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New technology uncovers never-before-seen human genome variations

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A new genome sequencing technology has uncovered many never-before-seen human genome variations.

The discoveries at the University of Washington close many human genome mapping gaps that have long resisted sequencing.

Evan Eichler, who led the team, said that the technique, called single-molecule, real-time DNA sequencing (SMRT), may now make it possible for researchers to identify potential genetic mutations behind many conditions whose genetic causes have long eluded scientists.

Eichler added that using the new approach in the hydatidiform genome, the researchers were able to identify and sequence 26,079 segments that were different from a standard human reference genome used in genome research.

He continued that most of these variants, about 22,000, have never been reported before and these findings suggest that there is a lot of variation they are missing.

The technique also allowed Eichler and his colleagues to map some of the more than 160 segments of the genome, called euchromatic gaps that have defied previous sequencing attempts and their efforts closed 50 of the gaps and narrowed 40 others.

Eichler further added that the gaps include some important sequences, including parts of genes and regulatory elements that help control gene expression and some of the DNA segments within the gaps show signatures that are known to be toxic to Escherichia coli, the bacteria that is commonly used in some genome sequencing processes.

He added that the gaps also carry complex sequences that are not well reproduced by standard sequencing technologies and the sequences vary extensively between people and are likely hotspots of genetic instability.

The study is published in the journal Nature.