

BROADCAST-Mapping the future for rare disease diagnosis

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Joint research between OSF Innovation and the University of Illinois Urbana Champaign could help primary care providers detect rare diseases earlier.

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A grant-funded project uses generative AI and something called knowledge graphs to maps signs and symptoms from a patient's medical record to detect 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.” (:22)

That's Dr. Adam Cross, who leads the OSF Children's Innovation Lab in Peoria. He wants to create a point-of-care tool for primary care providers to help patients get an early diagnosis of a rare disease – something that could improve treatment and outcomes.