Docket #: S15-348

Methods for interpreting whole-exome next generation sequencing

Stanford researchers in the KC Huang Lab have patented a method that identifies functionally conserved protein regions with recurrent genomic alterations in cohort studies using natural variations in genomic sequences, which allows for the discovery and further understanding of molecular mechanisms of action. The prototype identified shared molecular function between common and rare variants of known cancer genes for 20 cancer types. This method combines advanced bioinformatics and machine learning to improve variant prediction accuracy and diagnostic capability, making it an ideal tool for precision medicine, bioengineering, genomic biomarker calling, drug development, and more.

Stage of Development:
Prototype

Applications

- **Identification and Discovery:**
  - Large sequence datasets
  - Relevant variant clauses when applied to clinically annotated exomes
  - Functionally relevant coding changes
  - Novel genomic biomarkers related to patient states
- **Evaluation and Analysis:**
  - Risk scores for individuals
  - Recommend target drug candidates

Advantages

- **Currently no other methods exist to:**

    - Categorize functional relationships between exome positions in eukaryotic proteins
    - Interpret low-frequency coding variants using measurements of evolutionary coupling

Publications

- Colavin, A., Huang, K. C., & Araya, C. L. (2021). *U.S. Patent No. 10,886,007*. Washington, DC: U.S. Patent and Trademark Office.

Patents

- Published Application: 20170220734
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