

Rare variant discovered through deep whole-genome sequencing of 1,070 Japanese people

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Credit: Tohoku University

A research group at Tohoku Medical Megabank Organization (ToMMo) has successfully constructed a Japanese population reference panel (1KJPN), from the genome information of 1,070 individuals who had participated in the cohort studies of the Tohoku Medical Megabank Project.

ToMMo identified through this high-coverage sequencing (32.4 × on

average), 21.2 million, including 12 million novel, single-nucleotide variants (SNVs) at an estimated false discovery rate of

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