TRANSCRIPT OF MEDIA- Mapping the future for early diagnosis of rare diseases

Adam Cross, MD, a pediatric hospitalist, clinical informaticist who heads the Children's Innovation Lab at Jump Trading Simulation & Education Center. Assistant Professor, University of Illinois College of Medicine Peoria (UICOMP).

Dr. Cross says the grant-funded research project is using generative AI and medical knowledge graphs to improve diagnosis of rare diseases.

"We are trying to create a model that will take the notes that are already being written by primary care physicians and look for unique sets of signs and symptoms that might suggest a patient has an undiagnosed rare disease." Dr. Cross adds, "That approach can empower the physician with information regarding those diseases and prompt further testing if they choose to do so." (:25)

The Jump ARCHES research creates comprehensive knowledge graphs to visually map the relationships between various medical conditions and symptoms of rare diseases from existing resources.

"We did this so that we could give people a better visual example of just how complex these diseases are and how they're all connected," Dr. Cross observes while viewing an early version of a knowledge graph created by researchers that looks like a celestial constellation. "It's kind of beautiful when you see the simplicity and the complexity of all the different symptoms, but the simplicity of the structure. It gives you a sense of just how incredible these new technologies really are in terms of helping us find patterns in the chaos." (:26)

The effort involves sophisticated machine learning approaches that have changed with lightning speed, even since Dr. Cross and his UIUC colleagues began their work.

"Even in that year, there have been more advanced and more powerful methods that have come out that we've since adopted, and we continue to improve our own methods based on what's being developed globally." (:13)

As the approach is refined and improved, Dr. Cross also doesn't rule out the possibility of discovering new rare diseases.

"As we go along, it is certainly possible with the technology that we've created, maybe we'll start to see patterns of signs and symptoms that don't have a known diagnosis. Or maybe there are subtypes of diseases that are out there that really seem to have their own unique progression, and we could also maybe discover those in subsequent phases of the project." (:22)