UNIVERSITY OF CINCINNATI

June 24, 2003

I, Erin McCain,

hereby submit this as part of the requirements for the degree of:
Master of Science

in Medical Genetics

It is entitled Assessing the genetics knowledge and referral practices of pediatric cardiologists in Indiana, Ohio, and Kentucky

Approved by:
Elizabeth F. Sellers
Richard D. Meyer
Leah Doerfler
Thomas R. Koyhagen

ASSESSING THE GENETICS KNOWLEDGE AND REFERRAL PRACTICES OF PEDIATRIC CARDIOLOGISTS IN INDIANA, OHIO, AND KENTUCKY

A thesis submitted to the

Division of Research and Advanced Studies
of the University of Cincinnati

in partial fulfillment of the
requirements for the degree of

MASTER OF SCIENCE

In the Department of Genetic Counseling
Of the College of Allied Health Sciences

2003

by

Erin McCain

B.S., Baldwin-Wallace College, 2001

Committee Chair: Elizabeth Schorry, M.D.

Abstract

The purposes of this study were to a) determine how pediatric cardiologists utilize genetics and b) assess whether a correlation exists between their genetics knowledge and the strength of their relationships with community genetics services providers. Subjects were pediatric cardiologists practicing in one of six major children's hospitals throughout Indiana, Ohio, and Kentucky. Thirty-four physicians completed a study questionnaire. Physicians correctly answered an average of 70.1% of the knowledge items. We found that the quality of interactions with genetic counselors and medical geneticists in the community did not influence knowledge scores. Additional factors, such as date of graduation from medical school, also did not have correlations with physician knowledge scores. Twenty-eight of 34 (82.4%) physicians reported they refer less than 10% of their patients to genetics professionals. We concluded that pediatric cardiologists have a fairly high level of knowledge in genetics but are underutilizing the genetics services available to them.

Acknowledgements

I would like to thank my co-authors on this project: Dr. Schorry, Dr. Robert Hopkin, Dr. Richard Meyer, Leah Hoechstetter, and Dr. Tom Korfhagen. I would also like to thank Dr. Alan Rope and Dr. Howard Saal for their help in the development of the survey tool used in this study. Thank you also to all the pediatric cardiologists who agreed to participate in this project. Finally, I would like to thank my classmates and family for their never-ending support and encouragement.

Table of Contents

Physician Demographics	
Board Certified in Cardiology	29/34 (85.3%)
Year of Graduation From Medical School:	
Before 1960	3/34 (8.8%)
1960-1979	9/34 (26.5%)
1980-1989	12/34 (35.3%)
1990-1994	6/34 (17.6%)
1995-1999	4/34 (11.8%)
Attended U.S. Medical School	31/34 (91.2%)

Table 1

Number of Physicians Participating By Cardiology Center	
Children's Hospital Medical Center of Akron	7
Cincinnati Children's Hospital Medical Center	9
Kosair Children's Hospital (Louisville, KY)	3
Rainbow Babies and Children's Hospital (Cleveland, OH)	4
Riley Children's Hospital (Indianapolis, IN)	7
University of Kentucky Children's Hospital (Lexington, KY)	4

Table 2

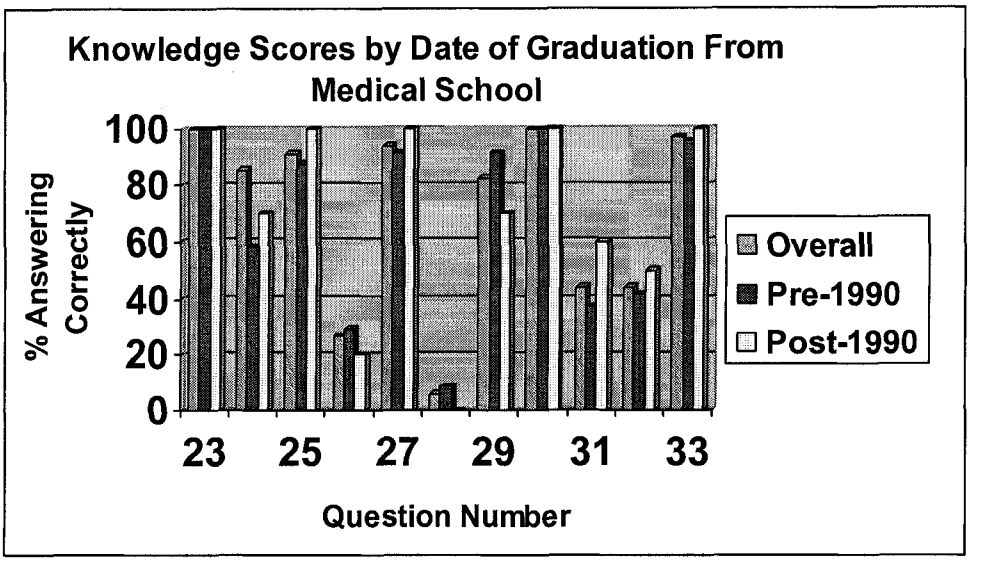

Chart 1

Introduction

With sequencing of the human genome now complete, advances in the field of medical genetics are having an increasingly large impact on the practice of clinical medicine. One of the fields where physicians regularly interact with patients having genetic syndromes is pediatric cardiology. However, to date, no studies have been done examining how knowledge of medical genetics influences the practices of pediatric cardiologists with their patients.

Objectives of this study were to 1) assess the knowledge of pediatric cardiologists in the area of dysmorphology and medical genetics; 2) study the ways pediatric cardiologists interact with the genetic service providers available to them; and 3) study the methods by which pediatric cardiologists obtain their knowledge about genetics and explore how comfortable they feel with this information.

Background

The interactions between primary care medicine and medical genetics have been studied extensively. A study done in 1993[1] found that genetics knowledge was increasing among family physicians, internists, pediatricians, obstetrician-gynecologists and psychiatrists. This study also found that physicians who dealt with genetic conditions in their practices on a regular basis were more likely to have a larger genetics knowledge base and to be interested in learning more. However, little study has been made of non-primary care medical specialists, who are often major providers of care for patients with complex congenital anomalies and genetic disorders.

Studies performed as early as 1974[2] have assessed the attitudes of physicians concerning genetics knowledge and genetic counseling for their patients. In 1974, less than 2% of primary care physicians felt themselves to be knowledgeable enough to provide genetic counseling to their patients. However, only 13% of these physicians were interested in learning more about genetics, stating that they had neither the time nor the interest. Many studies have since assessed these two areas in the primary care community. A study published in 1993[3] found that only 27% of primary care physicians felt confident enough about their knowledge of genetics to decide which patients should receive referrals to local genetics centers. In addition, 81.3% of these physicians felt that additional education in genetics would be useful in their practices. These studies have all shown that there is a growing need for education targeted at physicians and that these physicians are becoming increasingly more interested in expanding their knowledge of genetics.

One of the fields where advances in medical genetics are most applicable is pediatric cardiology. In a study performed by the Baltimore-Washington Infant Study Group,[4,5,6] researchers found that approximately 30% of congenital heart defects are associated with extracardiac malformations. In many cases, these children are first seen for treatment of their heart conditions. When an underlying genetic syndrome is present, a specific diagnosis can have implications in treating the other health problems associated with a particular syndrome. In the past, tests for many of these conditions did not exist. As these tests become available, health care providers will need to be well educated to correctly interpret results and provide complete, comprehensive patient care. The purpose of this study was to assess the attitudes and knowledge of pediatric cardiologists

about genetics and genetic syndromes with congenital heart disease as a common associated finding.

Methods

Subjects for this study were pediatric cardiologists who were practicing in one of six children's hospitals in Indiana, Ohio, and Kentucky. The centers included Children's Hospital Medical Center of Akron (Akron, Ohio), Cincinnati Children's Hospital Medical Center (Cincinnati, Ohio), Kosair Children's Hospital (Louisville, Kentucky), Rainbow Babies and Children's Hospital (Cleveland, Ohio), Riley Children's Hospital (Indianapolis, Indiana), and University of Kentucky Children's Hospital (Lexington, Kentucky). These centers were selected because they had separate departments focusing on pediatric cardiology and genetics and because of their proximity to the researchers in this study. This ensured that the physicians being surveyed would have genetics professionals available on-site and that they would have access to these individuals. A questionnaire was given to all study participants. Pediatric cardiologists in each institution were invited to participate.

The survey tool was composed of thirty-three questions that addressed three main areas: genetics knowledge of cardiac malformations and syndromes (11 questions), strength of relationship with genetics professionals at their respective institutions (14 questions), and physician demographics (8 questions). Questions were either multiple choice or used the Likert scale. The survey tool was developed by medical geneticists with input from a pediatric cardiologist at Cincinnati Children's Hospital Medical Center. The study also included questions based on a previously used survey tool focused on

primary care physicians[7]. The survey tool for this study was evaluated by four medical geneticists and one pediatric cardiologist.

After data collection was completed, all information from each institution was analyzed using Microsoft Excel XP and Statistical Analysis Software, Version 8. An overall knowledge score was created by assigning one point for each correct answer for a possible score of 11. The scores were analyzed for each individual physician and then each institution by using Spearman correlation coefficients and regression analysis to determine whether any variable used to assess strength of relationship was related to a higher score on the knowledge portion of the survey. The significance level was set at $p<0.05$.

We also asked these cardiologists how they feel personally about genetics. Using Likert scales, we asked the physicians about their comfort level with genetics, their interest in learning more about cardiac genetics, their assessment of the strength of their relationship with their local genetics center, and how adequate they feel their own understanding of genetics is in meeting their practice needs. These scores were then analyzed against other questions that served as concrete measures of the interactions between each cardiologist and the genetic service providers in their communities. The purpose of this was to see if the physician interest in genetics or comfort level correlated with referral practices. We also attempted to see whether a specific interaction type (i.e. referral to a genetic counselor, participation in multidisciplinary clinics, etc.) predicted a higher score in the area of physician knowledge using Spearman correlation coefficients and regression analysis. Significance levels were set at $p<0.05$.

Results

Overall, 53 physicians were eligible for this study. Thirty-four participated by filling out a questionnaire and completing a short knowledge test, giving a participation rate of 64.2%. No physicians declined to participate in this study. Those who did not fill out a questionnaire were not present at the meetings when the survey was taken mainly because of patient care commitments or because they were not in the office on that day. The number of physicians participating from each center is shown in table 1.

Demographics of Physicians

Of the 34 physicians who participated, 22 (64.7%) had graduated from medical school after 1980. Twenty-nine of 34 (85.3%) physicians were board-certified in cardiology. Of the five physicians who were not board-certified, three of these were recent graduates who had not been out of training long enough to sit for a board exam. Almost all of the physicians who participated had graduated from a medical school in the United States (31/34, 91.2%). The majority of the physicians (28/34, 82.4%) reported that they spend more than 50% of their time in patient care. Interestingly, spending more time caring for patients was not related to a higher knowledge score. All physicians reported that they see less than 100 patients per week, with the average being between 20 and 50 patients (22/34, 64.7%). Twenty of 34 physicians (58.8%) reported that more than 10% of the patients they care for have a genetic syndrome. This was based on physician perception, not practice records or other documented measures.

When asked about their most recent educational exposure to genetics, 24/34 (70.6%) reported their last class being during medical school. Seven of 34 physicians reported postgraduate training in genetics. Of these seven physicians, five had graduated from medical school before 1990. Twenty-four of 34 physicians (70.6%) also reported that they attend continuing medical education courses or read continuing education materials related to the field of genetics. All physicians (34/34, 100%) reported that they had exposure to genetics through academic journals, 31/34 (91.2%) attending professional meetings, 25/34 (73.5%) attending Grand Rounds presentations, 22/25 (88.0%) reading textbooks, 15/34 (44.1%) attending continuing medical education (CME) courses, and 5/34 (14.7%) using on-line computer services. Twenty-five of 34 physicians (73.5%) reported that they used other practitioners as a resource, but only 9 of 34 (26.5%) reported using their local genetics center as a resource for gaining new information about genetics. All physicians reported using more than one of the above techniques to enhance their knowledge of medical genetics.

Physician perception of knowledge and referral patterns were also assessed with ten being the highest level of interest, knowledge, or strength and one being the lowest. When asked about how would rate their knowledge of cardiac genetics, the average score was 6. The average score was 7.1 when physicians were asked to rate their relationship was with their local genetics center. Physicians reported that they felt their knowledge of genetics was fairly adequate in meeting their practice needs currently (average score of 6.7), but were less confident in estimating their practice needs five years from now without learning more (average of 5.7). The difference in these scores was not

statistically significant. Overall, pediatric cardiologists were interested in learning more about genetics (average score of 7.9).

When asked about referrals to genetic service providers, 28/34 (82.4%) reported that they refer less than 10% of their patients for genetic services. Two of the six cardiology centers did employ a genetic counselor. Each of these had a pediatric cardiologist focusing in the field of cardiac genetics. Referral patterns did not differ between centers that employed a genetic counselor and those that did not. Half (17/34) of the participants reported that they interact with a medical geneticist or genetic counselor on at least a monthly basis. This also did not differ between centers where a genetic counselor was employed directly through the department of cardiology. Ten of 34 (29.4%) reported being active in multidisciplinary clinics involving cardiology and genetics. Of these physicians, all but one had graduated from medical school before 1990 and all ten spent more than 50% of their time caring for patients. Overall, 19/34 (55.9%) physicians reported that they personally provide genetic counseling to a patient on a monthly basis or less frequently. Three of six centers reported being actively involved in research that focuses in molecular cardiology.

Genetics Knowledge

The mean overall score for all pediatric cardiologists was 70.1% correct (7.71/11). Scores ranged from 63.6-90.9%. When scores were compared from center to center, there were no statistically-significant differences found ($p>0.05$). The overall scores for each question (questions 23-33) are shown in the appendix. Four questions were answered incorrectly by more than half of the participants (26, 28, 31, and 32). Only 2

questions were answered correctly by all participants. No participant answered all the questions correctly.

Discussion

The average physician in our study graduated from a medical school in the United States after 1980, is board-certified in cardiology, and spends more than 50% of his or her time caring for patients. Of particular interest for our purposes was the fact that the majority of physicians reported that they see between 20 and 50 patients per week and that more than 10% of these patients have a genetic syndrome. This demonstrates the importance of genetics knowledge in the practice of pediatric cardiology.

One of the most striking findings in this study was the fact that physicians almost universally reported that they are interested in learning more about the genetics of cardiac malformations. Most physicians had not had formal exposure to medical genetics since medical school and were instead trying to find time to educate themselves. Pediatric cardiologists have very busy schedules and may not have easy access to education regarding advances in molecular medicine. This is shown in the ways that physicians report they stay abreast of new developments.

At the outset of this study, we hypothesized that pediatric cardiologists who had graduated from medical school after 1980 would score higher on the knowledge portion of the questionnaire than their colleagues who graduated earlier. However, this was not what we found. We also expected physicians to have more genetics knowledge if they had a strong relationship with their local genetics center. To assess the strength of this relationship, we included questions on the frequency of interactions with a medical

geneticist and/or genetic counselor, number of referrals to a genetics center, and questions targeted towards physician attitudes towards genetics professionals. We did not see any statistically-significant association between quality of interactions with a local genetics center and knowledge of genetics as measured by our survey tool. It is interesting that cardiologists who spend less than 50% of their time in patient care scored higher on the knowledge test than those who spent more time in clinical practice. Pediatric cardiologists who reported that they had postgraduate training in genetics also had higher scores. However, neither of these differences were statistically significant. Most physicians reported they had a good relationship with genetics and used their services appropriately. However, only a small percentage of cardiologists use genetics centers as a way to obtain knowledge about genetics. Other measures, such as referral to a genetics center and interactions with a genetic counselor or medical geneticist, indicate only sporadic use of genetic services.

The study participants had a relatively high level of genetics knowledge. Although our assessment tool has not been validated, we believe an overall score of 70% demonstrates that these pediatric cardiologists are reasonably well-informed about the genetic syndromes associated with cardiac malformations. Nevertheless, there were some gaps in knowledge as demonstrated by the 4 questions answered incorrectly by more than half of the participants. The depth and significance of these knowledge gaps could not be assessed in this study.

The majority of physicians reported more than 10% of their patient population had a genetic syndrome, yet they refer less than 10% of their patients to a genetic counselor or medical geneticist. Thus, it seems that more patients have underlying

genetic conditions than are referred for genetic services. The majority of pediatric cardiologists reported that they did not personally provide genetic counseling to patients on a regular basis. This study did not address whether or not families may be receiving counseling and /or genetic services through avenues other than referral from the cardiologist. Based on these results, it appears that the families of some patients with recognized genetic syndromes may not be receiving formal counseling about their underlying condition. In addition, it is important to note that recent studies have found evidence for genetic syndromes in approximately 30% of infants with congenital heart defects[4,5,6]. Thus, a number of syndromes may be going unrecognized. Thus, while pediatric cardiologists appear to be aware of genetic syndromes associated with heart disease, the patients and their families may benefit from more systematic referral practices to ensure evaluation and counseling for possible genetic syndromes. The emphasis of the genetic evaluation should place particular emphasis on addressing the non-cardiac manifestations and recurrence risks of these conditions.

One way of improving collaboration between pediatric cardiology and genetics may be participation in multidisciplinary clinics. Relatively few pediatric cardiologists in this study reported working with a genetics professional in a multidisciplinary clinic. These clinics bring multiple specialists together to treat children with conditions involving multiple body systems and are among the most effective ways of managing patients with complex medical needs[8,9,10,11]. Increasing participation in these clinics may be a feasible way of bridging the gap between the specialties and is likely to improve care.

Limitations

One of the major limitations of this study was its small sample size. We saw no correlations between level of genetics knowledge and any of the other variables we attempted to measure. However, because the study included only 34 physicians, it is impossible to determine whether there are truly no correlations present or if our study was simply not large enough to detect them. Our study design enabled us to detect a 20-point difference in knowledge scores between groups (α=0.10, β=0.70). Given a range of 63.6-90.9% for the knowledge portion of the study it is unlikely that we would have power enough to detect differences between subgroups of cardiologists. Another limitation was that our knowledge test was not validated and that the questions may not differentiate those with high knowledge from those with less. A third limitation of this study was the possible existence of a bias in the physicians who agreed to participate. We did not obtain information on the physicians who did not complete a questionnaire, and it is possible that this group of physicians may have differed from the study group. For example, we did not study physicians in private practice or practicing at smaller institutions. Perhaps these physicians would have presented us with different information and would have changed the outcomes seen in our study. Finally, the small scope of this study makes generalization of these results difficult.

In spite of these limitations this study is an important first step in defining the relationship of pediatric cardiologists with medical genetics. However, a larger study needs to be performed to determine what factors influence how pediatric cardiologists apply genetics knowledge to their practices and what types of interventions would be

most effective in improving the genetic services offered to families dealing with congenital heart defects.

Conclusion

We have demonstrated that, overall, the genetics knowledge of this group of pediatric cardiologists is fairly high. Furthermore, it is encouraging that the majority of physicians indicated that their interest in learning more about genetics is high. However, there appears to be a gap between knowledge and use of genetic services offered in clinical care. We believe that to ensure that their patients are being fully evaluated, counseled and treated for genetic conditions pediatric cardiologists and other pediatric subspecialists should encourage referral and interaction with genetics professionals. One way of facilitating this interaction may be the development of multidisciplinary teams involving members of the cardiology team and genetics professionals.

References

1. Hofman KJ, Tambor ES, Chase GA, Geller G, Faden RR, Holtzman NA. Physicians' knowledge of genetics and genetic tests. Acad Med 1993; 68 (8): 625-32.
2. Marino B, Digilio MC. Congenital heart disease and genetic syndromes: specific correlation between cardiac phenotype and genotype. Cardiovasc Pathol 2000; 9 (6): 303-15.

3. Ferencz C, Rubin JD, Loffredo CA, Magee CA. Epidemiology of Congenital Heart Disease. The Baltimore-Washington Infant Study 1981-1989. Futura Publishing Company, Inc.: Mount Kisco, New York: 1993.
4. Ferencz C, Loffredo CA, Correa-Villasenor A, Wilson PD. Genetic and Environmental Risk Factor of Major Cardiac Malformations. The Baltimore-Washington Infant Study 1981-1989. Futura Publishing, Inc: Mount Kisco, New York: 1997.
5. Hayflick S, Eiff M, Carpenter L, Steinberger J. Primary care physicians' utilization and perceptions of genetics services. Genetics Medicine. 1998. 1: 13-21.
6. Mountcastle-Shah E and Holtzman NA. Primary care physicians and their willingness to participate in testing and research. Am J Med Genet. 2000. 94 (5): 409-16.
7. Miller S, Saal H.
8. Tao LS, Hart P, Edwards E, Evans AT, Whitaker E, Smith P. Treatment of difficult-to-control blood pressure in a multidisciplinary clinic at a public hospital. J Natl Med Assoc. 2003. 95 (4): 263-9.
9. Walker MS, Ristvedt SL, Haughey BH. Patient care in multidisciplinary cancer clinics: Does attention to psychosocial needs predict patient satisfaction? Psychooncology. 2003. 12 (3): 291-300.

10. Soriano A, Castells A, Lacy AM, Ayuso C, Ayuso JR, Conill C, Delgado S, Fuster J, Garcia-Valdecasas JC, Gines A, Martin M, Maurel J, Miquel R, Molla M, Vilana R, Castellvi-Bel S, Elizalde JI, Pinol V, Pellise M, Biete A, Gascon P, Pique JM. Evaluation of the efficacy and efficiency of a multidisciplinary unit for the treatment of patients with colorectal cancer. Gastroenterol Hepatol. 2002. 25 (10): 579-84.

11. McDonald K, Ledwidge M, Cahill J, Quigley P, Maurer B, Travers B, Ryder M, Kieran E, Timmons L, Ryan E. Heart failure management: multidisciplinary care has intrinsic benefit above the optimization of medical care. J Card Fail. 2002. 8 (3): 142-8.

Appendix 1

A Survey of Genetics in Cardiology

Thank you for participating in this study. Your confidentiality will be maintained. By completing this questionnaire, you indicate your consent to participate in the study.

Section One

1. Are you board-certified in Cardiology?

   a. Yes b. No

2. Do you have a subspecialty?

   a. Yes b. No

   If yes, what is your subspecialty? _____

   Are you board-certified in that subspecialty? a. Yes b. No

3. When did you graduate from medical school?

   a. Before 1960
   b. Between 1960 and 1979
   c. Between 1980 and 1989
   d. Between 1990 and 1994
   e. Between 1995 and 1999
   f. After 1999

4. Where did you attend medical school?

   a. United States b. Canada c. Other _____

5. What percentage of your time is spent in patient care?

   a. Less than 10%
   b. Between 10% and 25%
   c. Between 26% and 50%
   d. Between 50% and 75%
   e. Greater than 75%

6. Does your cardiology group employ a genetic counselor?

   a. Yes b. No

7. Does your cardiology group employ a medical geneticist or other physician specializing in genetics?

   a. Yes b. No

8. Do you routinely participate in multidisciplinary clinics involving genetics professionals (medical geneticists, genetic counselors, etc.)?

   a. Yes b. No

   If yes, what are the names of these clinics and how often are they held?

   _____

   _____

   _____

9. Do you attend continuing education or educational seminars focusing on genetic information and its relationship to cardiology?

   a. Yes b. No

10. Does your department participate in research in molecular cardiology?

    a. Yes b. No

11. Which of the following describes your most recent educational exposure to genetics?

Human genetics course in college?	a. Yes	b. No
Human genetics course in medical school?	a. Yes	b. No
Postgraduate training in clinical genetics?	a. Yes	b. No

12. How would you rate your knowledge of the genetics of cardiac disease?

1	2	3	4	5	6	7	8	9	10
weak									strong

[Average score=6.0]

13. Approximately how many patients (inpatient and outpatient) do you see in one week?

    a. Less than 20
    b. Between 20 and 50
    c. Between 50 and 100
    d. Between 100 and 125
    e. More than 125

14. What percentage of your practice consists of caring for individuals with genetic disorders?
    a. Less than 5% **(11.8%)**
    b. Between 5% and 10% **(29.4%)**
    c. Between 10% and 20% **(38.2%)**
    d. Between 20% and 30% **(11.8%)**
    e. Greater than 30% **(8.8%)**

15. What percentage of your patients do you refer to a medical geneticist or genetic counselor?
    a. Less than 5% **(32.4%)**
    b. Between 5% and 10% **(50%)**
    c. Between 10% and 20% **(5.9%)**
    d. Between 20% and 30% **(8.8%)**
    e. Greater than 30% **(2.9%)**

16. How often do you personally provide counseling on inheritance risks and natural history to a patient with a known genetic disorder or a family history of a known genetic disorder?

    a. Several times daily **(2.9%)**
    b. Daily **(8.8%)**
    c. Weekly **(32.4%)**
    d. Monthly **(32.4%)**
    e. Rarely **(20.6%)**
    f. Never **(2.9%)**

17. From whom or where do you obtain knowledge and information about genetics? (Circle all that apply)

    a. Journals
    b. Professional meetings
    c. Other practitioners
    d. CME courses
    e. On-line computer services
    f. Local genetics centers
    g. Grand Rounds
    h. Pharmaceutical companies
    i. Medical/Genetics textbooks
    j. I have not obtained information recently about genetics

18. How strong do you feel your department's relationship is with your local genetics center?

    1 2 3 4 5 6 7 8 9 10
    weak strong
    [Average score=7.1]

19. How often do you interact with a medical geneticist or genetic counselor?
    a. Daily **(2.9%)**
    b. Weekly **(23.4%)**
    c. Monthly **(26.5%)**
    d. Rarely **(47.1%)**
    e. Never **(0%)**

20. How adequate do you feel your understanding of genetics is in meeting your current practice needs?

1	2	3	4	5	6	7	8	9	10
inadequate									very adequate

[Average score=6.7]

21. How adequate do you feel your understanding of genetics is in meeting your practice needs five years from now?

1	2	3	4	5	6	7	8	9	10
inadequate									very adequate

[Average score=5.7]

22. How interested are you in learning more about the role of genetics in medical practice?

1	2	3	4	5	6	7	8	9	10
not interested									very interested

[Average score=7.9]

Section Two

23. In a newborn female with coarctation of the aorta, what study should be ordered?
    a. FISH for Velo-cardio-facial syndrome (VCF)
    b. FISH for Williams syndrome
    c. Molecular study for Noonan syndrome
    d. Chromosome analysis for Turner syndrome* **(34/34 correct)**

24. In a newborn male with coarctation of the aorta, what study is indicated?
    a. None of the below* **(29/34 correct)**
    b. Chromosome analysis
    c. FISH for Williams syndrome
    d. FISH for Velo-cardio-facial syndrome (VCF)

25. Which of the following is **not** an indication for FISH for 22q11 deletion?
    a. Perimembranous VSD
    b. ASD secundum* **(31/34 correct)**
    c. Tetralogy of Fallot
    d. Interrupted aortic arch

26. In a five-year-old with pulmonic valvular stenosis, which would **not** raise additional suspicion for Noonan syndrome?

    a. Chronic abdominal pain with GE reflux
    b. Cryptorchidism
    c. Microcephaly* **(9/34 correct)**
    d. Learning disabilities

27. A three-month-old infant presents with direct hyperbilirubinemia. His father had pulmonary artery stenosis. What syndrome would you suspect?

    a. 22q11 deletion
    b. Alagille syndrome* **(32/34 correct)**
    c. Townes-Brock syndrome
    d. These are probably unrelated health problems

28. A three-week-old infant presents with radial anomalies and a ventricular septal defect. His family history is significant for his father having imperforate anus. The most likely diagnosis is:

    a. Ellis-van Creveld syndrome
    b. Fanconi's anemia
    c. VATER association
    d. Townes-Brock syndrome* **(2/34 correct)**

29. A newborn presents with tetralogy of Fallot. He looks otherwise normal. Which study would you order?

    a. None
    b. FISH for 22q11 deletion* **(28/34 correct)**
    c. Chromosome analysis
    d. Molecular study of the PTPN11 gene

30. The diagnosis of AV canal malformation is made by fetal echocardiogram in a second-trimester fetus. The most likely diagnosis is:

    a. Trisomy 13
    b. Trisomy 18
    c. Trisomy 21* **(34/34 correct)**
    d. CHARGE association

31. A seven-year-old presents with tetralogy of Fallot. He is performing well in school and has a normal physical exam. The probability of him having a previously undetected deletion of 22q11 is:

    a. Less than 2%* **(15/34 correct)**
    b. 5%
    c. 15%
    d. 25%

32. A child has an echocardiogram and aortic root dilitation is noted. Differential diagnoses besides Marfan syndrome include all of the following **except**:

    a. Ehlers-Danlos syndrome
    b. Beals syndrome (congenital contractural arachnodactyly)
    c. Turner syndrome
    d. Fragile X syndrome* **(15/34 correct)**

33. In a child presenting with Long QT syndrome, an appropriate additional medical screen would be:

    a. MRI of the pituitary
    b. Hearing ABR* **(33/34 correct)**
    c. Urinalysis
    d. Bone age testing