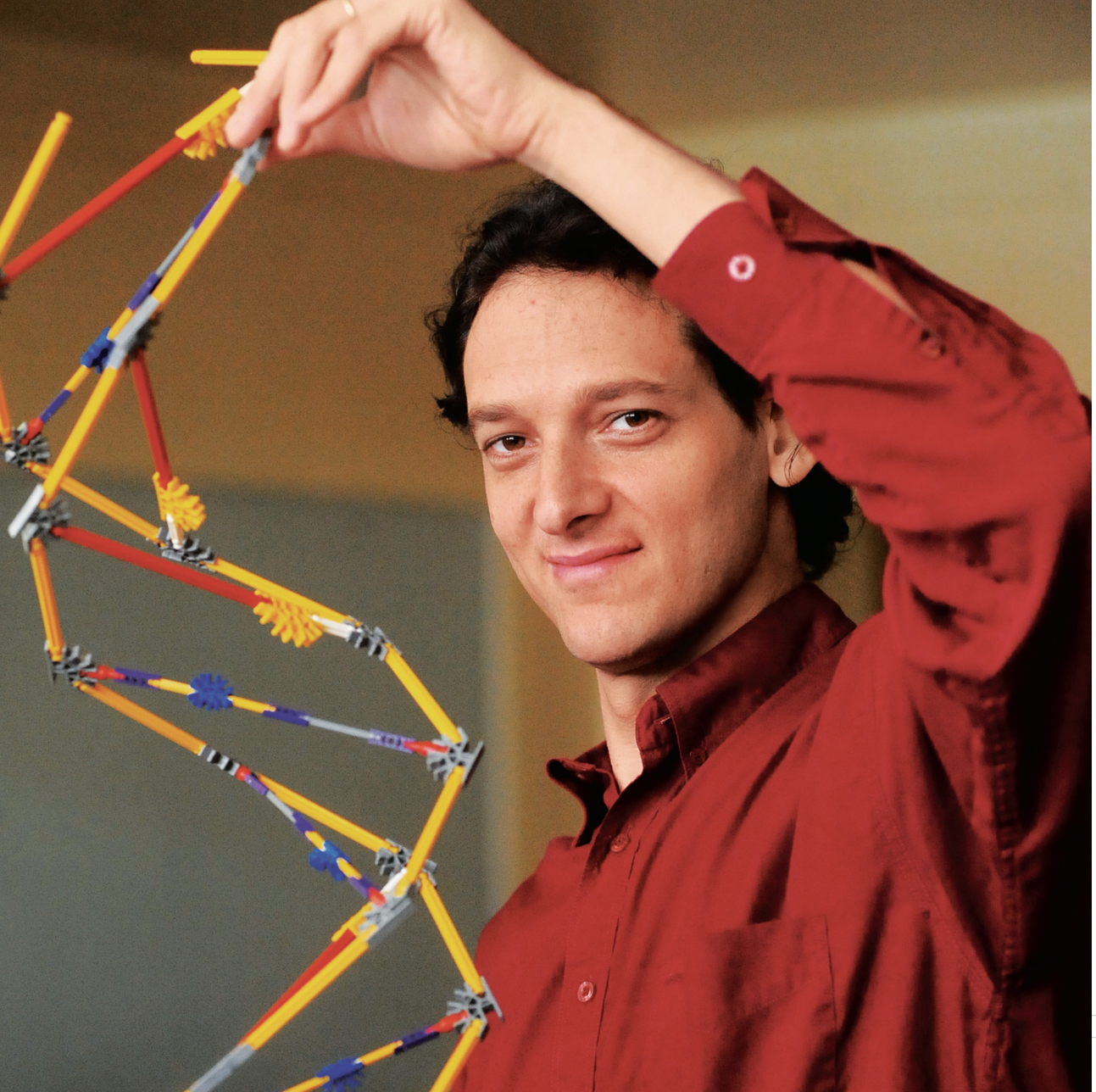

Discovering Origins of Diabetes

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Nearly 50 million people nationwide struggle with type 2 diabetes or high cholesterol, and rates are increasing annually. The clues to why some people are more susceptible than others are being discovered on a small Pacific Island, where Columbia Engineering researchers are discovering new genetic variation and associating it with metabolic disease.

Itsik Pe'er, who leads the Itsik Pe'er Lab of Computational Genetics, is developing analytical methods for analysis of DNA sequence variants. Recent technological breakthroughs now allow high-throughput observation of these genetic alterations along the genome (an individual's collection of genetic material).

Such heritable changes are thought to be responsible for 40 to 90 percent of population risk to a wide variety of health conditions, from diabetes to schizophrenia. The Pe'er group is studying a population from the Pacific Island of Kosrae, in the Federated States of Micronesia, which suffers from increased rates of metabolic disorders, such as obesity, type 2 diabetes, and high cholesterol.

The unique genetic makeup of the islanders, who have been isolated for thousands of years, makes them ideal for genetic studies, but their interrelatedness makes analysis of their DNA extremely complex.

The Pe'er group has developed computational tools to decipher remote family ties between individuals based on identity of genomic segments inherited by descent from a recent unknown ancestor. These analytical methods enabled examination of 500,000 polymorphic sites along the genomes of 3,000 Kosraeans, representing most of the adult population.

The lab was thus able to discover multiple new disease genes for health traits. Based on these disease associations, the researchers were able to sequence the entire genome of representatives of the Kosraean population, resulting in discoveries that have broad implications for anyone with these metabolic diseases.

The unprecedented scope and uniqueness of this recently completed dataset expose the effects of population isolation, and pinpoint severe mutations in individual genomes that are likely associated with disease. The combination of these genome sequences with the precompiled map of segments that are identical by descent delineates large groups of mutation carriers to confirm such associations.

Before joining Columbia, Pe'er was a postdoctoral researcher at several institutions, including the Weizmann Institute of Science and Massachusetts General Hospital. His research group is a home to an interdisciplinary team attracting diverse academic backgrounds, analytical talents, and skill sets to effectively promote computational understanding of human genetics.

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