From a Trio to a Duo: Non-Paternity Identified Through Diagnostic Exome Sequencing

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Family centered Diagnostic Exome Sequencing (DES) can reveal non-paternity in which a proband's attributed father is not the proband's biological father. Such findings can have ethical, psychological, and medical consequences for a family. While recommendations have been made, there is no clear consensus for if, when, and to whom non-paternity findings should be disclosed.

Non-paternity events detected through parent-proband trio DES from 1547 consecutive families were retrospectively reviewed. For each such case, referring clinicians were notified of non-paternity during analysis, and asked how they would like to proceed. After reporting, clinicians were re-contacted to determine if non-paternity was disclosed.

Among 1547 patients analyzed, non-paternity was detected and confirmed in 7 patients (0.45%). Two cases were excluded from the study as the possibility of non-paternity was brought up prior to testing, and DES confirmed the suspicion in these cases. Of the 5 remaining cases, clinicians chose to proceed with the analysis as a duo in 3 cases (60%) and elected to use an additional family member's sample for a trio in 2 cases (40%). Clinicians also requested modified reports in which pedigree and co-segregation data were removed in 4 cases (80%) and chose to receive an unaltered report in 1 case (20%). Furthermore, clinicians chose not to disclose the non-paternity findings to the family in 1 case (20%) and chose to disclose the findings to only the mother in 4 cases (80%).

Overall, despite subsequent limitations to analysis, the majority of clinicians elected to proceed with testing as a duo, and requested removal of information from the reports. While the majority of clinicians chose to disclose the non-paternity findings to the mother, none of the clinicians elected to disclose the findings to the father or the proband. Taken as a whole, these findings demonstrate a general trend towards avoiding disclosure of non-paternity findings. Non-paternity is a difficult situation, and genetic counselors are needed to discern how to respond to these findings.