

# Machine learning for damaging mutations prediction

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Next-generation sequencing technology has ushered in a new era in medicine, making it easier to identify a sequence of nucleotides in DNA or a sequence of amino acids in the proteins of a specific individual and

use this information for diagnosis and treatment. Minute alterations in these sequences can be indicative of a minor disorder, and sometimes a grave disease.

Scientists from Skoltech, the Technical University of Munich, St. Petersburg Polytechnic University and the Indian Institute of Technology Madras (Chennai, India) have developed a [machine-learning](#)-based method to analyze the atomic structures of proteins and predict the pathogenicity of mutations. The method is adapted for [transmembrane proteins](#) that account for 25 to 30 percent of all the proteins in a cell, and often serve as targets for drugs.

"In this study, we used a combination of 1-D information on the amino acid sequences of proteins and 3-D information on the protein's atomic structures to create an effective machine-learning-based model that helps identify disease-associated amino acid substitutions in membrane proteins," says the first author of the study and assistant professor at Skoltech, Petr Popov.

**More information:** Petr Popov et al, Prediction of disease-associated mutations in the transmembrane regions of proteins with known 3D structure, *PLOS ONE* (2019). [DOI: 10.1371/journal.pone.0219452](https://doi.org/10.1371/journal.pone.0219452)

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