Understanding the Practice of Genetic Result Communication to Extended Family Members by Participants in the Undiagnosed Diseases Network (UDN)

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Genetic testing results have implications not only for the individual, but also for their family members. Research on family communication of genetic results has primarily focused on families affected by adult-onset, dominant conditions as well as more common genetic diseases such as familial hypercholesterolemia, cardiomyopathies, and genetic hearing loss. This study therefore aimed to characterize the genetic result communication in the Undiagnosed Diseases Network (UDN), a study focused on providing diagnoses to individuals with undiagnosed conditions. The goals of this project were to (1) characterize the practice of genetic result communication to extended family members by UDN participants and parents who received a diagnosis through the UDN, and (2) identify factors that influence genetic result communication. A survey was distributed to 142 eligible participants to assess who participants communicated with, the content of their communication, motivations for communicating, and interactions with health care providers regarding what and how to communicate genetic results. All respondents shared genetic results with at least one person. Adult participants and parents/guardians of participants showed high levels of perceived understanding, comfort communicating, and utility of genetic results. Additionally, parents/guardians were more likely to disclose genetic results due to a general desire to share, while adult participants reported that they shared results to communicate risk to family members. Many respondents did not recall discussing what and how to communicate genetic results with a health care provider. The results of this study provide insight into the practice of result communication by participants in the UDN, which can ideally be utilized to develop more effective counseling strategies and guidelines to aid family communication.