

Genetic data may up diagnostic efficiency for rheumatic disease

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(HealthDay)—Converting genotype information into an interpretable

probability value for five different inflammatory arthritis-causing diseases could potentially improve the diagnostic efficiency of rheumatic disease, according to a study published in the May 27 issue of *Science Translational Medicine*.

Rachel Knevel, M.D., Ph.D., from Harvard Medical School in Boston, and colleagues developed a Genetic Probability tool (G-PROB) to calculate the probability of different diseases for a patient based on genetic risk scores. The G-PROB was tested for inflammatory arthritis-causing diseases ([rheumatoid arthritis](#), [systemic lupus erythematosus](#), spondyloarthropathy, psoriatic arthritis, and gout). After validation on simulated data, G-PROB was tested in three cohorts with 1,211, 245, and 243 patients.

The researchers found that calibration of G-probabilities with disease status was high, with regression coefficients varying from 0.90 to 1.08. Across the three cohorts, G-probabilities discriminated true diagnoses, with pooled areas under the curve of 0.69, 0.81, and 0.84, respectively. At least one disease could be ruled out for all patients; in 45 percent of patients, a likely diagnosis was identified with a [positive predictive value](#) of 64 percent. The clinician's initial diagnosis was incorrect in 35 percent of cases. Thirty-nine percent of the variance in final disease was explained by initial clinical diagnosis; after adding G-probabilities, this improved to 51 percent.

"Preexisting [genetic data](#) could be considered part of a patient's [medical history](#) given its potential to improve precision medicine in the modern outpatient clinic," the authors write.

One author disclosed financial ties to the biopharmaceutical industry; a second author disclosed consulting for a nutrition company.

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