

Press Release

The world's leading AI-based, diagnostic tool for medical professionals, SimulConsult, is now accessible on mobile devices

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SimulConsult announces a new version of its diagnostic decision support tool. The tool combines the power of curated human expertise and computational artificial intelligence (AI) to empower clinicians in diagnosis and workup of patients. It is used today in 118 countries. NIH-funded studies showed it can lower diagnostic errors by up to 75%.

The new version of the tool being released today has a completely new interface that allows it to run on mobile devices as well as on computers. It is fast to use and puts the diagnostic power into a clinician's hands whenever needed.

The tool is designed by doctors for doctors. It allows the user to compare a patient with its curated descriptions of diseases. The tool suggests useful findings (i.e., symptoms, signs and test results) based on the differential diagnosis (the probability-weighted candidate diseases), allowing the iterative approach that is the classic approach used by doctors for effective and efficient diagnosis. Its database already covers >6,900 diseases, including all well-described chromosomal disorders and all diseases with genes that have germline changes convincingly associated with human disease.

In many medical specialties, clinicians do not need much assistance to make diagnoses in their own specialty. However, genetics includes essentially all specialties, and includes thousands of diseases. All clinicians, including geneticists, welcome assistance in diagnosing genetic diseases, including the hundreds of new genetic diseases added each year. In addition, all specialists value the ability to get help with diagnosis in adjacent specialties. That is why we have built SimulConsult as a single tool across all specialties, so that the differential diagnosis includes both the diseases the clinician is expecting and unfamiliar diseases that should be considered as well. This makes it easier to identify the relevant specialists, select the most useful tests based on their relevance, cost and the treatability of the diseases.

SimulConsult achieves the once-elusive "Explainable AI". It automatically documents the justification for the diagnosis. It also documents justification for testing in a way that is so evidence-based that many insurers are accepting it as justification for reimbursement.

Three capabilities differentiate the SimulConsult tool. One is the iterative model for diagnosis (imagine if your web search would suggest questions helping narrow your search). A second is the use of time, including onset information for findings and speed of emergence, made possible by detailed curation of information about onset and disappearance of each finding in each disease. The third is the ability to use pertinent negative findings, made possible by the tool's detailed quantitative database.

The innovative Prognosis Table feature has been shown, in PCORI-funded research, to be valued for its ability to summarize any disease with a table of findings and how each evolves over time. This is a powerful resource for answering the question from patients and referring physicians "given the diagnosis, what should I expect".

The Genome-Phenome Analyzer version of the tool allows the user to import annotated genomic variant tables and then interpret them in the clinical context, answering the questions "which gene abnormalities fit with the clinical picture?" and "given the gene abnormalities found, what clinical findings or tests should I check to distinguish among the suspicious genes?" Copy Number Variants (CNV) can be analyzed together with Single Nucleotide Polymorphisms (SNP).

For more information and to subscribe, go to <https://simulconsult.com>. Users who subscribe during the ACMG meeting will get a special discount.