaining steam, both nationally -- from a federal initiative -- and also statewide, through a \$3 million California initiative to boost precision medicine efforts. Using breakthrough genomic techniques, researchers are able to diagnose the source of infections by sequencing an individual's DNA. And the California initiative has a specific goal: to affect actual health outcomes within two years. This is a report for *California Healthline*, a daily news service of the California HealthCare Foundation. I'm Ali Budner.

Julie Osborn is a stay-at-home mother of two in Cottage Grove, Wis. In 2012, her teenage son Joshua came back from a mission trip to Puerto Rico with unusual vision problems and a dramatic drop in his platelet count. Doctors kept the symptoms under control for a little while, but then Joshua got worse.

(Osborn): "That's when the meningitis set in and more problems with his eyes and bloody noses and extreme headaches."

His parents pulled him out of school, and he was admitted to the hospital. He developed severe encephalitis -- swelling of the brain -- and started having seizures. Eventually his doctors placed him in a coma to protect him, but they still couldn't pinpoint the cause of his symptoms.

(Osborn): "It was really scary. Especially when you bring him to the doctor and they don't have answers."

That's when a consult with UC-San Francisco physicians may have provided a breakthrough. They had pioneered a way to use DNA testing to diagnose acute infectious diseases, using a test called unbiased metagenomic next generation sequencing. The name is a bit of a tongue-twister, but ...

(Chiu): "It's actually a very simple idea. It's a sequencing test that is able ... to identify... all potential pathogens that cause disease. So it has the ability to detect all viruses, bacteria, fungi and parasites directly from clinical samples."

Charles Chiu, an infectious disease specialist at UCSF, helped develop this process for making speedy diagnoses. His lab developed a computer program to analyze the

data and process the sequencing information, which is called sequence-based ultra-rapid pathogen identification, or SURPI.

Using these technologies in tandem, Chiu was able to diagnose Joshua's illness in two days. He found the DNA of a bacteria called *Leptospira* in Joshua's cerebrospinal fluid.

(Chiu): "That's an organism for which there currently is no validated test to detect. So we were able to use this sequencing test to diagnose this and to make sure that he got appropriate treatment."

Joshua was cured with a high dose of one of the most common antibiotics: penicillin. The problem Joshua encountered, Chiu says, is that clinicians often rely on an empirical diagnosis -- basically meaning an educated guess -- to diagnose infectious diseases like pneumonia, sepsis and encephalitis. This can leave room for error, incorrect treatment and potential worsening of the patient's symptoms, Chiu said.

(Chiu): "By harnessing one of the key tools of precision medicine -- genomic testing -- we hope we can make a difference in infected patients by providing a more timely diagnosis."

Precision medicine treats and prevents disease by taking into account individual variability in genes, environment and lifestyle. Esteban Burchard, a UCSF researcher who is on the advisory team for President Obama's \$215 million Initiative to Advance Precision Medicine, said individual DNA profiles can go a long way toward treating them properly.

(Burchard): "So we are actively taking people's genetic material from biopsy and looking at which genes are turned on or turned off and using that to either risk-stratify patients in terms of high-risk or low-risk groups, and that will bring about certain therapies. Or we're using it to determine exactly which drugs you need."

Gov. Jerry Brown first proposed California's precision medicine plan during last year's State of the State address.

(Jerry Brown): "Just as California has led the way with stem cell research, so too we can pioneer the new fields of precision medicine, which uses genomics, medical devices, computer sciences and other fields to treat individual patients instead of broad populations."

Atul Butte of UCSF is the principal investigator for California's precision medicine initiative, and said the state took a different approach to funding projects.

(Butte): "What we have is a process where the 10 University of California campuses, each had a process to generate two proposals for precision medicine."

Butte and his panel announced the two winning proposals in early August. One of them is Chiu's DNA-based diagnostic test, the other is a study of genomics and childhood cancer at UC-Santa Cruz.

(Butte): "The total amount for this year is \$3 million, which is only a modest amount of money here. So it's really just a start, to show what can be done. The intent was to get patients benefitting in these projects within two years."

Medical researchers also have partnerships with computer technology and biotech companies. They help crunch the data, store it and develop the platforms for sharing it in a meaningful way with doctors and patients. According to Butte, California is the perfect place for that nexus between medicine and technology.

(Butte): "We're the birthplace of two major technologies in the United States. One is the computer revolution. And the other is the biotech revolution."

Precision medicine does have some limitations. There are questions about how patient privacy will be handled and how much this will all cost. Also, precision medicine can seem abstract and difficult to understand -- so there's the lingering question of whether physicians are qualified to make sense of genetic testing on a large scale or to truly translate that into better care for patients. Chiu wants to bridge that gap.

(Chiu): "Though many hospital staff may not be familiar with precision medicine, this is something that is rapidly changing."

Both precision medicine projects hope to produce tangible results by 2017. Chiu said he also plans to validate his DNA test in a clinical laboratory, and then run it on 300 patients over the next 18 months in parallel with conventional testing to determine what impact, if any, precision medicine will have on outcomes.

This has been a report for *California Healthline*, a daily news service of the California HealthCare Foundation. If you have feedback or other issues you'd like to have addressed, please email us at CHL@CHCF.org. I'm Ali Budner, thanks for listening.