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# SimulConsult Launches Software to Link Genomic, Clinical, and Phenotypic Data

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**SimulConsult,** a bioinformatics company based in Chestnut Hill, Mass., is marketing a new software tool called the Genome-Phenome Analyzer, which lets clinicians and research labs combine whole-genome and whole-exome sequencing data with clinical findings and phenotype information from the scientific literature.

Lynn Feldman, CEO, explained to *BioInform* that SimulConsult developed the software to automatically correlate patients' clinical findings and bioinformatics analysis results with phenotype information, a step that is usually done manually.

Genome-Phenome Analyzer, which was launched in March, enables clinicians to assess and prioritize which gene variants are most likely to account for the clinical findings, the company said.

It uses information from SimulConsult's curated phenome database, clinical findings, and a table of associated variants generated by whole-exome sequencing to compute a differential diagnosis for patients using proprietary statistical pattern-matching algorithms.

The software reads information in the variant table generated by the initial bioinformatics analysis of a patient's genomic information, and imports the Human Genome Nomenclature Committee code for the gene, the severity of each variant, and the zygosity of the variant. This information is then matched with the clinical findings for the patient as well as information culled from the literature.

With this tool users can detect autosomal dominant and autosomal recessive diseases as well as deal with complex situations involving compound heterozygosity, for instance. It can also be used to identify difficult-to-detect genetic variants such as trinucleotide repeats and large copy number variants.

Clinicians can then take the results generated by the system and run further tests, such as additional Sanger sequencing, to obtain more information about a variant in question, or they can check familial information in cases of inherited diseases, Feldman said.

Genome-Phenome Analyzer builds on medical decision-support software previously developed by the company to enable clinicians to combine clinical and laboratory findings to come up with a diagnosis for their patients.

The medical decision support system prioritizes diseases based on pertinent positive and negative findings, as well as the onset and disappearance of each finding, family history, and incidence of diseases.

That system also includes SimulConsult's database — focused primarily on genetic neurological and metabolic diseases, although it is being expanded — which is populated by information from about 1,400 sources including GeneReviews and GeneTests as well as several textbooks and articles.

Before public information is entered into the database, it is manually curated by medical specialists and sub-specialists, Feldman said. In addition to ensuring that the information entering the repository is of good quality, the manual curation step also ensures that clinical findings that may not be explicitly reported in the medical literature or that may be rare occurrences in patient populations are captured and added to the database, she said.

Clinicians can contribute information about clinical findings and diseases to the database, which is freely available. Contributed information is subject to the same curation process as public sources before it's entered into SimulConsult's database.

Since the tool's initial debut at American College of Medical Genetics annual meeting in March, Feldman said that it has received "considerable interest." So far the company has signed one contract with a CLIA lab and is in the process of signing additional contracts for evaluation pilots, although she could not disclose who these groups are.

She said the company offers seat-based licenses for small research labs and site licenses for larger CLIA labs but declined to provide pricing details.

SimulConsult was founded in 1998 by Michael Segal, a pediatric neurologist who formerly worked at the Children's Hospital Boston and now serves as the company's chairman and chief scientist. The firm launched its first product, a database and the decision support software, in 2006.

Although the use of the database is free, that could change, Feldman said. The company currently charges for integrating its resource with physician order-entry platforms, she said.

It is also working on integrating the system with an electronic health record system in a clinic but Feldman could not disclose additional details.

Currently, the company has less than ten employees and it plans to hire more staff including a chief medical officer and a sales person in the next few months, Feldman said.

The company also hires a large number of contractors to curate the information that goes into its database, she said.