Developing Genetic Education to Improve Postmortem Genetics - The NSGC Postmortem Working Group (PMWG) Collaborative Experience

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Abstract

Postmortem genetic testing provides an opportunity to clarify the etiology of sudden death cases and can clarify the risk of disease for family members. The unique challenges of this type of testing results in questions from the medical community and family members.. The NSGC Postmortem Working Group (PMWG) was formed to address queries about postmortem genetic testing in a consistent and efficient manner. The PMWG launched an email address in October 2014 and, has received and responded to 34 total queries, highlighting the need for additional education about postmortem genetic testing and providing a robust example of how to fulfill this need that may be applied to various medical subspecialties.

Introduction

Sudden death can be a devastating feature of inherited cardiac conditions. In up to 30% of sudden death cases among those 0-40 years of age, the cause of death remains unknown after postmortem examination.1 The detection rate of postmortem genetic testing in sudden unexplained death cases varies according to the number of genes analyzed, but can range from 15-20%.2,3 Published guidelines recommend collection of a DNA specimen and postmortem genetic testing in sudden death cases suspicious for channelopathies.4 Postmortem genetic testing is also helpful in autopsy positive sudden death cases including cardiomyopathies, aneurysms, pulmonary embolisms and metabolic causes among others. Subsequently, the last decade has seen an exponential increase in utilization of genetic testing in the area of forensic pathology. Postmortem genetic testing can identify the underlying cause of an individual's death and clarify disease risk for family members with the goal of prevention. Postmortem testing is unique in that it is performed on a limited DNA specimen in a complex setting. Particular challenges accompany this testing, including timing, correct specimen acquisition, proper specimen storage and shipment and a wide variety of legal and ethical issues. These challenges are best addressed with proper education of healthcare providers, medical examiners (MEs), coroners and family members. In an effort to address these educational needs the National Association of Medical Examiners (NAME) published a 2013 Position Paper entitled: Retaining Postmortem Samples for Genetic Testing.5 Genetic counselors are well-positioned to assist with the educational needs of the forensic community. However, the profession is relatively small with only 4,000 certified genetic counselors in the U.S. and Canada.6 Further, only 10% of GCs providing patient care do so in the cardiac setting.7 In an effort to improve access to genetic counseling support in the post-mortem setting, a Postmortem Working Group (PMWG) was formed through the National Society of Genetic Counselors (NSGC).

Methods

In 2013, shortly after the NAME publication, a Postmortem Working Group (PMWG) was formed through the National Society of Genetic Counselors (NSGC). The purpose of the PMWG is to increase awareness of proper specimen collection and storage following an autopsy and to act as a resource for all involved in the postmortem genetic testing/DNA banking process.

Resources

Anticipating existing questions within the ME field and amongst families members whose loved ones have died suddenly, the PMWG developed a webpage through the NSGC dedicated to postmortem genetic testing in October 2013 (http://www.nsgc.org/postmortem) as well as an email address (postmortem@nsgc.org) staffed by PMWG genetic counselors (GCs).

Staffing

Seven GCs rotate through a monthly call schedule replying to queries that are received through the PMWG email address. Staffing of the PMWG has expanded to include eight consultant GCs specializing in prenatal, epilepsy, and metabolism, reflecting the relevance of postmortem genetic testing across subspecialties.

Tracking

Queries to and replies from the PWMG email have been tracked since its inception. A Google Form and spreadsheet are used to track each query, including origin of the question, GC responder, nature of the query and subsequent referral to genetic services (if applicable). Actual email correspondence is recorded as well.

Results

Since launching the PMWG email in October 2014 to March 2017 (30 months), the PMWG has received 34 email inquiries from a variety of sources (see Figure 1).

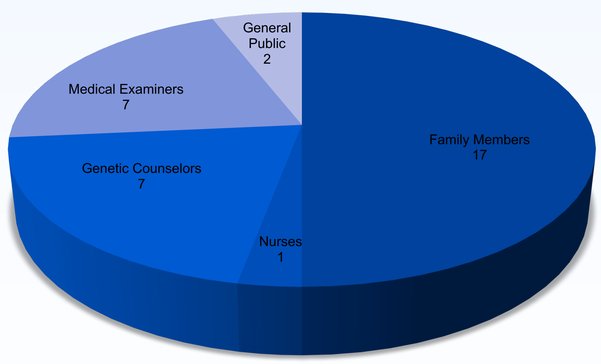

Figure 1 Thirty-four total inquiries to PMWG email categorized by source

Results

The majority of the inquiries were received from family members whose loved ones had died. Tables 1, 2 and 3 present details about the most common types of inquires as well as examples of each.

Family Members (17 queries)	**Overall nature of the queries involved requests for assistance facilitating genetic testing for their loved one.**
Example: "My husband (age 36) recently passed away. We are currently awaiting autopsy results. He did not have a family history of heart problems, but was having trouble breathing. His PCP suggested that it could be something that sounds very familiar to things your website describes. Is there anything I can do at this point to check if it was SADS?"	PMWG GC reply focused on importance of autopsy results to guide next steps and emphasized importance of genetic testing to clarify both cause of death and risk for family members. GC reply also discussed need for an appropriate specimen type to be saved by the ME office. Availability of local GC consultation was offered.
Example: Sister of decedent inquired whether a DNA sample would have been saved during autopsy from her brother who died several months earlier.	PMWG GC referred her to the NSGC Postmortem website for DNA banking facilities and referred her to a local GC to assist the family in navigating the postmortem testing process.

Table 1 Sample inquiries from family members of decedents and PMWG GC reply

Medical Examiners (7 queries)	**Overall nature of the queries involved assistance facilitating genetic testing, use of obscure specimen types and specimen handling.**
Example: ""I have a 17-yo girl with a negative autopsy and non-contributory toxicology. She has a history of a few "fainting spells," but died in a 20-minute window from being in the kitchen cooking to dead in bed. My coroner has authorized channelopathy testing. I need help deciding what lab test to order. We are spending money we do not have, so cost containment is an issue. Please have a counselor contact me."	PMWG GC reply focused on value of clinical DNA banking, the importance of clinical cardiac evaluation for family members of the decedent, availability of genetic counseling, and currently available genetic testing for channelopathies in various laboratories.

Table 2 Sample inquiries Medical Examiners and PMWG GC reply

Genetic Counselors and Nurses (8 queries)	**Overall nature of the queries involved finding an appropriate specimen, timeline for collecting specimens and identifying laboratories that perform postmortem genetic testing.**
Example from a nurse: "I was wondering if genetic testing would be plausible on a two year old who has been buried now for almost 6 years. Her father is falsely accused of child abuse. The mother apparently has some form of Ehlers-Danlos. If in fact postmortem testing is possible…..is it done on a liver biopsy?"	PMWG GC reply focused on some of the difficulties surrounding genetic testing performed on an exhumed body. Other options presented included genetic testing on living family members who may be affected, and the possibility of acquiring a blood-spot card from newborn screening for testing (as opposed to exhuming the body).
Example from multiple GCs: "What is the timeframe for collecting a postmortem blood specimen?"	PMWG GC reply advised collection at time of autopsy, as close to time of death as possible.

Table 3 Sample inquiries Genetic Counselors and Nurses and PMWG GC reply

Conclusions

The utilization of postmortem genetic testing provides a promising avenue to clarify the cause in sudden death cases. The use of postmortem genetic testing is increasing among the medical examiner community, and will likely continue to do so with continued educational efforts about the process and reduced costs of genetic testing. In addition, the application of postmortem testing to multiple subspecialties (including cardiology, neurology and metabolic), and protocols that are being developed to identify, standardize and understand the causes sudden death will lead to greater uptake.8 Genetic counselor access through the mechanism of email has shown to be effective in fielding calls from the postmortem community. Queries to the email most often involve guidance for family members of decedents seeking a cause of death for their loved one, and logistical expertise provided to medical examiners, genetic counselors and nurses navigating the complex path of postmortem testing. The PMWG will continue to track the email inquiries and continue to educate those involved in postmortem genetic testing to help make the process easier for all. This model is proposed as one method for genetic counselors to share their expertise with a specialty disease community in the setting of limited genetic counselor availability.

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