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PERSONALIZED ALLELE-SPECIFIC EXPRESSION PROFILING RNA PROBE REAGENTS & ALGORITHMS

DIAGNOSTICS

In situ hybridization probes, algorithms, and software to profile allele-specific expression in patient samples.

TECHNOLOGY TYPE

Biomarkers
Research Tools
Allele-Specific Expression
Genomic Imprinting

STAGE OF DEVELOPMENT

- Tested in leukemia, Ewing's sarcoma, and melanoma cell lines.

- Ongoing testing to optimize algorithm for detecting rapidly processed introns.

IP PROTECTION

Utility Patent Pending in the United States

Patent Pending in Canada

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Reference Numbers: U-6015, U-6202

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TECHNOLOGY SUMMARY

Understanding the allele-specific expression effects helps determine how inherited mutations may impact carriers and their offspring. Existing allele expression diagnostics, such as bacterial artificial chromosome (BAC) probes, only work on cultured cells, which increases the time required for testing.

The proposed invention uses in situ hybridization probes to detect allele specific expression in cells and tissues. Nuclear whole transcriptome RNA sequencing is used to provide an intron retention score from samples to resolve expression of target alleles. These tools can resolve epigenetic allelic effects, genomic imprinting and random X-inactivation to monitor health and disease progression, and detect disorders by profiling RNASeq data.

FEATURES AND BENEFITS

- Provides single-cell detection tools for mapping mono-allelic versus bi-allelic expression in blood or tissue biopsies.
- Offers a pipeline to compare RNASeq data and identify abnormal allelic expression to categorize individual patients.
- Large proprietary Atlas/Database of tissue specific human genes with allelic imprinting effects in healthy human body.
- Algorithm tools to discriminate epigenetic allelic effects from genetic defects (cis eQTLs).

RECENT PUBLICATIONS

Huang, W., Ferris, E., Cheng, T., Hörndli, C. S., Gleason, K., Tamminga, C. . . . Gregg, C. (2017). Diverse Non-genetic, Allele-Specific Expression Effects Shape Genetic Architecture at the Cellular Level in the Mammalian Brain. *Neuron*, 93(5). doi: [10.1016/j.neuron.2017.01.033](https://doi.org/10.1016/j.neuron.2017.01.033)

Bonthuis, P.J., Huang, W.C., Hörndli, C.S., Ferris, E., Cheng, T., Gregg, C. (2015). Noncanonical genomic imprinting effects in offspring. *Cell Press*. 12(6):979-991. doi: [10.1016/j.celrep.2015.07.017](https://doi.org/10.1016/j.celrep.2015.07.017)

INVENTOR PROFILE

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