MedicalXpress

Geneticists launch Matchmaker Exchange for rare disease gene discovery

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In a special issue of *Human Mutation*, a team including investigators at Brigham and Women's Hospital has announced the launch of the Matchmaker Exchange - a way for the rare disease community to share information and find new connections. Matchmaker Exchange connects databases of genetic information and symptoms that physicians and investigators can "match" with a patient's rare disease.

"In the past, searching for the cause of a rare genetic disease was like trying to find a needle in a haystack. There would be an occasional, serendipitous connection made by a clinical laboratory or individual investigator of two patients who shared the same rare disease, but there was no systematic way to find these matching cases," said Heidi Rehm, PhD, a molecular geneticist at BWH and director of the Laboratory for Molecular Medicine at Partners HealthCare Personalized Medicine. "Matchmaker Exchange offers a reliable, scalable way to find matching cases and identify their genetic causes."

Matchmaker Exchange (MME) 1.0 brings together multiple databases and programs and harnesses collective data from across rare disease repositories. The platform allows investigators to search the databases and uncover similar symptoms and genetic profiles, using standardized application programming interfaces (APIs) and procedural conventions. Using a federated network approach, MME protects the privacy and security of patient data while connecting the databases through APIs.

In the same issue of Human Mutation announcing MME's launch,

researchers also present case examples of discoveries already made using matchmaking approaches for uncovering rare disease genes and describe the matchmaking services and components that are already part of MME or are intended to be connected to it in the future.

Rehm and her colleagues will also be presenting the new platform at the American Society of Human Genetics Annual Meeting on October 7 at 7 p.m. For more details, please visit http://www.ashg.org/2015meeting/.

More information: Phillippakis, A et al. "The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery." *Human Mutation*. September 17, 2015. DOI: 10.1002/humu.22667

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