Postmortem medicolegal genetic diagnostics also require reporting guidance

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The recommendations by the working group (WG) of the Genetic Services Quality Committee of the European Society of Human Genetics (ESHG) for reporting results of diagnostic genetic testing (biochemical, cytogenetic and molecular genetic) published by Claustres *et al*[1] in the *European Journal of Human Genetics* are welcomed and definitely needed. The WG focused on clinical patients, but did not address reporting of genetic testing in deceased individuals, especially for those who have been subjected to a medicolegal autopsy. In the realm of medicolegal autopsy practice, samples are collected routinely for human identification purposes. However, beyond identity testing, there are cases where genetic diagnostics are being performed to establish the underlying or contributing causes (and manners) of death. Although conceptually still somewhat in its infancy, 'molecular autopsy' employs genetic markers and predictive power of risk to assist in cause and manner of death investigations.[2] The situations often considered include genetic testing in negative autopsies, that is, sudden unexpected deaths, that have no findings in standard autopsies and cases with unusual toxicology results (pharmacogenetic analyses).[3,4] At the same time, national legislations for reporting of genetic tests postmortem are diverse or nonexistent and, although some local practice guidelines have been established,[5] international recommendations concerning medicolegal and forensic postmortem genetic diagnostics are lacking.

The principles and recommendations advocated by the ESHG WG also provide a good framework by which to guide the reporting of postmortem genetic tests. However, the medicolegal setting encounters some aspects that differ from the standard clinical arena. First, a medicolegal autopsy, in most countries, is ordered by representatives of the judicial system (eg, police or court). Therefore, an autopsy report, including diagnostic results and accompanying information, is submitted to the judicial instead of the health care system. As a consequence, at least the following potential issues arise: (1) genetic information of deceased individuals resides in files of judicial institutions; (2) there is no institutional body within the judicial infrastructure who would naturally meet the medical/genetic consultation needs of the relatives, even if the relatives were to seek advice;[6,7] and (3) the genetic information in the reports can be elicited to public during court proceedings. Some type of coding of the variants/mutations in the report possibly could address privacy protection in the report (although the type of test itself may reduce the ability to protect privacy), but the proper practices of information flow from the judicial system to relatives is more cumbersome.

Second, postmortem sampling typically is a onetime opportunity. The possibility of use of histology or toxicology samples exists for some cases, but such suboptimal samples tend to be limited for large scale genomic analyses for some future question(s) that may arise.[8] Possible exhumations for resampling have occurred for human identification or in the investigation of serial poisonings,[9] but generally have not been entertained for other scenarios. Another situation to consider is that genetic testing may be requested only after the autopsy has been performed.[10] Some standardization of collection and storage of biological specimens (eg, blood) for such purposes may facilitate effective postmortem genetic diagnostics and reports. Indeed, such protocols and suitable materials already are available and have been tested for long-term storage.[10,11]

Finally, medicolegal experts (forensic pathologists or medical examiners) often do not have the means, especially a timely action, to inform relatives. Many of these professionals are not trained on how to interpret the results and details in genetic reports, and genetic consultations based on these reports are not a standard part of their job description. The use of a multidisciplinary team including at least, but not limited to, a clinical geneticist and a forensic pathologist/medical examiner is necessary to deliver proper information of a postmortem genetic test report to the relatives.[12]

Overall, there is a great potential value in postmortem genetic testing, which could be used effectively for the benefit of individuals, families and society. To enjoy the benefits of such testing an internationally agreed set of recommendations are needed on how to make proper use of the samples and report the results in a standardized way, a consultation team to communicate with the relative(s) of the deceased, and all practices be performed in a sensitive and private manner. Such recommendations could be added to the recommendations of the (WG) of the Genetic Services Quality Committee of the ESHG that would go far in establishing a sound and healthy foundation for postmortem medicolegal tests and the manner to report the results to all appropriate parties.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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Reply to Sajantila and Budowle

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We thank Drs Sajantila and Budowle[1] for raising this interesting and important topic. They correctly point out that our Recommendations for reporting results of diagnostic genetic testing[2] do not cover the special circumstances surrounding the reporting of post-mortem genetic testing. We did not consider this issue while preparing our recommendations, focusing instead on reporting of routine genetic testing (biochemical, cytogenetic and molecular genetic). Although some of our recommendations will apply to all reports of genetic testing, there may be important exceptions when post-mortem results are being reported.

The issues around post-mortem genetic testing and 'molecular autopsy' go far beyond the reporting of results, encompassing *inter alia* issues of consent, sample integrity, legal custody and retention/storage of tissues. We understand that the Professional and Public Policy Committee (PPPC) of the ESHG is currently considering these issues with a view of producing a policy statement. The Genetics Services Quality Committee fully supports this initiative and looks forward to commenting on the draft statement when available.

CONFLICT OF INTEREST

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