PA Competencies for Genomic Medicine

Natcher Conference Center
National Institutes of Health

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I hope to accomplish the following:

- Review some challenges to expanding and improving education for non-genetics health professionals
- Consider (briefly) different levels of genetic literacy as they relate to PA practice
- Solicit your thoughts on the integration of genetics into each of the Task Areas for PAs
- Review the NCHPEG/AAPA website now in development

“There is always a genetic contribution to all common and rare disorders….*The contribution of genetics knows no disciplinary boundaries and…affects all fields of medicine.”*

"The manner by which genetics will be integrated into medical practice is still largely undetermined. It is difficult to motivate students to learn things *on the promise that they will be important in the future*, *and it is difficult to find current case examples and role models."*

Harry Lesmana, fifth-year medical student, University of Indonesia, Jakarta, 24 March 2007

What we've been promising:

- A revolution in health care driven by the genome projects
  - Identification of genes that confer susceptibility to common, complex disease
  - Insights into disease processes
  - Individualized medicine
  - Prevention-based health care

A reality check:

"The prevalent response to the HGP has been:
'genes to be found; phenotypes to be predicted!'
(but) the HGP has provided information that signifies complexity rather than simplification."

Scriver, C.R. Why mutation analysis does not always predict clinical consequences: Explanations in the era of genomics. *J. Pediatrics* 140(5): 502-506, 2002.

Genetics and Prevention

"The availability of (genomic) information will demand a level of intimacy between doctor and patient that is rarely seen today. After all, it's one thing for patients to learn that they have a strep throat or a sprained ankle; it's another for them to comprehend the implications of a progressively more complex genetic risk profile.

Genetics and Prevention (cont'd.)

- This is not the kind of information amenable to a ten-minute office visit. Rather, it demands the sort of relationship that the eminent nineteenth-century physician, Sir William Osler, had in mind when he said, 'the good physician treats the disease, the great physician treats the patient.'"

Shaywitz, D.A. and Aisiello, D.A. "Back to the future: Medicine and our genes," *New York Times,* 16 April 2000

Genetic Alliance/NCHPEG Survey

Ask affected individuals and family members about their experiences with a variety of health professionals not trained in genetics

Genetic Alliance
Advocacy, Education & Empowerment

NCHPEG
National Coalition for Health Professional Education in Genetics

Respondents

- Recruited from members of Genetic Alliance: >600 organizations; 14M individuals; 1,000 conditions
- On-line questionnaire
- 5,915 respondents between December 2004 and August 2005

Respondents

- >80% reported 1-4 affected family members

  1 = 55% 3 = 8%

  2 = 14% 4 = 4%

- Majority diagnosed within five years of the survey

Survey data

(Now in press at *Genetics in Medicine*)

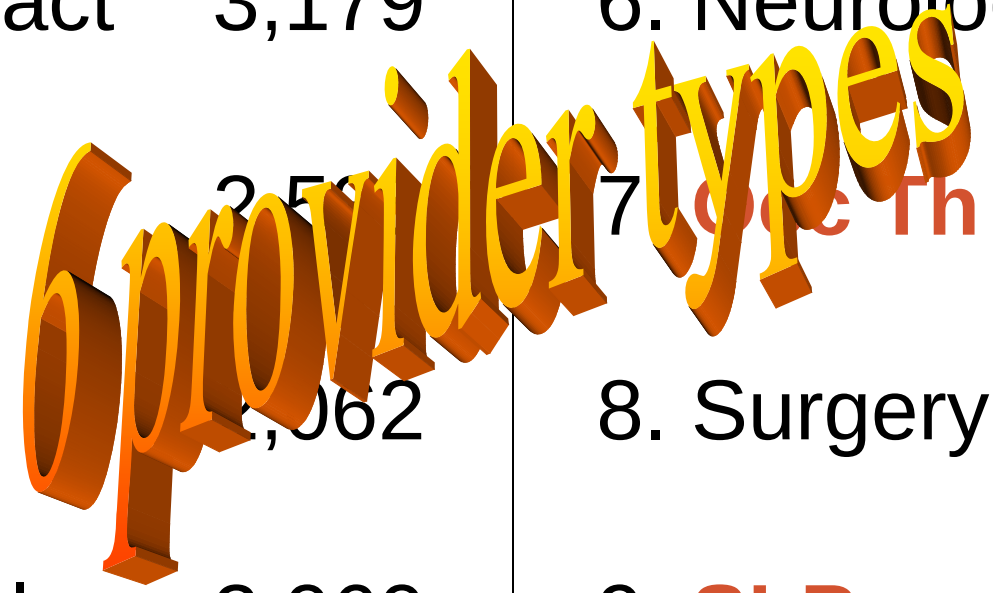
- Medical demographic data
  - **How many in family dx; types/no. of providers**
- Knowledge and skills of "most important" provider type (8 questions)
  - **Knowledge of condition in family; skills include management plan, referrals, research, psychosocial**
- Self knowledge of the condition in question
- Sources of genetics information
- Positive and disappointing experiences

10 Most Common Conditions

1. Marfan	465	6. SMA	148
2. PXE	260	7. Long QT	147
3. Alpha-1	243	8. 47, XXY	140
4. TSC	205	9. HHT	138
5. Albinism	148	10. BRCA 1/2	122

10 Most-Frequently Consulted Providers

1. Fam pract	3,179	6. Neurology	1,885
2. Peds	2,5[illegible]	7. Occ Th	**1,726**
3. Cardio	2,062	8. Surgery	1,620
4. Ophthal	2,060	9. **SLP**	**1,546**
5. **Phys Th**	**1,949**	10. Orthop	1,382



Knowledge Scores for Providers

Specialty	Consulted	Most Imp.	% Poor	% Good/Exc.
Fam. Prac.	3179 (53.7%)	1022 (17.2%)	39	34
Pediatrics	2530 (42.7%)	811 (13.7%)	27	47
Cardiology	2062 (35%)	635 (10.7%)	22	56
Neurology	1885 (31.8%)	578 (9.7%)	21	56
Ophthalmol.	2060 (34.8%)	336 (5.6%)	22.6	55
Surgery Emergency	1620 (27.4%) 1100 (18.6%)	115 (2%) 15 (<1%)	19.6 62	62.4 17.4

Providers' Knowledge

31.7%

Percentage who assigned their providers "poor" knowledge rankings for the condition in the family – averaged across all specialties

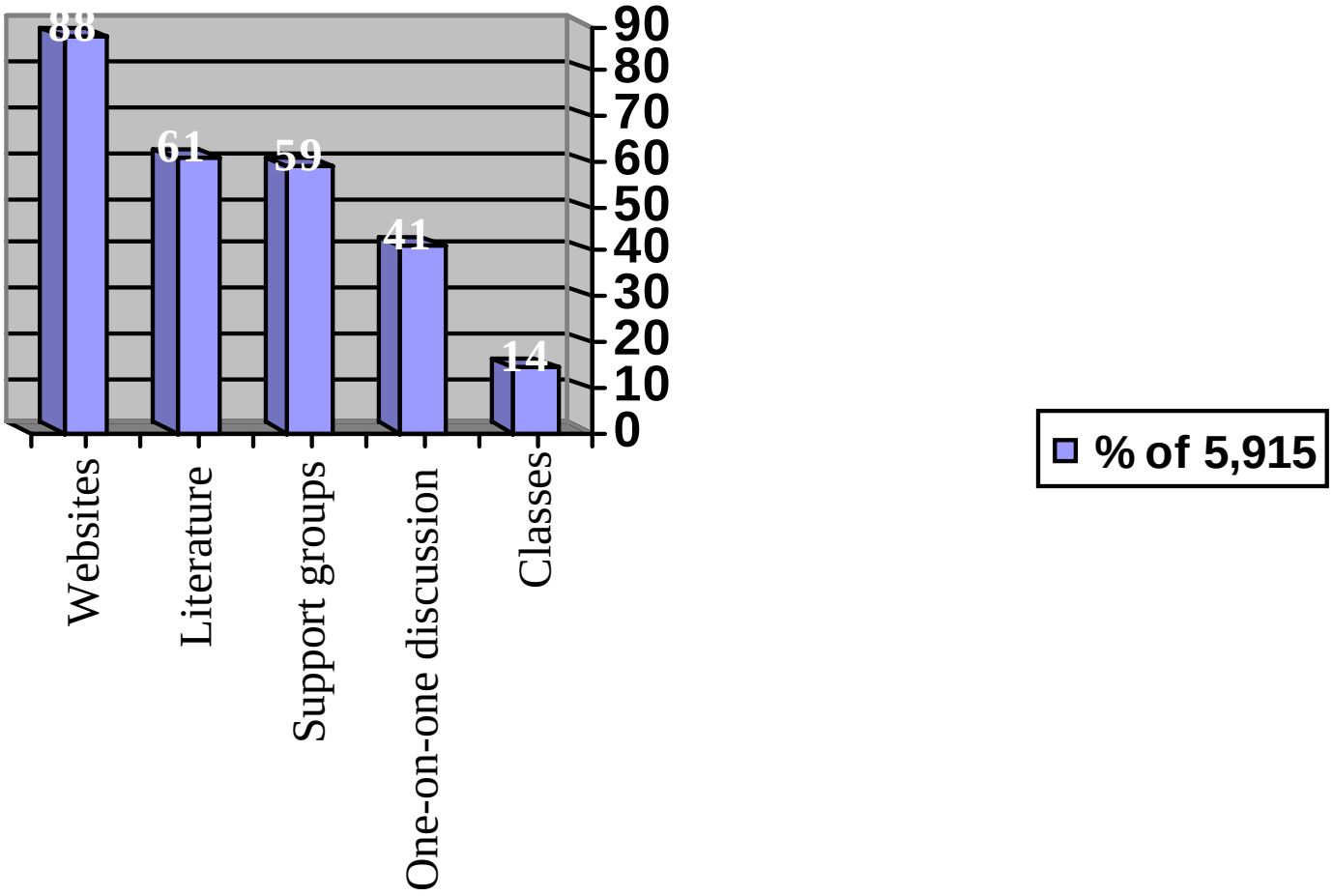
Sources of Genetics Information

78%

(3,769/4,821)

no genetics-education materials from provider designated as most important for management of the condition in the family

Sources of Genetics Information



“People say believe half of what you see, son, and none of what you hear.”

“I Heard it Through the Grapevine”

Norman Whitfield and Barrett Strong, 1967

Positive & Disappointing Experiences

- **“It's a relief when we get a doctor who has knowledge of Marfan syndrome or is cooperative about learning.”**
- **“When my child is sick, it's frustrating to have to explain the syndrome and its implications. I want to focus on my child, not educating the medical staff.”**
- **“Every time I walk into the ER it seems I am drawing a sketch of . . .chromosomes (t11/22) and giving a class… lol.”**

Major Positive Themes

- Knowledgeable, competent, generally pleased 684/1978 (35%)
- Happy with specialists/specialized institutions 319/1978 (16%)
- Engages in learning/does research 223/1978 (11%)
- Sympathetic, listens, explains 100/1978 (5%)

Major Negative Themes

- Provider lacks interest, willingness, knowledge: 1,307/3828 (34%)
- Personally did research and educated provider(s): 591/3828 (15%)
- Disease rarity limits information for everyone: 269/3828 (7%)
- Information offered was old, conflicting, or incorrect: 190/3828 (5%)

Conclusions

- Missed opportunities to educate patients and families
- Importance of primary care and allied health providers in care, and as sources of genetics-related information
- Importance of a coordinated, team approach to care
- Importance of a willingness to learn on the part of providers
- Individuals and families as sources of information for providers

Barriers to Genetics Education for Health Professionals

- Crowded curriculum
- Misconceptions about genetics
- Lack of knowledgeable faculty
- Disconnect between basic sciences and clinical experiences during training
- Failure to integrate genetics across the curriculum
- Inadequate representation of genetics on certifying exams

Some Barriers to the Integration of Genetics into Primary Care

- a dearth of genetics professionals
- lack of knowledge about genetics among primary-care providers
- lack of confidence
- inadequate family histories (time is an issue)
- lack of referral guidelines
- payment for genetics-related services (time is an issue)

Suther, S. and Goodson, P. 2003. Barriers to the provision of genetic services by primary care physicians: A systematic review of the literature. *Genetics in Medicine* 5(2): 70-76.

The challenge is to meld the practical and the conceptual:

"Primary care providers are asking for instruction on specific content, and there is no debate about this need. However, with the rapid pace of change in genetic medicine, specific content will fall short of what PCPs really need….A thoughtful, deliberate, and informed refinement of the 'usual' cognitive strategies will have the greatest impact on integrating genetics into all of health care."

Hayflick & Eiff. 2002. *Genetics in Medicine*. 4(2): 43-44.

Levels of Genetic Literacy: Where Should PAs Be?

- *Nominal*: identify terms and concepts as genetic in nature, provide naïve explanations of genetics concepts, hold some misconceptions
- *Functional*: use genetics vocabulary, define terms correctly, but memorize responses
- *Structural*: understand conceptual schemes of genetics, explain genetics concepts in one's own words
- *Multidimensional*: understand the relationship of genetics to other disciplines and to health and disease

Define the following terms:

- Chromosome
- Gene
- Haploid number
- Diploid number
- Autosomal dominant
- Autosomal recessive
- Penetrance

Family History: The First Genetic Test

Recognize RED FLAGS!

- Multiple affected individuals
- Early age at onset of disease
- Severity of disease
- Presence of disease in the less-frequently-affected sex, e.g., breast cancer in a male
- Recurrence of disease despite preventive measures
- Multifocal disease

Complex vs. Single-Gene Disorders

Characteristics	Complex	Single-Gene
Gene(s)	**segregate**	**segregates**
Disorder	**aggregates**	**segregates**
Gene products involved	multiple	primarily one
Role of environment	important	often over-ridden by effect(s) of gene mutation
Age at onset	older	younger
Risks for relatives of probands	smaller, less predictable	larger, more predictable
Health-care burden	high	low
Selection against	low	high

Genetic Literacy: Different for Each Discipline

What do you want PAs to

Know? Value? Do?

Task Areas

The list of tasks below include knowledge and skill areas that were identified as important to physician assistant practice through an intensive practice analysis. Many of these knoweldge areas and cognitive skills are covered on NCCPA's examinations.

Tasks	% of Exam Content
History Taking & Performing Physical Examinations	16
Using Laboratory & Diagnostic Studies	14
Formulating Most Likely Diagnosis	18
Health Maintenance	10
Clinical Intervention	14
Pharmaceutical Therapeutics	18
Applying Basic Science Concepts	10
	Total: 100%

Each question you encounter will address an organ system and a task area from the table at left.

History Taking & Performing Physical Examinations

Knowledge of:

- Pertinent historical information associated with selected medical conditions
- Risk factors for development of selected medical conditions
- Signs and symptoms of selected medical conditions
- Physical examination techniques
- Physical examination findings associated with selected medical conditions
- Appropriate physical examination directed to selected medical conditions
- Differential diagnosis associated with presenting symptoms or physical findings

Cognitive skills in:

- Conducting comprehensive and focused interviews
- Identifying pertinent historical information
- Performing comprehensive and focused physical examinations
- Associating current complaint with presented history
- Identifying pertinent physical examination information

Using Laboratory & Diagnostic Studies

Knowledge of:

- Indications for initial and subsequent diagnostic or laboratory studies
- Cost effectiveness of diagnostic studies or procedures
- Relevance of common screening tests for selected medical conditions
- Normal and abnormal diagnostic ranges
- Risks associated with diagnostic studies or procedures
- Appropriate patient education related to laboratory or diagnostic studies

Cognitive skills in:

- Using diagnostic equipment safely and appropriately
- Selecting appropriate diagnostic or laboratory studies
- Collecting diagnostic or laboratory specimens
- Interpreting diagnostic or laboratory studies results

Formulating Most Likely Diagnosis

Knowledge of:

- Significance of history as it relates to differential diagnosis
- Significance of physical findings as they relate to diagnosis
- Significance of diagnostic and laboratory studies as they relate to diagnosis

Cognitive skills in:

- Correlating normal and abnormal diagnostic data
- Formulating differential diagnosis
- Selecting the most likely diagnosis in light of presented data

Health Maintenance

Knowledge of:

- Epidemiology of selected medical conditions
- Early detection and prevention of selected medical conditions
- Relative value of common screening tests
- Appropriate patient education regarding preventable conditions or lifestyle modifications
- Healthy lifestyles
- Prevention of communicable diseases
- Immunization schedules and recommendations for infants, children, adults and foreign travelers
- Risks and benefits of immunization
- Human growth and development
- Human sexuality
- Occupational and environmental exposure
- Impact of stress on health
- Psychological manifestations of illness and injury
- Effects of aging and changing family roles on health maintenance and disease prevention
- Signs of abuse and neglect
- Barriers to care

Cognitive Skills in:

- Using counseling and patient education techniques
- Communicating effectively with patients to enhance health maintenance
- Adapting health maintenance to the patient's context
- Using informational databases

Organ Systems

The table below illustrates the approximate percentage of exam questions you'll encounter. Other content dimensions cross-sect these categories. For example, up to 20 percent of the questions on any exam may be related to surgery, and up to two percent may cover legal or ethical issues.

Organ System	% of Exam Content
Cardiovascular	16
Pulmonary	12
Endocrine	6
EENT (Eyes, Ears, Nose and Throat)	9
Gastrointestinal /Nutritional	10
Genitourinary	6
Musculoskeletal	10
Reproductive	8
Neurologic System	6
Psychiatry/Behavioral	6
Dermatologic	5
Hematologic	3
Infectious Diseases	3
	Total: 100%

Although not an exhaustive listing, click on each organ system to view a sample of the diseases, disorders and medical assessments you may encounter during the exam. These lists can provide a foundation for your exam preparation and serve as your blueprint to the exam content.

Each question you encounter will address an organ system from the table at left and a physician assistant practice task area.

Cardiovascular System

Cardiomyopathy
Dilated
Hypertrophic
Restrictive
Conduction Disorders
Atrial fibrillation/flutter
Atrioventricular block
Bundle branch block
Paroxysmal supraventricular tachycardia
Premature beats
Ventricular tachycardia
Ventricular fibrillation/flutter
Congenital Heart Disease
Atrial septal defect
Coarctation of aorta
Patent ductus arteriosus
Tetralogy of Fallot
Ventricular septal defect

Congestive Heart Failure
Hypertension
Essential
Secondary
Malignant
Hypotension
Cardiogenic shock
Orthostasis/postural
Ischemic Heart Disease
Acute myocardial infarction
Angina pectoris
- *Stable*
- *Unstable*
- *Prinzmetal's/variant*

Vascular Disease
Acute rheumatic fever
Aortic aneurysm/dissection
Arterial embolism/thrombosis
Chronic/acute arterial occlusion
Giant cell arteritis
Peripheral vascular disease
Phlebitis/thrombophlebitis
Venous thrombosis
Varicose veins
Valvular Disease
Aortic stenosis/insufficiency
Mitral stenosis/insufficiency
Mitral valve prolapse
Tricuspid stenosis/insufficiency
Pulmonary stenosis/insufficiency
Other Forms of Heart Disease
Acute and subacute bacterial endocarditis
Acute pericarditis
Cardiac tamponade
Pericardial effusion

Pharmaceutical Therapeutics

Knowledge of:

- Mechanism of action
- Indications for use
- Contraindications
- Side effects
- Adverse reactions
- Follow-up and monitoring of pharmacologic regimens
- Risks for drug interactions
- Clinical presentation of drug interactions
- Treatment of drug interactions
- Drug toxicity
- Methods to reduce medication errors
- Cross reactivity of similar medications
- Recognition and treatment of allergic reactions

Cognitive skills in:

- Selecting appropriate pharmacologic therapy for selected medical conditions
- Monitoring pharmacologic regimens and adjusting as appropriate
- Evaluating and reporting adverse drug reactions

Applying Basic Science Concepts

Knowledge of:

- Human anatomy and physiology
- Underlying pathophysiology
- Microbiology and biochemistry

Cognitive skills in:

- Recognizing normal and abnormal anatomy and physiology
- Relating pathophysiologic principles to specific disease processes
- Correlating abnormal physical examination findings to a given disease process
- Correlating abnormal results of diagnostic tests to a given disease process

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Assumptions about designing programs for adult learners

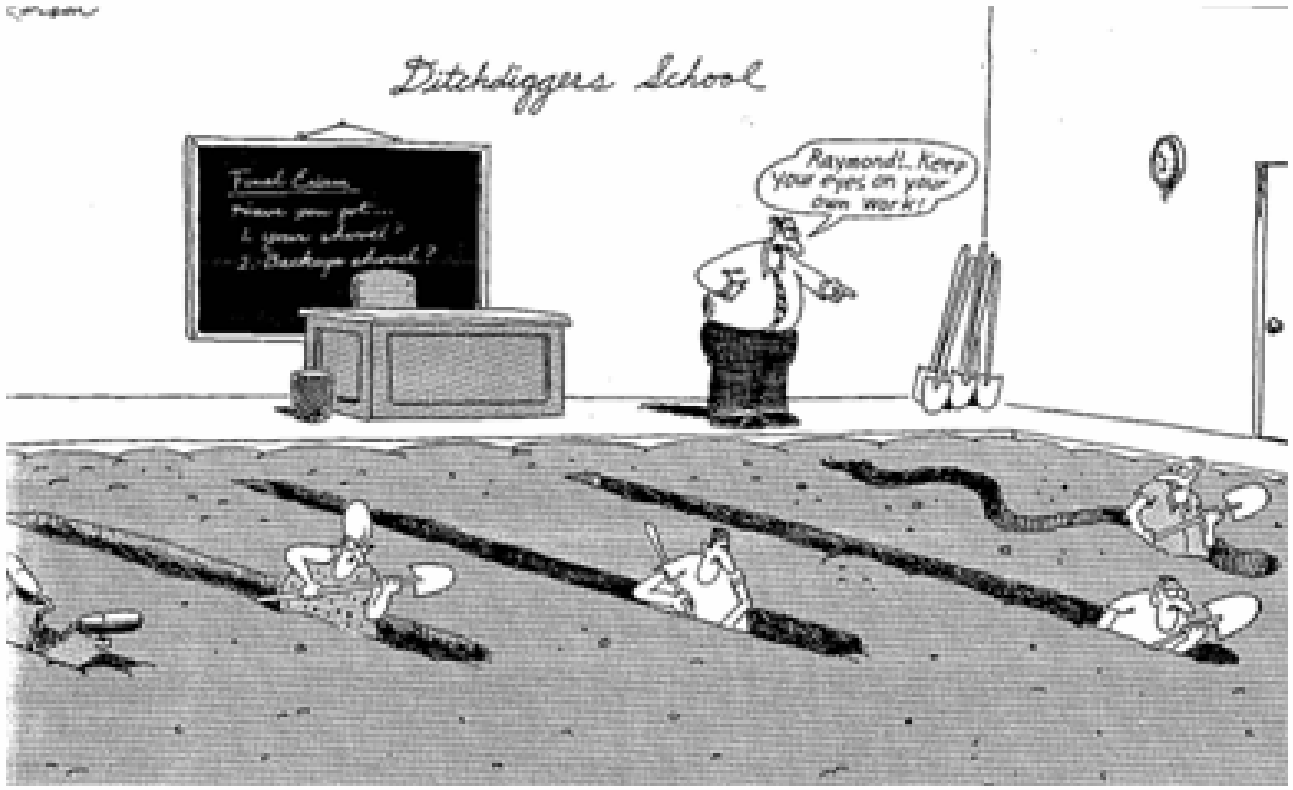
- Involve participants directly in the planning from the outset.
- Learners are a rich resource for learning; use them.
- Focus on immediate application of new knowledge.
- Use problem-centered instruction.

What works for health professionals?

- Case-based approaches, especially cases that reflect actual practice (genetics through the practitioner's lens)
- Layered content
- Relevant clinical guidelines
- Guidelines for referral
- Links to resources (people and information)

There is a difference between accurate and complete.

EVALUATION


Ditchdiggers School
Raymond!.. Keep your eyes on your own work!

Objectives for the NCHPEG/AAPA Program

When you have completed the case studies in this site, you should have an increased understanding of:

- Genetics vocabulary
- The utility of the genetic family history in practice
- Basic patterns of single-gene inheritance
- Red flags that signal a genetic contribution to disease
- Differences between the presentation of single-gene and complex disorders
- Some ethical, legal, and social issues related to the provision of genetics services

And be able to:

- Collect basic but informative family history information
- Identify patients and families who may benefit from genetic services
- Consult trusted resources about genetic contributions to speech language pathology and audiology findings
- Locate and refer to genetics professionals as necessary

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Welcome to Genetics in PA Practice

This site is designed to help you apply genetic family history information and work through genetic differential diagnoses in practice.

Click here for a video introduction.

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This site is designed to help you apply genetic family history information and work through genetic differential diagnoses in practice.

Click here for a video introduction.

Patient Name: Roberto Lopez

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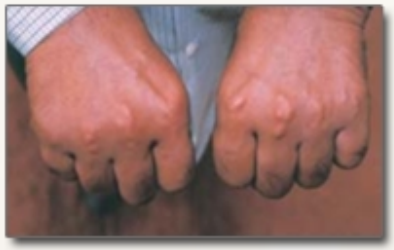
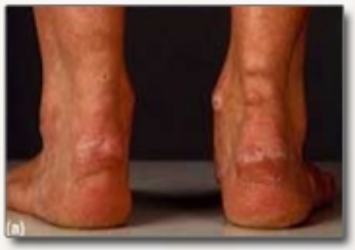
PE

BP:120/70 | HR:76 | RR:12 | Weight:148lb | Height:5'7" | BMI:23

General
Head
Eyes
Neck
Chest
Cardiovascular
Skin
Musculoskeletal
Neurological

Skin: Yellowish, soft, mobile lesions over MCP joints bilaterally.





Note:

Roberto has what appear to be Xanthomas on the extensor tendons of both hands and over his Achilles tendons, suggestive of familial hypercholesterolemia (FH). This finding in the context of his family history requires further evaluation, specifically a fasting lipid panel.

Patient Name: Roberto Lopez

Previous

FH The following family history information is provided on the intake form:

Family member	Name	Birth date	Living?/Healthy?
Mother	Maria Lopez	10/8/1950	Y/N
Father	Miguel Lopez	01/11/1950	Y/N
Sisters	Lola Lopez	06/28/1973	Y/Y
Brothers	Santos Lopez	09/03/1972	N
Son	Jose Lopez	06/15/1994	Y/Y
Daughter	Alicia Lopez	04/12/1991	Y/Y

Please place a check mark next to any condition that affects a close relative (parents, siblings, aunts/uncles, grandparents). For each check, explain who is affected below.

	Bleeding problems	X	Diabetes		Kidney problems		Sight /hearing problems
	Blood clots		Asthma		Learning disabilities		Mental retardation
X	Heart attack		Migraine	X	High blood pressure		Birth defects
	Stroke		Cancer		High cholesterol		Psychiatric disorders
	Eczema		Seizures	X	Obesity		Genetic disorders

My father had a heart attack when he was 50. My grandfather died of a heart attack. My mother and her sister have high blood pressure and my aunt has diabetes too. My uncle Manolo was found dead at home at age 40.

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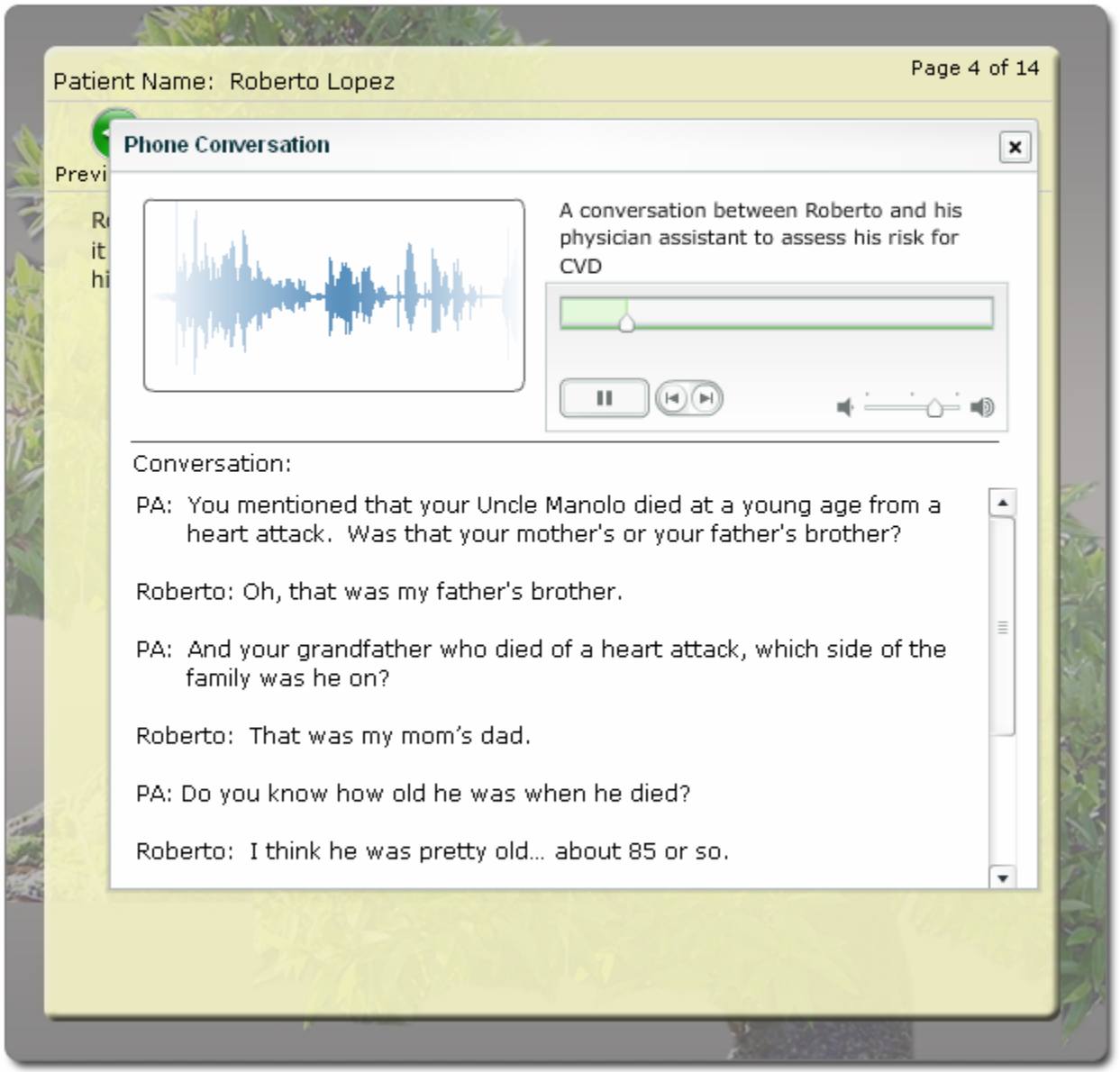

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Patient Name: Roberto Lopez

Roberto obviously has a stron
it is not his chief complaint, it
history information to assess

Talk to

Exercise 1

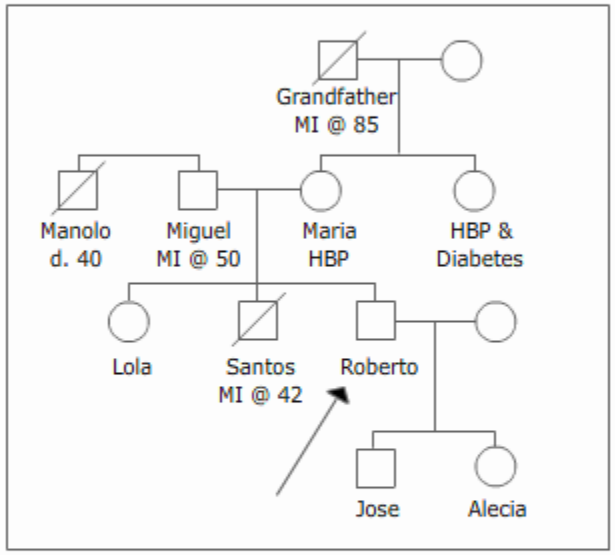
In the complete pedigree
considered "red flags" fo

1. Death from heart
2. Multiple family me
3. MI at a young age
4. Death at a young
5. Hypertension in overweight siblings.

Submit

Completed Pedigree

Minimize Window | Print Window



Genetics in PA Practice

Patient Name: Roberto Lopez

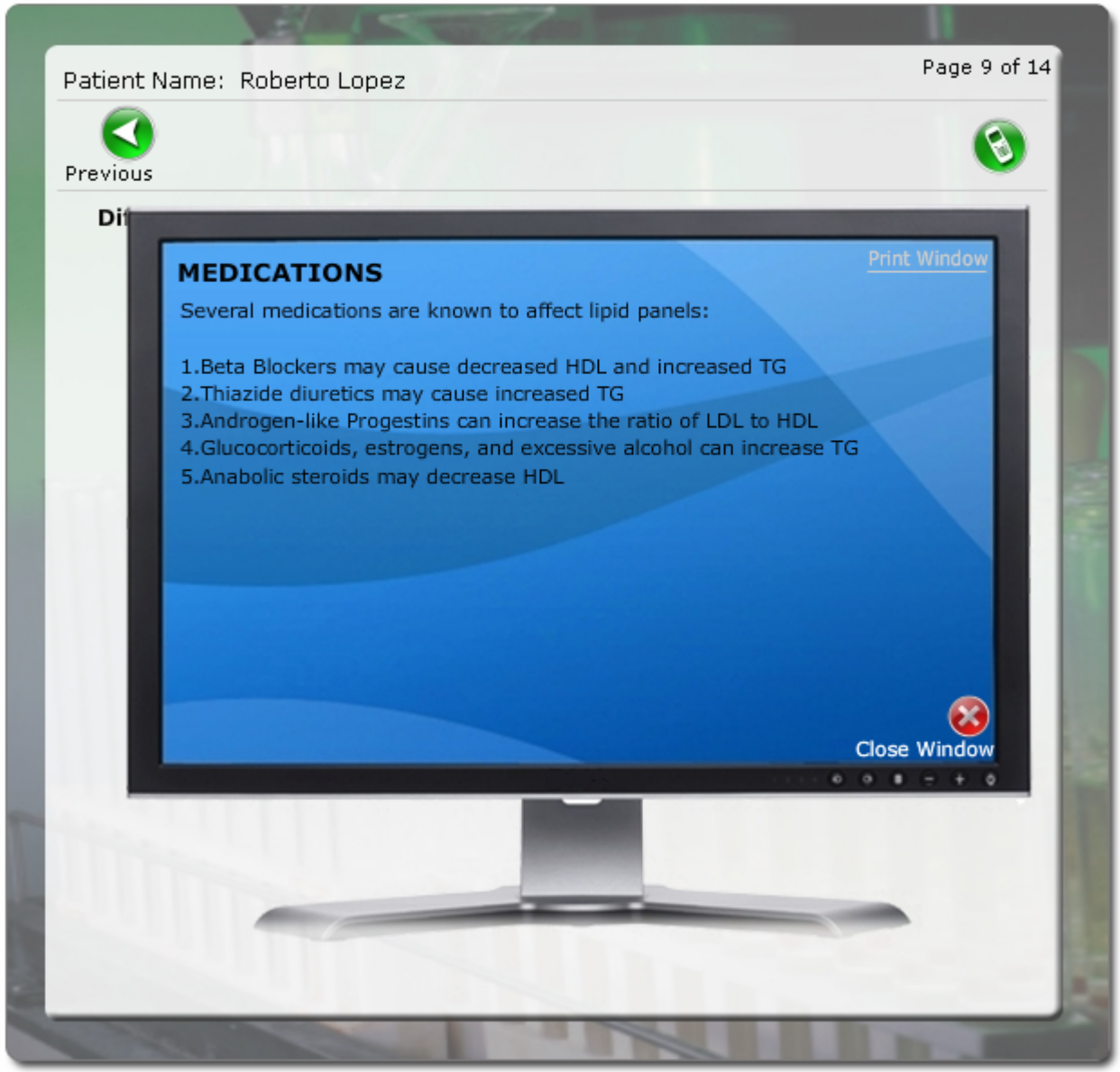
Previous

Differential diagnoses:

Based on Roberto's initial evaluation, examination, and lab results, you consider the following possible diagnoses:

- Achilles tendonitis
- Familial hypercholesterolemia
- Familial Dysbetalipoproteinemia
- Hypothyroidism (subclinical)
- Medication-induced hyperlipidemia

Genetics in PA Practice



Genetics in PA Practice

Genetic Testing

USES OF GENETIC TESTING

- Diagnostic
- Predictive
- Carrier
- Prenatal
- Preimplantation
- Newborn

TYPES OF GENETIC TESTING

- Graphic

GENETIC TESTING IN CHILDREN

- Guidelines

GENETIC TESTING TECHNOLOGY & METHODS

- FISH
- Karyotype
- PCR
- Southern Blot
- Chips & Microarrays

EVALUATING GENETIC TESTS

- Sensitivity & specificity Karyotype
- Positive predictive value
- Negative predictive value

A Modest Proposal To Help Integrate Genetics into Mainstream Health care

Stop using the terms “genetic disorder” and “genetic disease”