Communicating Exome Sequencing Results in the Clinic

○UCLA **Tanya Stivers**
UCLA **Stefan Timmermans**

Genomic tests such as exome sequencing have recently become an option for diagnosing patients. The tests allow clinical geneticists to sequence the majority of patients' disease causing genetic variants. As a new technology, exome sequencing confronts the question of what the benefit is of this increased genetic information. Against a narrow perspective of clinical utility that emphasizes tangible improvements in a patient's disease management, professional organizations have argued that genomic sequencing should be considered beneficial if it helps families and society. Based on video-recorded observations of the return of exome sequencing results to parents of a child with disabilities in the clinic and in-depth interviews with these parents, we examine how genomic test results become actionable in the clinical encounter. We find that parents and clinicians marshal exome results beyond biomedical diagnostic and management goals to address questions about guilt for causing the disabilities and to secure access to disability-related services. We argue that genomic actionability rests on the interaction between the biological characteristics of genetic results and the predicaments facing parents of children with disabilities.

Yet, despite a broad range of different sorts of results, news deliveries are striking in this genetic context. Whereas research on news deliveries has previously focused on how monovalently good or bad news and their associated interactional trajectories. We find that speakers offering bright sides against a backdrop of bad news work to achieve bivalent equilibrium – a state where speakers can reach agreement that the news is appropriately understood as a mix of bad with good elements. We propose that bivalent equilibrium facilitates affiliation through a two-step process that is distinct from affiliation to a monovalently positive or negative evaluative stance.

We draw from a corpus of 38 video-recorded consultations of families who had undergone exome sequencing by geneticists at a large academic clinic and were visiting to receive the results for their child. We augmented these data with observations and interviews with parents. Our methods include ethnography and conversation analysis.