Docket #: S11-082

Method and System for the Use of Biomarkers for Regulatory Dysfunction in Disease

Many human disease conditions, such as Rheumatoid Arthritis and Type 2 diabetes, are known to have components of regulatory dysfunction as part of the genetic and biomolecular basis of their pathogenesis. Underlying genetic variants or environmental factors can contribute to disease pathology through disruption of gene regulatory processes, which can result in altered downstream molecular physiology (e.g. insufficient levels of signaling proteins). Because the status of regulatory function in an individual cannot be determined from genetic sequence alone, comprehensive clinical assessment requires direct molecular profiling of regulatory components and processes. Current methods for disease risk assessment and diagnosis test for genetic variants as well as protein expression, but do not consider established knowledge of regulatory dysfunction in disease. This invention addresses the unmet need for regulatory biomarker profiling to incorporate regulatory functional status into diagnosis and prognosis. This invention allows for assessment of regulatory profiling from peripheral tissues, such as blood, which are easily and routinely collected as part of the typical course of clinical care.

Applications

- Markers that can be used in diagnostics for diseases where a transcription factor binding event plays a role.
- Used to adjust disease risk profiles for healthy individuals, as with typical genetic variants

Advantages

- Existing methods for disease risk calculations rely on genetic markers, which are one more degree separated from the biology involved in disease. For diseases where a single protein product is known to be altered in the disease, diagnostics can be done at the protein level, but these are difficult to develop in a high-throughput fashion. Transcription factor binding can be used as an effective, biologically-relevant biomarker that can be rapidly and cost-effectively developed.

Patents

- Published Application: 20130116931
- Published Application: 20180373838
- Issued: 9,946,835 (USA)

Innovators

- Michael Snyder
- Atul Butte
- Konrad Karczewski
- Joel Dudley
- J. Michael Cherry
- Alan Boyle
- Eurie Hong
- Julie Park

Licensing Contact

Hyunjin Kim

Licensing Manager, Life Sciences

Email