Preventing heart disease with machine learning and smartphone technology

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ABOUT THE LECTURE

Heart disease continues to be the world's leading cause of death, yet our ability to predict and lower our own disease risk remains limited. By combining advanced genomic and informatics technologies, Ali Torkamani, PhD, and his team are deciphering the human genome and developing smartphone app-based digital tools to predict people's individual cardiovascular disease risk and empower them with actionable health information. In his Front Row lecture, Torkamani described how genetic risk can be used to target prevention and improve long-term health outcomes.

TOP TAKEAWAY POINTS

1. Coronary artery disease, or CAD, the most common heart disease in the United States, is caused when **plaque builds up in blood vessels that supply the heart with blood and oxygen.** Age, lifestyle, other health conditions and genetics all play a role in determining our heart health.

2. There are two types of genetic risk—monogenic and polygenic. Monogenic risk is driven by variants in a single gene and is considered rare. **Polygenic risk is calculated based on the contribution of thousands of known variants** and can help identify who is at higher risk of developing disease and consequently who would benefit the most from treatments. Torkamani focuses on developing tools for calculating polygenic genetic risk scores for a number of diseases, including CAD.

3. The MyGeneRank study is the premier genetic risk score study from Torkamani's lab. It aims to determine how knowledge of genetic risk for CAD influences health decisions. Participants can download the free study app to their smartphone, link it with their genetic data from 23andMe and receive a personalized genetic risk score for CAD. By completing occasional surveys through the app, **participants are enabling scientists to gain a more precise understanding of how genetic risk impacts our health** while receiving valuable personal health information they can consult their doctor about.

4. Artificial intelligence is enabling scientists to expand the benefits of genetics even further. Torkamani's lab is developing predictive machine learning models known as "meta-prediction scores" to fine-tune the algorithms used to calculate genetic risk. Their meta-prediction score for CAD has been shown to provide a **more precise assessment of who is likely to experience a heart attack or stroke** in the next 10 years.

5. The team is planning on **expanding the scope of the MyGeneRank stud**y to include breast and prostate cancer genetic risk. Individuals who are interested in learning more should visit mygenerank.scripps.edu

Interested in hearing more? Tune in to a recent podcast episode where Torkamani shares insights about his work.

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