Start up example 2: Grail

first abstract

In 2013, Illumina acquired Verinata Health, a company focused on noninvasive prenatal testing (NIPT). That science came from the discovery that small fragments of fetal DNA cross the placenta during pregnancy. With sensitive tools, scientists learned how to isolate the fetal DNA to safely screen for any chromosomal abnormalities that might impact the infant's development.

By the time Illumina acquired Verinata, the prenatal test had been run over 100,000 times, Huber said. It worked extremely well, except for a handful of cases — around 20 — where a foreign signal was detected.

The researchers struggled to identify the source of the signal, theorizing that it could be some rare fetal abnormality. Several months later, the 20-odd babies were all born healthy and the team went back to the drawing board.

Around the same time, Illumina hired the former director of the National Cancer Institute, Rick Klausner. He took a look at the data in question and promptly declared that it was cancer, Huber recounted.

Unfortunately for the 20 women involved, Klausner was right. The prenatal test had detected tumor fragments known as cell-free DNA (cfDNA) circulating in the bloodstream.

"They were finding cancers with perfect specificity, literally 100 percent accuracy," Huber said.

Other NIPT companies and academic groups have reported similar findings. The catch was that the cancers were late-stage. The test couldn't change patient outcomes.

"And that led to the light bulb within Illumina of saying, OK, if this test that was developed for this other purpose, that doesn't have the kind of sensitivity that it would need to have but is still detecting the signal, what would it take to be able to go from the late-stage diagnoses that are happening here, to be able to detect cancer in its earliest stages," Huber said.

Illumina's R&D team began working on the science, embarking on a partnership with Memorial Sloan-Kettering that continues to this day. As the potential of the technology came to light, Huber said the discussions turned from, "wow this is a really good business idea," to the realization that they had a moral and ethical imperative to make this test a reality

Second abstract

Many companies are trying to develop early detection "liquid biopsy" tests that capture bits of DNA that cancer cells shed into blood.

On Thursday, Johns Hopkins University scientists launched a company called Thrive Earlier Detection Corp. to develop its CancerSEEK test, which yielded results similar to Grail's more than a year ago.

Third abstract

Backed by $1.5 billion from investors, Grail takes a very different approach to early detection of cancer. Rather than identifying genetic mutations in cells, Grail's technology will use machine learning and data crunching to identify epigenetic methylation signals, or rather which parts are used by the DNA to be transcribed to a protein. "We found it's these earlier signals that are more important," says Alex Aravanis, Grail's cofounder and chief scientific officer.