

Recontact Upon Reclassification of Previously Identified Variants of Unknown Significance – Assessing Current Practices and Challenges Facing Diagnostic Laboratories

Jennifer L Walsh, Samantha Baxter, Stephanie Jo Brewster, MaryAnn W Campion
Boston University School of Medicine, Boston, Massachusetts

Background: Variants of unknown significance (VUSs) pose a challenge for clinical genetics providers. With continual advances in technology and genetic knowledge it is possible for a previously identified VUS to be reclassified as pathogenic or benign, which has the potential to greatly impact patient care. This study aims to assess the current practice of diagnostic laboratories with regard to VUS reclassification and communication of this information to ordering physicians. **Methods:** Laboratory directors and genetic counselors working in clinical diagnostic laboratories were invited to participate in semi-structured interviews. Qualitative analysis was performed to extract common themes regarding the current practice and the challenges of recontacting in the event of VUS reclassification. **Results:** Of the 116 laboratories invited to participate, 19 responded (16%). A greater number of academic laboratories responded (63%) compared to commercial laboratories (37%). Seventy-six percent of the respondent laboratories recontacted providers upon VUS reclassification on a regular or case-by-case basis. Factors limiting the ability to recontact providers included limited resources, difficulty locating providers, and data management challenges. Fifty-three percent of respondent laboratories believed that the practice of recontact should be standard of care. **Conclusions:** This study fills in a gap in the literature regarding the current practices of diagnostic laboratories in recontacting providers upon VUS reclassification. Implications, limitations, and future research recommendations are addressed.