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Investigating Medical Examiners' Practices: Genetic Evaluation for Fatal Acute Aortic Dissection

Bradley Power

INVESTIGATING MEDICAL EXAMINERS' PRACTICES: GENETIC EVALUATION FOR FATAL ACUTE AORTIC DISSECTION

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INVESTIGATING MEDICAL EXAMINERS' PRACTICES: GENETIC EVALUATION FOR FATAL ACUTE AORTIC DISSECTION

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Acute thoracic aortic dissection (TAD) is a life-threatening event with a hereditary component. Currently, pathogenic variants in 11 genes associated with aortic aneurysm and dissection predispose to a heritable form of disease thereby conferring an increased risk for TAD. Genetic testing plays a pivotal role not only in diagnosis, but also in risk stratification for relatives and medical management to prevent premature death from dissection. Due to its high fatality rate, medical examiners and coroners (ME/Cs) may be the first to identify TAD cases and initiate genetic testing for the decedent and at-risk relatives. ME/Cs were surveyed using three clinical vignettes detailing two cases of early-onset TAD, one with features of Marfan syndrome and another without, and a later onset TAD case. Sixty respondents reported their likelihood to complete various actions related to their level of suspicion for a genetic cause and recommendations for relatives (e.g. collect sample for testing, recommend imaging for relatives). Additionally, respondents were queried about current practices and perceived barriers regarding genetics evaluations for TAD. Reported practices were compared to recommendations established by the National Association of Medical Examiners (NAME). Respondents were significantly more likely to perform all proposed actions in the two early-onset cases versus the late-onset, non-syndromic case. ME/C's were significantly more likely to speak with the decedent's next-of-kin (NOK) about increased TAD risk and refer for genetic counseling in the early-onset syndromic vignette compared to early-onset non-syndromic case. Experience, approximated by the number of TAD cases seen at practicing institution, did not impact respondents' choices, but access to a genetic counselor did. Cost of genetic testing was the most frequently reported barrier, followed by contacting NOK. Alignment with NAME guidelines varied, converging around sample collection, but diverging when communicating with NOK. Our results suggest that ME/Cs recognize the utility of postmortem genetic testing and the clinical risk factors for

hereditary TAD. However, ordering genetic testing and recommending aortic imaging for at-risk relatives is inhibited by concerns regarding cost of genetic testing or access to NOK. Increasing ME/C's access to genetic counseling services will be important for postmortem genetics evaluations in this population.

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Abbreviations

TAAD- Thoracic aortic aneurysm or dissection

TAD- Thoracic aortic dissection

NAME- National Association of Medical Examiners

IACME- International Association of Coroners and Medical Examiners

AHA/ACC- American Heart Association and American College of Cardiology

ME/C- Medical Examiner and Coroner

NOK- Next-of-kin

VAS- Visual analog scale

IQR- Inter-quartile range

Introduction

Acute thoracic aortic dissection (TAD) is a life-threatening event associated with a high fatality rate. Death occurs in up to 50% of affected individuals within the first 24 hours after symptom onset, most commonly from pericardial tamponade (1-3). Incidence of TAD is likely underestimated because up to 40% of individuals do not survive long enough to reach the hospital. However, it is estimated to range from 2/100,000/year to 6/100,000/year, reflecting the magnitude of individuals impacted. Due to the incidence and high disease fatality rate outside the hospital, medical examiners may be the first to make the diagnosis on autopsy (4).

It has been established for many years that Marfan syndrome, due to mutations in a single gene, *FBN1*, confers a high risk for TAD. Further, studies have shown that up to 20% of individuals with thoracic aortic aneurysm or dissection (TAAD) who have not been diagnosed with genetic syndrome, report a family history of TAAD (5, 6). Expert review of genes linked to hereditary TAAD in 2018 using a validated ClinGen framework determined that highly penetrant variants in *FBN1* and ten other genes lead to a heritable form of TAAD.(7). Burgeoning evidence demonstrates that the identification of such variants can be used to guide the treatment and management of TAD survivors and identify relatives who are at risk for dissection, thus preventing further fatalities (8). The majority of diagnostic genetic testing for TAD is completed via a multigene panel approach where the genes known to be associated with TAD are collectively assayed on a next-generation sequencing platform. The yield of genetic testing to identify the causative variant in an affected individual varies based on the presence of a family history of TAAD, syndromic physical features, and age of dissection onset. For example, 9.3% of early-onset (≤ 56 years) TAD cases with no family history have an identifiable genetic cause, while >30-40% of cases with a positive family history harbor a pathogenic variant in a known gene (9-11). Further, if syndromic features characteristic of Marfan or Loeys-Dietz syndromes are present, the likelihood of determining the genetic cause of TAD is even higher. However, classic syndromic features are not present in all individuals with Marfan syndrome (*FBN1*) or Loeys-Dietz syndrome-related genes (*TGFBR1*, *TGFBR2*,

SMAD3, *TGFB2*) (12, 13). Despite the diagnostic yield and clinical implications of genetic testing, its use by medical examiners and coroners (ME/Cs) for cases of TAD has not been investigated.

The National Association of Medical Examiners (NAME) established recommendations in 2013 to facilitate postmortem genetic testing and evaluation in cases with an increased suspicion for genetic etiologies, including TAD (14). Additionally, the American Heart Association and American College of Cardiology (AHA/ACC) guidelines for the diagnosis and management of patients with thoracic aortic disease advise on the utility of genetic testing for individuals with TAAD and recommend aortic imaging for first-degree relatives of all individuals with thoracic aortic disease (15). With over 2,000 ME/C offices in the United States, ME/C's are in a unique position to investigate genetic diagnoses in cases of fatal TAD. Genetic diagnosis in the decedent can lead to predictive genetic testing in other family members, which may inform them of their risk status, prompt relevant screening, and improve clinical outcomes for these families (16). Furthermore, data shows that hypertension is a major risk factor for aortic dissection and autopsy studies may reveal how pathogenic variation in aortic disease genes may operate in conjunction with environmental risk factors to lead to aortic events, something that can help inform clinical care and management for the wider patient base (3, 4, 17).

Here we aimed to evaluate: 1) ME/Cs' genetic testing and counseling practices for cases of TAD diagnosed at the time of autopsy, 2) identify barriers to providing genetic services in the ME/C setting, and 3) compare ME/Cs' reported practices to the NAME's recommendations for postmortem genetic testing. Aims were assessed by administering online surveys to ME/Cs though the NAME and the International Association of Coroners and Medical Examiners (IACME) membership listservs. Using three clinical vignettes, we report how often and in what clinical scenarios ME/Cs collect samples and/or order post-mortem genetic testing for TAD cases, describe the clinical features contributing to their decision to pursue a genetics evaluation, and identify whether reported practices are in accordance with established NAME guidelines.

Methods

Study respondents were members of NAME and/or IACME. The study was approved by the University of Texas Health Science Center at Houston Institutional Review Board (HSC-MS-01-251). The survey was distributed to an estimated 1,337 and 1,100 professionals through the NAME and IACME e-mail list-servs, respectively. Respondents were asked to complete the survey between October 2019 and February 2020. English-speaking ME/Cs who actively perform autopsies were eligible to participate in the study. Progression past the first question of the survey served as an acknowledgement of informed consent.

The survey, developed using Qualtrics XM software, consisted of 59 questions and was estimated to take 15 minutes to complete. The complete survey is available in Appendix C. Questions were designed to collect demographic information and evaluate three primary topics: 1) ME/Cs' recognition of the genetic contribution to TAD using three vignettes with different clinical presentations, 2) access to clinical genetic services and perceived barriers to genetic testing, and 3) ME/Cs' current practices regarding biospecimen collection and storage, and their interactions with family members. The first topic was assessed by employing a visual analog scale (VAS) to capture how likely the respondent was to perform a follow-up action for each of the three clinical vignettes (questions 1-7) and how suspicious they were for a genetic diagnosis (question 8) in the context of a fatal TAD (Table 1). Possible VAS scores ranged from 0 to 100, with 0 reflecting that the respondent would "never" perform the given task or had "no suspicion" for a genetic diagnosis and 100 reflecting that the respondent would "always" perform the specified action or was certain of a "definite genetic cause." Complete descriptions of the vignettes can be found in Table 1. Vignette 1 described a 30-year-old male decedent with syndromic features characteristic of Marfan syndrome (18); Vignette 2 described a 72-year-old male decedent with no syndromic physical features; and Vignette 3 described a non-syndromic 27-year-old male decedent with TAD. All respondents were asked questions regarding the second survey topic. Questions pertaining to the third topic were not asked to all respondents. The extent of the

questions asked to respondents was based on their responses to two questions, namely, if they collect samples for genetic testing and if they order genetic testing.

Table 1. Clinical Vignettes and accompanying Visual Analog Scale Questions

Vignette 1: A 30-year-old man dies suddenly at home. On post -mortem exam, he is 6' 8", thin, has a mild pectus carinatum and long, thin fingers. His medical record notes scoliosis. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

Vignette 2: A 72-year-old man complains of chest pain and dies suddenly in a restaurant. On post-mortem exam, he is 5' 9" with average body habitus. His medical record indicates her was healthy except mild hypertension treated with one drug. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

Vignette 3: A 27-year-old man collapses and dies suddenly after a run. On post-mortem exam, he is 5' 9", average body habitus, and the family reports he was healthy and had no medical problems. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

Visual analog scale prompt

On a scale from "I would always perform that task" to "I would never perform that task," how likely would you be to carry out the following tasks for this case?

Question 1) I or someone in my office would collect a biological sample from the deceased person that could be used for genetic testing.

Question 2) I or someone in my office would attempt to collect additional family history from relatives.

Question 3) I or someone in my office would provide information to the deceased person's relatives about an increased risk for thoracic aortic aneurysms and dissections.

Question 4) I or someone in my office would recommend thoracic aortic imaging for first degree relatives of the deceased person.

Question 5) I or someone in my office would order genetic testing for the deceased person.

Question 6) I or someone in my office would refer the deceased person's relatives for cardiology evaluation.

Question 7) I or someone in my office would refer the deceased person's relatives for genetic counseling.

Question 8) Based on the information provided, how suspicious are you that there is a genetic cause for Vignette #?

Legend: Respondents were asked to read three clinical vignettes and respond to questions using a visual analog scale prompt. Respondents used a sliding scale from 0-100 with demarcations for 0% (Never), 25%, 50%, 75%, and 100% (Always) to indicate how likely they would be to perform the action listed (questions 1-7) or how suspicious they were for a genetic cause (question 8) for each vignette.

Survey responses were collected into Microsoft Excel and statistical analyses were performed using STATA v.13.1. Analyses were performed using an α level of 0.05. Data were not normally distributed; therefore, non-parametric tests were used and medians and inter-quartile ranges (IQR) are reported. Wilcoxon signed-rank test was used to evaluate the difference in responses for each individual respondent between vignettes. Mann-Whitney U test was used to assess differences in responses between respondents who do and do not order genetic testing and between those with and without access to a genetic counselor. Kruskal-Wallis test with post-hoc Dunn test was used to assess differences in participant responses based on the number of cases of thoracic aortic dissections seen in 1 year and 5 years. Fischer's exact test was used to assess differences between perceived barriers in respondents with and without access to a genetic counselor and between those who do and do not order genetic testing.

Results

Characteristics of survey respondents

Between October 16, 2019 and February 5, 2020, 97 survey responses were collected. After excluding 28 responses due to unknown or lack of involvement in autopsies, 69 remained. A total of 60 respondents were included in the analysis for this study after excluding 9 additional responses due to lack of progression past the demographic portion of the survey (Figure 1). However, the total number of responses for a particular question varied due to survey completeness and skip logic.

Demographic information is summarized in Table 2. The majority of ME/C respondents practice in the United States (US) (90%, n=54/60), although responses were also collected from Australia (n=3/60), New Zealand (n=1/60), Belgium (n=1/60) and Canada (n=1/60). Responses were collected from ME/Cs practicing in 24 US states, with the majority of representation from California, Washington state, Ohio, and New York (Appendix A: Table A1). Eighty-five percent (n=51/60) of respondents identified as NAME members and most worked at a state- or county-owned facility (75%, n=42/60). A doctor of medicine or doctor of osteopathic medicine was the highest degree held by 85% (n=51/60). Fifty-five percent (n=33/60) identified as a medical examiner or forensic pathologist. Respondents reported seeing a median of 2 TAD cases in the past year (range: 0-25; IQR: 3) and a median of 8 cases in the past 5 years (range: 0-100; IQR: 10).

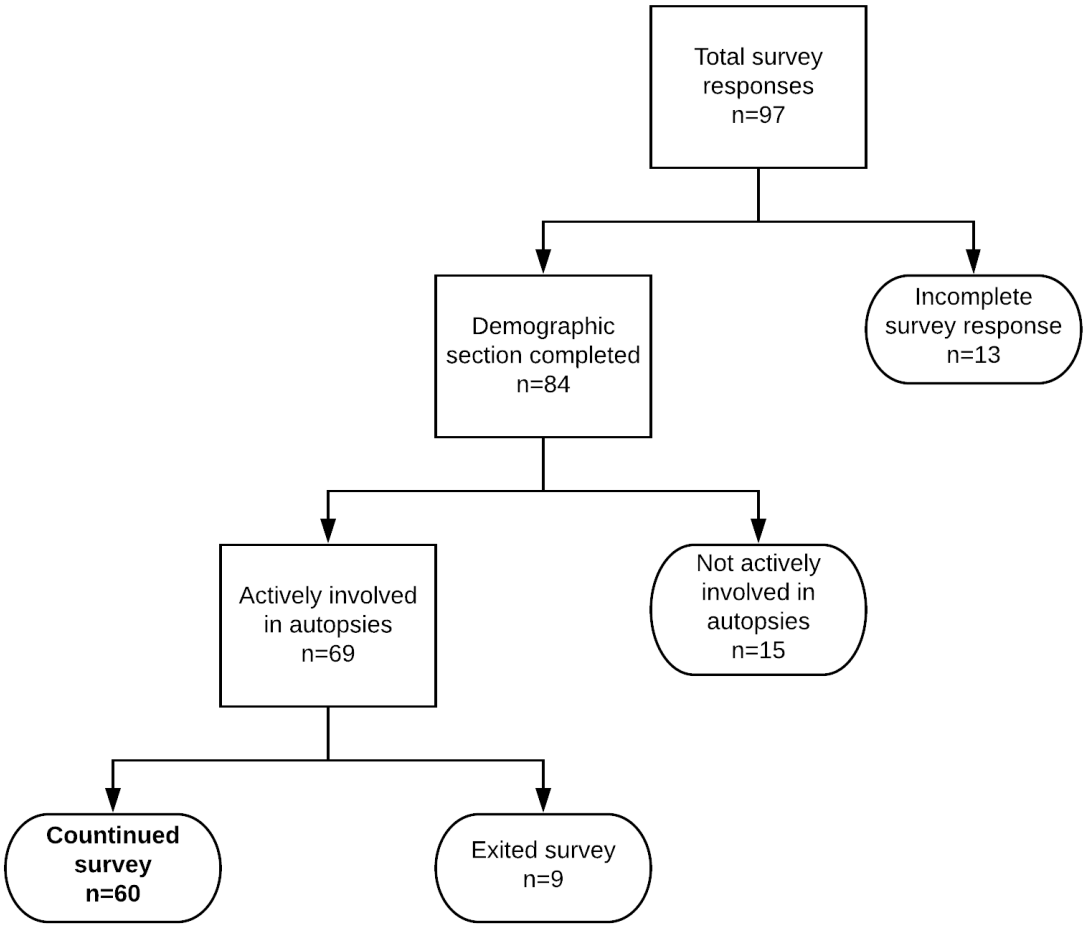


Figure 1. Survey Responses used in analysis. 97 total responses were collected, but after exclusions due to respondents not confirming that they actively involved in autopsies or proceeding past demographic questions, 60 responses were analyzed in this study.

Table 2. Demographics of Survey Respondents

Responses		% (n)
Country		
	United States	90.00 (54)
	Non-US	10.00 (6)
Organization		
	NAME	75.00 (45)
	IACME	13.33 (8)
	Both	10.00 (6)
	Neither	1.67 (1)
Facility		
	State or County Owned	70.00 (42)
	Academic Center	10.00 (6)
	Academic/State	5.00 (3)
	Private	8.33 (5)
	Other	6.67 (4)
Highest degree		
	MD/DO and PhD	3.33 (2)
	MD/DO	81.67 (49)
	DDS	1.67 (1)
	MBBS	3.33 (2)
	MS	1.67 (1)
	BA/BS	5.00 (3)
	AAS	1.67 (1)
	Other	1.67 (1)
Job Title(s)		
	Medical Examiner	50.00 (30)
	Coroner	18.33 (11)
	Forensic Pathologist	60.00 (36)
	Other	5.00 (3)
		n (median, min-max, IQR)
Number of thoracic aortic dissection seen on autopsy		
	Cases seen last year	59 (2, 0-25, 3)
	Cases seen over past 5 years	59 (8, 0-100, 10)

Legend: Demographic information reported by respondents (n=60). Highest degree was chosen from all degrees attained. Respondents were able to select one or more relevant job titles, with the use of each title reported in aggregate. Abbreviations: US United States; NAME National Association of Medical Examiners; IACME International Association of Coroners and Medical Examiners; MD Doctor of Medicine; DO Doctor of Osteopathic Medicine; PhD Doctor of Philosophy; DDS Doctor of Dental Surgery; MBBS Bachelor of Medicine; Bachelor of Surgery; MS Master of Science; BA Bachelor of Arts; BS Bachelor of Science; AAS Associate of Applied Science; Min minimum; Max maximum; IQR inter-quartile range

Postmortem genetic testing protocol for fatal thoracic aortic dissection

Fifty percent (n=27/54) of ME/Cs who collect samples for genetic testing reported that they order postmortem genetic testing if TAD is diagnosed on autopsy and 51% (n=29/56) collect a sample for genetic testing regardless of the decedent's age and sex. Others acknowledged age-specific cut-offs they use to inform their decision to collect a specimen for genetic testing. Notably, 5.3% (n=3/56) of respondents only collect a sample from decedents below age 30, which is not consistent with NAME's guidelines to collect from anyone age 40 and below. Additionally, 12% (n=7/56) never collect a sample or did not know at what age a sample is collected (Table 3).

Most ME/Cs who order genetic testing disclose all types of test results issued by the laboratory to the decedent's next-of-kin (NOK) (76%, n=20/26): positive results consistent with the identification of a pathogenic variant, negative test results, and variants of uncertain significance (VUS), which describe genetic variants with insufficient evidence to determine pathogenicity. Test result disclosure protocols differed by result type for other respondents as some report only positive results (7.7%, n=2/26), positive and VUS results (7.7%, n=2/26), or positive and negative results (3.8%, n=1/26). Only one respondent who orders genetic testing does not report genetic testing results to NOK (3.8%). Moreover only 39% (n=22/56) reported having access to a genetic counselor.

Table 3. Genetic Service Capabilities and Practices

Responses	% (n)
Access to genetic counselor (n=56)	
Yes, I have access to a genetic counselor	39.28 (22)
No, I do not have access to a genetic counselor	60.71 (34)
Sample collection for male decedent (n=56)	
Below 30 years	5.36 (3)
Below 40 years	17.86 (10)
Below 50 years	10.71 (6)
Below 60 years	1.79 (1)
I collect samples regardless of age	51.79 (29)
I do not collect samples for males for genetic testing	3.57 (2)
I do not know	8.93 (5)
Sample collection for female decedent (n=56)	
Below 30 years	5.36 (3)
Below 40 years	17.86 (10)
Below 50 years	8.93 (5)
Below 60 years	3.57 (2)
I collect samples regardless of age	51.79 (29)
I do not collect samples for females for genetic testing	3.57 (2)
I do not know	8.93 (5)
Ordering genetic testing for TAD (n=54)	
Yes, I order genetic testing	50.00 (27)
No, I do not order genetic testing	50.00 (27)
Type of result disclosed (n=26)	
Positive, Negative, and VUS	76.9 (20)
Positive and VUS	7.7 (2)
Positive and Negative	3.8 (1)
Positive only	7.7 (2)
Does not disclose results	3.8 (1)

Legend: Genetic counselor access, sample collection, genetic testing ordering, and genetics results disclosure practices reported by respondents. Abbreviations: TAD thoracic aortic dissection; VUS variant of uncertain significance

Clinical Vignettes

Using a sliding VAS for three clinical vignettes, respondents were asked how likely they were to perform a given action, from never to always. Individuals' scores, representing the percent chance they would perform a certain action, were compared between vignettes. Median VAS scores are summarized for each clinical vignette (Table 4), with statistical analyses reported in Appendix A Table A2. For the two vignettes with a young decedent, the 30 year-old TAD case with syndromic features (vignette 1) and the 27-year-old non-syndromic TAD case (vignette 3), ME/Cs were significantly more likely to perform all proposed actions (questions 1-7) compared to the 72-year-old non-syndromic TAD case (vignette 2) ($p<0.05$). Specifically, the majority of respondents report that they would *always* collect a biological sample for genetic testing for vignette 1 (73% n=42/57) and vignette 3 (78.18%; n=43/55) compared to only 56% for vignette 2 (n=32/57). Further, ME/Cs' behaviors were similar for both the early-onset syndromic case (vignette 1) and the early onset non-syndromic case (vignette 3) with regard to how likely they were to order genetic testing. However, respondents were more likely to provide information to relatives about increased risk for TAAD (question 3) and refer relatives for genetic counseling (question 7) in the early-onset syndromic case (vignette 1) than the early-onset non-syndromic case (vignette 3) ($p = 0.0006$, $p=0.0285$, respectively). ME/C's were asked how suspicious they were for a genetic cause of TAD for each clinical vignette (question 8). Respondents were significantly more suspicious of a genetic cause in the 30-year-old case with pectus carinatum, long fingers and other systemic features suggestive of Marfan syndrome (vignette 1) compared to the 72-year-old non-syndromic case (vignette 2) and the 27-year-old non-syndromic case (vignette 3) ($p<0.0001$, $p=0.0001$, respectively). However, respondents were still more suspicious for a genetic cause in the early-onset non-syndromic case (vignette 3) compared to the later-onset non-syndromic case (vignette 2) ($p<0.0001$).

We elicited the number of TAD cases respondents reported in their office within the last year or 5 years to evaluate the effect of ME/Cs' experience performing autopsies on TAD cases. The number of cases seen by ME/Cs was not significantly associated with how likely they were to

perform certain actions (questions 1-7) nor how suspicious they were for a genetic cause (question 8) across vignettes (Appendix A: Table A2). Similarly, responses were not significantly different based on whether or not the respondent orders genetic testing in practice, except in question 5, where respondents who order genetic testing in practice were significantly more likely to order genetic testing in the early-onset syndromic case (vignette 1) and the early-onset non-syndromic case (vignette 3).

Access to a genetic counselor impacted actions taken by ME/Cs for the early-onset syndromic case (vignette 1) and the early-onset non-syndromic case (vignette 3). In the early-onset syndromic case (vignette 1), those who had access to a genetic counselor were significantly more likely to collect a sample (question 1; p=0.0125), collect family history from relatives (question 2; p=0.0362), and provide information to relatives about an increased risk for TAAD (question 3; p=0.0401). This was different from the early-onset non-syndromic case (vignette 3), where those who had access to a genetic counselor were significantly more likely to order genetic testing (question 5; p=0.045), refer relatives for a cardiology evaluation (question 6; p=0.0134), or refer relatives for genetic counseling (question 7; p=0.0067). However, genetic counselor access did not impact ME/Cs' behaviors regarding their actions or level of genetic suspicion for the 72-year-old TAD case (Appendix A: Table A2).

Table 4. Summary of Clinical Vignette Visual Analog Scale Responses

	Vignette 1: Early-onset, Syndromic (Marfan)		Vignette 2: Later-onset, Non-syndromic		Vignette 3: Early-onset, Non-syndromic	
Question	n	Median (min-max; IQR)	n	Median (min-max; IQR)	n	Median (min-max; IQR)
1. Collect a biological sample for genetic testing	57	100 (0-100; 1)	57	100 (0-100; 88)	55	100 (0-100; 0)
2. Collect additional family history from relatives	60	100 (0-100; 24.5)	59	79 (0-100; 90)	56	100 (0-100; 17)
3. Provide information to relatives about increased risk for TAAD	60	100 (0-100; 19)	56	20 (0-100; 92.5)	53	100 (0-100; 50)
4. Recommend thoracic aortic imaging for first degree relatives	57	0 (0-100; 76)	55	0 (0-100; 1)	52	0 (0-100; 66)
5. Order genetic testing for the deceased person	57	5 (0-100; 72)	55	0 (0-100; 1)	54	2.5 (0-100; 96)
6. Refer relatives for cardiology evaluation	56	90 (0-100; 96.5)	55	0 (0-100; 26)	52	61 (0-100; 100)
7. Refer relatives for genetic counseling	56	75 (0-100; 100)	54	0 (0-100; 9)	52	45 (0-100; 100)
8. Suspicion for a genetic cause	60	97 (30-100; 15.5)	54	17.5 (0-100; 25)	55	80 (40-100; 35)

Legend: Median visual analog scale (VAS) scores for the eight questions posed in each vignette. VAS scores indicate the likelihood that an individual would do the action in question or the level of suspicion for a genetic cause with 0 being "never" or "no suspicion" and 100 being "always" or "definite genetic cause". Abbreviations: Min minimum; Max maximum; IQR inter-quartile range; TAAD thoracic aortic aneurysm or dissection

Barriers to ordering genetic testing

Respondents identified several barriers to ordering postmortem genetic testing for TAD cases (Table 5). Cost was the most frequently reported barrier with 42 of 55 (76%) respondents selecting it as a barrier and 36 of 51 individuals (70.6%) ranking it as the number one barrier. The next most common barrier selected was difficulty in contacting NOK of the decedent (n=11/55). Sixteen percent of respondents (n=9/55) reported that there were no barriers to genetic testing; these individuals were significantly more likely to have access to a genetic counselor (p=0.021). Collectively, issues surrounding genetic testing information were listed as barriers by 50.9% (n=28/55), specifically, difficulty finding an appropriate lab, not knowing what test to order, not knowing how to order genetic testing, and lack of information to provide to NOK.

Table 5. Barriers to Ordering Genetic Testing for a Deceased Person

Barriers	% (n)
Financial	
Cost of genetic testing	76.36 (42)
Inability to obtain insurance coverage	16.36 (9)
Testing	
Inability to collect samples for genetic testing	5.45 (3)
Difficulty finding an appropriate lab	16.36 (9)
Do not know what test to order	14.54 (8)
Do not know how to order genetic testing	12.72 (7)
Next-of-kin	
Difficulty in contacting next-of-kin of the deceased person	20.00 (11)
Inability to get informed consent from next-of-kin	16.36 (9)
Lack of information to provide next-of-kin	7.27 (4)
Staff	
Insufficient staffing	7.27 (4)
Lack of time	7.27 (4)
Lack of geneticists and/or genetic counselors in your area	10.90 (6)
Other	14.54 (8)
I do not see any barriers to genetic testing for a deceased person	16.36 (9)

Legend: Barriers to genetic testing for a deceased reported by respondents (n=55). Respondents could select one or more barriers as applicable.

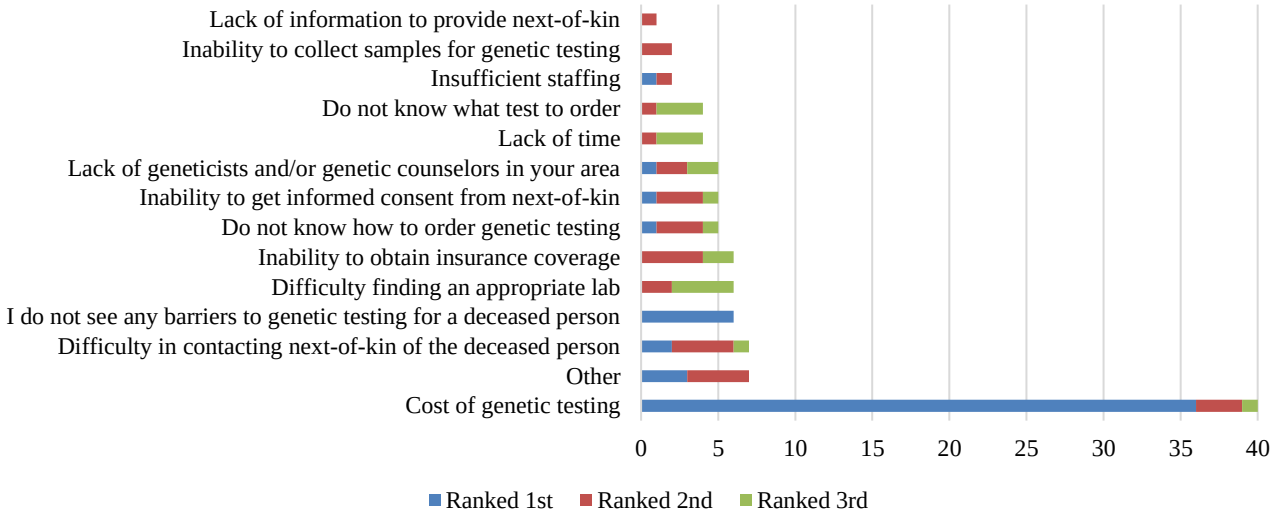


Figure 2. ME/Cs rankings of their top three barriers to genetic testing for a deceased person ordered by total combined frequency of selection (n=51)

Alignment of practices with NAME guidelines

Respondents' reported practices regarding sample collection, handling, and interactions with NOK in reference to genetic testing are summarized in Table 6. Sample collection protocols were largely consistent with guidelines established by NAME, as the majority of respondents collect whole blood (90.7%, n=49/54) in a purple top tube (K2 EDTA) (95.9%, n=47/49) for all individuals under the age of 40 years (82.1%, n=46/56). Regarding biospecimen storage and shipping protocols, respondents' current practices were more varied in their alignment with NAME recommendations, ranging from 36-53%. Finally, respondents' practices were least consistent with guidelines when it came to obtaining informed consent from families (38%, n=10/26), checking with families about religious and cultural objections (29%, n=16/54) and informing families of DNA banking options (24%, n=13/53).

Table 6. Alignment of Reported Practices for Genetic Services with NAME Guidelines

Recommendation	% (n)
Sample collection	
Sample collection age (<40 years) (n=56)	82.14 (46)
Sample type (whole blood) (n=54)	90.7 (49)
Blood tube (Purple top [K2 EDTA]) (n=49)	95.9 (47)
Sample handling	
Sample stored <1 month (4°C) (n=46)	41.3 (19)
Sample stored >1 month (-20°C to -70°C) (n=49)	53.06 (26)
Fresh sample shipping (overnight at room temperature) (n=26)	46.15 (12)
Frozen sample shipping (overnight on dry ice) (n=25)	36.00 (9)
Interactions with next-of-kin	
Obtaining informed consent (n=26)	38.46 (10)
Informing families of DNA banking options (n=53)	24.53 (13)
Checking with families about religious and cultural objections (n=54)	29.63 (16)

Legend: Percent of respondents whose survey answers were compliant with NAME guidelines for the given category. The NAME guidelines regarding sample collection and sample handling are noted in parentheses. Abbreviations: NAME National Association of Medical Examiners; EDTA ethylenediaminetetraacetic acid

Discussion

With high mortality rates and many individuals failing to reach the hospital in cases of TAD, ME/Cs are in a unique position to recognize cases at high risk for having a heritable form of disease and play a role in providing genetic counseling and testing, either directly or through referral (1-4). In the context of fatal TAD diagnosed by autopsy, a positive genetic test result will determine the cause of dissection and enable the identification of relatives at-risk for dissection through subsequent cascade genetic testing. The clinical utility of genetic testing for TAD has expanded extensively over the past decade as more aortic disease genes have been identified and technological advances enabled more accessible testing by reducing cost. Indications for genetic testing now extend beyond cases of suspected Marfan or Loeys-Dietz syndromes as 11 genes have been established to be strongly associated with TAAD with or without syndromic features (10). Here we sought to investigate the clinical factors that may currently contribute to a ME/C's suspicion for hereditary TAD and describe their practices with regard to postmortem genetic testing and counseling.

Several clinical determinants should raise suspicion for a genetic component to TAD; namely, an early age at diagnosis (< 60 years), a family history of TAAD or sudden death, and/or the presence of physical features (deemed "syndromic") associated with Marfan syndrome or, Loeys-Dietz syndrome. In this study, we did not ask respondents to propose a diagnosis for the decedent in any of the vignettes; however, ME/Cs appeared to recognize the syndromic features described in the vignette 1, which included pectus carinatum, arachnodactyly, and tall stature. Our intent was for these features to be suggestive of Marfan syndrome; however they also overlap with features of Loeys-Dietz syndrome so ME/Cs may have appropriately included this in their differential diagnosis. Regardless, respondents were significantly more suspicious of a genetic cause and more likely to refer the family for genetic counseling for this early-onset syndromic case (vignette 1) than they were for either the later-onset (vignette 2) or the early-onset (vignette 3) non-syndromic cases. In addition, age appeared to be a driving factor for some actions taken by ME/Cs. Across all vignette questions examining collection, referral, ordering, and information-giving practices, respondents were more likely to

perform these actions for the early-onset decedents both with (vignette 1) and without (vignette 3) syndromic features, compared to the later-onset non-syndromic decedent (vignette 2). This suggests that respondents recognize that both early age of diagnosis and syndromic physical characteristics are concerning for an underlying genetic etiology and should prompt genetic counseling and testing.

Highly penetrant pathogenic variants in eleven genes are confirmed to confer a risk for TAD; however, absence of a pathogenic variant in one of these does not eliminate genetic risk. While environmental factors, such as illicit drug use or hypertension, are associated with TAD and could be at play in a case with negative genetic testing, they do not preclude a genetic cause (3, 4, 19). Additionally, it is unlikely that all the genes predisposing to heritable TAD have been identified. For these reasons, the AHA/ACC guidelines for the diagnosis and management of TAAD patients recommend aortic imaging for first-degree relatives of all cases of TAAD regardless of age (15). Similar to cascade genetic testing, imaging of at-risk relatives promotes the identification of asymptomatic aortic aneurysms at an early stage, which can be repaired in a timely manner to prevent an acute dissection. However, for maximum efficacy in the postmortem setting, this requires collection of family history and communication with NOK by the ME/C, or referral to an appropriate healthcare provider. Involvement of ME/Cs in genetic testing and risk communication to relatives of the decedent has previously been explored, but has primarily centered on so-called "autopsy negative" conditions like cardiac arrhythmias (20-24). In our study, ME/Cs were more likely to perform these tasks in the early-set TAD cases than in the later-onset case. Regardless of the case at hand, median VAS scores indicate that ME/Cs would collect family history and provide information to NOK about an increased risk for TAAD, but the final step of referring NOK for a cardiology evaluation or recommending thoracic aortic imaging is less common. While the reason for this discrepancy was not explored, an issue complicating referral may be that communication with NOK to provide this complete care is not always easy. The second most commonly identified barrier was difficulty in contacting NOK. While this barrier was only listed by 20% of respondents, it represents a real issue in the delivery of results and obtaining follow-up care for NOK. Uptake of cascade

genetic testing already presents a challenge with only a 7-30% uptake in other studies (23, 24). Thus, having the ability to make direct contact, employ online educational tools, or use other means of communication with families is crucial to improve uptake of cascade genetic testing.

The importance of involving a multidisciplinary team that includes genetic counselors in postmortem genetic evaluations has been established and was further supported by ME/C responses in our study (25-27). Significant differences in participant responses were observed between those who had access to a genetic counselor and those who did not, indicating that genetic counselor access may impact actions and recommendations made by ME/Cs. For example, respondents who had access to a genetic counselor were more likely to order genetic testing (question 5), refer to cardiology (question 6), and refer to genetic counseling (question 7) for the early-onset non-syndromic decedent (vignette 3). While our study was not designed to evaluate knowledge regarding hereditary TAD, it may be that ME/Cs who have access to a genetic counselor are better informed regarding the features that should prompt genetic testing and how to execute the genetic testing process. Data from the cardiology setting indicate that genetic counselors aid in appropriate test selection, identify appropriate testing candidates, and provide appropriate follow-up care including genetic test result disclosure (27). In our study ME/Cs level of comfort with delivering genetic test results was not directly assessed, but with 76% of ME/Cs who order genetic testing disclosing all result types (positive, negative, variant of uncertain significance) to NOK (n=20/26) it is important that they have access to genetics professionals when needed. The benefits of this is further supported as respondents in our study who reported experiencing no barriers to genetic testing were significantly more likely to have access to a genetic counselor. Currently, genetic counselors are accessible remotely with recent studies finding that up to 68% of genetic counselors have participated in telemedicine, both as part of hospital systems as well as through companies specifically designed to provide genetic telehealth services, demonstrating that genetic counselors do not need to be a direct part of an ME/C's home institution to assist both the provider and the NOK (28).

Historically, genetic testing cost and/or insufficient insurance coverage has been a pervasive barrier to providing genetic services, even when medical guidelines support its utility. However, over the last five years, the industry has seen a dramatic reduction in cost, enabling increased access to genetic testing for individuals in all settings, including postmortem (29, 30). Yet, our study demonstrates that cost of genetic testing remains a major concern, as it was the most frequently identified barrier by 76% of respondents and top-ranked by 70%. This is higher than a previous study of medical examiners in Europe, which reported that 30.9% believed the cost of genetic testing was too high (20). It is worth noting that studies predating 2014 that investigated insurance coverage in the US for postmortem genetic testing reported very limited coverage; however, more recent data demonstrate coverage has improved (14, 31, 32). Therefore, the perceived cost barrier reported by our respondents may be both influenced by a lack of updates on recent improvements in payor coverage for postmortem testing and the healthcare payor system in the U.S., where the majority of our respondents practice (n=54/60). Moreover, ME/Cs may not be aware of more recent alternative payment options for genetic testing through many commercial laboratories, which provide lower-cost testing for those without insurance (D. Angeles, personal communication, March 2, 2020). Additional barriers to genetic testing were noted but appeared to be less impactful. Encouragingly, the ability to focus on one or two meaningful barriers will allow for targeted studies in the future and focused implementation of solutions.

Professional medical society guidelines in forensics and cardiology provide an important foundation and resource for practitioners to provide care for individuals with families with TAAD. In 2013, NAME published recommendations regarding the storage and collection of biospecimens for postmortem genetic testing, which were used to shape questions in this study (14). Overall, respondents demonstrated a good understanding of the appropriate age for genetic testing and best sample type to collect for that purpose, with 51% reporting that they would collect a sample for anyone with TAD regardless of age, and 90% appropriately collecting whole blood. However, there was a wider range of practices regarding the handling of samples after collection, such as the proper

temperatures to ship and/or store samples. While the NAME guidelines established general recommendations for postmortem specimen collection and handling, individual laboratories may have specific recommendations and should be considered. Lastly, respondents reported that they rarely contact NOK to obtain consent or discuss DNA banking options. Obtaining consent is not required by law in public autopsies, which may impact the likelihood of ME/Cs seeking informed consent from NOK (33). Furthermore, these reduced frequencies of contacting NOK about consent or DNA banking may not represent a lack of effort, as the second most common barrier reported was difficulty in contacting NOK. Overcoming these challenges will be crucial to ensuring that families are informed about their increased dissection risk and receive appropriate medical intervention.

This survey garnered a relatively low response rate, which did not enable us to model the data and determine the factors influencing ME/Cs decisions to engage in the proposed actions in the clinical vignettes. However, because the study topic was specific to fatal cases of TAD and dispersed to a busy provider cohort of ME/Cs, 60 useable response was adequate to establish trends with baseline data. It is possible that a low volume of TAD cases presenting to an ME/C's office dissuaded some from responding. More members of NAME responded than IACME, which likely led to lower responses from ME/Cs outside the US; therefore, findings cannot be generalized to reflect ME/Cs' practices globally. Additionally, using VAS scores allowed for more flexibility in participants' responses; however, respondents were not permitted to return to previous vignettes to modify their answers. Therefore, if an individual answered "100" for a particular question, meaning they always perform a particular task, their answer for the same question in subsequent vignettes could only stay the same or decrease, despite if a respondent determined they were more likely to do that task in a future vignette. This limitation may result in some discrepancies when analyzing the difference in responses for an individual between vignettes. Finally, responses were obtained by self-report, were not confirmed by medical record review, and therefore, subject to recall bias.

With thousands of deaths from acute aortic dissection every year, ME/Cs will continue to be among the first line of practitioners to evaluate these decedents and interact with their families.

Through a series of clinical vignettes, we demonstrate that ME/Cs appear to recognize the need for a genetic testing and evaluation in young patients with TAD. The likelihood of ordering genetic testing or providing further information and referrals to family members was impacted not by clinical experience, but by access to a genetic counselor, hinting at the importance of establishing multidisciplinary teams to provide care. Additional barriers to genetic testing exist, most notably cost and communication with NOK. Guidelines established by NAME provide a foundation for establishing appropriate postmortem testing protocols for TAD, and while many ME/Cs' practices align with sample collection and storage guidelines, there remains a gap regarding communication with NOK to provide informed consent and options for genetic testing. Moving forward, this study provides a basis for the investigation of clinical strategies to improve communication with NOK, to establish resources to assist with ME/C access to genetic services, and to increase ME/Cs' ability to take the necessary actions to diagnose the TAD decedent and aid their family. As syndromic features are not always present and family history information is not always available it would be reasonable to collect samples for genetic testing or DNA banking in all TAD cases given the established genetic component and the implications of identifying it. In addition, all first-degree relatives should be referred to genetic counseling and cardiology for appropriate screening and testing. Finally, based on the initial barriers identified in our study, future research could further examine those barriers with the aim of establishing programs or resources to aid ME/Cs in identifying affordable genetic testing and providing NOK with the appropriate care and referrals.

Appendix A: Additional Data

Table A1. State in which ME/C currently works

State	n (%)
Arizona	2 (3.7)
Arkansas	1 (1.85)
California	6 (11.11)
Colorado	2 (3.7)
Connecticut	3 (5.56)
Florida	3 (5.56)
Hawaii	1 (1.85)
Idaho	2 (3.7)
Illinois	2 (3.7)
Iowa	1 (1.85)
Kentucky	1 (1.85)
Maine	1 (1.85)
Michigan	3 (5.56)
Missouri	2 (3.7)
Nevada	1 (1.85)
New Jersey	1 (1.85)
New York	4 (7.41)
Ohio	5 (9.26)
Pennsylvania	3 (5.56)
Texas	2 (3.7)
Vermont	1 (1.85)
Washington	5 (9.26)
Wisconsin	1 (1.85)
Wyoming	1 (1.85)

Legend: States reported by respondents as their current employment location (n=60)

Table A2. P-values from several statistical tests comparing visual analog scores amongst vignettes or between distinct groups of respondents

Test	p-value							
	Question 1	Question 2	Question 3	Question 4	Question 5	Question 6	Question 7	Question 8
Sign-rank								
Vignette 1 vs vignette 2	0.0005*	0.0001*	0*	0.0024*	0*	0*	0*	0*
Vignette 2 vs vignette 3	0*	0*	0*	0.0004*	0*	0*	0*	0*
Vignette 1 vs vignette 3	0.2169	0.207	0.0006*	0.9045	0.079	0.058	0.0285*	0.0001*
Kruskal-Wallis with Dunn test								
Vignette 1 by number of cases seen in 1 year	0.7336	0.2349	0.1449	0.1234	0.2787	0.5901	0.572	0.4933
Vignette 2 by number of cases seen in 1 year	0.9028	0.2562	0.26	0.6223	0.5687	0.3878	0.2578	0.6736
Vignette 3 by number of cases seen in 1 year	0.7306	0.1181	0.8946	0.098	0.3454	0.4563	0.7303	0.1041
Vignette 1 by number of cases seen in 5 years	0.4228	0.0799	0.4739	0.0752	0.856	0.4995	0.4755	0.9114
Vignette 2 by number of cases seen in 5 years	0.6419	0.7421	0.6707	0.9742	0.7018	0.7853	0.6133	0.6125
Vignette 3 by number of cases seen in 5 years	0.1171	0.1384	0.8409	0.1568	0.925	0.9698	0.7352	0.4739
Mann-Whitney U								
Vignette 1 by Access to a genetic counselor	0.0125*	0.0362*	0.0401*	0.606	0.0826	0.1033	0.0524	0.1621
Vignette 2 by Access to a genetic counselor	0.2485	0.0524	0.0816	0.9338	0.94	0.7995	0.981	0.7726
Vignette 3 by Access to a genetic counselor	0.0541	0.0717	0.1047	0.9164	0.045*	0.0134*	0.0067*	0.2358
Vignette 1 by Order genetic testing	0.9516	0.8711	0.1889	0.2786	0.0014*	0.7812	0.9923	0.4264
Vignette 2 by Order genetic testing	0.804	0.5784	0.6139	0.4646	0.4624	0.0500	0.1254	0.1656
Vignette 3 by Order genetic testing	0.6317	0.8748	0.6093	0.6176	0.0014*	0.8442	0.4608	0.5781

* = Significant p-value using α level of 0.05

Appendix B: Survey Invitation

You are invited to take part in a research study called, "Genetic Testing and Counseling for Fatal Cases of Acute Aortic Dissections", conducted by Bradley Power, BA of the University of Texas Health Science Center at Houston. Dianna Milewicz, MD, PhD is the principal investigator. The purpose of this study is to evaluate medical examiners' practices and perceived barriers surrounding genetic testing and counseling in cases of thoracic aortic dissection. The purpose of this survey is not to assess for correct answers; answers will only be analyzed and reported in aggregate.

If you decide to take part in the study, the total time commitment is 15-20 minutes. You are invited to take part in this study because you are a member of the National Association of Medical Examiners (NAME) and/or the International Association Coroners and Medical Examiners (IACME). Participation in this study is voluntary and you may choose to stop taking the survey at any time. Of note, two of the questions involve data retrieval that may not be possible if you take the survey outside of your place of employment.

If you agree to take part in this study, the information you provide will help determine current practices and barriers surrounding post-mortem genetic testing ordered by medical examiners and coroners. You may not receive any benefit from taking part in this study. Possible risks are limited to breach of confidentiality. The information collected in the survey will not be used to identify participants and will be kept on a secure server.

There is no cost to participate and you will not be paid to take part in this study. You will not be personally identified in any reports or publications that may result from this study. Any personal information about you that is gathered during this study will remain confidential to every extent of the law. If you have any questions about this project please contact Bradley Power or Alana Cecchi, MS, CGC at 713–500-6715.

Proceeding with the survey implies that you have read and understand your rights and that you consent to participate in the study.

Appendix C: Survey Questions

Instructions: This survey contains questions about thoracic aortic aneurysms and dissections in the form of clinical vignettes and questions related to your current work practices. If you have already completed this survey, please exit now. Thank you for your time.

2. I am a member of ...

- o National Association of Medical Examiners (NAME)
- o International Association of Coroners and Medical Examiners (IACME)
- o Both
- o Neither

3. What country do you work in?

▼ United States of America ... Zimbabwe

Skip To: Q5 If List of Countries = United States of America

4. What city(ies) do you serve?

Skip To: Q7 If Condition: What city(ies) do you serve? Is Not Empty. Skip To: What type of institution do you work ....

5. In which state do you currently work?

▼ Alabama ... I do not reside in the United States

6. What county(ies) do you serve? For those who work in Louisiana, please enter parish name(s). For those who work in Alaska or New York City, please enter borough name(s).

7. What type of institution do you work at? (Check all that apply)

- ▢ Academic center
- ▢ Private Facility
- ▢ State or county owned facility
- ▢ Other (Please specify)

8. Are you currently actively involved in performing autopsies?

- o Yes
- o No

Skip To: End of Survey If Are you currently actively involved in performing autopsies? = No

9. What degrees have you attained? (Check all that apply)

- ▢ BA/BS
- ▢ MS
- ▢ PhD
- ▢ MD/DO
- ▢ DPT
- ▢ DDS
- ▢ JD
- ▢ Other (Please specify)

10. What is your job title? (Check all that apply)

- ▢ Medical Examiner
- ▢ Coroner
- ▢ Forensic Pathologist
- ▢ Other (Please specify)

Instructions: Please answer one of the following questions with an integer. The answer box will not accept ranges.

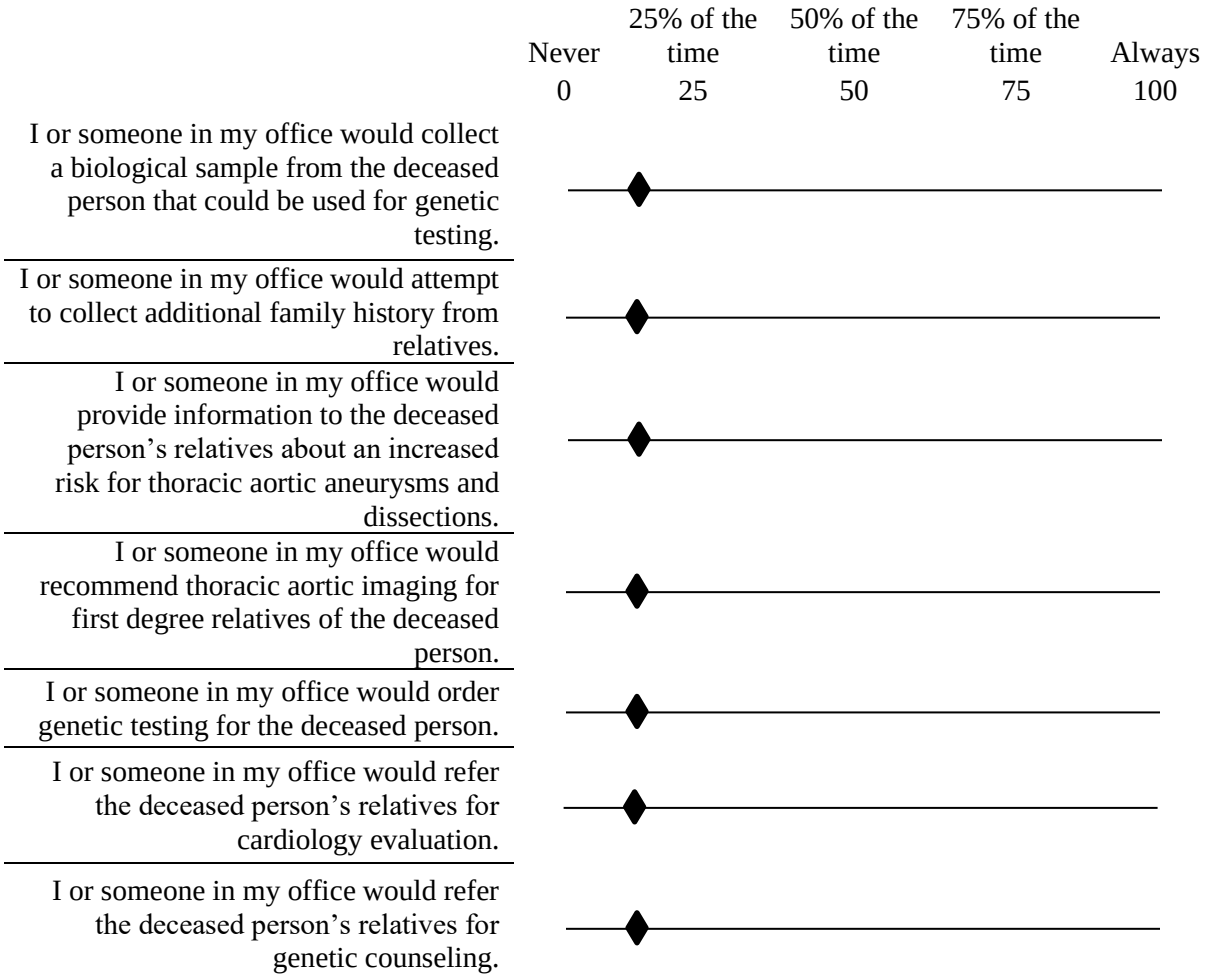
11. How many cases of thoracic aortic dissection have you seen in the past year?

12. How many cases of thoracic aortic dissection have you seen in the past 5 years?

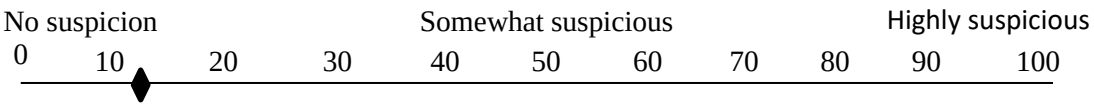
Instructions: The following are three clinical vignettes describing a potential case. Please read the vignette and respond to the following questions.

Vignette 1: A 30-year-old man dies suddenly at home. On post-mortem exam, he is 6' 8", thin, has a mild pectus carinatum and long, thin fingers. His medical record notes scoliosis. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

On a scale from "I would always perform that task" to "I would never perform that task," how likely would you be to carry out the following tasks for this case? **Please move the slider to register your response.**

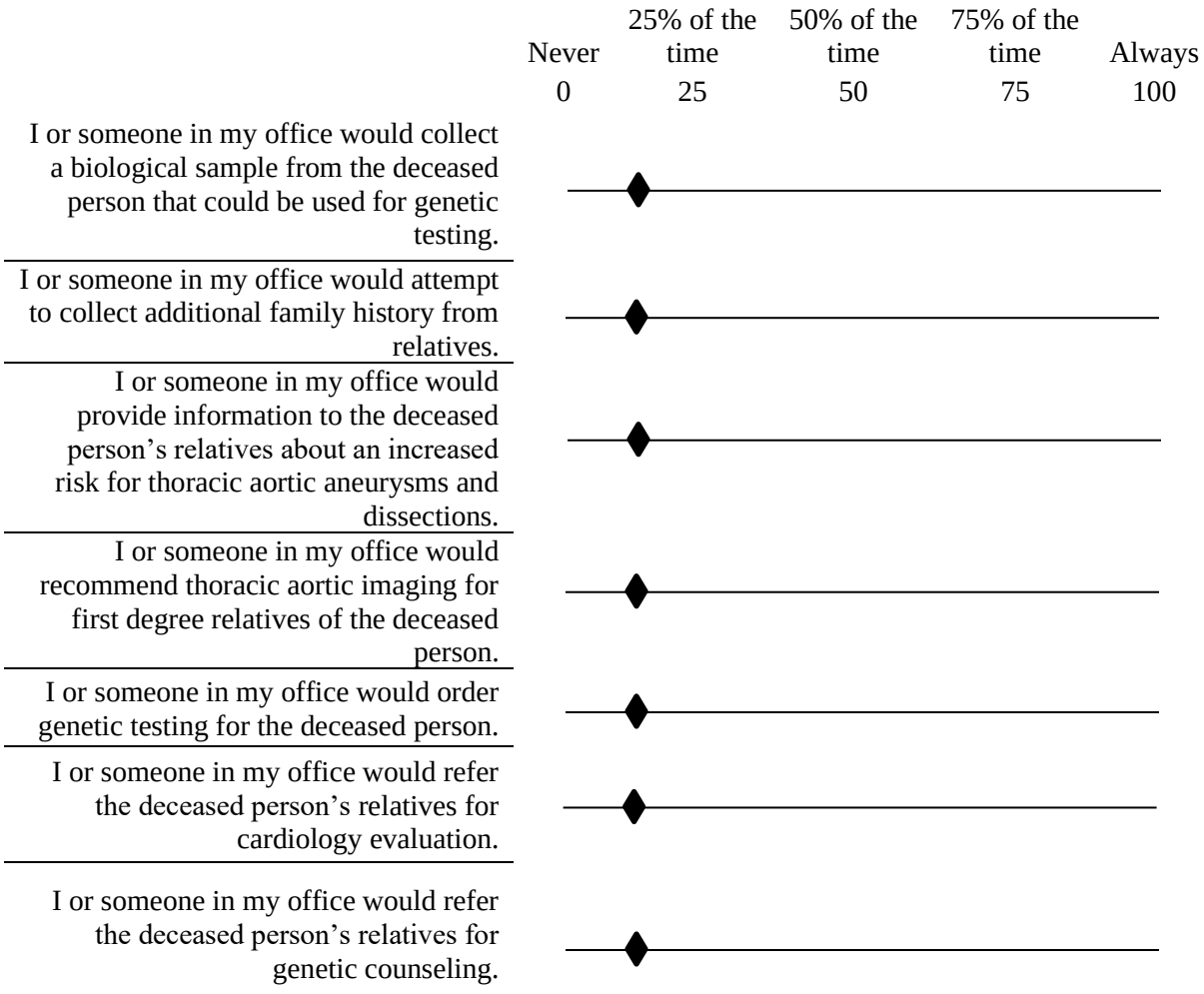


Based on the information provided, how suspicious are you that there is a genetic cause for Vignette 1?

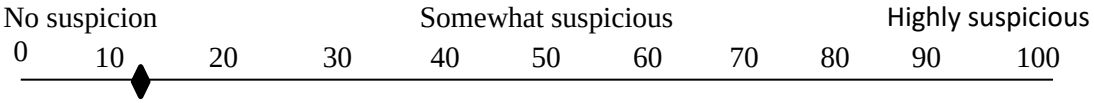


Vignette 2: A 72-year-old man complains of chest pain and dies suddenly in a restaurant. On post-mortem exam, he is 5'9" with average body habitus. His medical record indicates he was healthy except mild hypertension treated with one drug. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

On a scale from "I would always perform that task" to "I would never perform that task," how likely would you be to carry out the following tasks for this case? **Please move the slider to register your response.**

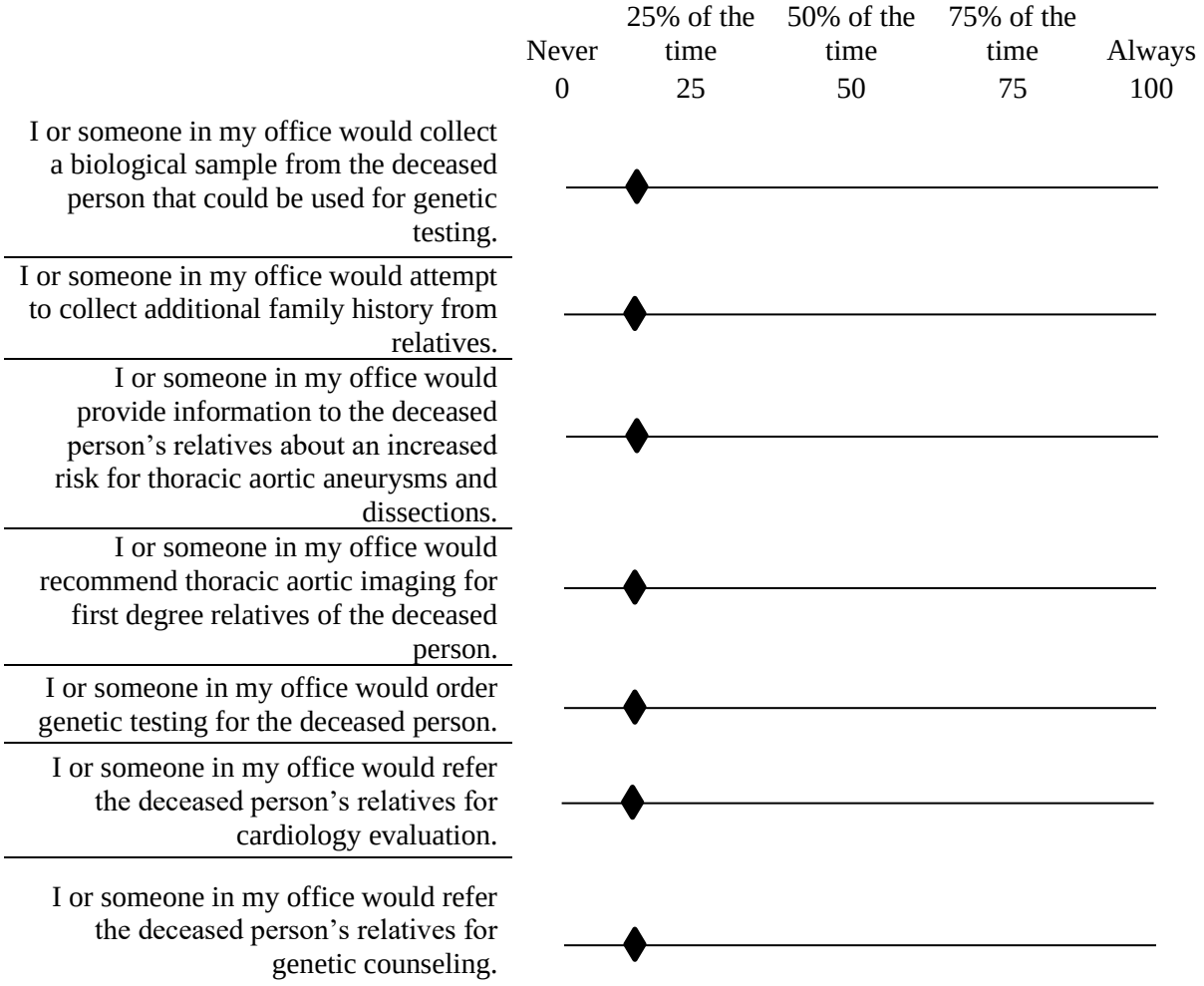


Based on the information provided, how suspicious are you that there is a genetic cause for Vignette 2?

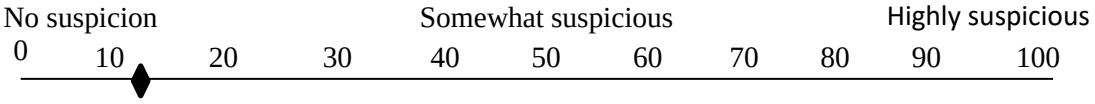


Vignette 3: A 27-year-old man collapses and dies suddenly after a run. On post-mortem exam, he is 5'9", average body habitus, and the family reports he was healthy and had no medical problems. On autopsy, he is found to have type A aortic dissection with pericardial tamponade.

On a scale from "I would always perform that task" to "I would never perform that task," how likely would you be to carry out the following tasks for this case? **Please move the slider to register your response.**



Based on the information provided, how suspicious are you that there is a genetic cause for Vignette 3?



13. Do you have access to a genetic counselor?

- o Yes, I have one I can contact and/or refer to.
- o Yes, I have one I can contact and they work at my institution.
- o No, I do not have one I can contact, but I know how to find one.
- o No, I do not have one I can contact and I do not know how to find one.

14. Are there any additional services you would provide for the deceased person or next-of-kin in the above vignettes? e.g. referral to research studies

- o Yes (Please specify)
- o No

Instructions: The following questions apply to cases of **thoracic aortic dissection** or **aneurysm**.

15. At what age of death do you collect samples for genetic testing in **males**?

- o Below 80 years
- o Below 70 years
- o Below 60 years
- o Below 50 years
- o Below 40 years
- o Below 30 years
- o Below 20 years
- o Below 10 years
- o I collect samples regardless of age.
- o I do not collect samples for males for genetic testing in cases of thoracic aortic aneurysm or dissection.
- o I do not know

16. At what age of death do you collect samples for genetic testing in **females**?

- o Below 80 years
- o Below 70 years
- o Below 60 years
- o Below 50 years
- o Below 40 years
- o Below 30 years
- o Below 20 years
- o Below 10 years
- o I collect samples regardless of age.
- o I do not collect samples for females for genetic testing in cases of thoracic aortic aneurysm or dissection.
- o I do not know

Instructions: Please answer the following questions regarding sample collection practices for the purpose of genetic testing. Questions about sample submission will follow.

17. When collecting a sample, do you check with next-of-kin about religious or cultural objections?

- o Yes
- o No
- o Sometimes

18. What samples do you collect? (Check all that apply)

- ▢ Hair
- ▢ Whole blood
- ▢ Skin
- ▢ Tissue (Please specify type of tissue)
- ▢ Blood card
- ▢ Muscle (non-cardiac)
- ▢ Other (Please specify)

Skip To: Q22 If What samples do you collect? (Check all that apply) != Whole blood

19. When you collect whole blood, what type of tube(s) do you use? (Check all that apply)

- ☐ Purple top (K2 EDTA)
- ☐ Green top (sodium heparin)
- ☐ Royal blue top (no additive)
- ☐ Gold top (serum separator)
- ☐ Other (Please specify)

20. If the whole blood sample is kept for **less than a month**, at what temperature is the sample stored?

- o 4°C
- o -20°C
- o -70°C or colder
- o I do not know

21. If the whole blood sample is kept for **longer than a month**, at what temperature is the sample stored?

- o 4°C
- o -20°C
- o -70°C or colder
- o I do not know

Instructions: In the following questions we define genetic testing as the clinical sequencing and/or deletion/duplication analysis of genes associated with thoracic aortic aneurysms and dissections.

22. Do you ever order genetic testing?

- o Yes
- o No

Skip To: Q31 If Do you ever order genetic testing? = No

23. How do you ship **fresh** whole blood samples to a genetic testing laboratory?

- o Overnight in a Styrofoam container at room temperature
- o Overnight in a Styrofoam container with dry ice
- o Other (Please specify)

24. How do you ship **frozen** whole blood samples to a genetic testing laboratory?

- o Overnight in a Styrofoam container at room temperature
- o Overnight in a Styrofoam container with dry ice
- o Other (Please specify) _____

25. Do you or someone else in your office seek consent from the deceased person's next-of-kin for genetic testing?

- o Yes
- o No

Skip To: Q27 If Do you or someone else in your office seek consent from the deceased person's next-of-kin for gen... = No

26. Which members of the office obtain consent from next-of-kin? (Check all that apply)

- ▢ I, myself, obtain consent.
- ▢ Social worker
- ▢ Genetic counselor
- ▢ Nurse
- ▢ Nurse Practitioner
- ▢ Physician's assistant
- ▢ Other (Please specify)

27. Do you or someone else in your office disclose the results of genetic testing to next-of-kin?

- o Yes
- o No

Skip To: Q30 If Do you or someone else in your office disclose the results of genetic testing to next-of-kin? = No

28. Which members of the office disclose genetic testing results to next-of-kin? (Check all that apply)

- ☐ I, myself, disclose genetic testing results.
- ☐ Social worker
- ☐ Genetic counselor
- ☐ Nurse
- ☐ Nurse Practitioner
- ☐ Physician's assistant
- ☐ Other (Please specify) _____

29. Which of the following type(s) of genetic testing results do you disclose to next-of-kin? (Check all that apply)

- ☐ Positive results (gene mutation identified)
- ☐ Negative results (No gene mutation identified)
- ☐ Variants of uncertain significance

30. What financial means have you used to cover the cost of genetic testing? (Check all that apply)

- ☐ Paid out of pocket by next-of-kin
- ☐ Insurance of the deceased person
- ☐ Research grant
- ☐ The county government covers the full or partial cost of testing.
- ☐ The state government covers the full or partial cost of testing.
- ☐ The federal government covers the full or partial cost of testing.
- ☐ Testing is covered by the office budget
- ☐ I collect samples and refer to other healthcare providers to order genetic testing.
- ☐ Other (Please specify) _____

31. Do you or someone else in your office inform the next-of-kin of options for DNA banking? Here we define DNA banking as the long term storage of DNA for future analysis or testing.

- ○ Yes
- ○ No

Skip To: Q33 If Do you or someone else in your office inform the next-of-kin of options for DNA banking? Here we... = No

32. Which members of the office inform next-of-kin of DNA banking options? (Check all that apply)

- ☐ I, myself, inform the next-of-kin of DNA banking options.
- ☐ Social worker
- ☐ Genetic counselor
- ☐ Nurse
- ☐ Nurse Practitioner
- ☐ Physician's assistant
- ☐ Other (Please specify)

33. What do you see as barriers to ordering genetic testing for a deceased person? (Check all that apply)

- ☐ Difficulty in contacting next-of-kin of the deceased person
- ☐ Do not know how to order genetic testing
- ☐ Inability to get informed consent from next-of-kin
- ☐ Inability to collect samples for genetic testing
- ☐ Difficulty finding an appropriate lab
- ☐ Lack of time
- ☐ Inability to obtain insurance coverage
- ☐ Do not know what test to order
- ☐ Cost of genetic testing
- ☐ Lack of information to provide to next-of-kin
- ☐ Insufficient staffing
- ☐ Lack of geneticists and/or genetic counselors in your area
- ☐ Other (Please Specify)
- ☐ I do not see any barriers to genetic testing for a deceased person.

Carry Forward Selected Choices - Entered Text from "What do you see as barriers to ordering genetic testing for a deceased person? (Check all that apply)"

34. Please rank your top 3 barriers you selected from most important (1) to least important (3).

Display This Question:

If Do you ever order genetic testing? = No

35. Besides any barriers selected above, are there any other reasons you do not order genetic testing?

Display This Question:

If At what age of death do you collect samples for genetic testing in males? = I do not collect samples for males for genetic testing in cases of thoracic aortic aneurysm or dissection.

And If

At what age of death do you collect samples for genetic testing in females? = I do not collect samples for females for genetic testing in cases of thoracic aortic aneurysm or dissection.

36. Besides any barriers selected above, are there any other reasons you do not collect samples for genetic testing?

End of Survey

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Vita

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