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Decoding the Evolutionary Histories and Functions of Human Accelerated Region

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Human accelerated regions (HARs) are sequences that have been highly conserved through millions of years of vertebrate evolution and then changed dramatically in the human genome since divergence from our common ancestor with chimpanzees. This evolutionary signature suggests that HARs play important roles and that their functions may have been lost or changed in our ancestors, making HARs exciting candidates for understanding the genetic basis for what makes us human. However, it has been challenging to determine what HARs do and why the evolutionary forces constraining HAR sequences in other species suddenly changed in our lineage. In this talk, I will describe updated methods for identifying accelerated regions in any lineage using large multiple sequence alignments and machine learning approaches that have shed light on the evolutionary histories of HARs. These modeling approaches are generating new hypotheses about the fastest evolving regions in the human genome, which we are testing using high-throughput genomic tools for functional characterization of non-coding sequences. This prediction-first strategy exemplifies my vision for a proactive, rather than reactive, role for data science in biomedical research.

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