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The Landscape of Human Evolution

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In the pregenomic era, evolutionary genetics was a painstaking process. From observations of the natural world, scientists hypothesized instances of selection and sought confirmation on a case-by-case basis. As of 2000, only a handful of such cases had been identified. Technological and analytical advances in the past decade, however, have enabled us to progress from hypothesis-testing to hypothesis-generating science. Rather than examining single-candidate genes, we can scan the entire genome to identify variants under natural selection. In the initial phase of the postgenomic era, we have confirmed earlier hypotheses of evolution for malaria resistance, skin pigmentation, and lactose tolerance, and we have identified new adaptations for the formation of hair, resistance to trypanosomes, and response to high altitude. The challenge now is to uncover how hundreds of newly discovered candidate loci have shaped our evolution. In my laboratory's own recent scans, we identified more than 200 loci with strong evidence of selection. Of these, roughly half point to genes, and the other half point to large, intervening, noncoding RNAs (lincRNAs), other regulatory elements, and many yet-unknown regions. It is intriguing that whole new adaptive pathways are coming into view, such as those regulating sensory perception and thermoregulation in Asia, and metabolism and infectious disease in all populations. In the next decade, scientists can look forward to investigating these pathways and many other new hypotheses being generated through genome scans to uncover the vast landscape of human evolution.