Physician Assistants and Genomic Medicine
September 19, 2007

Meeting Summary

The second "Physician Assistants and Genomic Medicine" meeting was held at the Natcher Conference Center of the National Institutes of Health (NIH) in Bethesda, MD on September 19, 2007. The conference was organized and supported by the National Human Genome Research Institute (NHGRI) with participation of leadership from the Accreditation Review Commission on Education for the Physician Assistant (ARC-PA), the American Academy of Physician Assistants (AAPA), the National Commission on Certification of Physician Assistants (NCCPA), and the Physician Assistant Education Association (PAEA). Representatives of the American College of Medical Genetics (ACMG), the National Coalition for Health Professional Education in Genetics (NCHPEG), the National Society of Genetic Counselors (NSGC), and the Office of the Surgeon General (OSG) were also in attendance.

The goals of this meeting were to: 1) provide an opportunity for the Physician Assistant organizations to share information regarding their activities in the arena of genetics and genomics since the March meeting, 2) identify gaps and means to close them, 3) discuss potential roles for the participating organizations in expanding the knowledge base of Physician Assistant faculty, students and graduates regarding the application of genomics to healthcare, and 4) plan next steps for all of the organizations in attendance.

The NHGRI is pleased to present this summary of the meeting proceedings. NHGRI would like to thank all of the presenters and participants for their active participation and for their thoughtful contributions that form the basis for this summary.

The body of this report summarizes each session and concludes with the next steps proposed by each of the participating organizations and recommendations from the meeting. Included also are Appendix A (meeting agenda), Appendix B (meeting participants), and Appendix C (PowerPoint presentations).

Welcome and Introductions:
The conference opened with welcoming remarks from Dr. Alan Guttmacher of NHGRI and Michael Rackover, PA-C, M.S. of Philadelphia University. Both recognized the work of the Physician Assistant (PA) organizations and others since the meeting in March. They underscored that the four PA organizations work very well together and can serve as role models to other health professional organizations.

A series of talks were then presented, with interactive discussion periods following each talk. Brief summaries of the talks and discussion points follow:

Update on the Genetic Information Non-discrimination Act (GINA) and implications for PA practice—Alan Guttmacher, M.D.
It has been a dozen years since genetic nondiscrimination legislation was first introduced in Congress. The Senate passed a similar version of GINA in both 2003 (vote of 95-0) and 2005 (vote of 98-0), but both times the House did not take action on the bill. The good news is that the House passed GINA by a vote of 420-3 (H.R. 493) in April of this year. The Senate bill (S. 358) has passed the HELP committee but has not been acted upon. It is reported that Senator Coburn of Oklahoma, and possibly one additional Senator, has a hold on the bill which keeps it from moving to a vote. There are ways to get around holds in the Senate (one is via cloture), but they may not be viable options at this time. An additional option is to attach GINA to another bill that will probably be passed, but there are possible problems with this approach, as well. It is fairly clear that if GINA actually comes to a vote, it would pass and the President would sign it.

If GINA is not brought to a vote before the end of the year, it could be brought up again next year; however, since next year is an election year, it may be difficult to move it then.

Discussion:
<u>What is the impact of GINA on health care providers?</u>
- There are few cases of genetic discrimination so far, but it may happen more in the future as technology and genetic applications to health advance. People already commonly cite genetic discrimination as a reason that they will not participate in scientific research. Without protective legislation, this may also have an increasing effect on patients and their willingness to have genetic testing.
- GINA's aim is not to curtail the insurance companies, but rather to curtail fears of the public. If the bill is passed, health care providers will need to allay the fears of the public regarding genetic discrimination to assure them that it is now safer to participate in scientific research as well as have genetic testing.

<u>What is the effect of concern about genetic discrimination in health care and employment on Genetic Counseling?</u>
- There is not much of an impact in the prenatal setting, but definitely in the adult medicine setting. Passage of the bill would allow Genetic Counselors to reassure their patients about genetic discrimination.

Update on Genome-Wide Association Studies (GWAS) — Teri Manolio, M.D., Ph.D.
Since 2005, over 30 genome-wide association studies have identified robust associations with genetic variants for nearly 20 complex diseases and traits, including age-related macular

degeneration, QT interval prolongation, neovascular AMD, inflammatory bowel disease, Type 2 diabetes, Crohn's disease, and obesity. This year, researchers have consistently replicated associations found for celiac disease, colorectal cancer, childhood asthma, multiple sclerosis, and many more.

A GWA study is a method for interrogating all 10 million variable points (single nucleotide polymorphisms – or "SNPs") across the human genome. Variation tends to be inherited in groups of DNA, or blocks, so not all 10 million points have to be tested. Technology now allows studies to use ~300,000 - 500,000 markers to represent the entire human genome adequately. Progress in genotyping technology has decreased the cost considerably in the past few years. The cost of genotyping 300,000 – 500,000, or even more, markers is now about $500 per person. For example, performing a genome-wide association study in 2,000 people cost about $20 billion in 2001; in 2007, the same study would cost about $1 million (due to decrease in number of SNPS needed as well as cost per SNP).

GWA studies have provided a tsunami of data. The GWA approach is unique since it permits examination of inherited genetic variability at unprecedented level of resolution and it permits "agnostic" genome-wide comparison. Most robust associations found in GWA studies have not been with genes previously suspected of being related to the disease, and some associations have been found in regions not even known to harbor genes.

NIH currently hosts two databases that store GWA data: the Database of Genotype and Phenotype (dbGAP), run by the National Center for Biotechnology Information; and the Cancer Biomedical Information Grid (caBIG), run by the National Cancer Institute.

Discussion:

<u>How can a PA use risk information in practice?</u>

This information can be best used to convey risk reduction information, rather than predictive information. People's behaviors may change based on this information, and we may be able to tailor this information for families/individuals at some point in the future.

If prostate cancer genes do indeed have some significant effects together - could you imagine doing a genetic test as a precursor to a PSA? Yes, that might be one way it could be used, or perhaps it would prove even more helpful post-PSA. PSA testing is currently very grey…could this new information help change that? Yes.

Many of the pathways that GWAS points us to will become targets for drug design.

<u>How do you get physicians to agree that they need to know this information?</u>

Physicians are action oriented; therefore, it is important for educators/speakers to share examples that have actual results. We are still on the hunt for effective examples, since this research currently has few practical applications yet.

Update from the Accreditation Review Commission on Education for the Physician Assistant — Laura Stuetzer, MS, PA-C

The ARC-PA protects the interests of the public, including current and prospective PA students, and the PA profession by defining the standards for PA education and evaluating PA educational programs within the territorial United States to ensure their compliance with those standards.

The ARC-PA has been in existence as a free standing organization since 2001. There are currently 139 accredited PA programs in the US (entry level PA programs). There are currently no accredited residency programs for PA education. The last accreditation meeting was held a few weeks ago, and three new programs were added to the accreditation list.

The ARC-PA develops and maintains the standards for the profession, which are competency based. The standards do not prescribe a specific degree or method for meeting the standards. The standards include a requirement for instruction in basic medical science, to include genetic and molecular mechanisms of health and disease. The standards require curriculum to include core knowledge about the established and evolving biomedical and clinical sciences and the application of this knowledge to patient care. The standards also require the curriculum to be of sufficient breadth and depth to prepare the student for the clinical practice of medicine.

During the accreditation review process, programs are cited when they do not rigorously meet the required standards. A citation includes a letter to the Dean of the program's institution, and the program is required to report back to the ARC-PA within a certain timeframe explaining how they are addressing the issue. Citation topics are used to develop workshops throughout the year.

Update from the National Commission on Certification of Physician Assistants — William Kohlhepp, MHA, PA-C

Activities regarding genetics/genomics at the NCCPA have taken place in the following three areas since the March meeting:

1) Exam content -

Though genetics is not featured explicitly as a subcategory on NCCPA's exams, questions regarding genetics-related issues are included in the exam in several areas. The next Practice Analysis will be conducted in 2009-2011, which will offer an opportunity to ask new and more explicit questions about what PAs are doing in the areas of genetics and genomics. In turn, that will position NCCPA to change the way those topics are incorporated into the exams. In the meantime, the NCCPA is beginning to code new items on the exam with a genetics code when applicable. NCCPA is also conducting a review of its item bank and including genetics coding in that review. A new item writer with experience in genomics will be added in 2008. Someone with genomics experience will also be included on the next Practice Analysis Committee.

2) Promotion -

NCCPA has taken steps to educate their Board about genetics by providing them with written reports in both May and August. They will also provide a more in-depth presentation to the group at their meeting in November and would welcome a talk by either Francis Collins or Alan Guttmacher at that Board meeting. NCCPA is also promoting genomics through other ongoing projects. The NCCPA Foundation is promoting the U.S. Surgeon General's Family History Initiative through their website.

3) Research -

The NCCPA Foundation has set aside research funds for their next RFP to be released in January. Genetics/genomics could be included as a topic of interest in the RFP.

Update from the Physician Assistant Education Association — Anita Duhl Glicken, MSW
In follow-up to the March meeting, PAEA published a summary of the meeting in the PAEA Networker. They also confirmed Dr. Collins' presentation, "Physician Assistants and Personalized Medicine" at the PAEA Annual Education Forum in October 2007.

Over the summer, results from the PAEA member program genetics survey were published in the *Journal of Physician Assistant Education* (vol. 18, no. 2, July 2007). Additionally, an editorial by Bruce Korf was published in the same journal edition.

In the fall, PAEA coordinated the "Put a Face on Genetics Campaign" for the 2007 PAEA education forum. They also announced the launch of the NCHPEG PA Website for educators. Additionally, a workshop presentation by Rackover and Healy has been scheduled entitled "Developing Instruction in Genetics and Genomics."

Future activities include the push to use the PA profession as a lab for educating health care providers regarding genetics. Evaluation and outcomes will be important in developing educational/curriculum resources (faculty development, curriculum resources, resource sharing).

***Update from the American Academy of Physician Assistants — Robert McNellis,* P.P.H., PA-C**
The AAPA has engaged their membership in genetics/genomics activities in several ways over the past six months. At their 2007 annual conference, AAPA conducted a survey of its House of Delegates that included genetic-oriented questions (see "Presentation and discussion of PA survey results," below) and provided exhibit hall space for NCHPEG and OSG. At least eight hours of continuing medical education offered at the conference included information on medical genetics. AAPA has included several articles focused on genetics in its newsletter over the past six months, in addition to adding genomics information on its website. AAPA has promoted the NCHPEG web-based CME to its membership and will continue to do so throughout the year. To date, the web-based CME has received nearly 3,000 visits with 77 PAs having completed Case #1, 59 PAs having completed Case #2 and 51 PAs having completed Case #3.

In preparation for its 2008 annual conference, AAPA plans to work with NCHPEG to develop a medical genetics track that will provide a daily session with key genetic content. A session on race and genetics is also being planned (co-sponsored by AAPA's African Heritage Caucus, its Committee on Diversity, and NCHPEG).

Other activities include the continuation of Doug Scott's "Genomics series" in AAPA News, the addition of pertinent survey questions in the Annual Conference Survey (over 2000 respondents), development of a needs assessment tool, review of AAPA policy statements, engaging the *JAAPA* editorial board, and development of expanded relationships with other genetics organizations, advisory committees, etc. AAPA has also been involved in legislation, policy, and partnership activities regarding genomics.

NHGRI activity update — Greg Feero, M.D., Ph.D.
NHGRI has been involved in many new activities since the meeting in March, including:

1) Personalized Healthcare Workgroup (part of the American Health Information Community). The goal is to bring some standardization to how Electronic Health Record systems deal with family history and genetic test information, and to facilitate clinical decision support.

2) Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics. To date, 48 nursing groups have endorsed the competencies. A meeting was held last week to discuss how to build a toolkit for nursing faculty.

3) "Have you seen Mary…or Fred…or Bob?" Five draft poster-format messages have been developed to target PA's focus on family history. The posters address hereditary breast and ovarian cancer syndrome, diabetes, coronary artery disease, HNPCC, preconception care (Tay-Sachs). Greg Feero welcomes feedback on the posters' content and format. He also welcomes any thoughts regarding appropriate venues for distribution.

4) Top 12 list: How do we move forward with these topics? Who are the targets, what is the distribution venue? Who will further populate the list? The group discussed the list and agreed it would be best reformulated to again be a "top 10" list.

Other tools and areas to consider:
- Poster presented at NCHPEG regarding building a model curriculum for family medicine residency education
- eDoctoring Website for primary care students, residents, practitioners.
- PDA support
- Virtual clinic tool
- Article series
- Video CME

Presentation and discussion of PA survey results — Michael Rackover, PA-C, M.S., and Robert McNellis, P.P.H., PA-C

A survey of the AAPA's 2007 House of Delegates was conducted in May 2007. The 19-question survey (3.5 minutes to complete) was handed out on the last day of the three-day meeting. Murugu Manickam, a Medical Genetics Fellow on rotation at NHGRI, analyzed the relevant data from the 113 surveys returned. A brief summary of the results (see attached PowerPoint for more detailed data) is as follows:

Of PAs surveyed, most believe genetics is important. A majority of the respondents were not aware of NCHPEG. The PAs surveyed are open to a variety of educational media : print, electronic, or short in-person conferences. Paper is still widely used for tools in practice.

The group suggested exploring the topics below for inclusion in next year's survey:
- View of genetic counseling
- How often do you take a three-generation pedigree?
- How often do you use family history?
- How do you document family history?
- If your patients brought you a print-out from a standardized family history software, would you use it?
- How often do your patients bring a question to you regarding genetics?

There was also discussion of possibly holding focus groups focused on the vignettes at the meeting.

A quick tour of ClinSeq and Multiplex as examples of NIH translational research — Greg Feero, M.D., Ph.D., Jean Jenkins, R.N., Ph.D., Michael Rackover, M.S., PA-C
Genomic discoveries relevant to common disease diagnosis and management are coming at an increasing rate, and basic discoveries are leading to the development of clinical application; however, there is a gap between developing clinical applications and improved healthcare. Bench scientists, clinical scientists, clinicians, and patients all need to work together to fill the gap.

NGHRI currently funds these three major projects that address translational questions:
1) The Multiplex Project is designed to develop a prototype for multiplex genetic susceptibility testing (how to consent and how to provide feedback and support) and to create an infrastructure to facilitate public health research.

2) ClinSeq is a translational research project in clinical genetics. The aims of ClinSeq are to develop a robust infrastructure for the generation and use of large-scale medical sequencing (LSMS) in a clinical research setting, to use LSMS data to develop novel approaches to clinical biomedical research, and to understand how to interact with subjects re LSMS.

3) The Health Professionals' Understanding of Human Genetic Variation Study (PUHGV) aims to investigate health professionals' knowledge of human genetic variation and their beliefs about biological and genetic differences based upon their patients' race and ethnicity and the impact of these on clinical practice. Vence Bonham is interested in involving the PA community in this survey.

Translation of genomic discoveries to primary care – A role for the PA?— Moderated discussion
Is adult medicine ready for the transition of care from pediatric medicine to adult medicine for those with single gene disorders? This could be the focus of a research project.

We need one or two good examples of that will become the standard of care as a result of genomic medicine. One example is Warfarin and the change in its labeling by the FDA to allude to the fact that genotyping of two genes provides an important guide for selection of dosing. Genetic tests are now being offered associated with this.

We need an opportunity to engage the PA community in family history. The idea of patient use of the Surgeon General's My Family Health Portrait Tool in advance of clinic visit needs to be instituted.

It would be useful to collect real world examples from PAs regarding genetics and patients they have seen in practice.

Soon, there will be a massive marketing program by 23andMe and Navigenics to market the opportunity for members of the public to have their genomes sequenced. As people begin to take advantage of these offers, they may bring the results to their health care providers for guidance.

The nursing community is hoping to hold a state of the science conference regarding integration of genetics/genomics into nursing care and whether or not this actually makes a difference to outcomes of care.

NHGRI is working with the NIH Office of Medical Applications of Research (OMAR) to organize a state of the science conference on use of family history as a screening tool in primary care. The planning committee is currently being formed.

A baseline survey could be conducted at the 2008 AAPA meeting to get information from workshop attendees. A more in-depth follow-up could involve a chart review to see if family history data is being collected.

Three is a need to build infrastructure and partnerships within PA organizations to do research. In coming years, it will be critical to engage PAs in research in order to learn if what we are doing is accomplishing its goal. The establishment of a research oversight group will be added to the agenda for the four-PA organization meeting in December.

What if NHGRI decided to find out how the family history approach using a standardized tool operates in practice, and could offer ~$100,000 in funding? Is there a structure within the PA community that could respond to such an RFP? Yes, PA organizations have collaborated on other federal contracts/grants in the past. An opportunity like that could allow the PA organizations to form a research group.

Develop a toolkit for PA instructors to use to teach genetics?

Discussion of next steps — Francis Collins, M.D., Ph.D.

Each of the four PA organizations presented their plans for the future:

- *Accreditation Review Commission on Education for the Physician Assistant - Laura Stuetzer, MS, PA-C*
  Laura would like to see ARC-PA look at the information it is collecting from the accredited programs to learn how they are integrating genomics. ARC-PA could work with PAEA and, possibly, receive funds from NCCPA to accomplish this. ARC-PA can also tally up the citations issued regarding the new standard to learn where the programs need improvement.

- *National Commission on Certification of Physician Assistants – William Kohlhepp, MHA, PA-C*
  NCCPA plans to continue review and coding of their exam questions to identify those that address genetics or genomics issues, add a member with genetics expertise to their test writing committees and to the next Practice Analysis Committee, as well as continue to educate their board members about work in this area with a presentation from Alan Guttmacher about genetics and genomics at their November 2007 Board meeting. The NCCPA Foundation will release a call for research proposals at the beginning of next year seeded with genomics topics.

- *Physician Assistant Education Association – Timi Agar Barwick*
  PAEA plans to move forward with possible development of a genetics/genomics evaluation tied to their annual conference, as well as an evaluation of tools used in the classroom to teach genetics/genomics concepts. They will work to identify key contacts in other health care professions to begin a dialogue regarding genetics/genomics. They will also work to further the dissemination of information regarding the importance of family history in care.

- *American Academy of Physician Assistants – Robert McNellis,* P.P.H., PA-C *and Lawrence Herman* MPA, RPA-C
  AAPA plans to institutionalize genetics/genomics activities within their organization. It will continue to include genomics articles in its publications, enhance the genomics information on its website, as well as work with *JAAPA* to get them more interested in the topic. AAPA will move forward with making plans for genetics/genomics in its 2008 annual conference. Together with NCHPEG, it will develop a genomics track for workshops at the conference, as well as work to develop CME lectures on the Top 10. It will also develop genetics/genomics focused questions for the conference survey. At the December meeting of the four-PA organizations, it will support the idea of developing a research oversight group.

- *National Coalition for Health Professional Education in Genetics - Joseph McInerney, MA, MS*
  NCHPEG plans to work with AAPA to develop a genomics track for the 2008 annual conference in San Antonio. Possible topics include the genetics of common disease, ELSI issues, race and genetics, psychiatric genetics, and family history. It will work with PAEA to develop a "train the trainer" workshop to be held at the 2008 PAEA Annual meeting. It will also begin development of a tool regarding colorectal cancer for the Department of Veterans Affairs and look for opportunities to use this to benefit PAs.

- *National Society of Genetic Counselors - Catherine Wicklund*
  The NSGC offers to have its 2,300 members interact with the PA community through lectures, consultations, etc. It could also gather volunteers to attend the AAPA annual meeting to interact with PAs through workshops, mock counseling sessions, and the "student bowl." It can also help to tackle the barriers preventing PA referral to genetic counselors.

- *American College of Medical Genetics -Michael Watson, Ph.D., FACMG*
  Mike Watson suggests that the PA groups need to bring specificity to what they do and don't know about genetics. Visibility of genetics/genomics can be increased by adding questions that include genetic concepts as "distractors" on the exam ACMG is able to interact with the PA organizations by providing genetics experts to attend annual meetings, sharing educational resources, and developing more "tele-health" opportunities for genetics services.

Closing comments — Francis Collins, M.D., Ph.D.
Opportunities for genetic testing and sequencing are moving forward at a rapid pace. We need to prepare for the time when every day applications of genomics begin to show up in practice.

NHGRI is serious about the possibility of providing a research opportunity for data gathering about the effectiveness of the use of the U.S. Surgeon General's My Family Health Portrait in practice.

We will plan to re-convene this group in twelve months. We will hold a teleconference of the group in about six months to catch up on ongoing activities.

Action items from our discussions today include:
1) NHGRI will update the PA organizations and other participants regarding plans to promote family history this Thanksgiving.

2) NHGRI will distribute electronic versions of the "Have you seen Mary?" campaign posters to the group for feedback. Group members are asked to pay specific attention to the diabetes poster, as it may be used as a promotional item for this Thanksgiving's National Family History Day.
3) NHGRI, with help from others, will further refine the Top 10 list.
4) NHGRI will share a list of contacts from other disciplines with the group.
5) NHGRI will distribute Vence Bonham's e-mail address to the group, for contact regarding the Health Professionals' Understanding of Human Genetic Variation Study: bonhamv@mail.nih.gov
6) Group members are asked to feed any genetics/genomics story ideas to Doug Scott of AAPA at dscott@aapa.org.

APPENDIX A

Physician Assistants and Genomic Medicine
September 19, 2007

Natcher Conference Center, C1/C2
National Institutes of Health, Bethesda, MD

Goals:

1) Share activities to date
2) Identify gaps and means to close them
3) Discuss potential roles for the Physician Assistant Organizations in expanding the knowledge base regarding the application of genomics to healthcare
4) Plan next steps

AGENDA

8:00 am	**Refreshments**
8:30	**Welcome and introductions** Michael Rackover and Alan Guttmacher
8:45	**Update on the Genetic Information Non-discrimination Act (GINA) and implications for PA practice** Alan Guttmacher
9:00	**Update on Genome-Wide Association Studies (GWAS)** Teri Manolio
9:30	**Discussion**
9:50	**Break**
10:05	**PA organization activity updates (15 minutes each)**
11:05	**Discussion**
11:25	**NHGRI activity update** Greg Feero
11:55	**Discussion**
12:15 pm	**Lunch** (From cafeteria)
12:30	**Working lunch: Presentation and discussion of PA survey results** Michael Rackover and Bob McNellis
1:15	**A quick tour of ClinSeq and Multiplex as examples of NIH translational research** Greg Feero
1:30	**Translation of genomic discoveries to primary care – A role for the PA?** Moderated discussion: Greg Feero, Jean Jenkins and Michael Rackover
2:30	**Discussion of next steps** (5-10 minutes each PA organization, NSGC, ACMG, NHGRI) Francis Collins
3:30	**Adjourn**

APPENDIX B

Physician Assistants and Genomic Medicine
September 19, 2007

National Institutes of Health, Bethesda, MD

PARTICIPANT LIST

Timi Agar Barwick
Executive Director
Physician Assistant Education Association

Mary Beth Bigley, Dr.Ph, MSN, ANP-C
Senior Health Fellow
Office of the US Surgeon General

Francis S. Collins, M.D., Ph.D
Director
National Human Genome Research Institute

Ragan Cohn
Managing Director
National Commission on Certification of Physician Assistants Foundation

Greg Feero, M.D., Ph.D.
Senior Advisor to the Director for Genomic Medicine
Chief, Genomic Healthcare Branch
National Human Genome Research Institute

Anita Duhl Glicken, MSW
President, Physician Assistant Education Association
Professor of Pediatrics
Interim Director, CHA/PA Program
University of Colorado Health Sciences Center

Alan Guttmacher, M.D.
Deputy Director
National Human Genome Research Institute

Lawrence Herman, MPA, RPA-C
Clinical and Scientific Affairs Council
American Academy of Physician Assistants

Jean Jenkins, Ph.D., RN, FAAN
Senior Clinical Advisor to the Director
National Human Genome Research Institute

William Kohlhepp, MHA, PA-C
Immediate Past Chairman
National Commission on Certification of Physician Assistants

Teri Manolio, M.D., Ph.D.
Senior Advisor to the Director for Population Genomics
National Human Genome Research Institute

Joseph McInerney, M.A., M.S.
Executive Director
National Coalition for Health Professional Education in Genetics

Robert J. McNellis, P.P.H., PA-C
Director, Science and Education
American Academy of Physician Assistants

Michael Rackover, PA-C, M.S.
Program Director & Associate Professor
Physician Assistant Program
Philadelphia University

Laura J. Stuetzer, MS, PA-C
Associate Director, Accreditation Review
Commission on Education for the Physician Assistant

Michael S. Watson, PhD, FACMG
Executive Director
American College of Medical Genetics
American College of Medical Genetics Foundation

Catherine A. Wicklund
President, National Society of Genetic Counselors
Associate Director, Northwestern University Graduate Program in Genetic Counseling

APPENDIX C

Update on Genome-Wide Association Studies: We Live in Interesting Times

U.S. Department of Health and Human Services
National Institutes of Health
National Human Genome Research Institute

Teri A. Manolio, M.D., Ph.D.
Director, Office of Population Genomics
Senior Advisor to the Director, NHGRI,
for Population Genomics

September 19, 2007

We Live in Interesting Times…

"'May he live in interesting times.' Like it or not we live in interesting times."

--Robert Kennedy, June 7, 1966

May you come to the attention of those in authority.

May you find what you are looking for.

Wikipedia, accessed 11Sep07

May You Live in Interesting Times…

Since 2005, over 30 genome-wide association studies have identified robust associations with genetic variants for nearly 20 common, complex diseases and traits:

- 10 Mar 2005: Age-related macular degeneration
- 30 Apr 2006: QT interval prolongation
- 19 Oct 2006: Neovascular AMD
- 26 Oct 2006: Inflammatory bowel disease
- 11 Feb 2007: Type 2 diabetes
- 5 Mar 2007: Crohn's disease
- 12 Apr 2007: Obesity

Genome-wide association study of prostate cancer identifies a second risk locus at 8q24

Meredith Yeager[1,2], Nick Orr[3], Richard B Hayes[2], Kevin B Jacobs[4], Peter Kraft[5], Sholom Wacholder[2], Mark J Minichiello[6], Paul Fearnhead[7], Kai Yu[2], Nilanjan Chatterjee[2], Zhaoming Wang[1,2], Robert Welch[1,2], Brian J Staats[1,2], Eugenia E Calle[8], Heather Spencer Feigelson[8], Michael J Thun[8], Carmen Rodriguez[8], Demetri
Walter
Edward
David J

Multiple regions within 8q24 independently affect risk for prostate cancer

Christopher A Haiman[1], Nick Patterson[2], Matthew L Freedman[2,3], Simon R Myers[2], Malcolm C Pike[1], Alicja Waliszewska[2,4,5], Julie Neubauer[2,4], Arti Tandon[2,4], Christine Schirmer[2,4], Gavin J McDonald[2,4], Steven
David
Kathlee
Brian E

Genome-wide association study identifies a second prostate cancer susceptibility variant at 8q24

Julius Gudmundsson[1,17], Patrick Sulem[1,17], Andrei Manolescu[1,17], Laufey T Amundadottir[1,17], Daniel Gudbjartsson[1], Agnar Helgason[1], Thorunn Rafnar[1], Jon T Bergthorsson[1], Bjarni A Agnarsson[2], Adam Baker[1], Asgeir Sigurdsson[1], Kristrun R Benediktsdottir[2], Margret Jakobsdottir[1], Jianfeng Xu[3], Thorarinn Blondal[1], Jelena Kostic[1], Jielin Sun[3], Shyamali Ghosh[1], Simon N Stacey[1], Magali Mouy[1], Jona Saemundsdottir[1], Valgerdur M Backman[1], Kristleifur Kristjansson[1], Alejandro Tres[4,7], Alan W Partin[5], Marjo T Albers-Akkers[6], Javier Godino-Ivan Marcos[7], Patrick C Walsh[5], Dorine W Swinkels[8], Sebastian Navarrete[9], Sarah D Isaacs[5], Katja K Aben[10], Theresa Graif[11], John Cashy[11], Manuel Ruiz-Echarri[4],

Nature Genetics, 1Apr2007

A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants

Laura
Willi
Anne
Tianl
Andr
Craig
Thom
Gonç
Jaakk

Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels

Diab
and

Replication of Genome-Wide Association Signals in UK Samples Reveals Risk Loci for Type 2 Diabetes

Eleftheria Zeggini,[1,2*] Michael N. Weedon,[3,4*] Cecilia M. Lindgren,[1,2*] Timothy M. Frayling,[3,4*] Katherine S. Elliott,[2] Hana Lango,[3,4] Nicholas J. Timpson,[2,5] John R. B. Perry,[3,4]

Sciencexpress, 26Apr2007

A Common Allele on Chromosome 9 Associated with Coronary Heart Disease

Ruth McPherson,[1*†] Alexander Pertsemlidis,[2*] Nihan Kavaslar,[1] Alexandre Stewart,[1] Robert Roberts,[1] David R. Cox,[3] David A. Hinds,[3] Len A. Pennacchio,[4,5] Anne Tybjaerg-Hansen,[6] Aaron R. Folsom,[7] Eric Boerwinkle,[8] Helen H. Hobbs,[2,9] Jonathan C. Cohen[2,10†]

A Common Variant on Chromosome 9p21 Affects the Risk of Myocardial Infarction

Anna Helgadottir,[1*] Gudmar Thorleifsson,[1*] Andrei Manolescu,[1*] Solveig Gretarsdottir,[1] Thorarinn Blondal,[1] Aslaug Jonasdottir,[1] Adalbjorg Jonasdottir,[1] Asgeir Sigurdsson,[1] Adam Baker,[1] Arnar Palsson,[1] Gisli Masson,[1] Daniel F. Gudbjartsson,[1] Kristinn P. Magnusson,[1] Karl Andersen,[2] Allan I. Levey,[3] Valgerdur M. Backman,[1] Sigurborg Matthiasdottir,[1] Thorbjorg Jonsdottir,[1] Stefan Palsson,[1] Helga Einarsdottir,[1] Steinunn Gunnarsdottir,[1]

Sciencexpress, 3May2007

Genome-wide association study identifies novel breast cancer susceptibility loci

Dougla
Dennis
Shahar
Christo
Suleep
Hui-Ch
Sheila
Børge
Jolanta
Daehe

A genome-wide association study identifies alleles in *FGFR2* associated with risk of sporadic postmenopausal breast cancer

David J
Sholom
Nilanja
Saundra
Richard
Gilles T

Common variants on chromosomes 2q35 and 16q12 confer susceptibility to estrogen receptor–positive breast cancer

Simon N Stacey[1], Andrei Manolescu[1], Patrick Sulem[1], Thorunn Rafnar[1], Julius Gudmundsson[1], Sigurjon A Gudjonsson[1], Gisli Masson[1], Margret Jakobsdottir[1], Steinunn Thorlacius[1], Agnar Helgason[1], Katja K Aben[2,3], Luc J Strobbe[4], Marjo T Albers-Akkers[5], Dorine W Swinkels[3], Brian E Henderson[6], Laurence N Kolonel[7], Loic Le Marchand[7], Esther Millastre[8], Raquel Andres[8], Javier Godino[9], Maria Dolores Garcia-Prats[10], Eduardo Polo[11], Alejandro Tres[8], Magali Mouy[1], Jona Saemundsdottir[1], Valgerdur M Backman[1], Larus Gudmundsson[1], Kristleifur Kristjansson[1], Jon T Bergthorsson[1], Jelena Kostic[1], Michael L Frigge[1], Frank Geller[1], Daniel Gudbjartsson[1], Helgi Sigurdsson[12], Thora Jonsdottir[12],

Nature and *Nature Genetics*, 27May2007

Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls

The W

Sequence variants in the autophagy gene *IRGM* and multiple other replicating loci cont

susc

Miles Pa
Mark Tr
Roland

We followed up on 37 SNPs from 31 distinct loci, associated at $P < 10^{-5}$ on initial analysis of the WTCCC data set. Support for some of these markers diminished in the final WTCCC analysis after extensive data filtering[5]. We selected two markers for each locus where low linkage disequilibrium (LD) between associated SNPs in

Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes

John A Todd[1], Neil M Walker[1,9], Jason D Cooper[1,9], Deborah J Smyth[1,9], Kate Downes[1], Vincent Plagnol[1], Rebecca Bailey[1], Sergey Nejentsev[1], Sarah F Field[1], Felicity Payne[1], Christopher E Lowe[1], Jeffrey S Szeszko[1], Jason P Hafler[1], Lauren Zeitels[1], Jennie H M Yang[1], Adrian Vella[1,8], Sarah Nutland[1], Helen E Stevens[1], Helen Schuilenburg[1], Gillian Coleman[1], Meeta Maisuria[1], William Meadows[1], Luc J Smink[1], Barry Healy[1], Oliver S Burren[1], Alex A C Lam[1], Nigel R Ovington[1], James Allen[1], Ellen Adlem[1], Hin-Tak Leung[1], Chris Wallace[2], Joanna M M Howson[1], Cristian Guja[3], Constantin Ionescu-Tîrgovişte[3], Genetics of Type 1 Diabetes in Finland[4], Matthew J Simmonds[5], Joanne M Heward[5], Stephen C L Gough[5], The Wellcome Trust Case Control Consortium[6], David B Dunger[7], Linda S Wicker[1] & David G Clayton[1]

Nature and *Nature Genetics*, 7Jun2007

2007: The Year of GWA Studies?

Consistently replicated associations found for:

- 10 Jun 2007: Celiac disease
- 1 Jul 2007: Atrial fibrillation
- 8 Jul 2007 : Colorectal cancer
- 15 Jul 2007: Gallstones
- 18 Jul 2007: Periodic limb movements in sleep
- 19 Jul 2007: HIV viral setpoint
- 26 Jul 2007: Childhood asthma
- 29 Jul 2007: Multiple sclerosis
- 1 Aug 2007: Amyotrophic Lateral Sclerosis
- 9 Aug 2007: Exfoliation glaucoma
- 2 Sep 2007: Height
- 5 Sep 2007: Rheumatoid arthritis
- 18 Sep 2007: ??

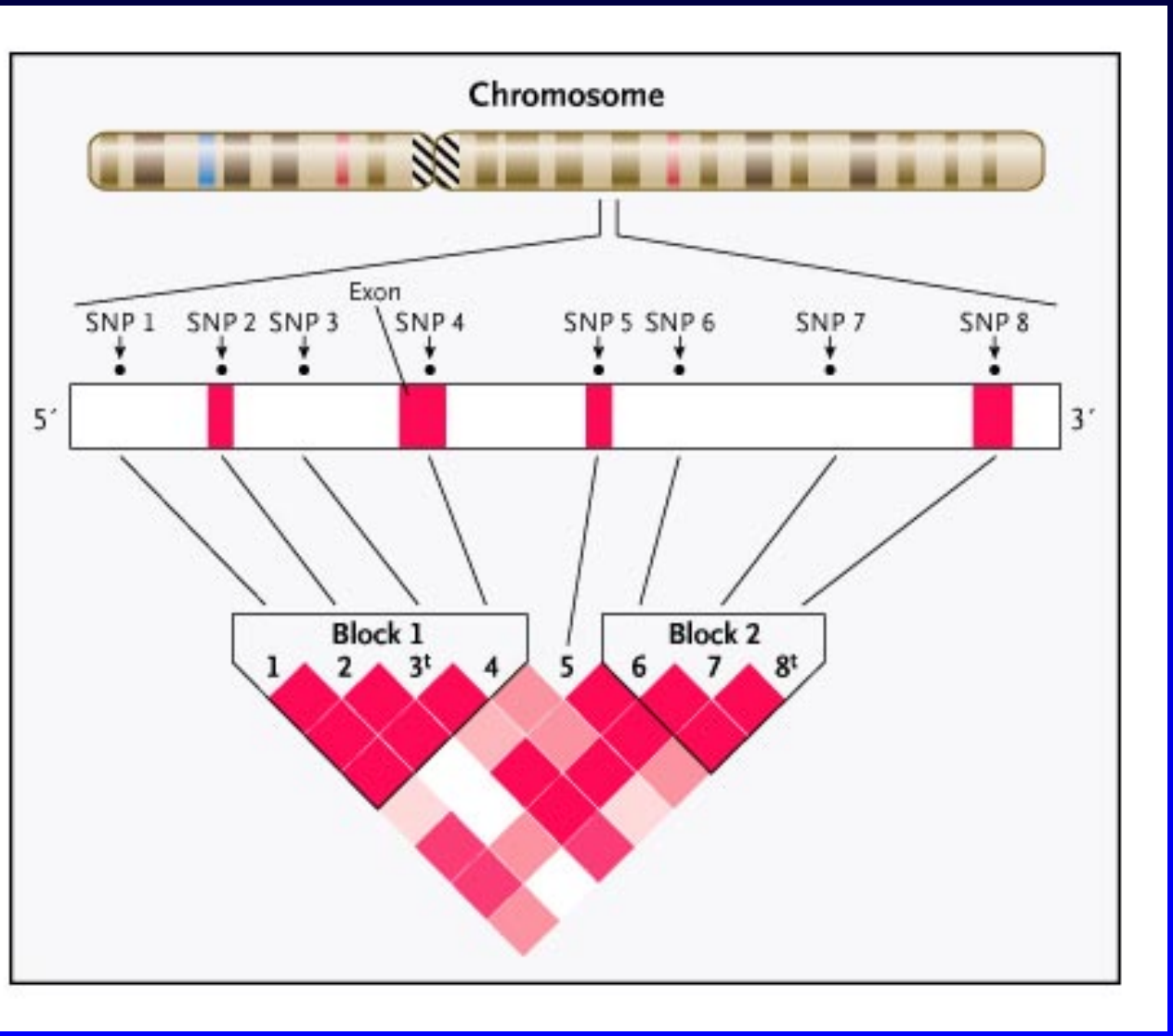
What is a GWA Study?

- Method for interrogating all 10 million variable points across human genome
- Variation inherited in groups, or blocks, so not all 10 million points have to be tested
- Blocks are shorter (so need to test more points) the less closely people are related
- Technology now allows studies in unrelated persons, assuming ~10,000 base pair lengths in common (300,000 - 500,000 markers)

DNA on Chromosome 7

SNPs 1 / 300 bases

Mapping the Relationships Among SNPs



Christensen and Murray, *N Engl J Med* 2007; 356:1094-1097.

Distances Among East Coast Cities

	Boston	Provi-dence	New York	Phila-delphia	Balti-more
Providence	59				
New York	210	152			
Philadelphia	320	237	86		
Baltimore	430	325	173	87	
Washington	450	358	206	120	34

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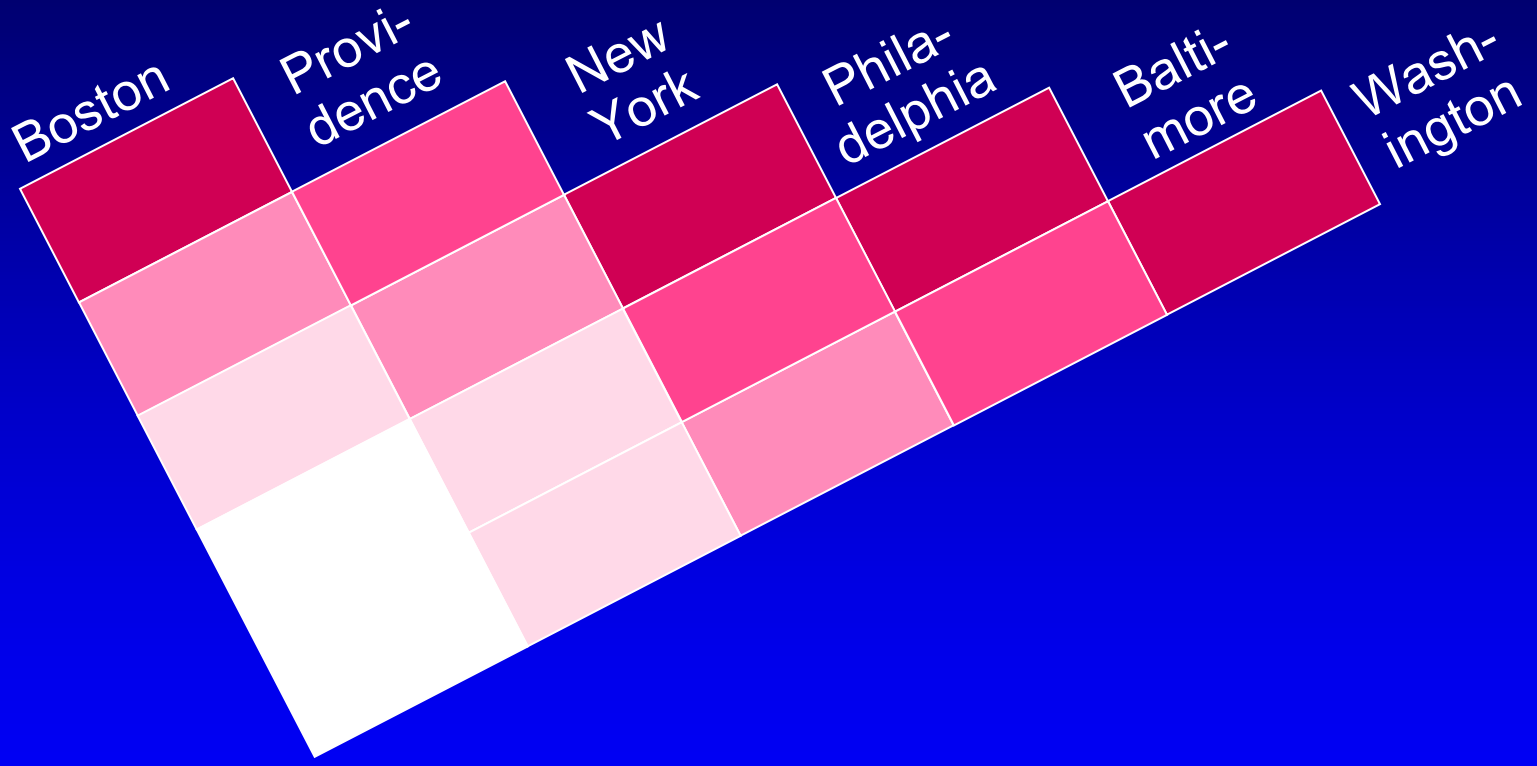
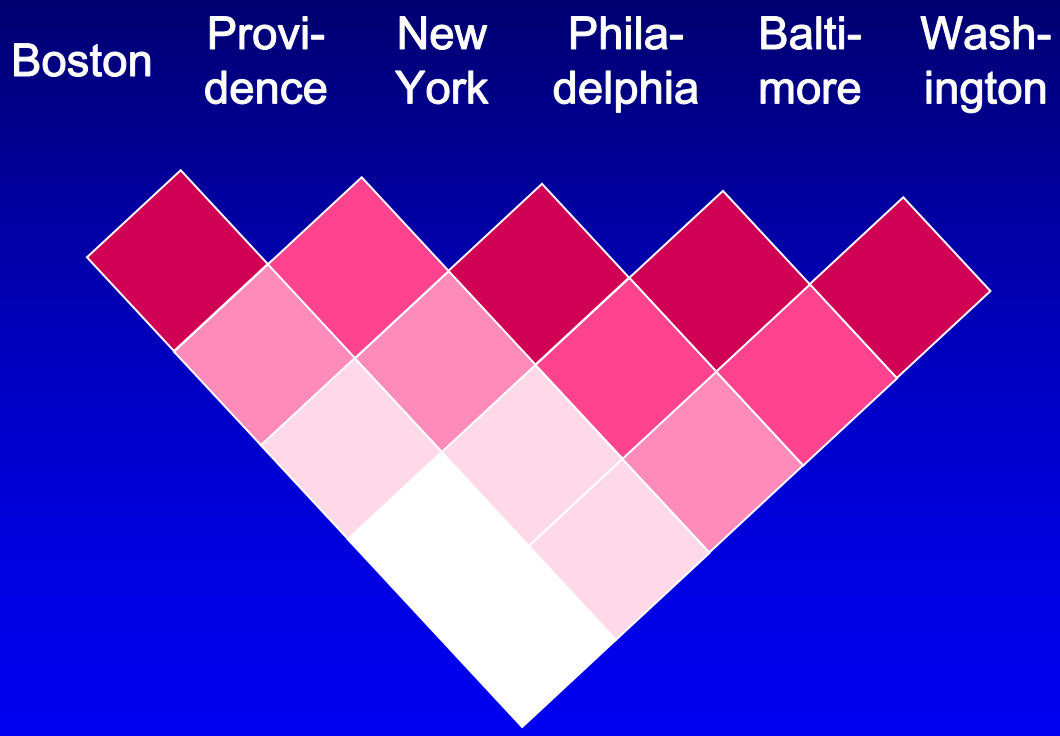
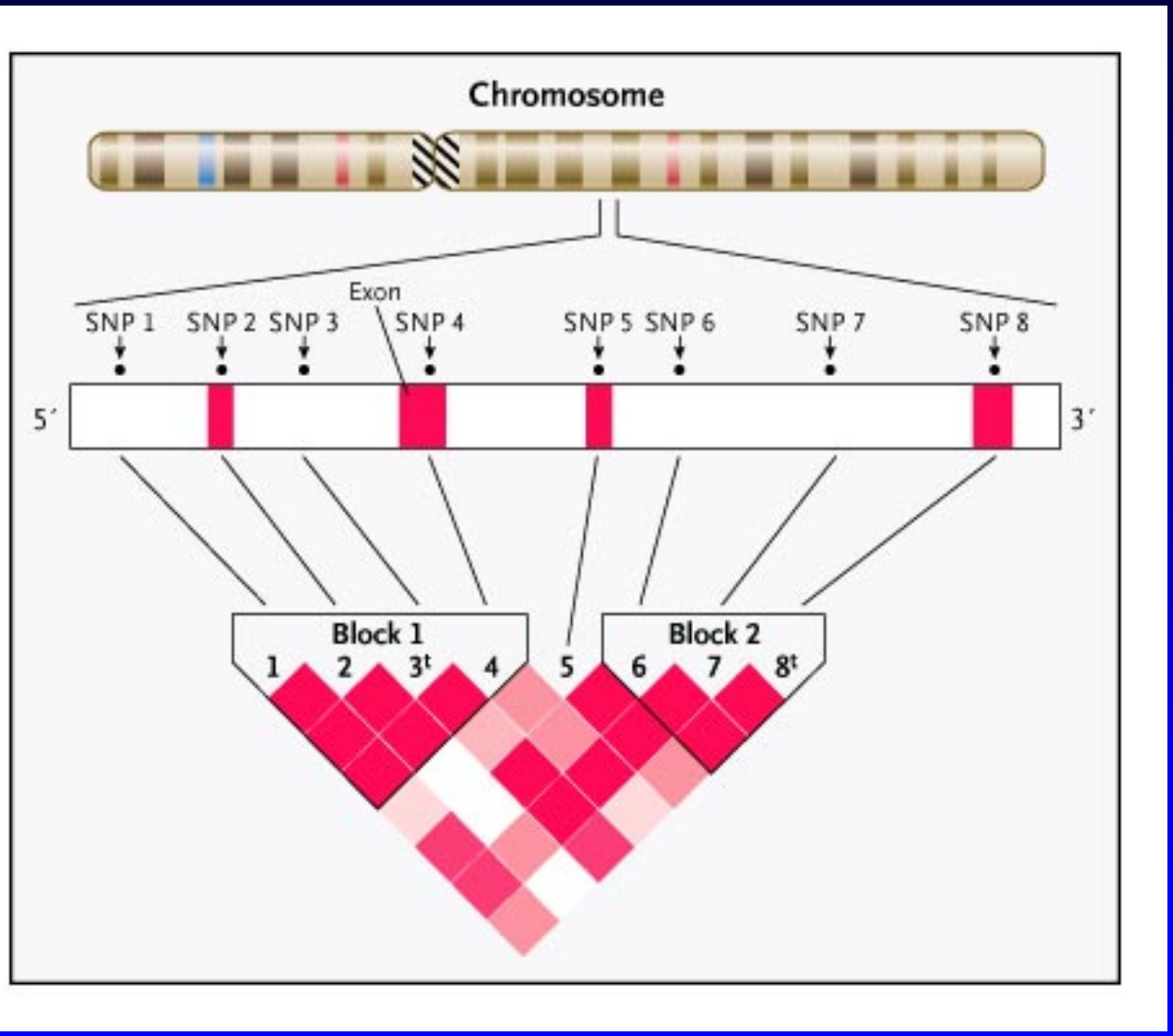


Distances Among East Coast Cities

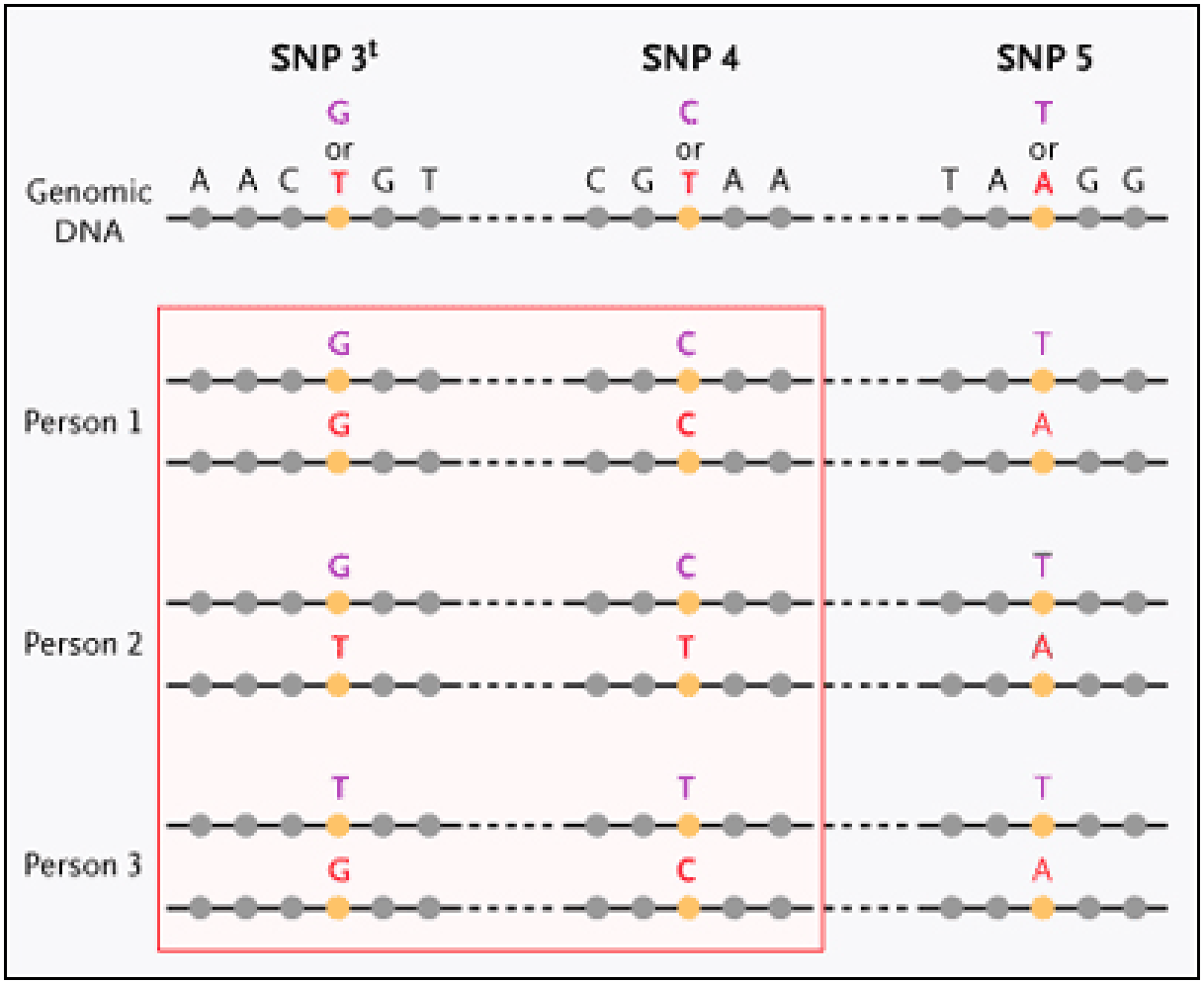


Mapping the Relationships Among SNPs



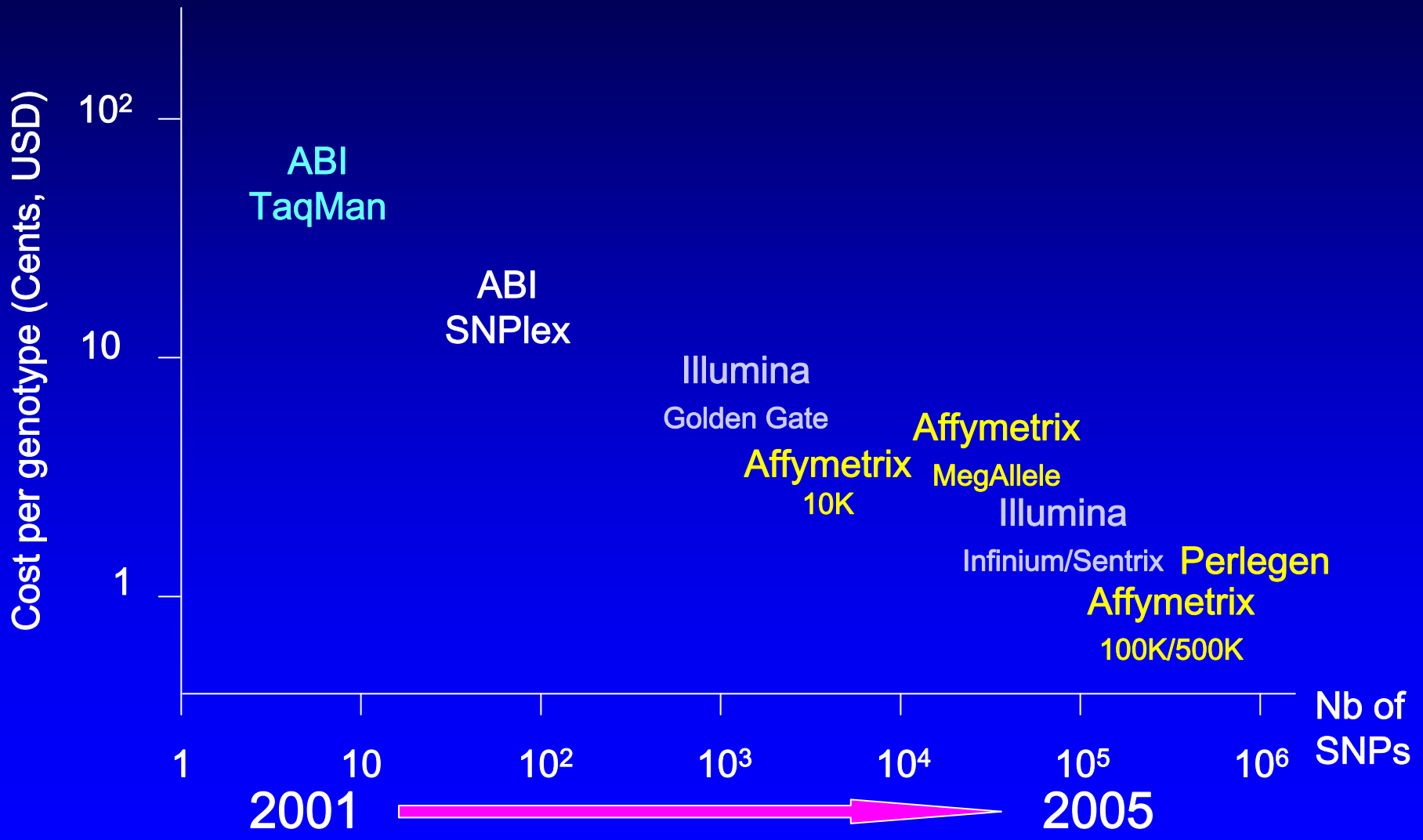
Christensen and Murray, *N Engl J Med* 2007; 356:1094-1097.

One SNP May Serve as Proxy for Many



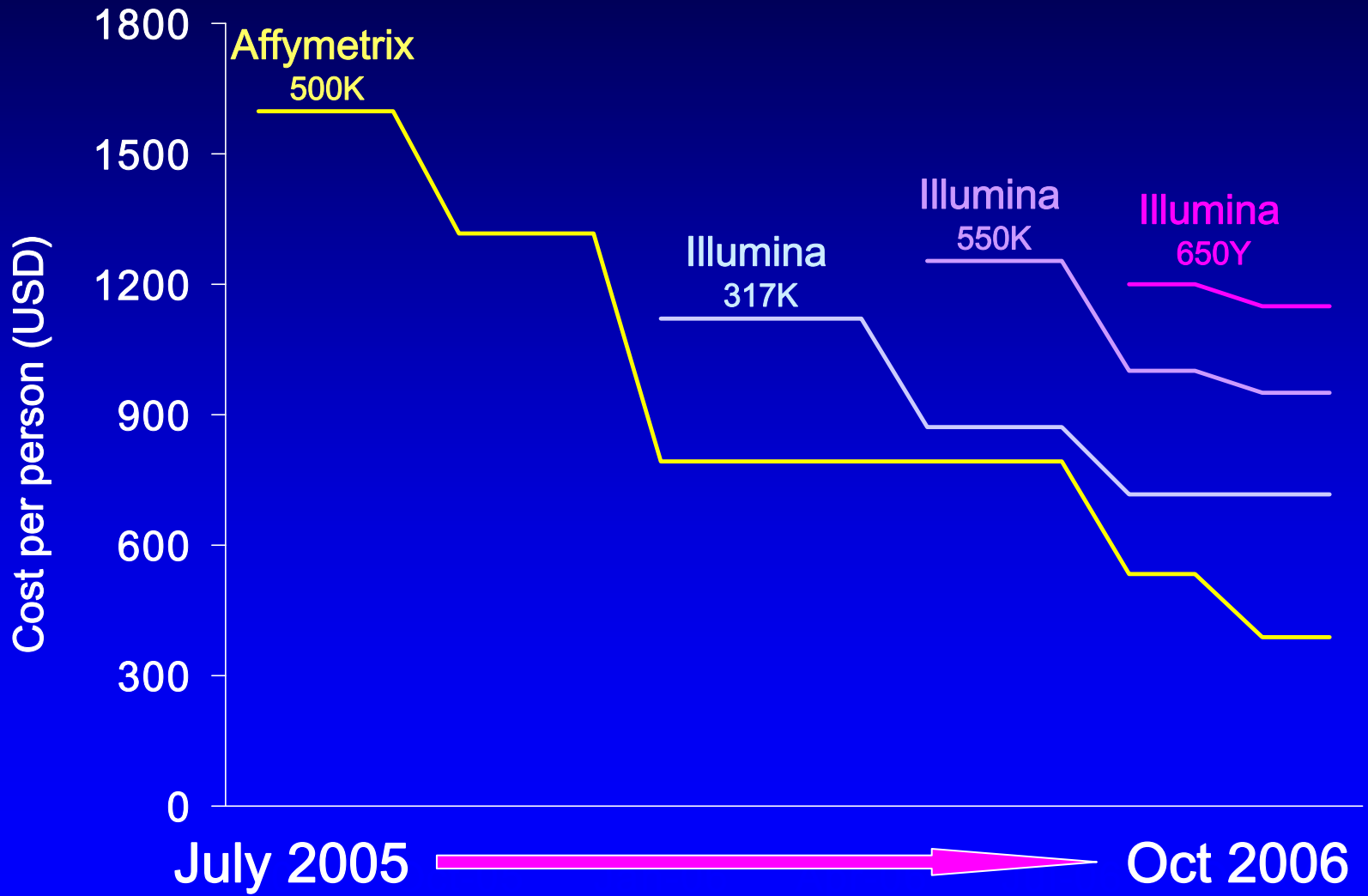
Christensen and Murray, *N Engl J Med* 2007; 356:1094-1097.

Progress in Genotyping Technology



Courtesy S. Chanock, NCI

Continued Progress in Genotyping Technology



Courtesy S. Gabriel, Broad/MIT

Cost of a Genome-Wide Association Study in 2,000 People

Year	Number of SNPs	Cost/SNP	Cost/Study
2001	10,000,000	$1.00	$20 billion
2007	500,000	0.1¢	$1 million

GWA Genotyping Data, Chromosome 22, Parkinson's Study

Study ID	Case/ Control Status	rs5747620		rs2236639	
		Allele 1	Allele 2	Allele 1	Allele 2
14	Case	T	T	G	G
20	Case	T	C	G	G
41	Case	T	C	G	G
412	Control	T	C	G	G
592	Control	C	C	G	G
665	Control	T	C	A	G

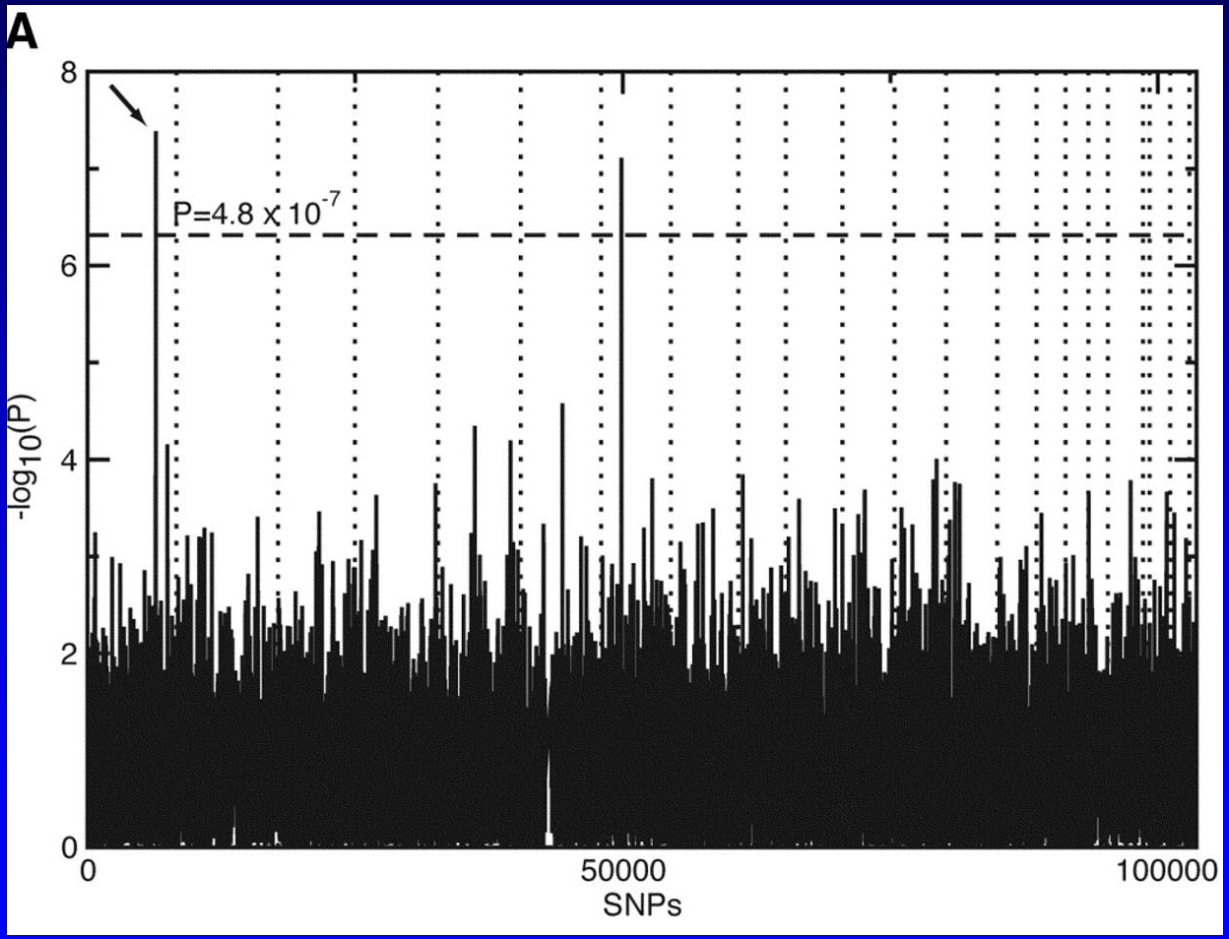
http://ccr.coriell.org/ninds/

Association of rs2236639 Alleles with Development of Parkinson Disease (Made Up!)

Variant Allele (A)	Development of Disease: Develop Disease	Development of Disease: Do Not Develop Disease	Total
Present	10	70	80
Absent	40	880	920
Total	50	950	1,000

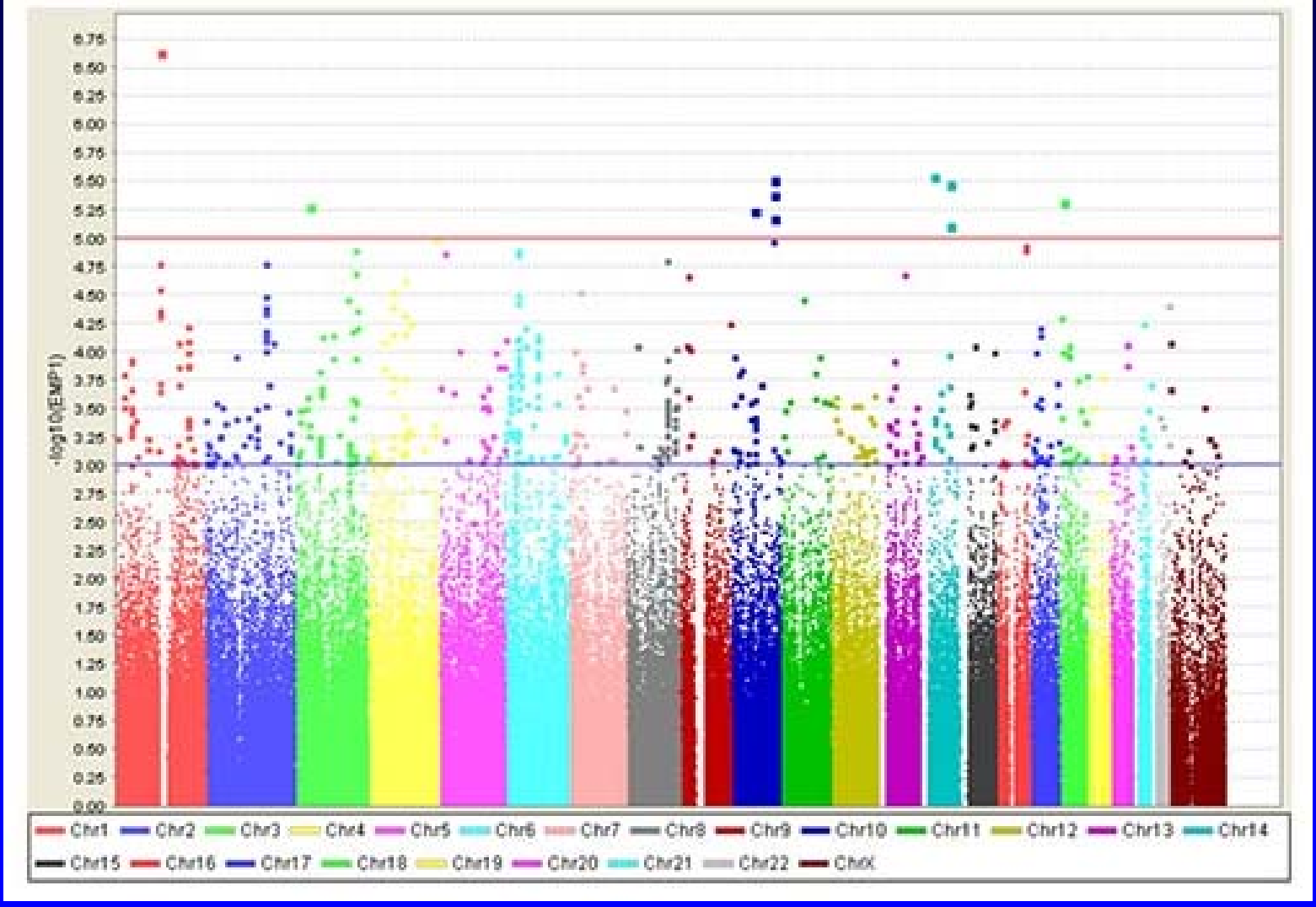
$$\text{Relative Risk} = \frac{\text{Risk in Exposed}}{\text{Risk in Unexposed}} = \frac{10/80}{40/920} = \frac{12.5\%}{4.3\%} = 2.9$$

P Values of GWA Scan for Age-Related Macular Degeneration



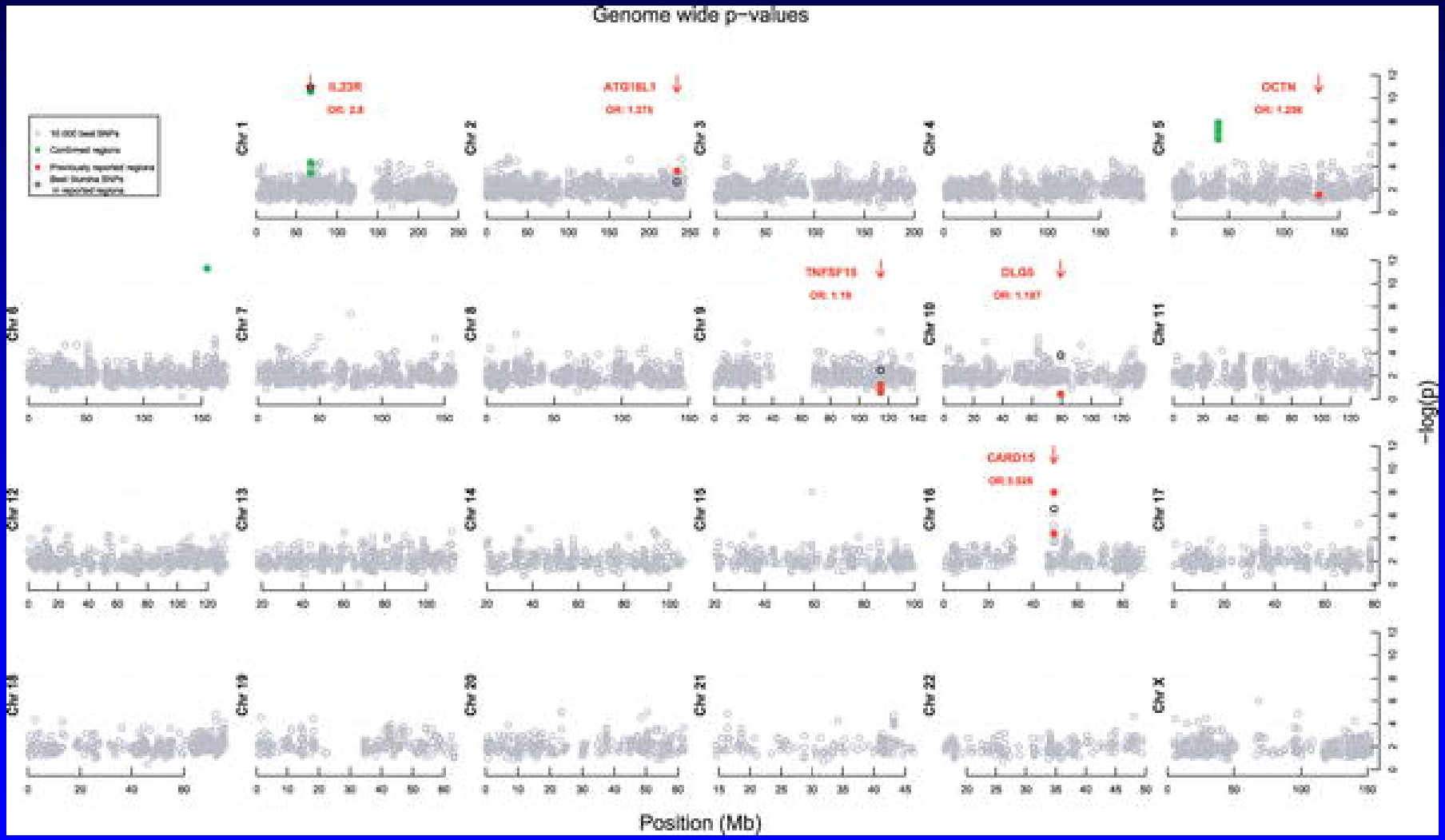
Klein et al, *Science* 2005; 308:385-389.

Genome-Wide Scan for Type 2 Diabetes in a Scandinavian Cohort



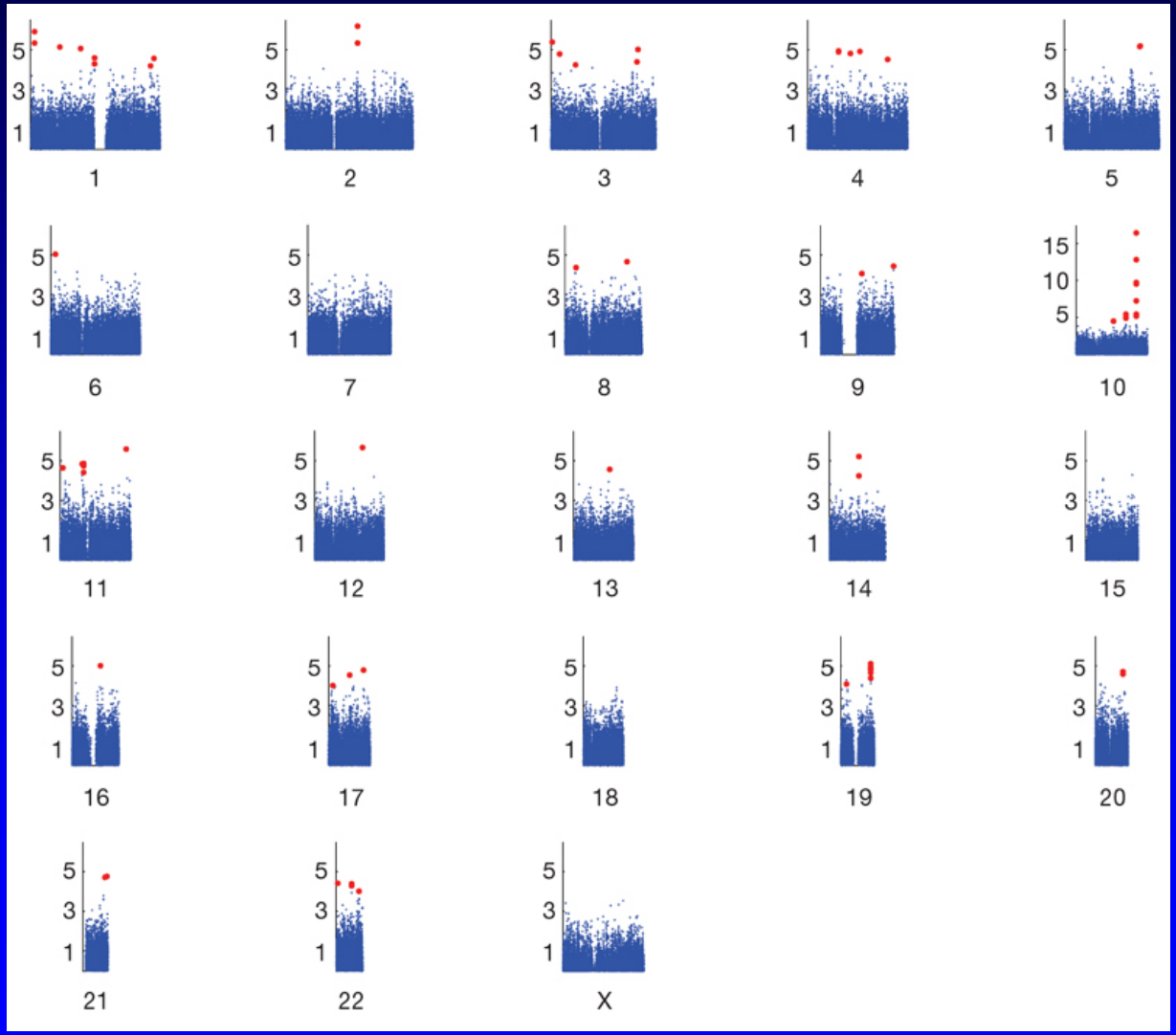
http://www.broad.mit.edu/diabetes/scandinavs/type2.html

Genome-Wide Scan for Crohn Disease in Belgian Cases and Controls



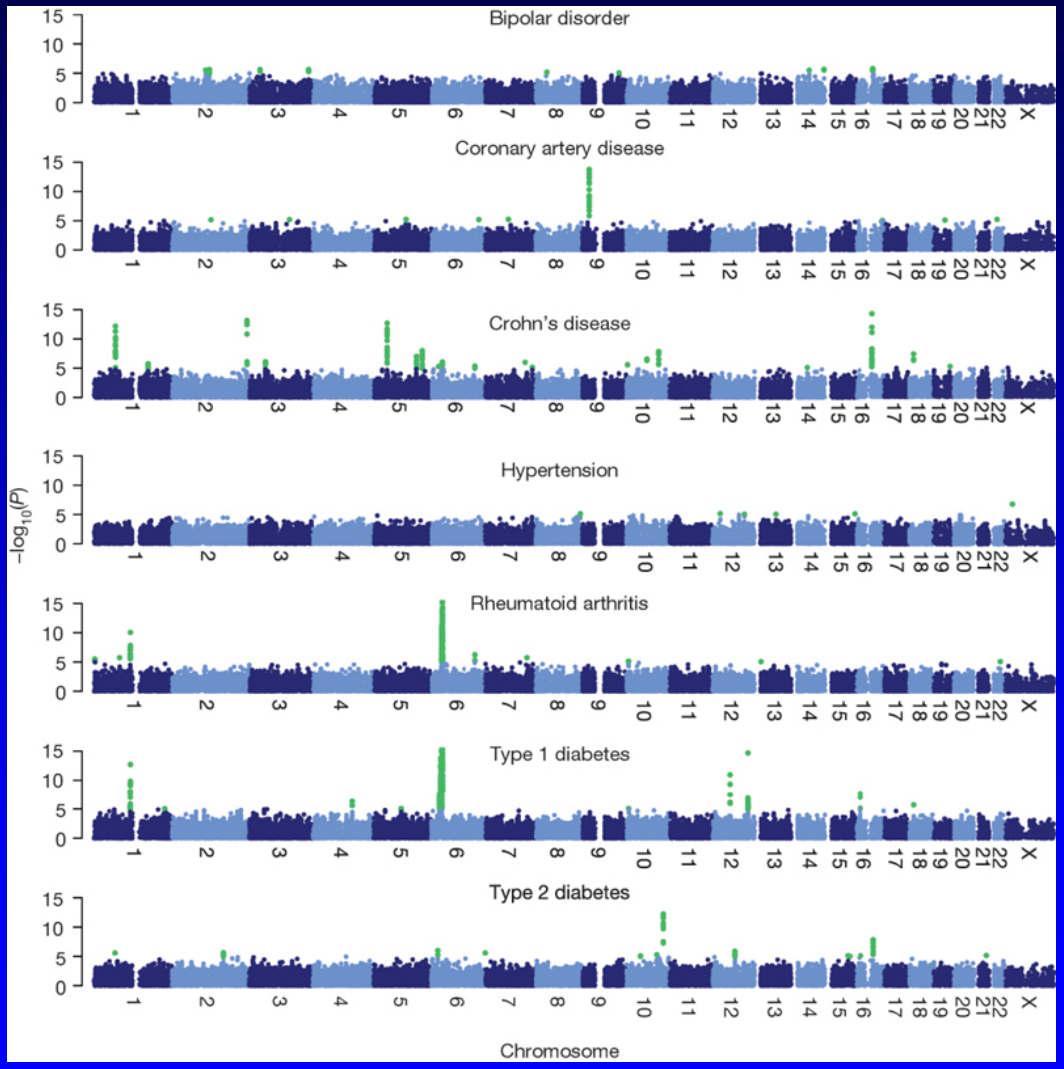
Libioulle C et al, *PLoS Genet*; 2007 Apr 20;3(4):e58.

Genome-Wide Scan for Type 2 Diabetes in French Case-Control Study



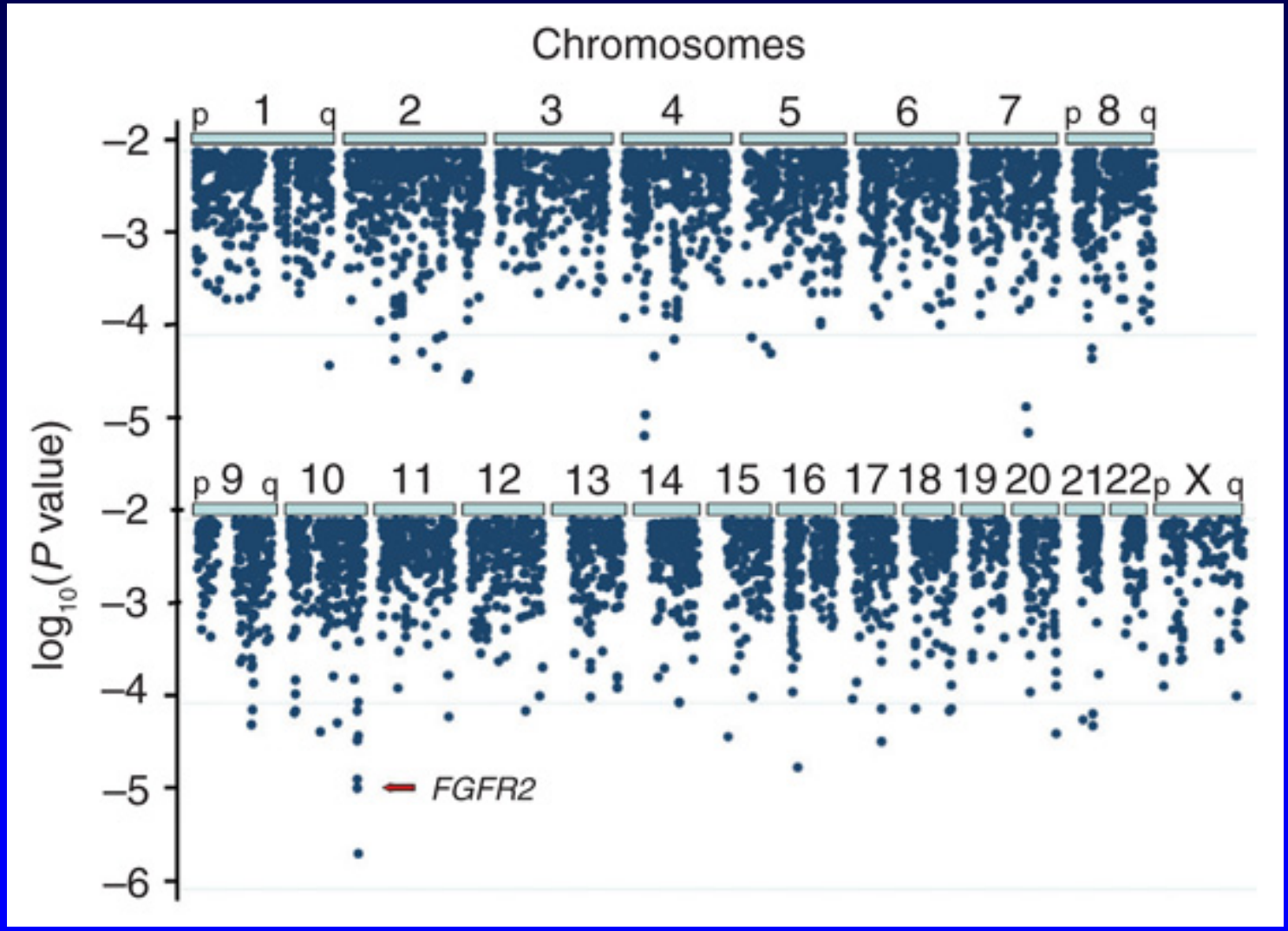
Sladek R et al, *Nature* 2007; 445, 881-885.

Wellcome Trust Genome-Wide Association Study of Seven Common Diseases



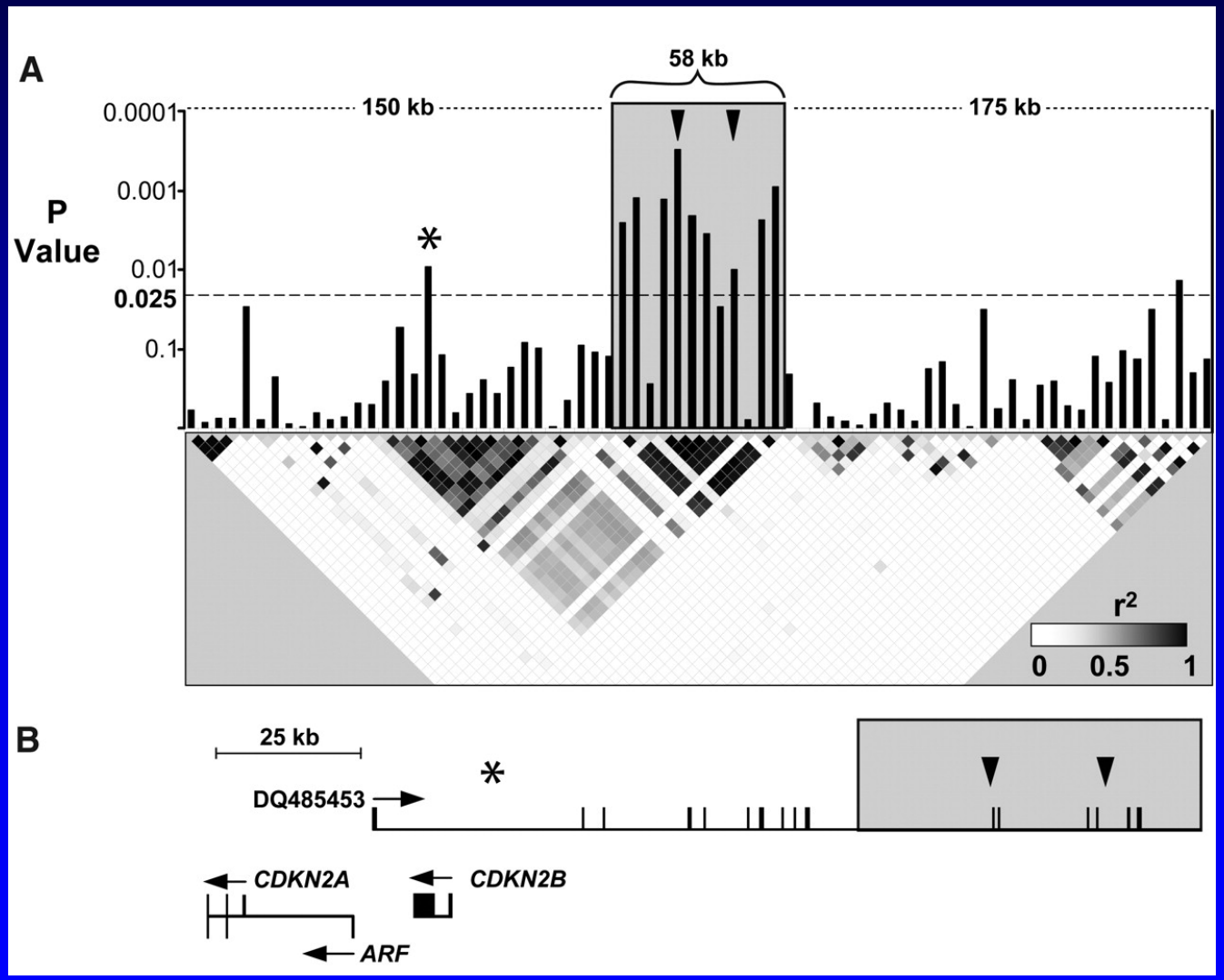
WTCCC, *Nature* 2007; 447:661-678.

Genome-Wide Scan for Breast Cancer in Postmenopausal Women



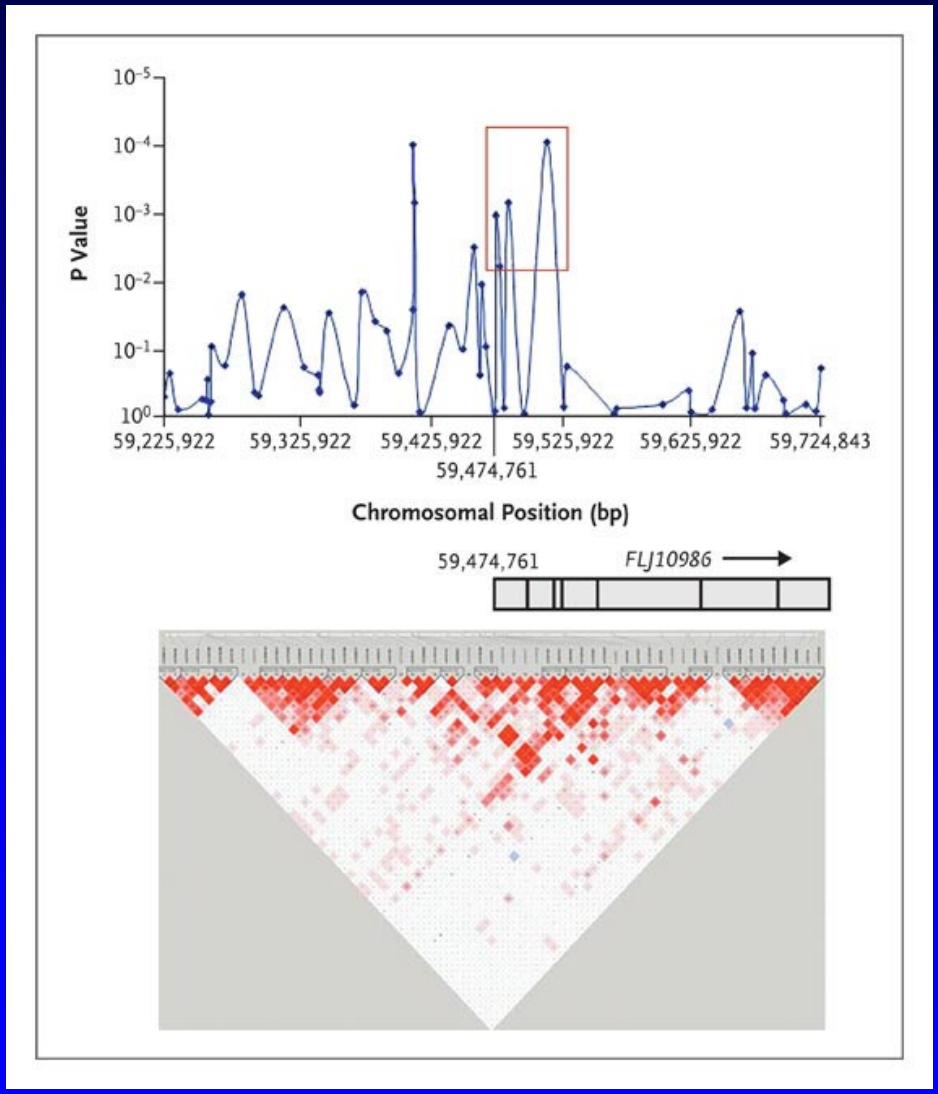
Hunter DJ et al, *Nat Genet* 2007; 39:870-874.

Genome-Wide Scan for Coronary Heart Disease in Ottawa Case-Control Study



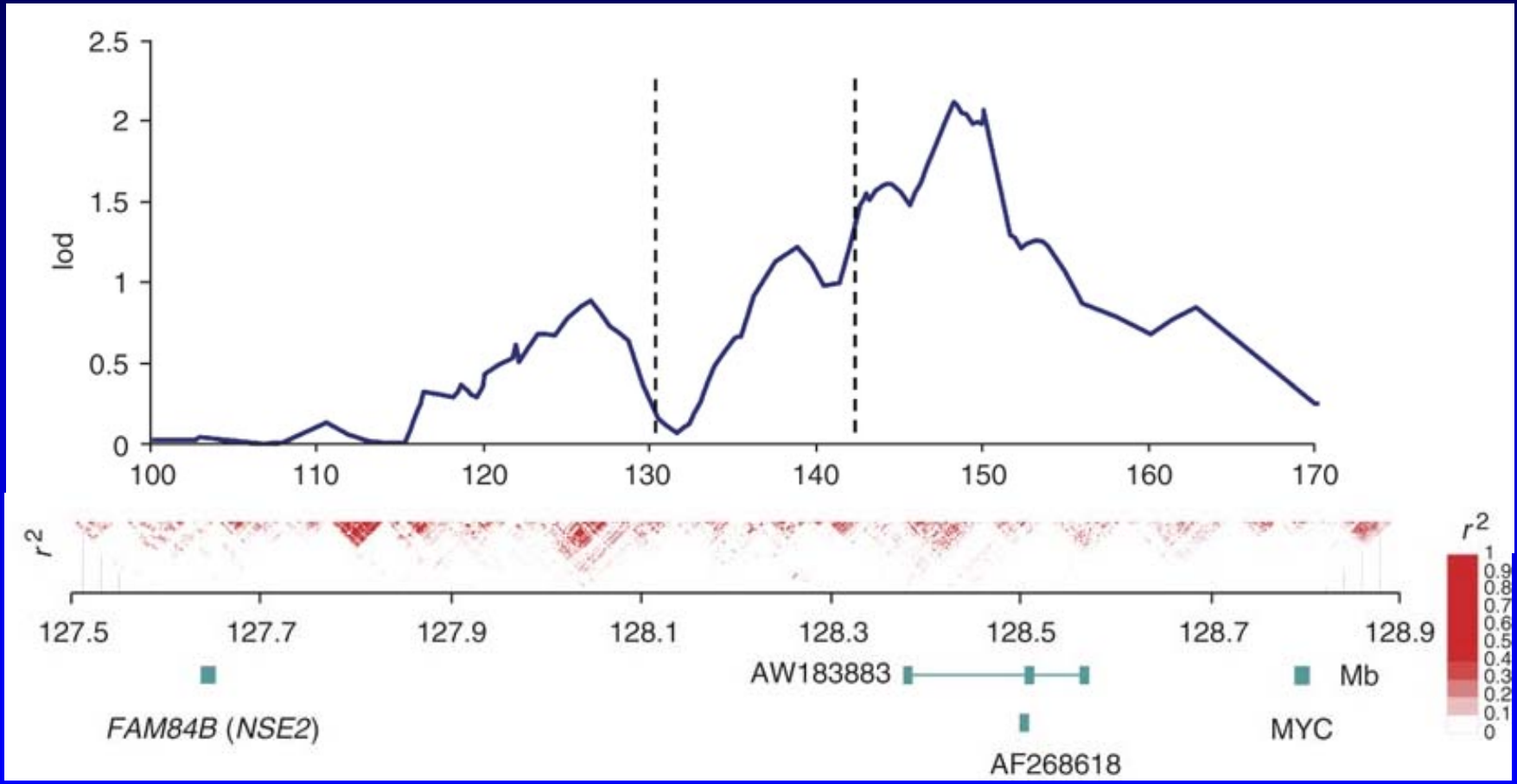
McPherson R et al, *Nature* 2007; 316:1488-1491.

Genome-Wide Scan for Sporadic Amyotrophic Lateral Sclerosis



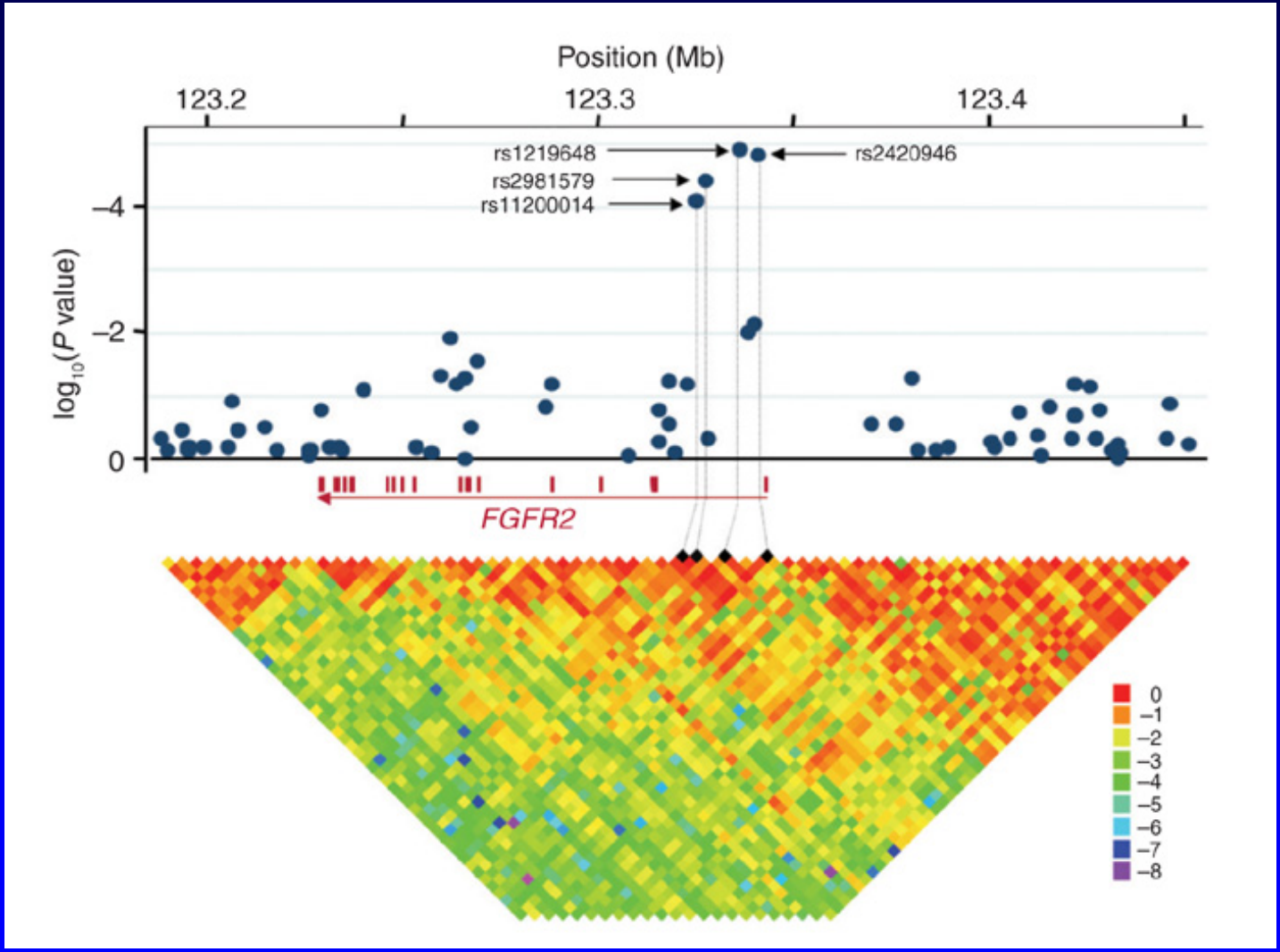
Dunckley T et al, *N Engl J Med* 2007; 357:775-788.

Genome-Wide Scan for Prostate Cancer

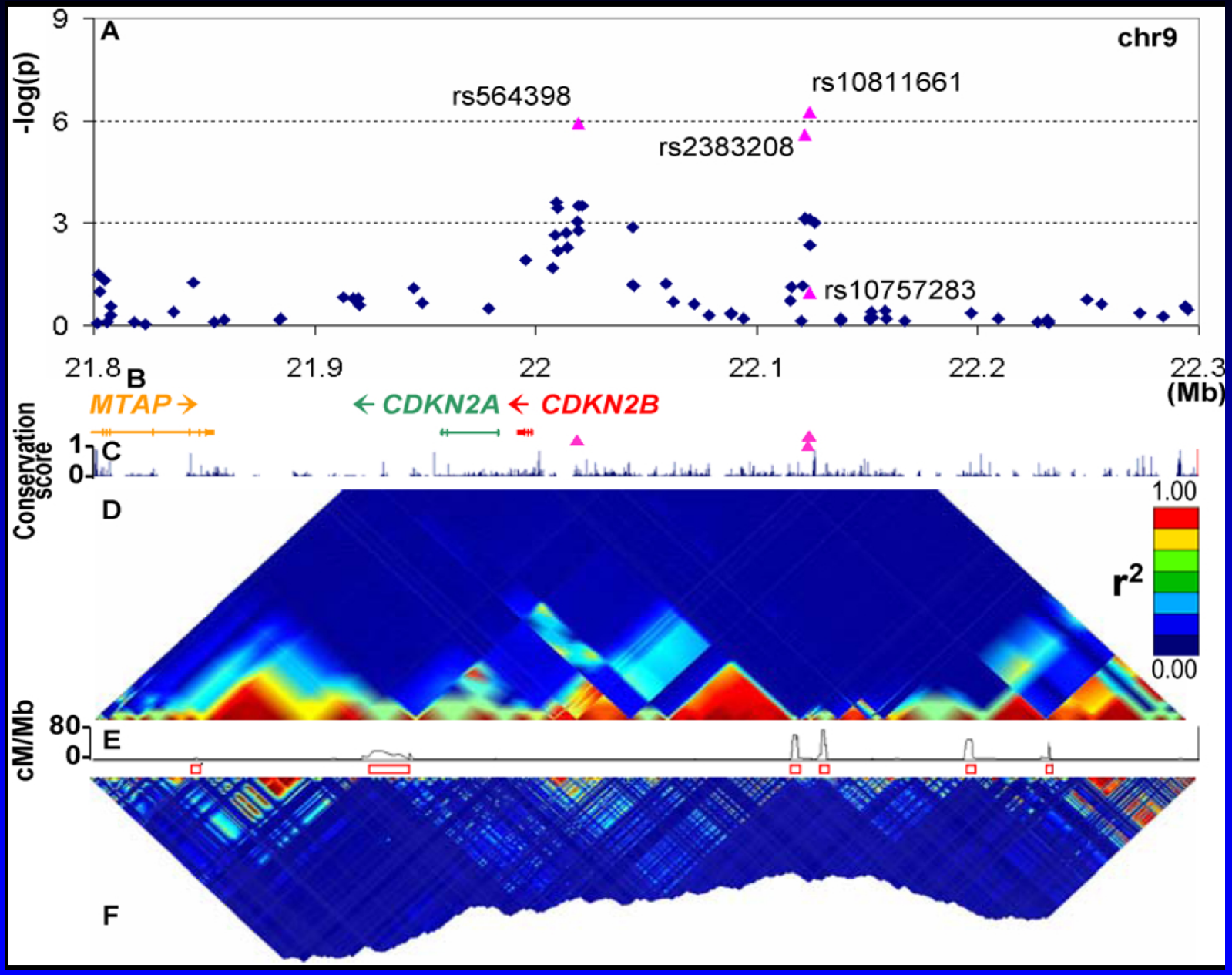


Gudmundsson J et al, *Nat Genet* 2007; 39:631-637.

Association Analysis of SNPs across FGFR2



Hunter DJ et al, *Nat Genet* 2007; 39:870-874.



Courtesy, F. Collins

Lessons Learned from Initial GWA Studies

Signals in Gene "Deserts"	
Prostate Cancer	8q24
Crohn's Disease	5p13.1, 1q31.2, 10p21
Signals in Common	
Diabetes, CHD, Melanoma	*CDKN2A/2B*
Prostate, Breast, Colon Cancer	8q24 region
Crohn's Disease, Psoriasis	*IL23R*
Crohn's Disease, T1DM	*PTPN2*

STATISTICS AND MEDICINE

Drinking from the Fire Hose — Statistical Issues in Genomewide Association Studies

David J. Hunter, M.B., B.S., and Peter Kraft, Ph.D.

Related article, page 443

"There have been few, if any, similar bursts of discovery in the history of medical research…"

The past 3 months have seen the publication of a series of

ating the need for guessing which genes are likely to harbor variants

The main problem with this strategy is that because of the

ost stud-
ained in
samples
ower to

and in this issue of the *Journal*, coronary artery disease (reported by Samani et al., pages 443–453). These genomewide association studies have been able to examine interpatient differences in inherited genetic variability at an unprecedented level of resolution, thanks to the development of microarrays, or chips, capable of as-

lated to the disease. Some of these associations have been found in regions not even known to harbor genes, such as the 8q24 region, in which multiple variants have been found to be associated with prostate cancer.[2] Such findings promise to open up new avenues of research, through both the discovery of new genes rele-

generate P values as small as 10^{-7}. In addition, most variants identified recently have been associated with modest relative risks (e.g., 1.3 for heterozygotes and 1.6 for homozygotes), and many true associations are not likely to exceed P values as extreme as 10^{-7} in an initial study. On the other hand, a "statistically significant" finding

Hunter DJ and Kraft P, *N Engl J Med* 2007; 357:436-439.

Unique Aspects of GWA Studies

- Permits examination of inherited genetic variability at unprecedented level of resolution
- Permits "agnostic" genomewide comparison
- Most robust associations in GWA studies have not been with genes previously suspected of being related to the disease
- Some associations in regions not even known to harbor genes

"The chief strength of the new approach also contains its chief problem: with more than 500,000 comparisons per study, the potential for false positive results is unprecedented."

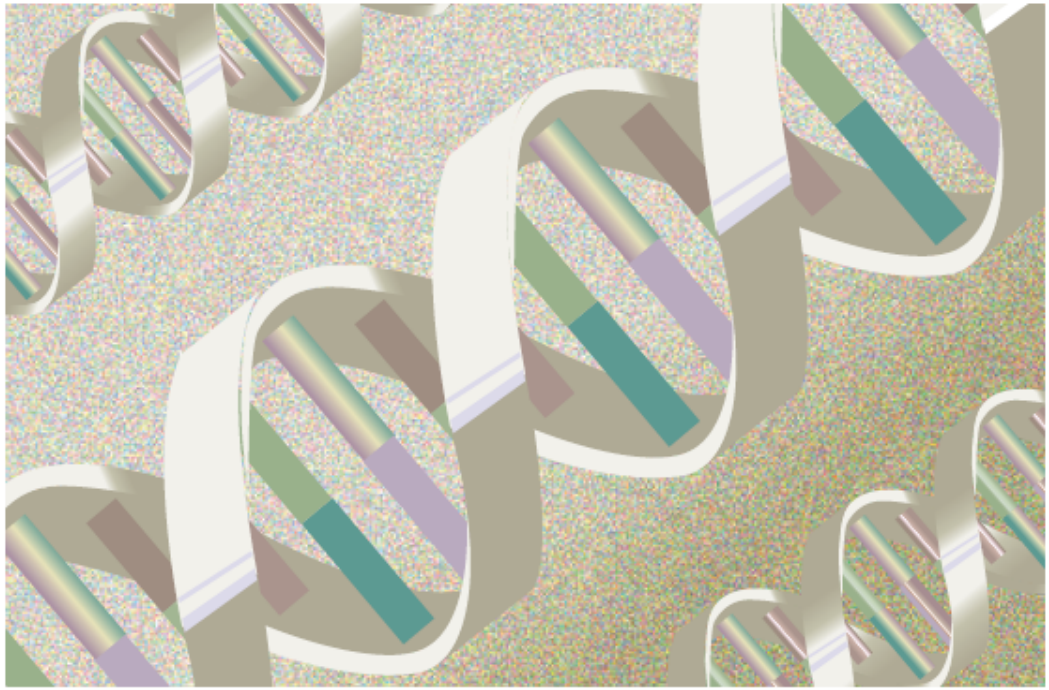
N Engl J Med 2007; 357:436-439.

FEATURE

Replicating genotype–phenotype associations

What constitutes replication of a genotype–phenotype association, and how best can it be achieved?

NCI-NHGRI Working Group on Replication in Association Studies



The study of human genetics has recently undergone a dramatic transition with the completion of both the sequencing of the human genome and the mapping of human haplotypes of the most common form of genetic variation, the single nucleotide polymorphism (SNP)[1–3]. In concert with this rapid expansion of detailed genomic information, cost-effective genotyping technologies have been developed that can assay hundreds of thousands of SNPs simultaneously. Together, these advances have allowed a systematic, even 'agnostic', approach to genome-wide interrogation, thereby relaxing the requirement for strong prior hypotheses.

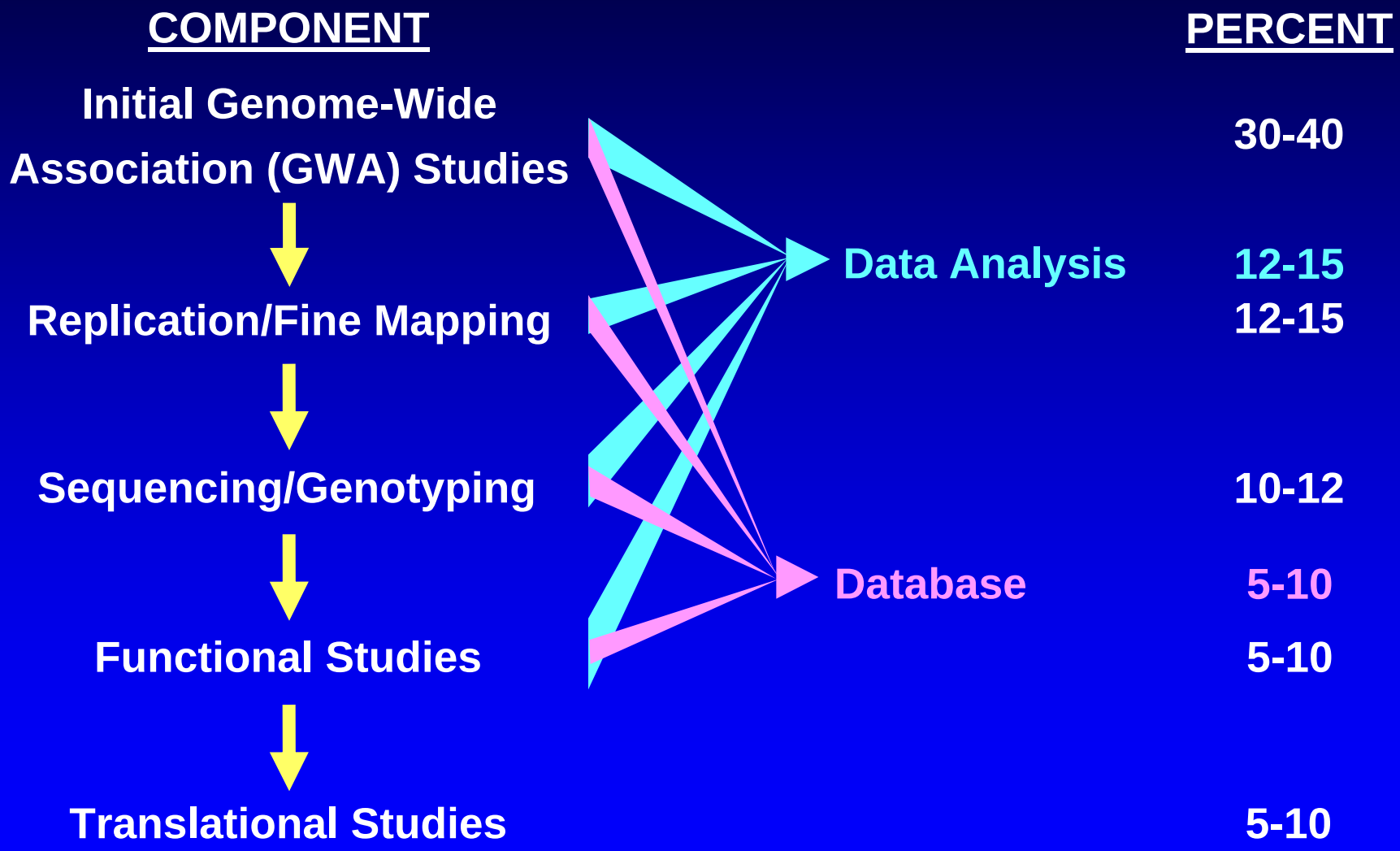
So far, comprehensive reviews of the published literature, most of which reports work based on the candidate-gene approach, have demonstrated a plethora of questionable genotype–phenotype associations, replication of which has often failed in independent studies[4–7]. As the transition to genome-wide association studies occurs, the challenge will be to separate true associations from the blizzard of false positives attained through attempts to rep-

studies because of issues in either the initial study or the attempted replication[4–6,32,33]. Small sample size is a frequent problem and can result

conclusion from the literature because follow-up studies have not consistently analysed the same markers or those in perfect linkage dis-

Chanock et al, *Nature* 2007; 447:655-660.

Flow of Investigation: From Genome-Wide Association to Clinical Translation



Availability of GWA Data in NIH Databases: Current

- Database of Genotype and Phenotype (dbGaP): http://www.ncbi.nlm.nih.gov/sites/entrez?db=gap
- Cancer Biomedical Information Grid (caBIG) and Cancer Genetic Markers of Susceptibility (CGEMS): https://caintegrator.nci.nih.gov/cgems/

Possible Implications of Many Variants of Small Effect

- Need not carry all of them to develop disease
- Probably need to carry more than one, unless very strong environmental interaction
- Some may affect same pathways and be duplicative
- Others may affect different pathways, so some key combination(s) needed
- Should be possible to identify “clusters” of variants carried by different groups of cases
- May be possible to classify on molecular basis

“The more we find, the more we see, the more we come to learn.

The more that we explore, the more we shall return.”

Sir Tim Rice, *Aida*, 2000

ARC-PA

September 19, 2007

Mission

- The ARC-PA protects the interests of the public, including current and prospective PA students, and PA profession by defining the standards for PA education and evaluating PA educational programs within the territorial United States to ensure their compliance with those standards.

Standards

- outline the requirements for accreditation of programs.
- are competency based.
- have specificity regarding curriculum requirements.
- do not prescribe a specific academic degree..

Standards

- Include one specific requirement for instruction in genetic and molecular mechanisms of health and disease.
- Include other areas where discussion of genetics may occur.

Standards

- Instruction in the professional phase of the program must include instruction in the following basic medical sciences - the genetic and molecular mechanisms of health and disease. (B2.02e)

Standards

- The curriculum must include core knowledge about the established and evolving biomedical and clinical sciences and the application of this knowledge to patient care (B1.01)

- The curriculum must be of sufficient breadth and depth to prepare the student for the clinical practice of medicine. (B1.02)

Standards

- For each didactic and clinical course, the program must provide a published syllabus that defines expectations and guides student acquisition of expected competencies. (B1.06)

Accreditation Application

- Programs must list in which courses content is found.
- Programs must include in an appendix: All expected student competencies/learning objectives and samples of student evaluation instruments, (i.e. exams), for genetic and molecular mechanisms of health and disease.

Genomics and PA Certification

An Update from the National Commission on Certification of Physician Assistants

William Kohlhepp, MHA, PA-C
Immediate Past Chairman

Areas of Current or Future Activity

- Exam content
- Promotion
- Research

Integration of Genomics in NCCPA Exams

The Practice Analysis

- Scientific approach to determining the weight of content areas of NCCPA's exams
- Identifies what PAs are actually doing in practice
- Last conducted in 2004; next in 2009-2011
- Opportunity to ask more specific questions to tease out how genomics has been integrated in PA practice

The Content Blueprint

- Foundation from which exams are constructed
- Application of the information gleaned from the practice analysis
- Two dimensions
  - Organ systems
  - Knowledge, task and skill areas

Current Genomics Content

- Do we have any questions on genetics or genomics?

Yes.

Current Genomics Content

Mapped content blueprint against GeneTests list of teaching cases.

- Diabetes mellitus
- Postpartum hemorrhage
- Coronary heart disease
- Huntington disease
- Sickle cell anemia
- Breast cancer
- Hearing loss
- Cystic fibrosis
- Renal failure

Current Genomics Content

Mapped content blueprint against the “Top Dozen.”

1. Breast cancer
2. Colorectal cancer
3. Other cancer
4. Children’s health
5. Family history
6. Making a referral
7. Genetics of common disease
8. Prenatal diagnosis
9. Pharmacogenetics
10. Thrombophilias
11. Psychiatric genetics
12. Nutrigenics

Current Genomics Content

- To what degree are we covering those areas?
- We don’t know…yet.
- Beginning to code new items with a genetics code when applicable.
- Conducting review of item bank and are including this step in that review.
- Will know more next year.

Further Integration of Genomics in Exams

- Adding a new item writer with experience in genomics in 2008.
  - Generate new items
  - Review existing items
- Will include someone with genomics experience on next practice analysis committee.
  - Ensure that we're asking the right questions to find out what PAs are doing in this area.

Promotion of Genomics

Educating the NCCPA Board

- First step for us: Bringing the rest of the organization along.
- Written reports to the NCCPA Board in May and August.
- More in-depth presentation in November.

Promoting the Family History Project

- Existing Family History Project a natural entry point for PAs.
- Featuring the Web tool in the “Don’t Miss” section of the NCCPA Foundation Web site (www.paexcellence.org).
- Willing to do more in this area if it seems appropriate.

Promotion of Genomics Through Other NCCPA Projects

- Procedures logging project in development and will use CPT and ICD9 codes.
- “Pedigree” not among them, but many address taking histories.
- Reviewing definition of PA competencies for gaps.

Research

New Opportunities in Research

- NCCPA Foundation research grants program
- $5k-$10k grants to support research ($50k total per year)
- This topic could be featured as a “topic of interest” in the next RFP.
- Looking for ideas for research topics while we’re here to seed the RFP.

Questions?

Physician Assistant Education Association (PAEA)

300 N. Washington Street
Alexandria, VA 22314
(703) 548-5538
www.PAEAonline.org

PAEA
PHYSICIAN ASSISTANT EDUCATION ASSOCIATION
Scholars. Leaders. Partners.

Follow-up

- Published summary of March Genetics Meeting in PAEA *Networker,* April 2007
- Confirmed Dr. Collins' presentation, "Physician Assistants and Personalized Medicine" at PAEA Annual Education Forum, October 2007
- Identified primary leadership contacts:
  - Anita Duhl Glicken, MSW, Genomics Workgroup
  - Connie Goldgar, MS, PA-C, NCHPEG

Education and Promotion

Summer 2007

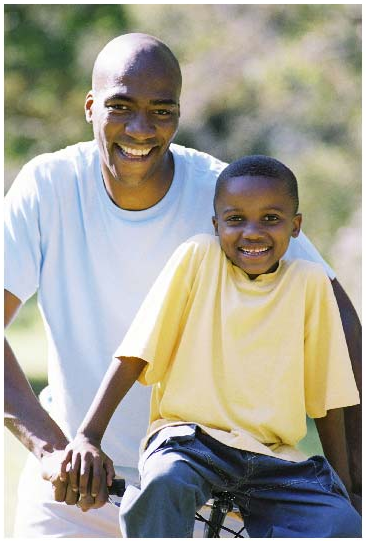
- Published results of PAEA member program genetics survey in the *Journal of Physician Assistant Education,* vol. 18, no. 2, July 2007
- Published editorial, “What Should Physician Assistants Know About Genetics and Genomics”, Bruce R. Korf, MD, PhD, *Journal of Physician Assistant Education*, vol. 18, no. 2 July 2007

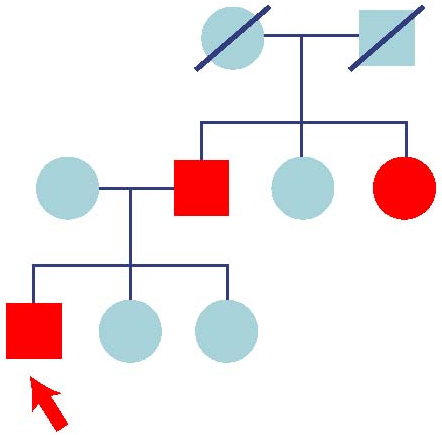
Education and Promotion

Fall 2007

- Plenary and workshop presentation, “Genomics and the Future of Medicine,” Dr. Francis Collins, scheduled for October 2007 PAEA Annual Education Forum
- Coordinated “Put a Face on Genetics Campaign” for October 2007 PAEA Education Forum
- Announced and scheduled launch of NCHPEG PA Web site link for educators, October 2007
- Workshop presentation scheduled, “Developing Instruction in Genetics and Genomics: A Workshop for PA Educators,” October 2007 PAEA Education Forum

Are YOU John's primary care provider?





Why did John, who is 35, have a colonoscopy?

John and his son bike 5 miles a day. He eats a low fat diet. John has a great job, loves his kids, and next Spring plans to hike the Appalachian Trail. John has one more reason to smile. He had a normal colonoscopy yesterday.

Why was he happy to have a colonoscopy? Because John's primary care provider took a thorough family history at his physical last fall. This history revealed that John's father was diagnosed with colon cancer when he was only 45, and that John's paternal aunt had endometrial cancer at 30. John's primary care provider realized that this put his patient at increased risk for a condition known as hereditary non-polyposis colorectal cancer syndrome (HNPCC), or Lynch syndrome.

This condition causes about 3-5% of all cases of colorectal cancer and greatly increases the risk of early onset colon and endometrial cancer in affected individuals. John's primary care provider referred John to a specialist familiar with this disorder. John and his family underwent genetic counseling and had testing that revealed that John and several of his relatives carry the gene alteration causing HNPCC. Armed with this knowledge, John and the others in his family with this gene alteration now get regular cancer screening beginning at an early age. Early and frequent colon cancer screening for people with HNPCC has been shown to save lives, and it may save John's.

U.S. Surgeon General's
Family History Initiative
www.surgeongeneral.gov/familyhistory

EDITORIAL

What Should Physician Assistants Know About Genetics and Genomics?

Bruce R. Korf, MD, PhD
Wayne H. and Sara Crews Finley Professor of Medical Genetics
Chair, Department of Genetics, University of Alabama at Birmingham

It is 54 years since the structure of DNA was discovered and 4 since the complete sequence comprising the human genome was elucidated. The driving force for this massive scientific undertaking was the promise of improving human health by providing tools to understand the genetic factors that contribute to both rare and common disease. The pace of discovery — and of translation to clinical practice — is rapidly accelerating, raising the critical question across all of health professional education: What does the practicing professional need to know about this new area of medicine? This question is especially relevant for physician assistant (PA) education, in part because it raises the usual question of how can more and more curriculum content be crammed into a training program of fixed size? The relevance to PAs, though, goes deeper, and reframes a challenge into an exciting opportunity: How can a profession that is on the front lines of clinical care help to serve as the fulcrum that changes the course of medical practice?

Genetics is not new to the PA curriculum — molecular genetics has long been taught as the basic science underpinning of cell biology. What is new is a major shift in focus — from genetics as a basic science of clinical relevance only for rare disorders to a major driver in our approach to common diseases. What is needed, then, is not so much the addition of new content to the curriculum, but rather a shift in emphasis. I believe that the overall goal of education of PAs about genetics should be the creation of a "genetics dashboard" to help PAs recognize where genetics fits into medical practice, how to wisely use the new tools that genetics is providing, and how and when to call on medical genetics specialists for help. We can build this dashboard by recognizing three overarching principles of how genetics is likely to be integrated into routine medical practice.

First, family history can be a clue to genetic risk assessment, and therefore a gateway to recognizing the genetic factors that create risk of disease. Family history has long been part of the PA armamentarium; it is used, for example, in identifying patients who should be screened for risk of cardiovascular disease or diabetes, and for interpretation of abnormal laboratory values such as in the lipid profile. Now we can go farther, however. Family history is the key to recognizing individuals who are at risk of breast and ovarian cancer or colon cancer, for example, leading to genetic testing of those at high risk.

This is far from an academic exercise: identification of individuals who are carriers for *BRCA* gene mutations, for example, can lead to life-saving interventions, including surveillance and prophylactic surgery. Individuals at risk for hemochromatosis based on family history can be offered monitoring of iron stores and phlebotomy to reduce those stores if they reach a level that would otherwise cause irreversible cirrhosis or cardiomyopathy. The list of not-so-rare disorders in which a significant proportion of individuals is at high risk on the basis of family history and can be diagnosed by genetic testing is increasing. The Surgeon General has championed a Web-based tool for gathering family history information. The PA is in an ideal position to move this effort to the next step by helping patients to gather family history information, recognizing major red flags, and referring those who are eligible for further testing to appropriate specialists.

Second, clinical decisions will increasingly rely on the results of genetic tests. Most common disorders, such as diabetes or hypertension, are the result of an interplay of multiple factors, including genetic and environmental. With recent advances in understanding the structure of the human genome, we are entering a golden age of discovery of genes that contribute to risk for common disorders. Identification of these genes will open a new era in disease prevention, helping providers to recognize those at high risk and opening the door to strategies of risk reduction — including both avoidance of environmental exposures that are particularly harmful to specific individuals and use of medications that target disease pathways to either prevent disease or reduce its burden.

Moreover, the manner by which drugs are absorbed and metabolized, as well as the manner in which they interact with their targets, are all under some degree of genetic control. Many of these genes are already known and can be tested to avoid harmful side effects in at-risk indi-

PA Genetics Lab

- PA Education: A place to innovate
  - Flexibility and Adaptability
  - Shorter Curriculum, rapid transfer to clinical practice
  - Connections with Other Professions: medical schools, schools of allied health, schools of health professions, community colleges

Evaluation and Outcomes

- Developing Educational/Curriculum Resources
  - Faculty Development
  - Curriculum Resources
  - Resource Sharing
- Defining Outcomes
- Collecting Data
  - When to get started
  - What are the data points
- Evaluation

NHGRI Activity Update

Greg Feero, M.D., Ph.D.

Sept. 19, 2007

NHGRI Activity Update

1) Personalized Healthcare Workgroup
2) Nursing Tool Box Meeting
3) Posters
4) Top 12 list

Personalized Healthcare Workgroup

Part of American Health Information Community

- Alan Guttmacher, M.D. is on the workgroup
- Greg Feero M.D., Ph.D. is an advisor to the workgroup

Goal (greatly simplified):

To bring some standardization to how EHR systems deal with family history and genetic test information, and to facilitate CDS

HHS Priorities
for America's Health Care

Secretary Leavitt's Priorities

- **Personalized Health Care**
- Medicaid Modernization
- Health Information Technology
- Medicare Rx
- Prevention
- Health Care Value Incentives
- Emergency Response
- Pandemic Preparedness
- New Orleans Health Care System
- Global Health

Secretary's Vision

- **Personalized Health Care:** "Health care is tailored to the individual. Prevention is emphasized. Propensities for disease are identified and addressed through preemptive intervention. Discovery and innovation move drugs to the market and to medical practice faster and at lower cost."
- **The Long Term Objective:** Advances in basic research have positioned us to harness new and increasingly affordable potential in medical and scientific technology. With clinical tools that are increasingly targeted to the individual, our health care system can give consumers and providers the means to make more informed, individualized, and effective choices.
- **The Secretary's 2-year Objective:** Establishes concepts and priorities that support health care system transformation to achieve long term objectives.

American Health Information Community (AHIC)

HHS Priorities for America's Health Care

AHIC is the public-private collaborative that sets priorities and oversees and/or endorses HIT standards, certification, the National Health Information Network, and policies on a national level.

- Supported through the Office of the National Coordinator for Health Information Technology
- Chaired by Secretary Leavitt and Dr. David Brailer
- Seven work groups are now established involving over 100 experts and stakeholders – Biosurveillance, Electronic Health Records, Chronic Care, Consumer Empowerment, Confidentiality, Privacy and Security, Quality, and Personalized Health Care
- Work groups develop recommendations to the AHIC and subsequently to the Secretary for action
  - Example: Executive Order requiring adoption of certification standards for electronic health records

Goals : PHC Initiative

Goal 1: Link Clinical and Genomic Information to Support Personalized Health Care

- Establish an interoperable public/private data partnership of networks to deliver information on individual medical outcomes and linking findings to genetic laboratory test.
- Establish Common Pathway for Data Integration through Electronic Personal Health Records

Goal 2: Support the Appropriate Use of Genetic Information

- Protect individuals from genetic discrimination
- Encourage policies and practices that provide sufficient protections to consumers that genetic test information is used only for their medical benefit
- Provide oversight of genetic testing to assure analytical and clinical validity
- Standardize access policies to federally funded databases of genetic information

AHIC PHC Working Group

HHS Priorities for America's Health Care

John Glaser	Partners HealthCare
Douglas Henley	American Academy of Family Physicians
Carolyn Clancy	Agency for Healthcare Research and Quality
Beryl Crossley	American Clinical Laboratory Association, Quest
Paul Cusenza	23andMe
Andrea Ferreira-Gonzalez	Virginia Commonwealth University
Becky Fisher	Patient Advocate
Felix Frueh	Food and Drug Administration
Emory Fry	Department of Defense
Alan Guttmacher	National Institutes of Health/NHGRI
Kathy Hudson	Genetics and Public Policy Center
Betsy Humphreys	National Institutes of Health/NLM
Charles Kennedy	WellPoint
Joel Kupersmith	Department of Veterans Affairs
Stephen Matteson	Pfizer
Deven McGraw	National Partnership for Women and Families
Amy McGuire	Baylor College of Medicine
Mark Rothstein	University of Louisville
Steve Teutsch	Merck
Janet Warrington	Affymetrix
Andrew Wiesenthal	Permanente Federation
Marc Williams	Intermountain Healthcare

Near Term Priorities

Genetic/Genomic Tests

- Inclusion of relevant genetic/genomic test results in the EHR
- Information to describe analytical validity, clinical validity, and clinical utility of genetic/genomic tests
- Incentives for development and evaluation of new genetic/genomic tests
- Consumer education about the potential benefits and risks associated with genetic/genomic tests
- Harmonization of standards for submission of clinical pharmacogenomics data and state-mandated newborn screens

Near Term Priorities (cont.)

Family Health History

- Consumer and clinician entry of family health history information in the interoperable PHR and EHR
- Support clinician use of consumer entered family health history information
- Standardization of nomenclature for family relationship and other data
- Characterization of the validity and utility of use of family health history in making clinical decisions

Long Term Priorities

Clinical Decision Support

- Development of approaches to informing the clinician of the clinical utility of test results
- Development and assessment of genetics/genomics predictive algorithms
- Development and assessment of genetics/genomics-based CDS to guide treatment and medication dosing decisions
- Incentives for development and incorporation of clinical decision support tools in EHRs

Long Term Priorities (cont.)

Confidentiality, Privacy, and Security

- Technical solutions and policy considerations to ensure that genetic/genomic information will be used appropriately
- Capabilities to link large datasets to generate large-scale, individual-level genetic/genomic data with sufficient protections and limits for use
- Balancing the desires of the research community to have secure and consented access to clinical databases with the privacy and confidentiality rights of the consumer and clinician
- Understanding the risks associated with certain types of genetic/genomic information:
  - Contextual access criteria limits to necessary information
  - Ensuring privacy and confidentiality rules apply to all collection/exchange of health information
  - Research to assess CPS of the NHIN and consumer confidence

PHC Workgroup Next Steps

Short Term

- Two subgroups
  - Genetic/Genomic Tests
  - Family Health History
- Recommendations approved by the AHIC at July 31, 2007 meeting

Longer Term

- PHC-CPS Subgroup
  - Coordinate activities with AHIC Confidentiality, Privacy, and Security Workgroup
- CDS Ad-hoc Workgroup
  - Coordinate activities across AHIC Electronic Health Records, Personalized Health Care, Population Health and Clinical Care Connections, and Quality Workgroups

Recommendation Approved by AHIC July 31, 2007

Recommendation 1.0:

The Community should advance the area of Personalized Health Care as a Priority for Use Case Development.

Recommendation 2.0:

An extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting) should be developed to address the specific information needs in the pre-analytic, analytic, and post-analytic phases of genetic/genomic tests. This extension to the use case should additionally address the need for integrated data flow across the pre-analytic, analytic, and post-analytic phases of genetic/genomic testing and address both the EHR and Laboratory Information Systems.

Recommendation 3.0:

A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to develop a core minimum data set and common data definition available for primary care collection of family health history information.

Recommendations Cont:

Recommendation 3.1:

Additionally, studies should be performed as part of this collaboration as an evidence-base to determine the validity and utility of family health history risk assessment and management tools, clinical decision support tools, and how clinicians view this information as helpful for informing their medical decisions.

Recommendation 3.2:

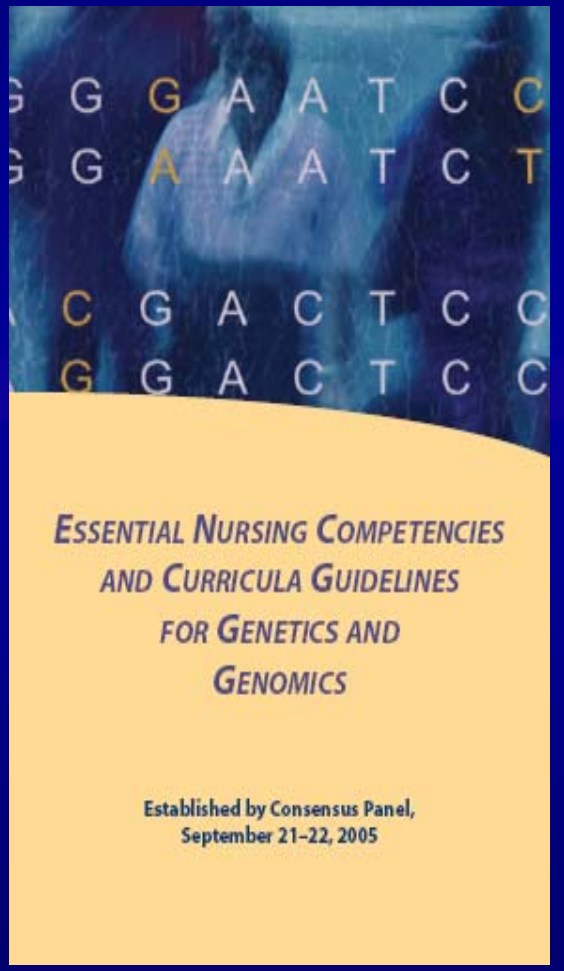
Federal agencies in conjunction with private health care organizations with similar interests and expertise sponsoring pilots in the area of family health history should be used to evaluate the core minimum data set and evidence-base developed through Recommendations 3.0 and 3.1. Health care providers involved in these pilots should also examine the feasibility of consumer-clinician exchange of family health history information between PHR and EHR systems. When possible, the pilots should test and implement the standards and architecture identified in the HITSP developed use case.

Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics

- Meeting held Sept. 2005 to define essential genetic and genomic competencies for all registered nurses - endorsed by 48 professional groups.

Implementation

- Strategic Implementation Plan meeting held October 22-24, 2006
  - Stakeholders from educational institutions, professional associations, certifying bodies and regulatory agencies met to plan for next steps.



Academia: Toolkit for nursing faculty (consider interdisciplinary)

- Meeting held September 14, 2007
- In collaboration with the American Association Colleges of Nursing
- Discussed faculty needs; available resources; gaps; and product options.
- Next steps: finalize what to include, how to package, and funding options.

KEY COMPONENT

- Awareness of relevancy of genetic and genomic information for quality healthcare services
- Consider marketing campaign that includes patient stories

Have you seen Mary…or Fred…or Bob?*

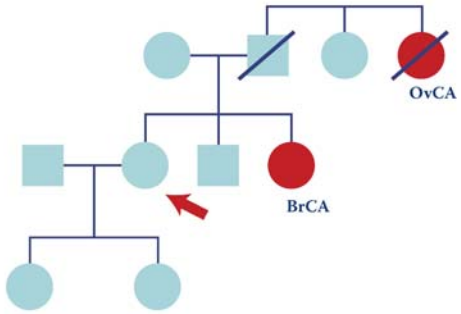
Five poster-format messages targeting PA's, focusing on family history

- Hereditary breast and ovarian cancer syndrome
- Diabetes
- Coronary artery disease
- Hereditary non-polyposis colorectal cancer syndrome
- Preconception care (Tay-Sachs)

*Jane Ades, NHGRI

Caring for Mary in Your Practice

Discovering Hereditary Breast and Ovarian Cancer



Mary is a 38-year old healthy mother of two who likes to jog, doesn't smoke, doesn't drink, watches her diet, and has faithfully gone each year to her primary care provider in her old home town. In short, Mary seems a woman with little risk for serious disease.

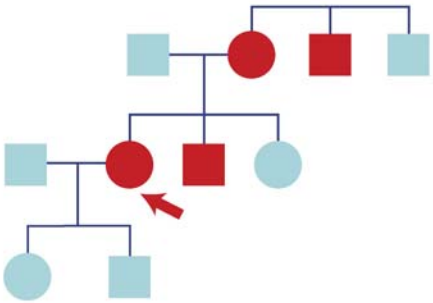
Five years ago, Mary's then 42 year old sister was diagnosed with early stage breast cancer. Fortunately, her cancer was localized and, with treatment, she did well. Unfortunately, Mary's primary care provider never asked Mary about her family history.

This year Mary will move and begin seeing a new primary care provider. Her new provider will take her family history and learn about the cancer in Mary's sister. Additionally, her new provider will discover that Mary's paternal aunt died at age 28 from ovarian cancer, before Mary was born. Mary's new provider will recognize that these facts from her family's medical history put Mary and Mary's family at risk of having hereditary breast and ovarian cancer syndrome. This affects about 1 in 800 individuals and increases the risk of developing certain cancers at a very early age. Mary's new provider will counsel Mary to speak to her sister about seeking additional information about her family's risk. Together Mary and her sister will undergo genetic counseling and learn through genetic testing that they carry a change in the BRCA 1 gene known to cause hereditary breast and ovarian cancer. Each of them will choose slightly different proven health care options that reduce their risk of potentially fatal disease.

YOU are Mary's new primary care provider. Congratulations – **YOU** will quite possibly save Mary's life, and perhaps that of her mother, sister, and children.

U.S. Surgeon General's
Family History Initiative
http://www.hhs.gov/familyhistory

Do You Take That Extra Step Caring for Your Patient?



Vanessa, 35, has just finished walking with her daughter and feels great. These walks are now part of her daily routine, and her healthcare provider tells her she won't need medication for her diabetes in the foreseeable future.

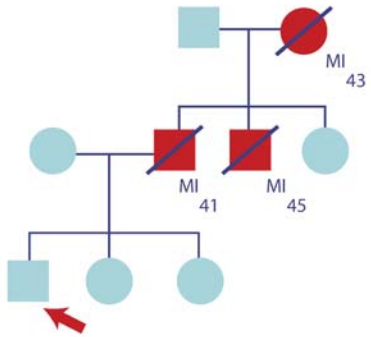
The outlook for Vanessa might not have been this good. All too often diabetes goes undiagnosed for years, with high blood sugars silently affecting vulnerable organs like the eyes, kidneys, and heart. By the time symptoms appear, organ damage has occurred. Luckily for Vanessa, her healthcare provider asked her about her family history at her last physical and found that her mother, uncle, and brother all developed diabetes in their early 40's. This prompted Vanessa's provider to check her fasting blood sugar, which was abnormal. With diet changes and exercise, Vanessa's sugars are in the normal range, and she is helping the rest of her family adopt a healthy lifestyle.

Obtaining your patient's family history can make a real difference to them and their family's health. The U.S. Surgeon General's My Family Health Portrait tool can help your patients to gather and organize their family history before they come to your office.

U.S. Surgeon General's
Family History Initiative
http://www.hhs.gov/familyhistory

Henry, 34, Likes His Morning Coffee

Today Henry can enjoy his coffee feeling confident that there are many more mornings like this one to come. Sadly, Henry's father wasn't so lucky. His life was cut short by a heart attack when he was only 41. As happens with about one in three heart attacks, it came almost without warning. Almost, save a strong family history of heart disease.



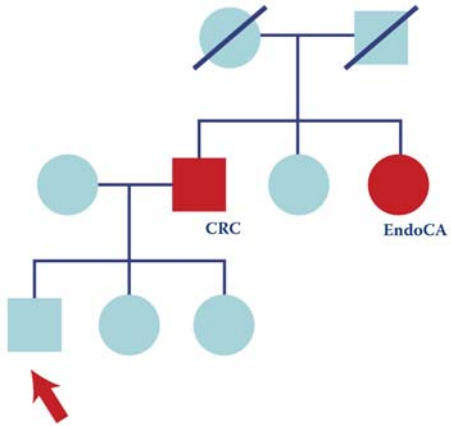
Unlike his father, Henry and his healthcare provider discussed Henry's family history at his last complete physical exam. Henry's healthcare provider learned of the tragedy of Henry's father and as well that Henry's grandmother and uncle both had heart attacks when they were in their early 40's. A quick lab test revealed that Henry's total cholesterol was 300.

As a result, Henry's lifestyle has undergone some recent changes, including a new medication, giving up smoking, and starting a new diet and exercise program. It hasn't been easy. But Henry's not complaining; after all it is a good cup of coffee.

The next time you see Henry, spend some time obtaining a family history, it can make the difference of a lifetime.

U.S. Surgeon General's
Family History Initiative
http://www.hhs.gov/familyhistory

Are YOU John's primary care provider?



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Family History Initiative
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Baby's First Visit Comes Early

Prenatal Care and Genetics

Jane and Bob are adding on to their home. Why? Because Jane and Bob will soon experience the joy of bringing a healthy newborn girl into their family. Excellent preconception care helped to assure that the health of their baby was not left to chance.

This will be Jane's first child. For this pregnancy Jane chose a new health care provider, who took a thorough family history at a preconception visit. Jane's family history revealed that both she and Bob were of French Canadian ancestry. Jane's provider recognized that this put Jane at elevated risk of having a baby with Tay-Sachs disease, a lethal inherited disorder affecting the nervous system and explained this to Jane and Bob. Jane and Bob chose to have genetic counseling and genetic testing and learned that they were both carriers of gene alterations that could cause Tay-Sachs. Jane and Bob then elected to have prenatal genetic testing to determine if their baby would be affected. To everyone's joy the testing revealed that their baby would not inherit Tay-Sachs.

Are **YOU** the provider helping Jane to bring her new daughter into the world?

To learn more about how genetics is relevant to your practice visit www.genome.gov

HIS FAMILY MEDICAL HISTORY WILL HELP YOU TREAT HIM TODAY. ASK ABOUT IT.

Obtaining a child's family history can help optimize care and treatment; it can also be an important tool for preventive medicine and public health. More accurately identifying children at risk for conditions including diabetes, cardiovascular disease, and single gene disorders such as cystic fibrosis and fragile X syndrome can change the primary care clinician's approach to pediatric medicine. A family history can be used as:

1. A diagnostic tool and guide to testing and evaluation
2. A way to identify patterns of inheritance
3. A patient education tool

To find out more about the importance of family history in pediatric care, please click here.

Additional resources:
CDC's Pediatric Genetics Information
CDC's Family History Resources & Tools
U.S. Surgeon General's My Family Health Portrait

CDC

Mary con't.

- Content?
- Format?
- Venues?
- Target audience?
- Other topics
  - Broad (e.g. pharmacogenomics)
  - Specific (e.g. other family history topics)

Top 12 Topics For PA Education In Genetics

1. Hereditary Cancer Syndromes
   a. breast cancer genetics
   b. colorectal cancer genetics
2. Cancer Genetics
3. Lifestage Genetics
   a. Pregnancy & the perinatal period
   b. Infancy
   c. Childhood and adolescence
   d. Adulthood
4. Family Pedigree History
5. Genetic Counseling & Making a Genetics Referral
6. Genetics of common disease
7. Genetic Testing
8. Pharmacogenomics
9. Thrombophilias
10. Psychiatric genetics
11. Nutrigenetics
12. Basic Concepts in Genetic Science

Top 12 topics for PA education in genetics

How do we move forward with these topics

- Who are the targets?
  - Educators
  - Students
  - PA's in practice
- What venue?
  - Courses?
  - Publications?
  - Web-based?
- Who will populate list?
  - Geneticists
  - Other specialists
  - PA's?

GENETIC TOOLS: Building a Model Curriculum for Family Medicine Residency Education

Gregory Feero, MD, PhD[1], Nancy Stevens, MD, MPH[2], Kelly Fryer-Edwards, PhD[3,4], Susan Brown Trinidad, MA[3], and Wylie Burke, MD, PhD[3,4]

[1] *Maine-Dartmouth Family Practice Residency Program, Augusta, ME (Currently at the National Human Genome Research Institute, NIH, Bethesda, MD)*
[2]*Family Medicine, University of Washington, Seattle, WA*
[3]*Institute for Public Health Genetics, University of Washington, Seattle, WA*
[4]*Medical History and Ethics, University of Washington, Seattle, WA*

Developed in part by funds from the Maternal and Child Health Bureau, Health Resources and Services Administration

Background & Purpose: Genetics Education in Family Medicine

Most Family Medicine residencies do not formally incorporate genetics into the curriculum. Genetics topics are addressed as they arise in clinical practice.

A number of educational resources for genetics education in primary care are freely available on the Internet. However, little guidance exists to assist primary-care faculty in making use of available resources in teaching. In particular, strategies are lacking for the opportunistic integration of this material into the existing curriculum.

Key requirements for the curriculum were to:
(1) Develop a modular approach to ensure flexibility for different topics and teaching formats,
(2) Ensure peer review by teaching faculty,
(3) Account for relevant ACGME and RRC requirements for Family Medicine, and
(4) Ensure ease of access, downloads, and updates.

The Model Curriculum Working Group

The Model Curriculum Working Group was established in November 2005 to develop strategies to help faculty integrate existing tools for genetics teaching into local curricula. The Working Group convened twice to:

(1) Review existing online resources,
(2) Review applicable ACGME and RRC requirements, and
(3) Develop strategies for incorporating genetics into the existing Family Medicine curriculum.

Tools identified for use:
GeneticTools, www.genetictools.org
GeneTests, www.genetests.org
AAFP CME unit, www.aafp.org/online/en/home/clinical/acf/genomics/acfgenomics.html

Thanks to the members of the Model Curriculum Working Group: Susie B
Feero, MD, PhD, Maine-Dartmouth Family Practice Residency Program, K
Washington, Valerie Ross, MS, University of Washington, Kerry Silvey, MA, Children's Development & Rehabilitation Center, Oregon Health & Science University, Nancy Stevens, MD, MPH, University of Washington, Michael Stehney, MD, MPH, Middlesex Hospital, Susan Brown Trinidad, MA, University of Washington, Adam Wilikofsky, PhD, Lancaster General Hospital, Calanthe Wilson-Pant, MD, McLaughlin Research Institute

Suggested Teaching Approaches Template

Suggested Teaching Approaches were developed for autism/developmental delay; Alzheimer disease/dementia; breast and ovarian cancer; colorectal cancer; hereditary hemochromatosis; family history; newborn screening; and pharmacogenomics.

With this modular approach, faculty can select the most appropriate tools and teaching points, whether they have 5 minutes at the bedside or a full hour-long didactic presentation. Suggestions for self-directed learning are also offered. Relevant ACGME/RRC competencies are identified.

Suggested Teaching Approaches: Hereditary Colorectal Cancer

Format 1: Point of Care	(~ 5 minutes) Also available: Suggested teaching approaches for Morning Report (~20 min), Noon Conference/Didactic Session (~60 min), and Self-Directed Learning
Setting & approach—where and how will this information be used by faculty?	These suggestions are intended to help faculty be prepared for questions that come up in clinical teaching, and to respond to learners' requests for additional information
Suggested pre-reading for faculty and/or discussion leaders	At-A-Glance topic on colorectal cancer, www.genetictools.org Suggested sites: www.ccalliance.org; http://ghr.nl,.nih.gov, www.genereviews.org, UpToDate, MDConsult Suggested search terms: FAP, hereditary colorectal cancer, HNPCC
Suggested mini-topics	***Medical Knowledge*** What percentage of CRC cases may be hereditary? What are "red flags" suggesting the possibility of an inherited CRC syndrome? What are the three major entities of inherited CRC? Compare and contrast HNPCC and FAP. Discuss evaluation options for suspected cases. Discuss screening options/recommendations for those at increased risk for CRC, and implications for other family members. Discuss prophylaxis/treatment options for those identified as having HNPCC or FAP ***Interpersonal & Communication Skills*** How might one integrate a discussion of hereditary cancer risk into a routine health maintenace visit? How might one best elicit a patient's concerns regardng a family history of colon cancer? ***Professionalism & Ethical, Legal, Social, and Cultural Implications*** What is the role (obligation?) of the physician to family members that may be at risk for hereditary CRC? What are the potential economic and social impacts of the diagnosis of HNPCC or FAP on the patient?
Suggested additional reading/information for learners	At-A-Glance topic on colorectal cancer, www.genetictools.org GeneticTools case 8: a woman unaware of her family history of colorectal cancer GeneticTools case 9: colorectal cancer in a 28-year-old woman NCI's Genetics of Colorectal Cancer PDQ: www.nci.nih.gov/cancertopics/pdq/genetics/colorectal/healthprofessional
Outcome—main teaching points or desired behavior change	Increased recognition of the contribution of hereditary factors to the burden of CRC Improved understanding of the evaluation and management of patients at risk for, or having, hereditary CRC Recognition that significant economic, ethical, and psychosocial issues surround a diagnosis of a hereditary CRC syndrome

Format 1: Point of Care (~ 5 min)	
Setting and approach Where and how will this information be used by faculty?	These suggestions are intended to help faculty be prepared for questions that come up in clinical teaching, and to respond to learners' requests for additional information.
Suggested pre-reading for faculty and/or discussion leaders Resources and/or references you would recommend to help faculty/discussion leaders feel comfortable with this topic	▸ At-A-Glance topic on Colorectal Cancer, www.genetictools.org ▪**Suggested Sites:** o www.ccalliance.org o http://ghr.nlm.nih.gov o www.genereviews.org o UptoDate o MDConsult ▸ **Suggested search terms: FAP, hereditary colorectal cancer, HNPCC**
Suggested mini-topics Small "chunks" of information that address questions that commonly arise in clinical precepting and point-of-care teaching. Preceptors and other faculty should be able to quickly scan these topics, then choose an appropriate mini-topic or two (based on current cases, residents' questions, or faculty interest) to incorporate in teaching.	***Medical Knowledge*** ▸ What percentage of CRC cases may be hereditary? ▸ What are "red flags" suggesting the possibility of an inherited CRC syndrome? ▸ What are the three major entities of inherited CRC? ▸ Compare and contrast HNPCC and FAP. ▸ Discuss evaluation options for suspected cases. ▸ Discuss screening options/recommendations for those at increased risk for CRC, and implications for family members. ▸ Discuss prophylaxis/treatment options for those identified as having HNPCC or FAP. ***Interpersonal & Communication Skills*** ▸ How might one best integrate a discussion of hereditary cancer risk into a routine health maintenance visit? ▸ How might one best elicit a patient's concerns regarding a family history of colon cancer? ***Professionalism & ELSI*** ▸ What is the role (obligation?) of the the physician to family members that may be at risk for hereditary CRC? ▸ What are the potential economic and social impacts of the diagnosis of HNPCC or FAP on the patient?
Suggested additional reading/information for learners Resources and/or references you would recommend for those who want to learn more about this topic	▸ At-A-Glance topic on Colorectal Cancer , www.genetictools.org ▪GeneticTools case 8, a year-old woman unaware of her family history of colorectal cancer, http://www.genetests.org/servlet/access?id=8888892&key=4ogu0lp2Lwiio&fcn=y&fw=CZHV&filename=/tools/cases/colorectal-8/index.html ▸ GeneticTools case 9, Colorectal cancer in a 28-year-old woman, http://www.genetests.org/servlet/access?id=8888892&key=4ogu0lp2Lwiio&fcn=y&fw=sZ0B&filename=/tools/cases/colorectal-9/index.html ▸ National Cancer Institute's "Genetics of Colorecetal Cancer PDQ," http://www.nci.nih.gov/cancertopics/pdq/genetics/colorectal/healthprofessional
Outcome What are the main teaching points or desired behavior changes to be addressed?	▸ Increased recognition of the contribution of hereditary factors to the burden of CRC. ▸ An improved understanding of the evaluation and management of patients at risk for or having hereditary CRC. ▸ Recognition that significant economic, ethical, and psychosocial issues surround a diagnosis of a hereditary CRC syndrome.

Format 2: Morning Report (~ 20 min.)	
Setting and approach Where and how will this information be used by faculty?	Brief informal discussion of case-based materials presenting major points regarding Hereditary Colorectal Cancer. Could be led by faculty or chief/senior resident. Advance preparation is expected.
Suggested pre-reading for faculty and/or discussion leaders Resources and/or references you would recommend to help faculty/discussion leaders feel comfortable with this topic	▸ At-A-Glance topic on Colorectal Cancer, www.genetictools.org ▸ GeneticTools case 8, a year-old woman unaware of her family history of colorectal cancer, http://www.genetests.org/servlet/access?id=8888892&key=4ogu0lp2Lwiio&fcn=y&fw=CZHV&filename=/tools/cases/colorectal-8/index.html ▸ GeneticTools case 9, Colorectal cancer in a 28-year-old woman, http://www.genetests.org/servlet/access?id=8888892&key=4ogu0lp2Lwiio&fcn=y&fw=sZ0B&filename=/tools/cases/colorectal-9/index.html
Suggested approaches Your suggestions for how to teach this material in the Morning Report setting, within the available time, and offering developmentally appropriate learning opportunities for both junior and senior house staff.	***Approach 1: Build on Mini-Topics*** ▸ Some of the 5-minute mini-topics suggested for Point of Care teaching (above) can be expanded, modified, or combined to work in a 20-minute teaching discussion. Add a 1 or 2-sentence case at the start to introduce the topic, and spend more time asking questions and discussing each point. ***Approach 2: Case Presentation*** On appropriate inpatient teaching service (Family Practice, Internal Medicine, Surgery) with morning report or teaching rounds, select a patient with CRC for presentation. Assign a team member to briefly present the case (3–5 minutes). Assign team pre-reading and set the date for case presentation. Direct discussion towards the issues selected from above Mini-Topics.
Suggested discussion topics/questions Your suggestions for guiding discussion of key issues about the topic and/or case.	***Medical Knowledge*** ▸ What percentage of CRC cases may be hereditary? ▸ What are “red flags” suggesting the possibility of an inherited CRC syndrome? ▸ What are the three major entities of inherited CRC? ▸ Compare and contrast HNPCC and FAP. ▸ Discuss evaluation options for suspected cases. ▸ Discuss screening options/recommendations for those at increased risk for CRC, and implications for family members. ▸ Discuss prophylaxis/treatment options for those identified as having HNPCC or FAP. ***Interpersonal & Communication Skills*** ▸ How might one best integrate a discussion of hereditary cancer risk into a routine health maintenance visit? ▸ How might one best elicit a patient’s concerns regarding a family history of colon cancer? ▸ What is his/her reaction to, and comprehension of, the material surrounding hereditary CRC? ▸ How might further evaluation and genetic testing be helpful? Harmful? ***Professionalism & ELSI*** ▸ What is the role (obligation?) of the the physician to family members that may be at risk for hereditary CRC? ▸ What are the potential economic and social impacts of the diagnosis of HNPCC or FAP on the patient?

Format 3: Noon Conference / Didactic Session (~ 1 hour)	
Setting and approach Where and how will this information be used by faculty?	This section helps faculty prepare a formal lecture-style presentation of information on Colorectal Cancer, in a Noon Conference or other didactic classroom-style setting. Suggestions for faculty pre-reading, as well as learner preparation, are offered.
Suggested pre-reading for faculty and/or discussion leaders Resources and/or references you would recommend to help faculty/discussion leaders feel comfortable with this topic	Audience: ▸ At-A-Glance topic on Colorectal Cancer, www.genetictools.org Presenter: ▸ National Cancer Institute's "Genetics of Colorecetal Cancer PDQ," http://www.nci.nih.gov/cancertopics/pdq/genetics/colorectal/healthprofessional
Suggested approaches Your suggestions for how to teach this material in the Noon Conference / Didactic setting, within the available time	Select a noon conference slot every 3 years for this topic. Assign faculty with interest in colorectal cancer to review slide sets and update if needed. Selected faculty will need to be familiar with the topic in order to effectively present and field questions. Limit presentation to 45 minutes, allowing 15 minutes for discussion of questions and/or cases of interest. (See below for suggested discussion topics/questions)
Suggested classroom materials If you know of teaching tools (eg, handouts, slide sets, websites) that can be used effectively in this setting, please note them here.	▸ At-A-Glance topic on Colorectal Cancer, www.genetictools.org, as handout ▸ Slide sets available from CDC genetic educational materials web site (e.g., those posted by the University of North Carolina). This may require permission (which this Working Group should obtain) and updating/tailoring to audience. http://www.sph.unc.edu/nccgph/tools/colon_cancer2_files/frame.htm ▸ AAFP Annual Clinical Focus Colorectal Cancer video module (***Note:*** requires high-speed Internet access), http://www.aafp.org/x30164.xml
Suggested discussion topics/questions Your suggestions for guiding discussion of key issues about the topic and/or case	***Medical Knowledge*** ▸ What percentage of CRC cases may be hereditary? ▸ What are "red flags" suggesting the possibility of an inherited CRC syndrome? ▸ What are the three major entities of inherited CRC? ▸ Compare and contrast HNPCC and FAP. ▸ Discuss evaluation options for suspected cases. ▸ Discuss screening options/recommendations for those at increased risk for CRC, and implications for family members. ▸ Discuss prophylaxis/treatment options for those identified as having HNPCC or FAP. ***Interpersonal & Communication Skills*** ▸ How might one best integrate a discussion of hereditary cancer risk into a routine health maintenance visit? ▸ How might one best elicit a patient's concerns regarding a family history of colon cancer? ▸ What is his/her reaction to, and comprehension of, the material surrounding hereditary CRC? ▸ How might further evaluation and genetic testing be helpful? Harmful? ***Professionalism & ELSI*** ▸ What is the role (obligation?) of the the physician to family members that may be at risk for hereditary CRC? ▸ What are the potential economic and social impacts of the diagnosis of HNPCC or FAP on the patient?

http://edoctoring.ncl.ac.uk/System_Check/

Live Search

Dogpile Web Search Home P... | eDoctoring

Page | Tools

Login | Contact us

home | about eDoctoring | Gallery | Feedback | Portfolio

Welcome to the eDoctoring website.

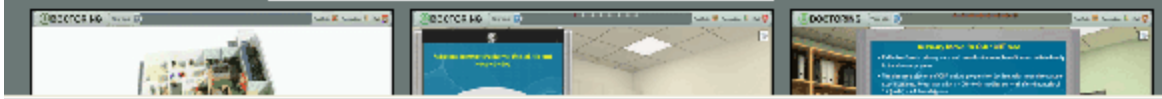
The project is a collaboration between the Newcastle University and the University of California. Its aim is to provide an online support tool for the Doctoring curriculum through the use of highly interactive case based material.

Requires login **Request Login**
You need to download flash 8 to use eDoctoring **Click here to install plug-in**

News

- New end of life cases are being developed and deployed within the system,
- New graphics for the whole site have been applied,
- New content delivery system applied to allow the multiple eDoctoring systems to operate in a new hospital based enviornment,
- New 3D graphics applied to the prostate / genetics cases, including new animated office interactions,
- Multiple genetics cases available online,
- Take a peek at some early eDoctoring screenshots in the **'Gallery'**.

Screenshots View more screenshots in the gallery



Internet | 100%

e-Doctoring screenshots from March 2007 - release version. Comments from Dan Plummer, programmer.

The eDoctoring system uses a portfolio to monitor your progress through the cases.

On your first login to the system you are given a virtual tour of the enviroment.

Patients / Cases are assigned according to your course and cover a wide variety of material from Genetics to Screen tests.

eDoctoring uses videos to help visualise the cases.

The cases are narrated by professor agents and use graphical displays to emphasise points in the learning material.

To make the material more interactive, questions are used through out to test your knowledge.

At the end of each case, a summary screen highlights the important points made about the patient and condition.

Some of the more important aspects of the cases are supported by seperate tutorials that provide more in-depth material.

[Older Screenshots]

Top 12 topics for PA education in genetics

Approaches:

- PDA support (GeneFacts - NCHPEG)
- Virtual clinic tool (NCHPEG, eDoctoring, Dartmouth)
- Article series (Rocky and others)
- Video CME (AAFP ACF)

Conclusions

- NHGRI is currently engaged in a variety of activities that will affect PA practice
- Key points for collaboration include awareness campaigns and development of educational resources
- Opportunities exist for inter-disciplinary synergy in these key areas

Physician Assistants and Genomic Medicine: Update from AAPA

September 19, 2007

Bob McNellis

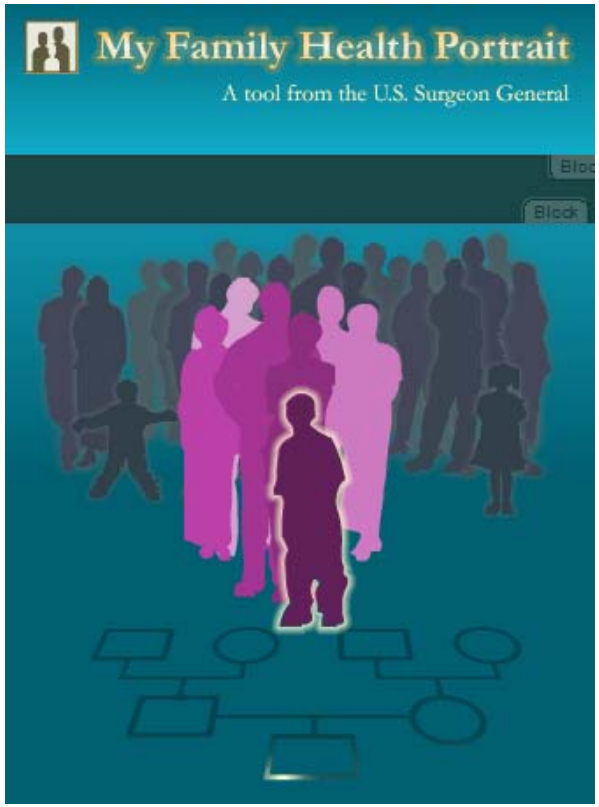
Director, Science and Education

Areas of activity

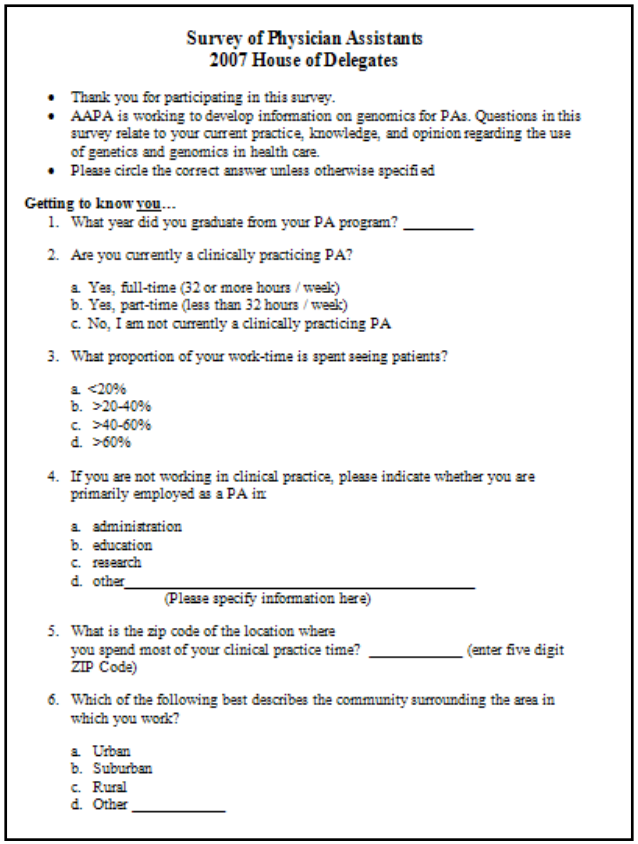
- 2007 Annual Conference
- AAPA News articles + JAAPA editorial
- Web page and post-tests
- Legislation, policy and partnership
- 2008 Annual Conference
- Other future activities
- Ponderings

2007 Annual Conference

Provided exhibit hall space for NCHPEG and the Office of the Surgeon General



A 19 question survey was distributed in AAPA's House of Delegates



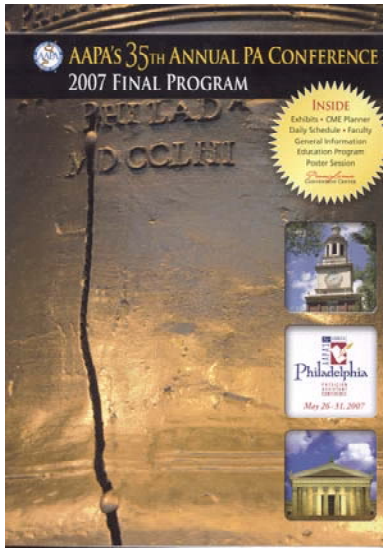
Survey of Physician Assistants
2007 House of Delegates

- Thank you for participating in this survey.
- AAPA is working to develop information on genomics for PAs. Questions in this survey relate to your current practice, knowledge, and opinion regarding the use of genetics and genomics in health care.
- Please circle the correct answer unless otherwise specified

Getting to know you…

1. What year did you graduate from your PA program? _____
2. Are you currently a clinically practicing PA?
   a. Yes, full-time (32 or more hours / week)
   b. Yes, part-time (less than 32 hours / week)
   c. No, I am not currently a clinically practicing PA
3. What proportion of your work-time is spent seeing patients?
   a. <20%
   b. >20-40%
   c. >40-60%
   d. >60%
4. If you are not working in clinical practice, please indicate whether you are primarily employed as a PA in:
   a. administration
   b. education
   c. research
   d. other_____
   (Please specify information here)
5. What is the zip code of the location where you spend most of your clinical practice time? _____ (enter five digit ZIP Code)
6. Which of the following best describes the community surrounding the area in which you work?
   a. Urban
   b. Suburban
   c. Rural
   d. Other _____

2007 Annual Conference Program



At least <u>eight</u> hours of continuing medical education included information on medical genetics

- Headlines and heredity
- Personalized medicine: Integration of medical genomics into clinical practice
- Gender differences in cardiovascular disease
- Sex and gender in the clinical encounter: The importance of considering both
- Ovarian cancer early detection, treatment and support: Issues for PAs
- Cancer genetics and genomics: Implications for PA practice
- Using new modalities to improve outcomes in macular degeneration
- Race-based therapeutics? What's the BiDil (Big Deal)?

Conference Daily

PAs Can Explore New World of Personalized Medicine

By Doug Scott

There is a growing misconception in the medical community that to integrate genomics and genetic medicine into a clinical practice, a clinician needs to be highly educated and trained in this field. Some providers also believe that genomics and genetic medicine are not applicable to their clinical practice.

But this is not true, according to PA Michael "Rocky" Rackover, who recently completed a four-month sabbatical at the National Human Genome Research Institute (NHGRI), where he worked alongside two world-renowned experts in the field of genetic medicine: Francis Collins, M.D., director of the NHGRI, and Alan Guttmacher, M.D., deputy director.

"We are entering a time when the dream of personalized medicine will become a reality," said Rackover, who is program director and associate professor at the Philadelphia University PA Program. "Instead of treating patients in a one-size-fits-all fashion, a PA will be able to use information technology and medical genetics to provide a focused, individualized approach to health care.

"I have heard many PAs say that they fear using genomics and genetic medicine in their clinical practice because they do not have the education. But a common sense utilization of basic genomic indicators, such as taking an informative, three-generation family history; recognizing patterns in family history; using available resources; and knowing when, where, and how to refer to a geneticist or genetic counselor, will allow PAs to treat their patients more individually and effectively."

Using this information, he said, will help patients become more informed, personal, preventive medicine stakeholders in their own health.

In a CME session called Personalized Medicine: Integration of Medical Genetics into Clinical Practice, Rackover and PA Connie Goldgar will explain through common case studies that by using what they term "personalized medicine," a PA can integrate genomic interventions that are targeted to individuals based on their risk to provide a more coherent and focused approach to the patient's care.

The presentation is scheduled for tomorrow at 8:00 a.m. in the Pennsylvania Convention Center Room 111.

Rackover and Goldgar describe personalized medicine as preventive care, diagnostic care, and therapeutic interventions. First, a

Michael Rackover

Connie Goldgar

Click to zoom out

ersal principles of understanding human risk factors are defined through family history or, eventually, their genetic make-up. Then strategies for prevention, detection, and treatment of disease can be tailored to that individual.

Goldgar, associate director of the University of Utah PA Program, explained that the two


Arcadia


National Medical
Challenge Bowl


ASSISTANT
CONFERENCE
20

AAPA News articles

January 30, 2007

PA Helps Take Profession into Next Age of Medicine

By DOUG SCOTT

When medical historians look back at the beginning of the 21st Century, they will surely conclude that it was the end of the age of antibiotics, imaging, and anesthesia, and the beginning of the age of genomic medicine.

Francis Collins, M.D., director of the National Human Genome Research Project (NHGRP), who is noted for his landmark discoveries of disease genes and visionary leadership in the development of the Human Genome Project, recently told *AAPA News* that genomics is going to change the way clinicians practice medicine.

Whether regarding a patient's risk of breast cancer or heart disease, Collins said, the most important thing a PA can do to incorporate genetic thinking into his or her daily practice is to recognize that all diseases have hereditary contributions. Today, he said, the best method is to take a good family history that will help predict the patient's risk of disease. In the future, however, the patient history will be augmented by specific genetic testing.

Michael "Rocky" Rackover recently spent four months as a visiting scholar at the National Human Genome Research Institute in Bethesda, Maryland.

Genetic Tools Will Help Providers, Educators — page 6

"I think it's great that PAs are really out in front in recognizing the relevance of genomic medicine," said Collins, speaking from his office at the National Institutes of Health (NIH) in Bethesda, Maryland.

"We are well aware that unless the new knowledge, tools, and approaches that have come from the Human Genome Project are integrated into clinical practice, the project's huge potential to improve health will have been merely a false promise. Rocky [Rackover] has been particularly helpful to us in thinking about

See NEXT AGE on page 6

January 30, 2007

New Web-based Tool Designed to Help Providers, Educators Teach Genetic Medicine

By DOUG SCOTT

A Web site launched last fall now provides PA and medical educators with concepts and skills, teaching cases, background information, implications, and links to other resources about genetics in a primary care setting.

The new resource, Genetic Tools, sponsored by the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA) and developed by the University of Washington School of Medicine, offers principles of genetic medicine that will help health care providers and educators do a better job of teaching their students.

"If PAs want to do a better job of diagnosing, preventing, and treating most of the common medical conditions that fill our clinics and hospitals, it is essential to have a general grasp of genetic principles, and it will become even more essential in the coming years," said Francis Collins, director of the National Human Genome Research Institute.

As a measure of its importance to PA education, the Accreditation Review Commission on Education for the Physician Assistant last September included genetics and molecular mechanisms of health and diseases in the core curriculum of PA education.

"The target of our tool is to assist primary care practitioners and faculty and to put genetics into their curriculum," explained Wylie Burke, M.D., project director of Genetic Tools and professor and chair of the Department of Medical History and Ethics at the University of Washington School of Medicine.

Burke said that, in developing the Web site, she found that many medical and PA programs had limited genetics content. She said that faculty need to know or refresh their knowledge of genetics and they need to recognize teachable moments in genetics. Educators, she said, have the perception that genetics is only associated with rare, single-gene disorders.

"The amount of genetics training in medical and PA school curriculum is going up," Burke noted, "but most clinicians in primary care practice today have received very little formal genetics training and some almost [none]. We know genetics issues do come up when [clinicians] see patients, but too many health care providers are not comfortable taking a three-generation family history or [lack] the time to interpret the findings or [don't know] whom to call for help.

"What we hope to do with Genetic Tools is to change the way clinicians think about genetics in their primary care practice and, to do that, we need to start training the next generation of medical and PA students."

The Genetic Tools site contains four major categories. The first, Genetic Concepts and Skills, provides health care providers, educators, and students with general background information to "think genetically," she said. It includes understanding modes of inheritance, taking a family history, using genetic tests, the rationale for genetic test screening, and using evidence-based medicine in genetics.

The essence of the Web site can be found in the Teaching Cases category, which includes 41 cases, ranging from Alzheimer's disease to Thrombophilia, illustrating how genetic conditions and questions arise in a primary care setting. Burke said that each of the 41 cases provides learning objectives; identifies family history; and reviews clinical care issues, risk assessment, and testing, including relevant ethical, social, legal, and cultural issues. Links to other relevant Web sites are provided.

"When we came to developing the cases for this Web site, we came to the realization that every case should be structured in a predictable way and hit each of the main areas of content that our educators are looking for," said Burke, who was assisted by 36 team members and project officers who are recognized as experts in primary care and genetics.

A separate category dealing with implications of genetic diagnosis, entitled Ethical, Legal, Social, and Cultural Issues, provides helpful information on such topics as unwanted genetic information, potential for genetic discrimination, informed consent, prenatal testing and pregnancy termination, confidentiality and privacy issues, and race and ethnicity issues.

The fourth category, Other Resources, gives educators and students links to professionally reviewed on-line resources, such as *GeneReviews*, at-a-glance snapshots that provide single-page summaries of topics and conditions most commonly taught in primary care, approaches to clinical teaching, and faculty resources.

"What a lot of educators have not realized is that the genetics train has already left the station and if they don't jump on board now, when it really speeds up, it is going to be hard to catch up," said Collins.

Before releasing Genetic Tools in September, it was tested by the Brody School of Medicine at East Carolina University, New York Medical College, University of Washington Family Medicine Network, and Baylor College of Medicine. There is a feedback section on the Web site for faculty and students to use, and Burke said that all the responses have been positive.

Genetic Tools can be found on the Gene Test Web site at *www.genetests.org* by clicking on Visit Genetic Tools in the lower-right-hand corner or by going directly to *www.genetests.org/servlet/access?id=INSERTID&key=INSERTKEY&fcn=y&filename=/tools/index.html*.

"Every one of the 41 cases are dated, so ideally we would like to have an editor review each case on a rolling, two-year schedule," said Burke. "Obviously, in addition to new cases, there are plenty of opportunities to develop additional curriculum guides."

May 15, 2007

NIH, Genomics Experts, PA Organizations Meet To Plan a Future for PAs in Genomics

Physician Assistants Poised to Introduce Genomic Medicine in Their Practices

By Doug Scott

An historic conference in March between the leaders of four PA organizations, the National Institutes of Health (NIH), and leading experts in the field of genomics was the first step in planning a future where PAs can take a leadership role in introducing genomic medicine in their practices.

The goal of the two-day meeting, held at NIH headquarters in Bethesda, Maryland, was to develop an outline for how PAs could utilize current and anticipated knowledge of genetics and genomics as a basis for improving clinical care and making personalized medicine a regular part of patient care.

"What came out of this meeting was a remarkably detailed and ambitious agenda," said Francis Collins, M.D., director of the National Human Genome Research Institute. "It was created by this group in a fashion that showed remarkable collaborative capabilities to push the agenda of genomic medicine with physician assistants as change agents in a way that I did not think possible before this meeting."

The conference, entitled Physician As-

PHOTO COURTESY OF MAGGIE BARTLETT, NHGRI

Leaders of AAPA and other PA organizations joined representatives from the National Human Genome Research Institute, other genomics experts, and Acting U.S. Surgeon General Kenneth Moritsugu to define how PAs can best introduce genomics in patient care.

May 30, 2007

Genetics Web Tool for PAs to Be Unveiled at Annual Conference

By Doug Scott

At the National Institutes of Health's National Human Genome Research Institute's (NHGRI) recent meeting on Physician Assistant Competencies for Genomic Medicine: Where We Are Today and How to Prepare for the Future, NHGRI Director Frances Collins, M.D., asked, "What is it that PAs need to know in terms of implementing genetics and genomics in their daily practice?"

One approach to answering that question is the development of the first-ever Web-based program written for and by PAs that is devoted to the role of genetics and genomics in primary care. The program, Genetics in a Physician Assistant's Practice, was developed through a $20,000 grant that AAPA received last year from the National Coalition for Health Professional Education in Genetics (NCHPEG).

Practicing PAs, PA students, and PA educators can all use the program. Through examples of common primary care cases, users can explore genetic and genomic skills and concepts, learn to perform risk assessments, gain an understanding of genetic counseling and testing, hear about new treatment options, and receive links to valuable genetic resources. Organized like a typical medical office, the site walks the user through a case from chief complaint to management plan, including data collection, physical examination, laboratory findings,

Organized like a typical medical office, the new Web-based site — Genetics in a Physician Assistant's Practice — walks PAs through a case from chief complaint to management plan, including data collection, physical examination, laboratory findings, and consultations.

opportunity to earn CME credit.

The site aims to provide PAs with the knowledge to collect basic but informative family history, identify patients and families who may benefit from genetic services, locate and refer patients to genetics professionals, and consult and collaborate with genetics professionals in patient management.

"What we tried to do," explained PA Connie Goldgar, principal investigator and lead author, "was aim this at a level of PA utility. We wanted to give just enough underpinnings of genetic information that makes it understandable to what a PA does in their practice."

site, PAs can enhance their family history-taking skills. Users can listen to or read a simulated conversation between a PA and a patient; a consultation between a PA, genetic counselor, and supervising physician; or a review of laboratory tests with the patient.

The group that produced this resource did "a good job of identifying [genomic] cases that PAs are going to relate to, and I am optimistic that this site is going to work well," said Joe McInerney, executive director of NCHPEG, who has helped design similar genomic sites for other health professions. "I really like the look and feel of it now, and I believe that it is a little more engaging and easily navigable than some of the other sites that we have done."

"We wanted this to be user-friendly and practical for the PA," Goldgar said. Users can collect a patient's history, with a focus on the family history. Red flags may indicate a potential genetic condition, then a differential diagnosis is formulated and narrowed with information from the physical exam and the laboratory — just as PAs do in everyday clinical problem solving. The PA needs to know what steps must occur next in the management of these common health conditions with a genetic element.

The site will be unveiled at AAPA's 35th Annual PA Conference in Philadelphia at the NCHPEG booth in the Exhibit Hall. PAs who visit the booth will be able to navigate the program with the assistance of the PA designers of the project. McInerney expects the site to be fully operational this summer, when it will be available at *www.nchpeg.org* or *http://pa.nchpeg.org*.

"This is a really important first step for PAs in learning genomics and genetics," said McInerney. "We now have this wonderful educational material, which should have extraordinary content validity. AAPA leadership and the PA community have really done a tremendous amount of work in bringing genetics to their constituents, and PAs should be very proud of that."

The primary authors of the project are Goldgar, associate director of the University of Utah PA Program; Chantelle Wolpert, PA and genetic counselor at Medical Genetics at Duke University; Kristine Healy, PA and assistant professor at Midwestern University PA Program; Karen Clark, PA and genetic counselor, Midwestern University PA Program; and Erin Harvey, NCHPEG project manager.

"This has been a lot of fun, a huge learning experience, and a lot of work," said Goldgar. "But the bottom line is that we hope that we have a product that gets PAs to say, 'This is information that I need to know now, and it is going to positively impact the care of my patients.' "

July 30, 2007

GINA Bill to Play Major Role in Advancing Genomics And Genetic Medicine to Patients

First in an occasional series on genomics and genetic medicine

BY DOUG SCOTT

In 1997 Sen. Olympia Snowe (R-Maine) received a letter from Bonnie Lee Tucker, a Maine resident, who feared that her daughter would have to take a BRAC Analysis genetic test for breast cancer. Having nine members of her immediate family diagnosed with breast cancer, and a survivor herself, Tucker feared for her daughter's future. She knew that genetic testing was available, but was afraid that the results would lead to discrimination affecting her daughter's insurance coverage and future employment.

In each of the past six Congresses, Snowe has introduced a version of the Genetic Information Nondiscrimination Act (GINA), which is aimed at assuring that patients and their families who participate in genetic research, testing, and therapies will not suffer from discrimination. The bill twice has passed unanimously in the Senate, but failed in the House of Representatives.

On April 25, GINA finally was passed by the House, 420-3. Key provisions call for federal standards that support the extension of medical privacy and confidentiality rules to genetic information, make it unlawful to discriminate against an employee or deprive an individual of employment opportunities because of genetic information, and prohibit the collection and distribution of genetic information.

"GINA will make discriminatory practices illegal by prohibiting health insurers from denying coverage or charging higher premiums to a healthy individual because of a genetic predisposition to develop a disease in the future," Rep. Louise Slaughter (D-N.Y.), who sponsored the legislation in the House, told *AAPA News*.

"It also bars employers from using genetic testing information for hiring, firing, job placement, or promotional decision. Simply put, GINA will stamp out a new form of discrimination."

Support from the PA Community

AAPA has supported the GINA bill and the advancement of genomics and genetic medicine. In March, AAPA joined the National Commission on Certification of Physician Assistants (NCCPA), the Accreditation Review Commission on Education for the Physician Assistant (ARC-PA), and the Physician Assistant Education Association (PAEA) for an historic meeting organized and supported by the National Human Genome Research Institute (NHGRI) that was entitled Physician Assistant Competencies for Genomic Medicine: Where We Are Today and How to Prepare for the Future (*AAPA News*, May 15).With new developments making genetics increasingly important to the practice of health care, the meeting concluded that PAs are poised to be a vital part of the health care team in integrating existing genomic tools, such as family history, into practice.

"We all benefit through the advancement of medical science," said AAPA Immediate Past President Mary Ettari, "and the GINA bill provides the necessary protections for patients to embrace the science of genetics."

Patients are going to need access to genetic tests after the bill passes, and PAs are in a position to spend more time with patients to be able to deliver the kind of information that patients are going to need, said Sharon Terry, president and CEO of Genetic Alliance, a coalition of more than 600 advocacy organizations that has lobbied for passage of GINA for the past 12 years.

"If we look at how genomics and genetics are going to be integrated into medicine, one of the first lines of defense will be the collection of a good family history and the second is the genetic test, which will do everything from diagnosing somebody to helping to determine what testing," added Susannah Baruch, director, Reproductive Genetics, Genetics and Public Policy Center at the Johns Hopkins University.

"We have seen and heard reports of people's reluctance to undergo genetic tests because they fear that whatever privacy they are promised without a law in place [will come] back to bite them."

A 2007 study done by the Genetics and Policy Center found that 86 percent of participants expressed some or a lot of trust in their clinician's having access to their genetic test. Only 24 percent and 16 percent expressed such trust regarding their insurer and employer, respectively.

"I assume that a lot of PAs have seen or will start to see patients where this is a question in their mind," said Baruch, "and [passage of] this bill will help PAs to reassure their patients."

Chances of Becoming Law

The White House said in a statement to *AAPA News*: "The administration favors enactment of legislation to prohibit the improper use of genetic testing information in health insurance and employment and supports House passage of H.R. 493."
A White House spokesperson said that once GINA is passed by the Senate, the president will sign the bill that both houses adopt.

Easy passage is expected again in

August 30, 2007

Personalized Medicine: the Right Prescription for Your Patient

Second in an occasional series on genomics and genetic medicine.

By Doug Scott

One of the most confusing things about the term "personalized medicine" is that most PAs will tell you that is what they are doing right now. PAs are listening to a patient's responses while they are writing prescriptions or treatment plans. To suggest otherwise would imply that PAs do not understand their patient's health care needs.

Yet learning and embracing the concept of personalized medicine is the first step to incorporating or *translating* genomic and genetic medicine into a PA's practice.

"Personalized health care encompasses prevention, diagnosis, and therapy, with a patient's risk for disease defined through genetics as well as through a family history," said Michael Rackover, program director and associate professor at the Philadelphia University PA Program who recently completed a sabbatical at the National Human Genome Research Institute (NHGRI).

"Most providers still assume that genetics and genomic medicine are not applicable to their current practice. Yet, an understanding of what can be learned about personalized medicine now and throughout their clinical careers will help PAs use genetic information to diagnose common disease sooner when treatment is more successful and to identify how disease can be managed more effectively."

Instead of treating patients in a one-size-fits-all fashion, Rackover said that PAs using personalized medicine will be able to use information technology and genomics to provide a more focused, individualized approach to health care.

"What we as clinicians tend to be dependent on is sort of what you might call therapeutic trials," explained Mark Williams, M.D., director of the Intermountain Healthcare Clinical Genetics Institute in Salt Lake City, Utah, where he runs a clinic for evaluating adults with mental retardation, birth defects, and genetic disorders.

> *"... We have these new tools for new gene discoveries, and this year alone, there have been some major discoveries in genes for a lot of common diseases ... The real question for all medical practitioners is: What do you do with [genetic] information? The next step for us is what we call 'translation.' The translation phase involves research, education, and developing tools and guidance for health care professionals like PAs.*
>
> — Muin J. Khoury, M.D.

Muin J. Khoury, M.D.

"Let's say you have a patient who has diabetes or hypertension or depression. There are a whole bunch of medicines that you could choose to treat them with, but you're going to start with this particular medication and there are many different reasons why it is chosen. It might be the one that is least expensive, it might be the one that is on the insurance formula, or it might be the one that the PA has the most experience with. But all these decisions are, for the most part, made without necessarily taking into account specific patient characteristics.

"Personalized medicine in the genomic sense is this: We know that a lot of the many variations or differences in response to medication and other interventions are related to changes at a genomic level. And if somehow we were able to understand those differences, then maybe we might have more information with which to choose the right medication that would be more effective and have a lower risk for development of side effects. That is the promise of personalized medicine."

Now What?

In February 2001, the Human Genome Project published the initial analysis of the human genome sequence. Already, more than 12,000 human genes have been discovered in relation to many diseases, and more than 1,000 genetic tests are available. Scientists at NHGRI will learn the sequence of 50,000 human genes over the next few years.

The question that the health community is asking is: The human genome is mapped, now what? What impact do these discoveries have in the everyday practice of PAs treating their patients?

Muin J. Khoury, M.D., director of the National Office of Public Health Genomics at the Centers for Disease Control and Prevention, said that finding the answer lies in professional health care providers ensuring the laboratory quality of genetic testing, the appropriateness of utility of genetic information in preventing disease and improving health, and the training of a workforce to meet those growing demands.

"Premature or inappropriate use of tests can lead to misdiagnosis, ineffective and confusing interventions, and a host of ethical, legal, and social issues," Khoury said. "Knowing 'now what' is a very tough question to answer, but we have these new tools for new gene discoveries, and this year alone, there have been some major discoveries in genes for a lot of common diseases like diabetes, cancer, heart disease, obesity, etc. The real question for all medical practitioners is: What do you do with [genetic] information? The next step for us is what we call 'translation.' "

The translation phase involves research, education, and developing tools and guidance for health care professionals like PAs.

"There are two areas of translation that PAs can be involved in," said Khoury. "One is the general education and literacy around genomics and potential applications. PAs need to be comfortable with the competencies of genomics so that they can apply and interpret it to themselves and their patients. The second thing they need is to be astute in providing the information around the utility of these new general applications."

This is where personalized medicine comes into play.

Grounded in Science

Based on DNA, things like eating habits, exposure to environmental factors, and types and amount of stress vary from person to person. Many of these variations play a vital role in health and disease. Variants found in genes could influence the risk of developing a certain disease as well as one's physical response to a disease. A combination of these variants across several genes can affect a patient's risk of developing a disease and can be one of the reasons that one drug works for one person and not for another.

The Personalized Medicine Coalition is an independent nonprofit organization that works to advance the understanding and adoption of personalized medicine into clinical practice. Edward Abrahams, the coalition's executive director, said the goals of personalized medicine are "to better manage a patient's disease or predisposition toward a disease, and to achieve optimal medical outcomes by helping clinicians and patients choose the disease management approaches likely to work best in the context of a patient's genetic and environmental profile."

The methods used may include genetic screening programs that more precisely diagnose diseases and their sub-types to help PAs select the type and dosage of medication best suited to a certain group of patients.

Abrahams said the integration of personalized medicine is going to change how a PA practices medicine by creating a greater reliance on genetic science, information management, and electronic health records, and a greater access to data than they ever had before.

"That should help make the medical decisions clearer and more scientifically grounded without minimizing the important role of judgment," said Abrahams.

National Office of
Public Health Genomics

Genomics & Health Weekly Update

This weekly update provides information about the impact of human genetic research on disease prevention & public health.

Get email updates

SPOTLIGHT

Family History in Pediatric Primary Care and Public Health

A new supplement published in Pediatrics summarizes the findings of a CDC-sponsored workgroup meeting on the use of family medical history information in pediatric primary care and public health.

- Access the supplement
- **CDC links on genetics, family history, and pediatrics**
  - Family history resources
  - Pediatric genetics

GENOMICS ANNOUNCEMENTS

New article published in the American Journal of Epidemiology on Turning the Pump Handle: Evolving Methods for Integrating the Evidence on Gene-Disease Association on September 5, 2007 (70KB)

Recent article published by AAPA News focuses on personalized medicine and the role of family history, genetics, and genomic applications in patient care. See quotes by Dr. Muin Khoury of CDC's National Office of Public Health Genomics and other experts. (576KB)

September public health genomics seminar presented by CDC
"How do we monitor the impact of genomics on population health?"
Sep 20, 2007, Rockville, MD (ENVISION – Atlanta, GA)

SECTIONS

UPCOMING EVENTS

First Annual NCI Meeting on Clinical Proteomic Technologies Initiative for Cancer

Association of State and Territorial Health Officials Annual Meeting

Eyes on the prize: Truth telling about genetic testing

LET'S GO SURFING

Eye on DNA - How's it going to change your life?

October 15, 2007 (in progress)

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Words:·1,179¶
Reviewed·by:·Rackover,·Goldgar,·Wolpert¶
Technical·Review·by:·McNellis¶
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Caption:·N/A¶
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How·to·Become·a·Good·Genetic·Detective¶
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Third·in·an·occasional·series·on·genomics·and·genetic·medicine¶
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→ According·to·*Risk·Assessment·for·Hereditary·Cancer·Syndromes,·A·Physician's·Guide·to·Clinical·Genetic·Testing·and·Medical·Management*,·a·report·released·by·the·American·Medical·Association·(AMA)·in·2006,·identifying·and·managing·patients·at·risk·for·hereditary·cancer·syndromes,·like·the·one·that·took·to·life·of·Jay·Monahan,·has·become·an·integral·part·of·clinical·medicine·today.·Clinicians·—·including·PAs·—·can·significantly·impact·the·future·health·of·their·patients·and·their·families·who·have·an·increased·cancer·risk·by·identifying,·educating,·counseling,·referring·to·genetic·

Journal of the American Academy of Physician Assistants
May 2007

EDITORIAL

Michael A. Rackover, PA-C, MS, is Program Director and Associate Professor in the PA program at Philadelphia University, Philadelphia, Pa. He recently completed a sabbatical at the National Human Genome Research Institute.

Embracing the new world of personalized medicine

We are entering a time when the dream of personalized medicine will become a reality. Instead of treating patients in a one-size-fits-all fashion, clinicians will use information technology and medical genetics to provide a focused, individualized approach to health care.

Personalized health care encompasses prevention, diagnosis, and therapy, with a patient's risk for disease defined through genetics as well as through clinical and family histories. Scientists have begun to understand diseases at the molecular level, allowing clinicians to see how those diseases manifest in individual patients and to tailor treatment more effectively. Genetic predisposition testing and pharmacogenetic testing will allow asthma treatment to be optimized, for example, or warfarin dosing to be better managed. In a personal communication, Alan Guttmacher, MD, Deputy Director of the National Human Genome Research Institute, told me that "genomics will irrevocably change how we practice medicine, enabling us for the first time to treat each patient truly as the individual person he or she is and thus dramatically [improve] the quality of health care."

Genomic medicine at present can be compared to the computer industry in the 1980s. As time passes, we will witness the transformation of genomic and proteomic science. These scientific advances, combined with progress in information technology, are giving new life to researchers, health care providers, and patients, forever changing the methods used for preventive, diagnostic, and therapeutic activities.[1]

Yet we are right to be cautious about this seemingly expansive future. Personalized health care is largely prevention-based, in contrast to the current delivery model, which services the acutely ill. Reimbursement will be problematic under the new model—and disparities in access to care and the need for universal health insurance still must be addressed. Also of concern is the use of genetic profiles to predict disease.

Racial and ethnic disparities in health are based on access to quality care, health risk behaviors, psychosocial factors, acculturation, biological/genetic factors, environmental and occupational exposures, and socioeconomic status. According to the National Coalition for Health Professional Education in Genetics Race, Genetics and Healthcare DVD, race may be mistakenly used to indicate ancestral origin. For example, a clinician may assume that a patient is African-American when, in reality, that person is Dominican. The two may have very different health care needs based on ancestral inheritance. When a patient's group identity might raise health issues, *ancestry* should replace *race* in discussions.

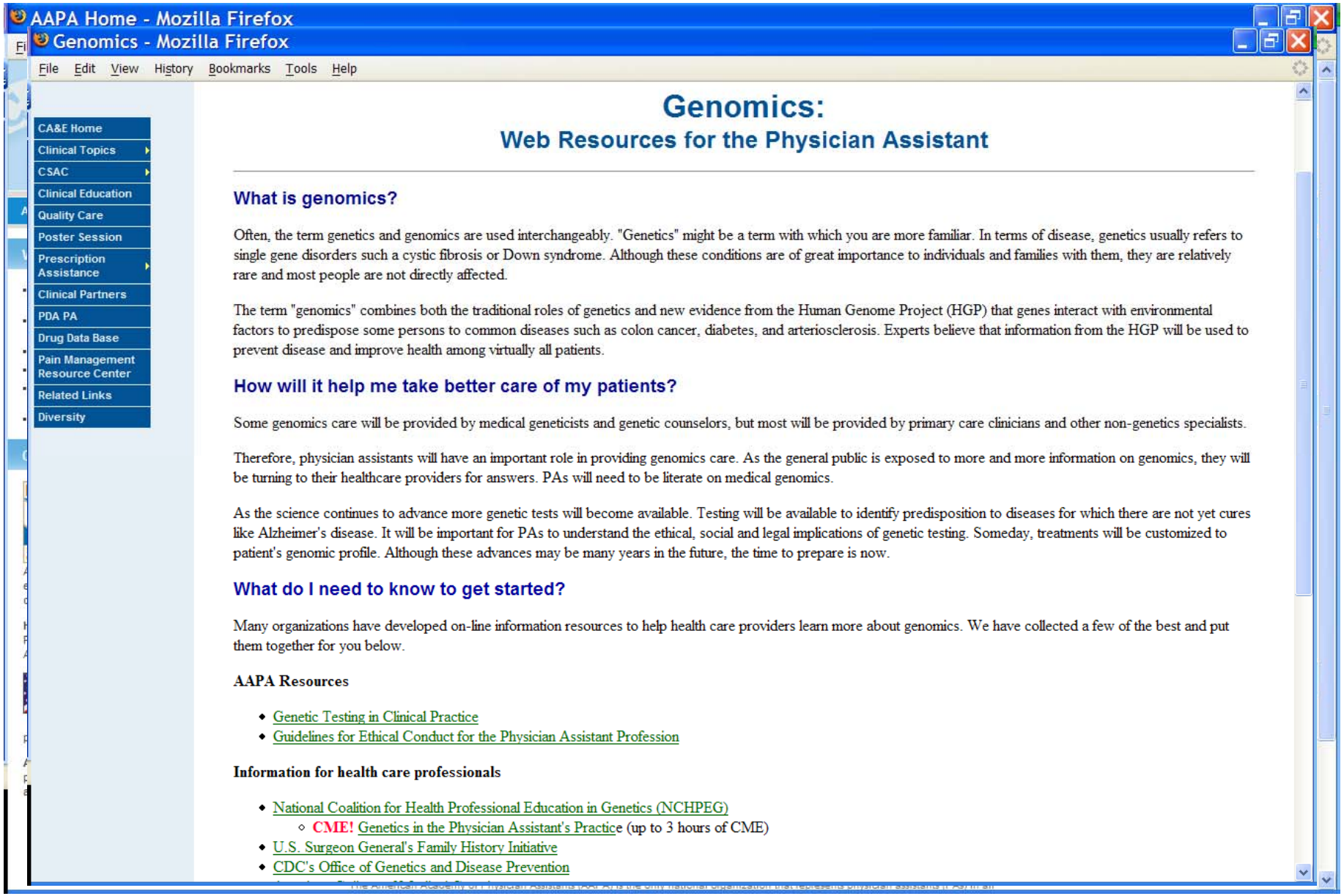
As predicted in the *Mayo Clinic Proceedings*, "the possibility of loss of confidentiality, issues of group stigmatization, and the difficult issue of genome predestination must all be addressed if humankind is to reap the full benefits of the potential of this exciting new science."[2] To protect an individual's personal information, Rep. Louise Slaughter (D-NY) introduced HR 493, the Genetic Information Nondiscrimination Act of 2007. This bill prohibits discrimination on the basis of genetic information with respect to health insurance and employment. Its passage would remove a potential barrier to the use of genetic testing and other genetic services.

The Personalized Medicine Coalition is an independent, nonprofit group that works to advance the understanding and adoption of personalized medicine for the ultimate benefit of patients. They have stated that "in considering whether personalized medicine has a viable future, a look into the past reveals that it has always been here in some form. But in its modern manifestation, which relies on molecular analysis and proactive care, personalized medicine will require an extensive system of support. This system will include new regulatory approaches, revamped medical education curricula, integrated health information systems, legislation to protect against genetic discrimination, insurance coverage for sophisticated molecular diagnostic tests, and a reimbursement system that encourages proactive care. Because of the many hurdles before it, some experts have questioned whether personalized medicine will become a dominant trend in healthcare, or just a passing phase."[3]

Most providers still assume that genetics and genomic medicine are not applicable to their current practice. Yet an understanding of what can be learned about personalized medicine now and throughout their clinical careers will help PAs to use genetic information to diagnose common diseases sooner, when treatment can be most successful, and to identify how diseases can be managed more effectively. For this process to be a success, however, we must commit ourselves to ongoing learning about how this new knowledge will help us take care of patients. JAAPA

REFERENCES

1. IBM Executive Brief. Personalized healthcare 2010: are you ready for information-based medicine? Available at: http://www-03.ibm.com/industries/healthcare/doc/content/resource/insight/992304105.html. Accessed April 10, 2007.
2. Weinshilboum RM. The genomic revolution and medicine. *Mayo Clin Proc.* 2002;77(8):745-746.
3. Personalized Medicine Coalition. The case for personalized medicine. November 2006. Available at: http://www.personalizedmedicinecoalition.org/communications/TheCaseforPersonalizedMedicine_11_03.pdf. Accessed April 10, 2007.


AAPA Home - Mozilla Firefox
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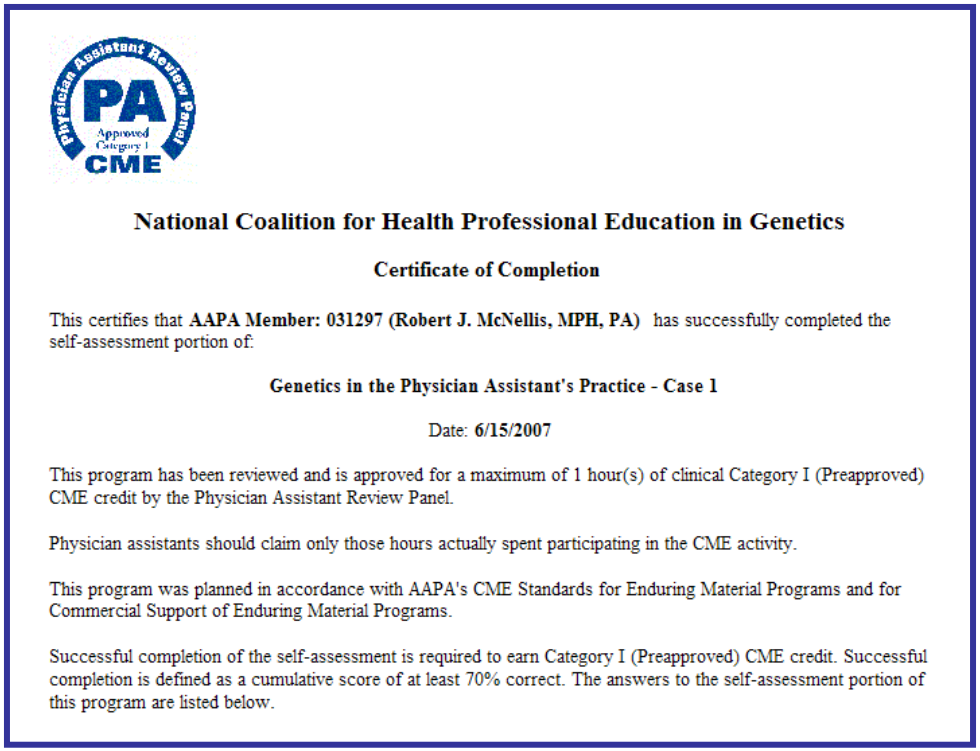
CA&E Home
Clinical Topics
CSAC
Clinical Education
Quality Care
Poster Session
Prescription Assistance
Clinical Partners
PDA PA
Drug Data Base
Pain Management Resource Center
Related Links
Diversity
Genomics:
Web Resources for the Physician Assistant
What is genomics?
Often, the term genetics and genomics are used interchangeably. "Genetics" might be a term with which you are more familiar. In terms of disease, genetics usually refers to single gene disorders such a cystic fibrosis or Down syndrome. Although these conditions are of great importance to individuals and families with them, they are relatively rare and most people are not directly affected.
The term "genomics" combines both the traditional roles of genetics and new evidence from the Human Genome Project (HGP) that genes interact with environmental factors to predispose some persons to common diseases such as colon cancer, diabetes, and arteriosclerosis. Experts believe that information from the HGP will be used to prevent disease and improve health among virtually all patients.
How will it help me take better care of my patients?
Some genomics care will be provided by medical geneticists and genetic counselors, but most will be provided by primary care clinicians and other non-genetics specialists.
Therefore, physician assistants will have an important role in providing genomics care. As the general public is exposed to more and more information on genomics, they will be turning to their healthcare providers for answers. PAs will need to be literate on medical genomics.
As the science continues to advance more genetic tests will become available. Testing will be available to identify predisposition to diseases for which there are not yet cures like Alzheimer's disease. It will be important for PAs to understand the ethical, social and legal implications of genetic testing. Someday, treatments will be customized to patient's genomic profile. Although these advances may be many years in the future, the time to prepare is now.
What do I need to know to get started?
Many organizations have developed on-line information resources to help health care providers learn more about genomics. We have collected a few of the best and put them together for you below.
AAPA Resources
Genetic Testing in Clinical Practice
Guidelines for Ethical Conduct for the Physician Assistant Profession
Information for health care professionals
National Coalition for Health Professional Education in Genetics (NCHPEG)
CME! Genetics in the Physician Assistant's Practice (up to 3 hours of CME)
U.S. Surgeon General's Family History Initiative
CDC's Office of Genetics and Disease Prevention

NCHPEG's Web-based CME

- The post-tests for the CME program reside on AAPA's site
- As of September the number of post-test certificates awarded:
  - Case 1: 77
  - Case 2: 59
  - Case 3: 51



National Coalition for Health Professional Education in Genetics

Certificate of Completion

This certifies that **AAPA Member: 031297 (Robert J. McNellis, MPH, PA)** has successfully completed the self-assessment portion of:

Genetics in the Physician Assistant's Practice - Case 1

Date: **6/15/2007**

This program has been reviewed and is approved for a maximum of 1 hour(s) of clinical Category I (Preapproved) CME credit by the Physician Assistant Review Panel.

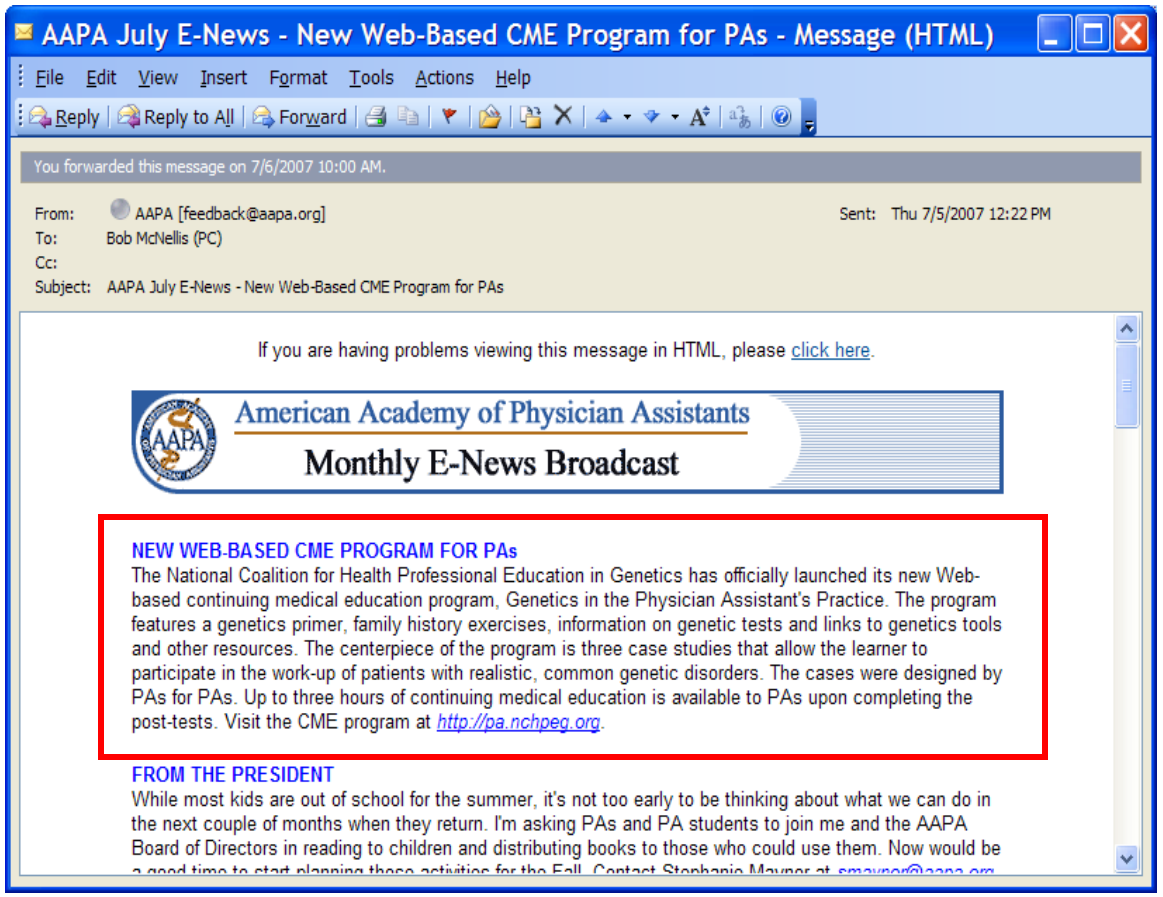
Physician assistants should claim only those hours actually spent participating in the CME activity.

This program was planned in accordance with AAPA's CME Standards for Enduring Material Programs and for Commercial Support of Enduring Material Programs.

Successful completion of the self-assessment is required to earn Category I (Preapproved) CME credit. Successful completion is defined as a cumulative score of at least 70% correct. The answers to the self-assessment portion of this program are listed below.

NCHPEG's Web-based CME

- Promoted in the e-News in July & August
- Delivered to over 20,000 addresses
- July:
  - 339 click thrus
  - (222 Annual Conference photos, 144 ACP diabetes care web page)
- August:
  - 68 click thrus
  - (346 NPI data, 93 PA legislation)



Legislation, policy, partnerships

- AAPA sent a letter to Congress urging support of the Genetic Information Nondiscrimination Act
- Our Clinical and Scientific Affairs Council will undertake a rewrite of AAPA's genetic testing policy paper
- AAPA nominated a PA for the EGAPP Stakeholders group
- Wrote a letter of support for an NCI grant application by Fox-Chase Cancer Center for an e-learning program in cancer genetics

Future Activity

2008 Annual Conference

- AAPA is working with NCHPEG to help coordinate a medical genetics track which would provide a daily session with key genetic content
- A session on race and genetics is being planned which would be co-sponsered by AAPA's African Heritage Caucus, Committee on Diversity and NCHPEG

Other activities

- Continuation of Doug Scott's "Genomics series" in AAPA News
- Addition of survey questions on the Annual Conference Survey (over 2000 respondents)
- Currently developing a needs assessment tool
- Review of AAPA policy statements
- Engage the JAAPA editorial board
- Expanded relationships with other genetics organizations, advisory committees, etc
- Support Rocky's junkets

What does it all mean?

- Are PAs reading Doug's articles?
- Are they heeding Rocky's advice?
- How many are going to genetics lectures?
- Why haven't more PAs completed the NCHPEG education program?
- What do we need to do to get their attention and completely engage them?
- How can we give them a way to respond, and get involved in the conversation?

Genetics Survey of Physician Assistants: 2007 House of Delegates

September 19, 2007

Bob McNellis

Director, Science and Education

Methods

- Survey developed in cooperation with Vence Bonham and Greg Feero
- Replicated existing categories of data where able (i.e. specialty, setting)
- Took approximately 3 minutes to complete
- Administered to AAPA's House of Delegates on the last day of a three day meeting
- Approximately 250 surveys were distributed, 113 were returned
- Survey responses entered into a Filemaker database, exported to Excel for analysis
- Thanks to Murugu Manickam, MD for his analyses (and Cara McNellis for her data entry)

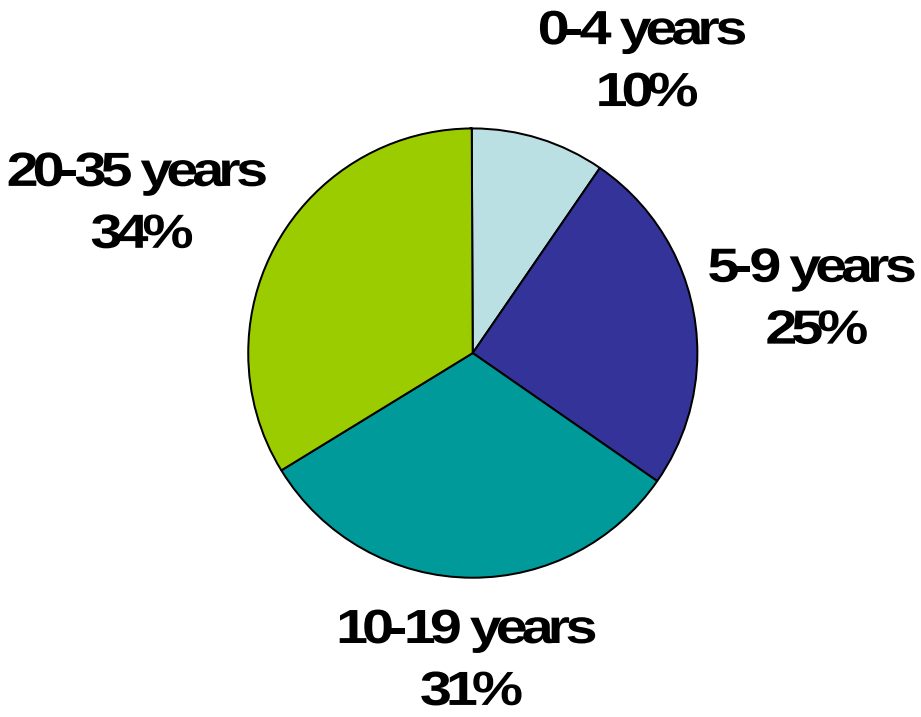
Demographics

Clinically practicing

Community characteristics

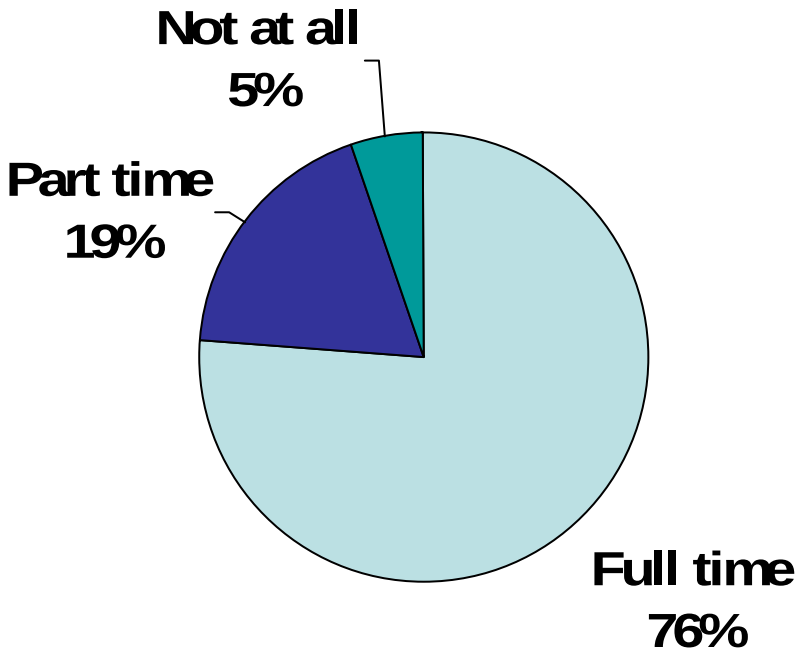
Practice setting and specialty

What year did you graduate from your PA program?



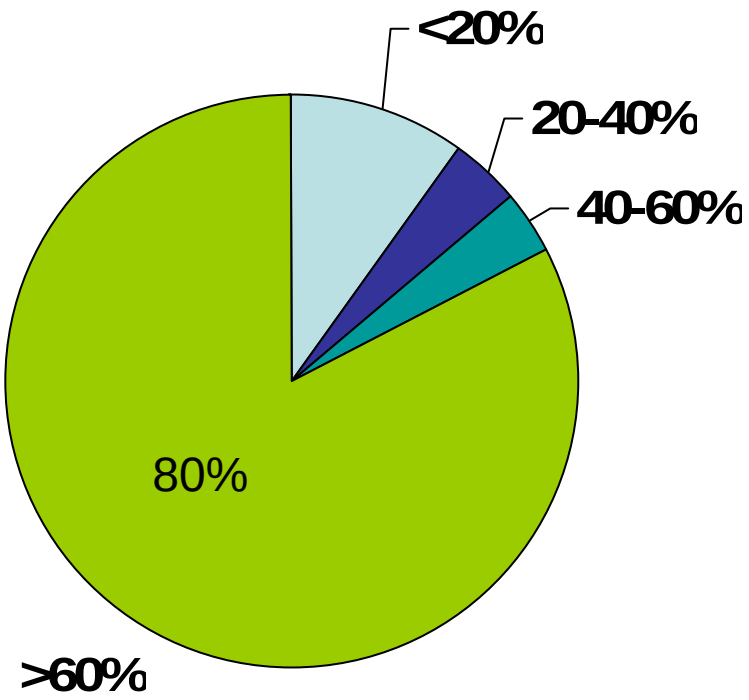
Mean = 16 years

Are you currently a clinically practicing PA?

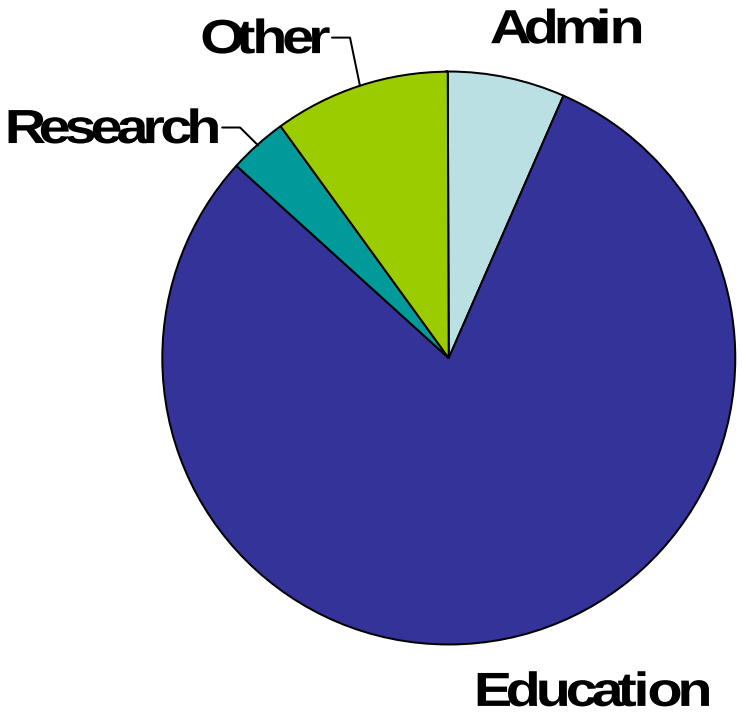


2006 AAPA Census – 90.8% in clinical practice

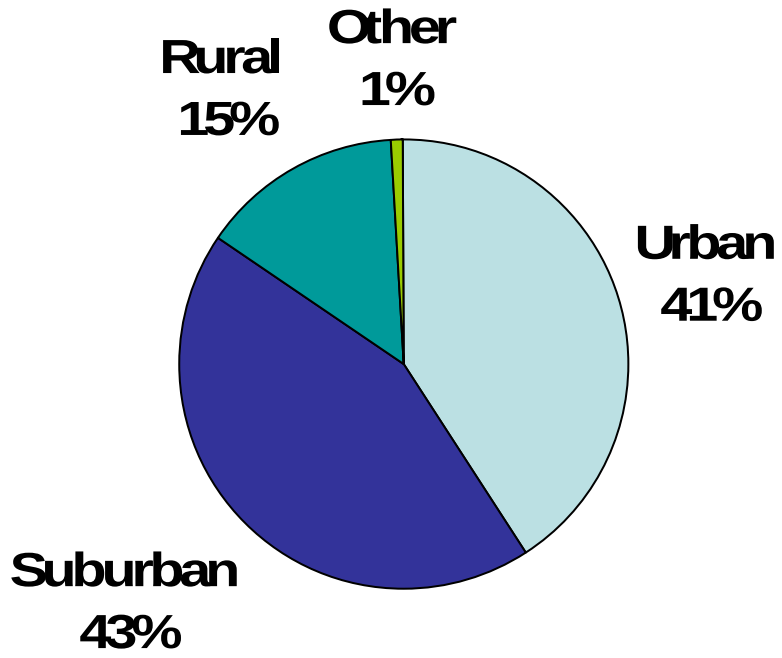
What proportion of your work-time is spent seeing patients?



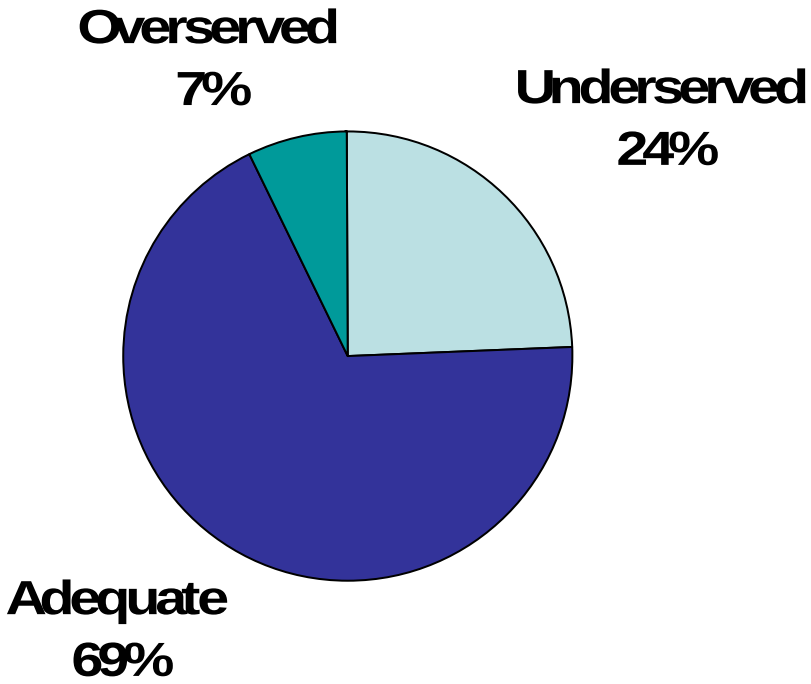
If you are not working in clinical practice...



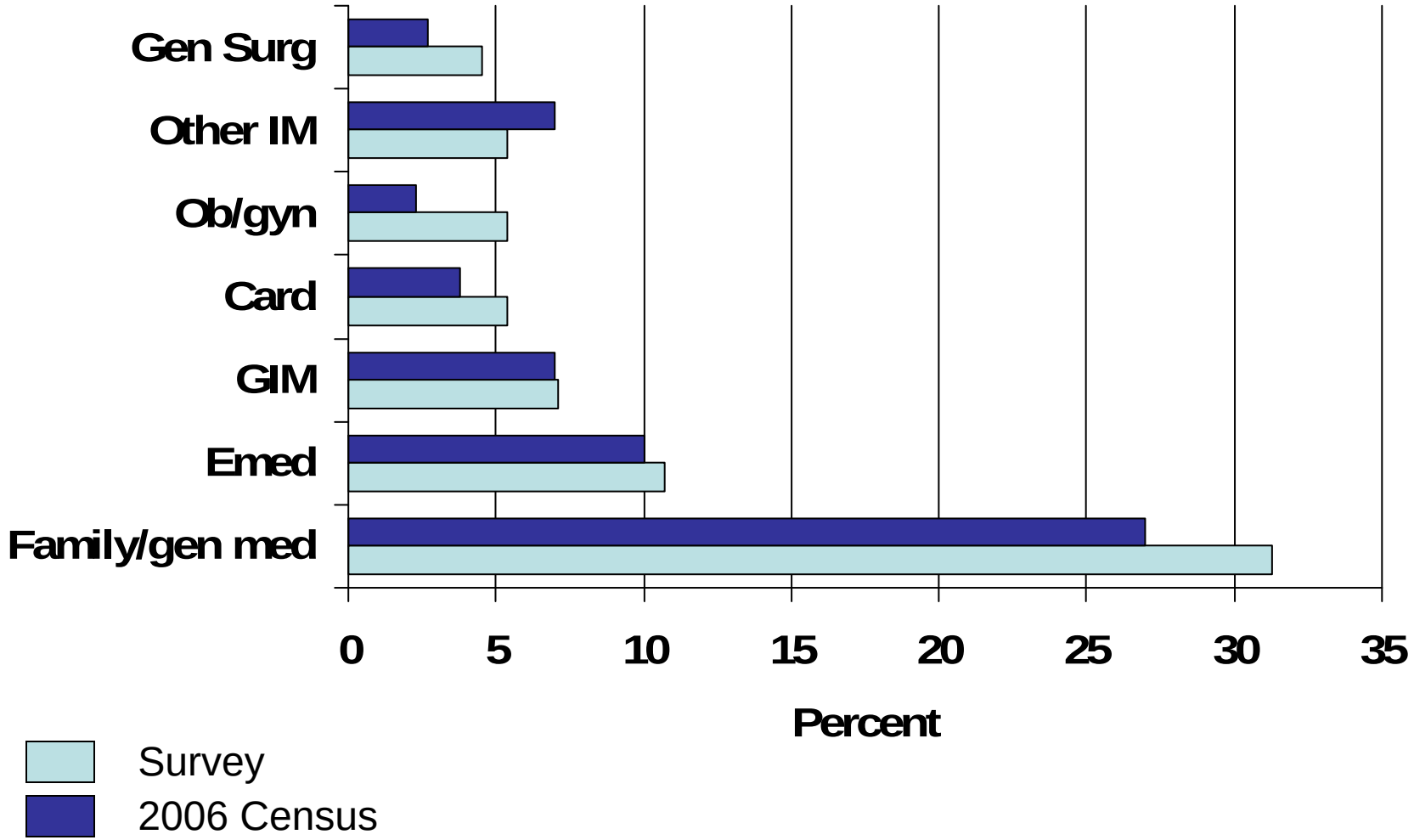
Which of the following best describes the community surrounding the area in which you work?



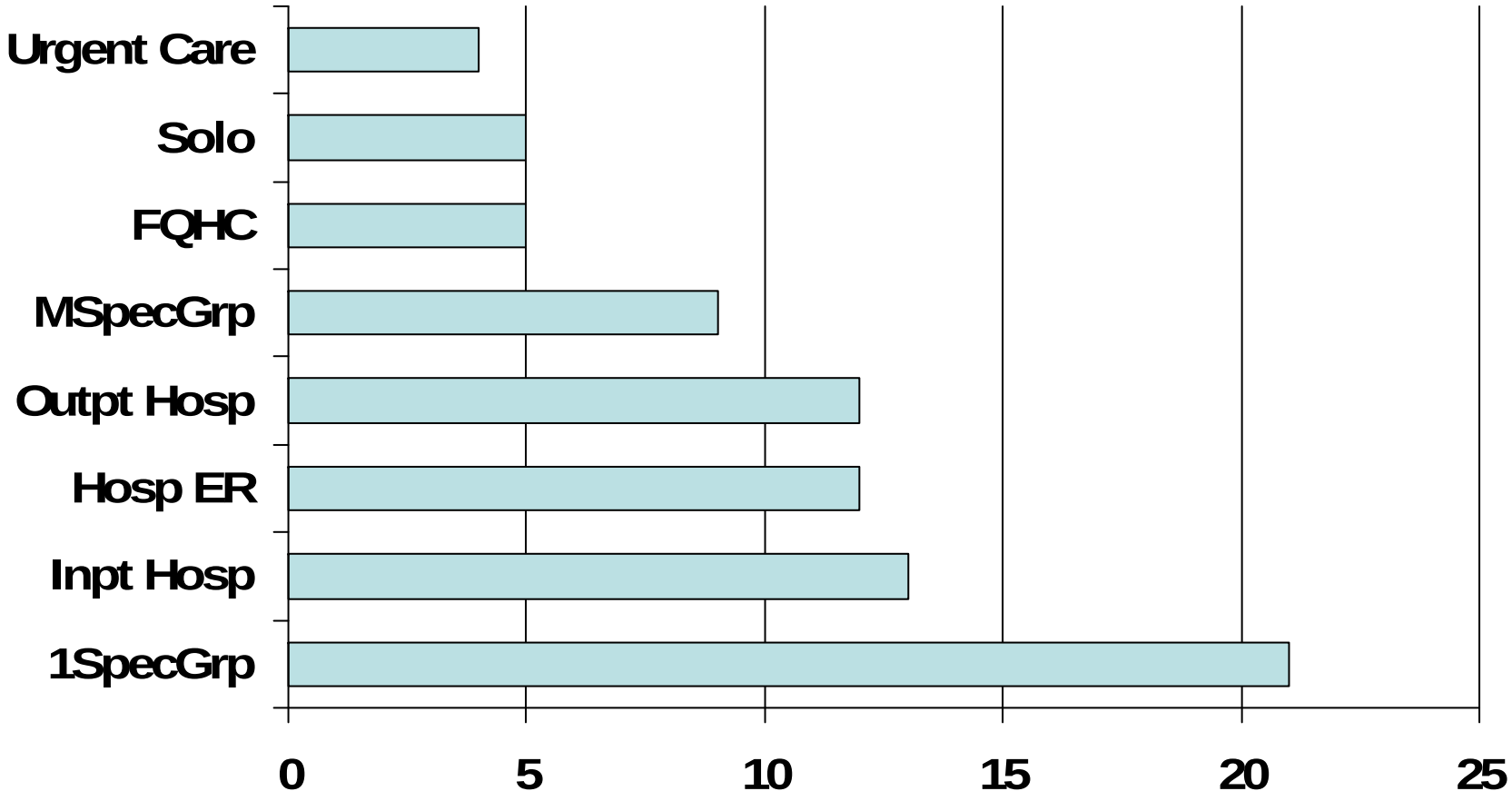
How would you describe the availability of general medical services in and around your practice location?



Current primary specialty



Primary setting



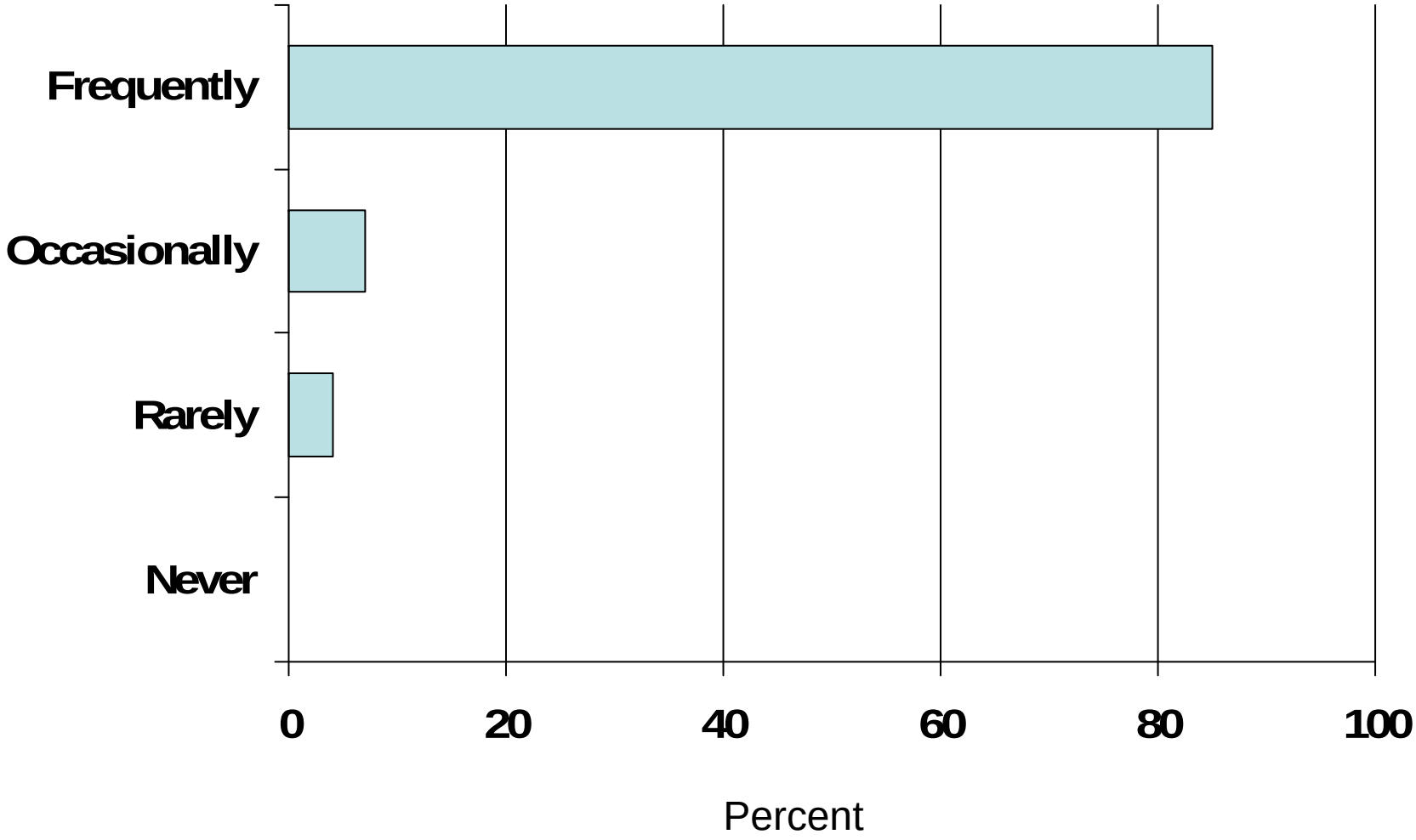
Use of genetics in practice

Family history

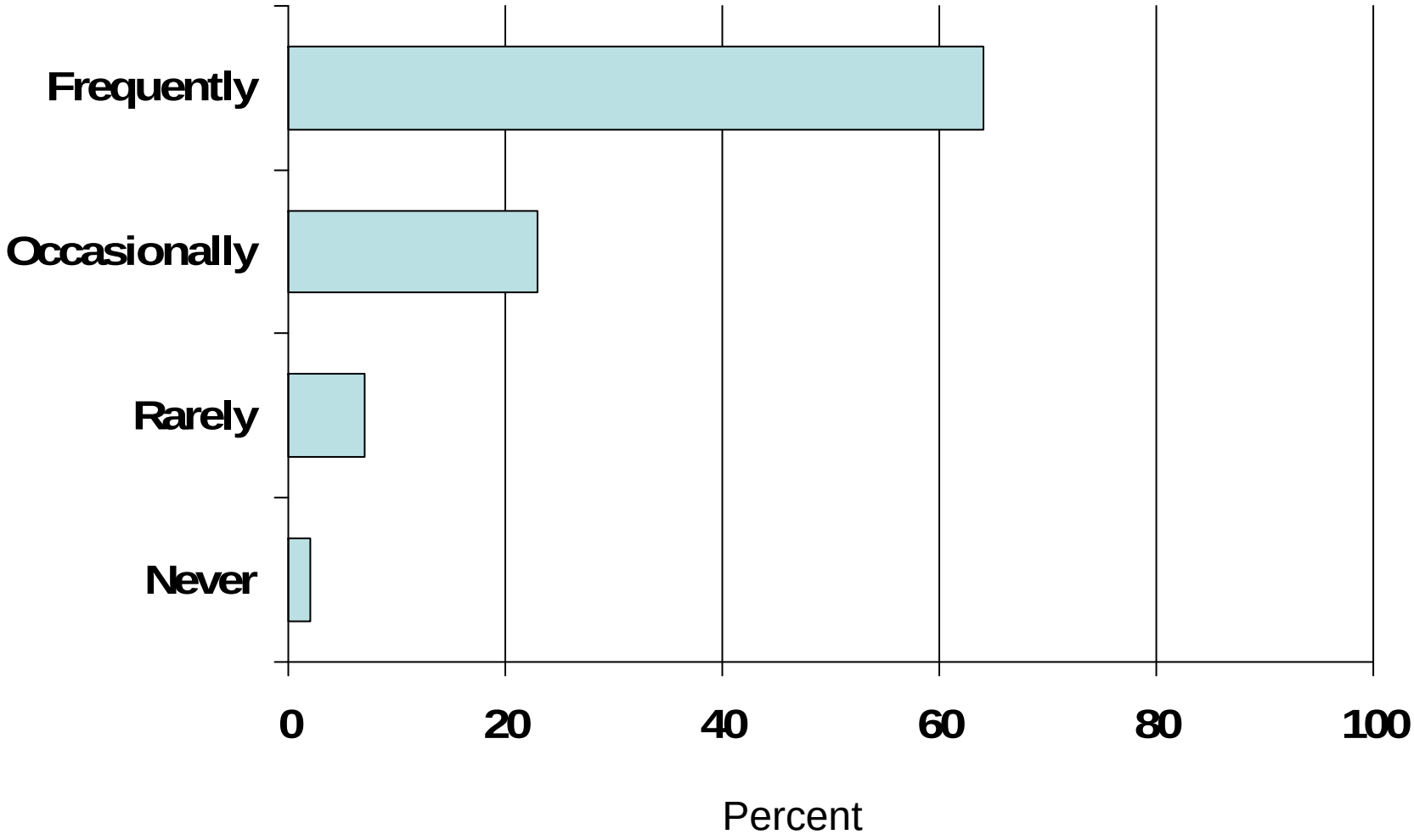
Genetic consults

Perception of physician knowledge

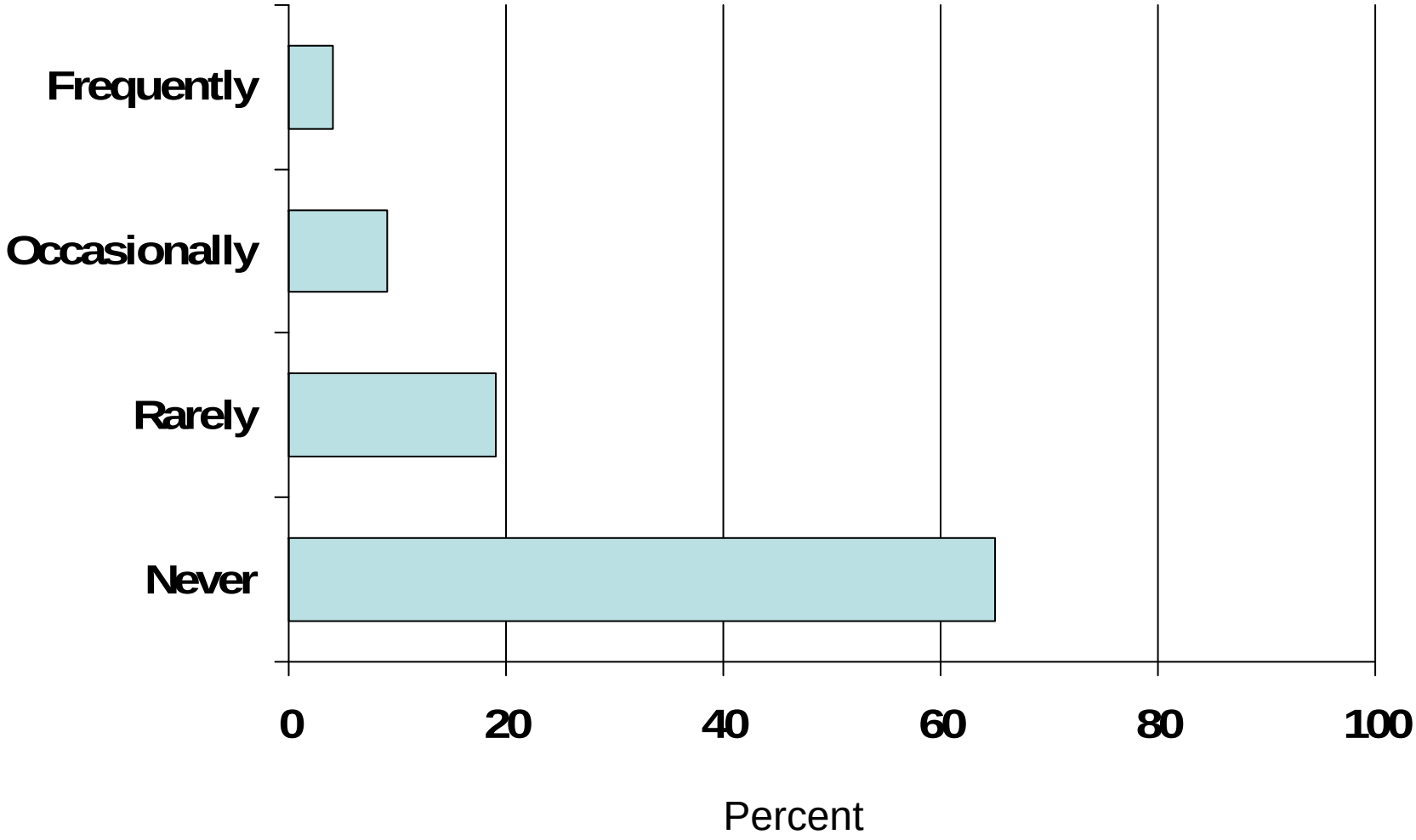
In the past 6 months approximately how frequently have you gathered family history information in the context of patient visits?



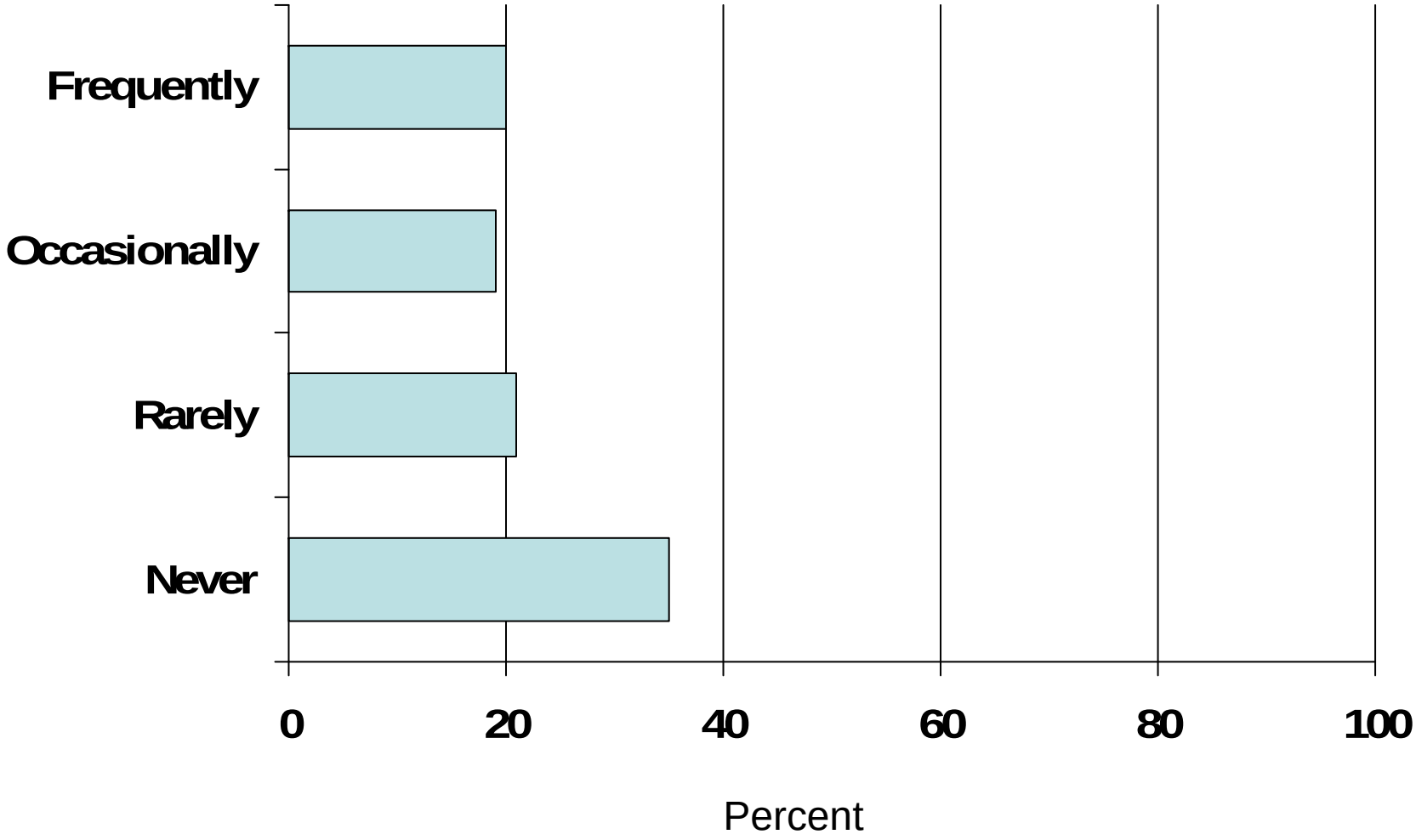
In the past 6 months, how often have you used family history information when making clinical decisions or recommendations for your patients?



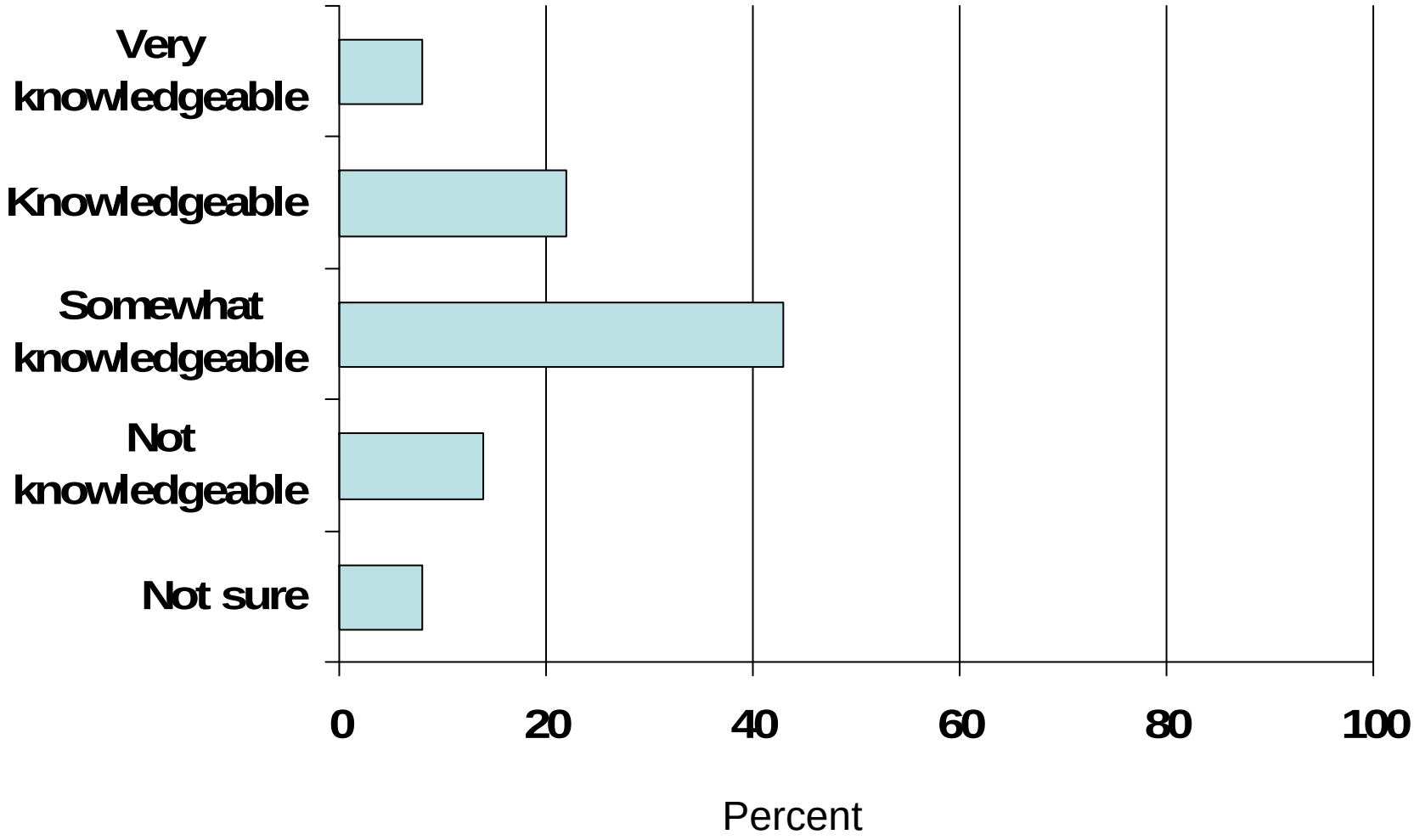
In the past 6 months, how often have you contacted a genetics specialist (a medical geneticist, genetics counselor, or advance practice nurse geneticist) for a formal or informal consultation or referral?



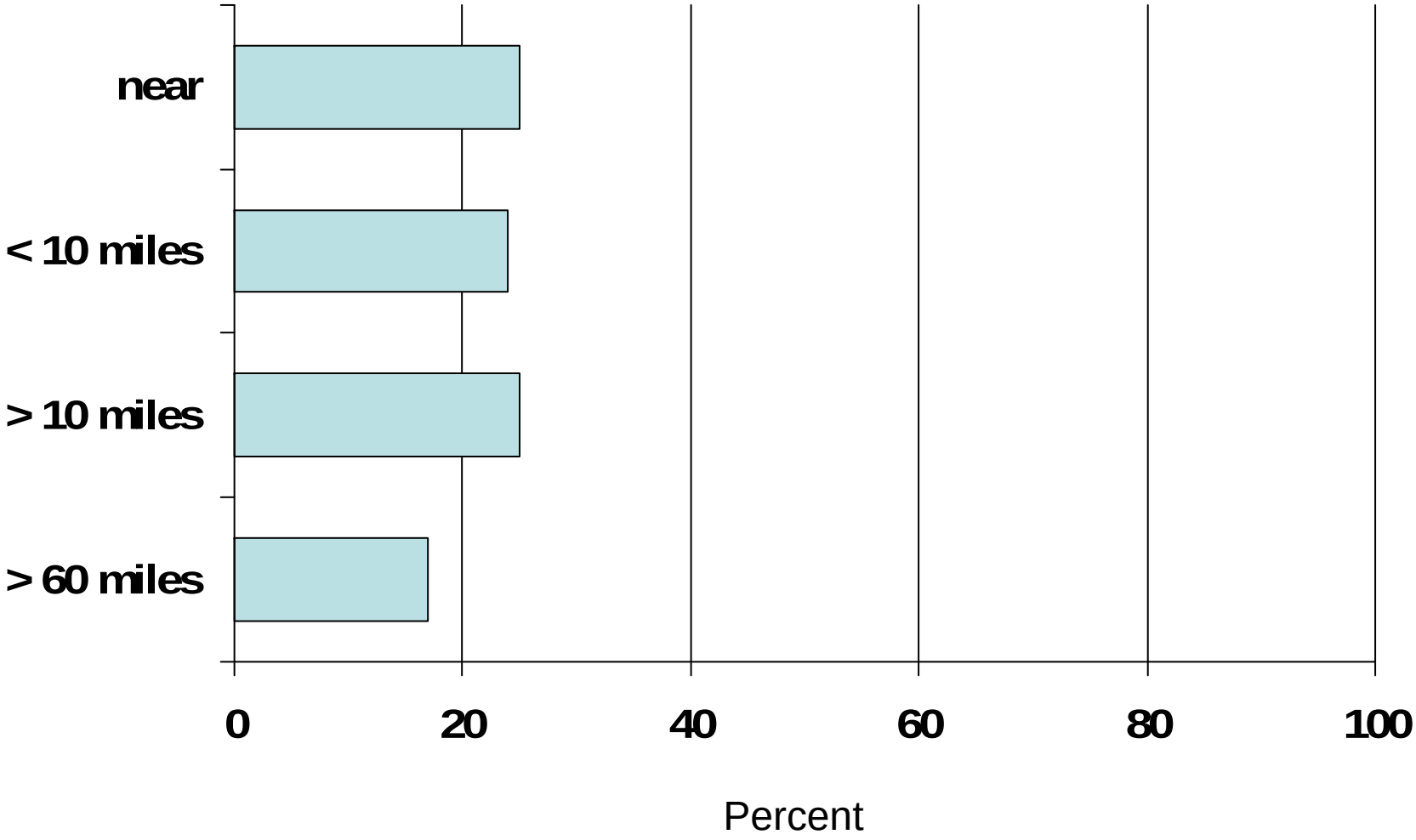
In the past 6 months, how often have you contacted a non-genetics specialist (e.g., a cardiologist, gastroenterologist, etc.) for a formal or informal consultation or referral on a genetics issue?



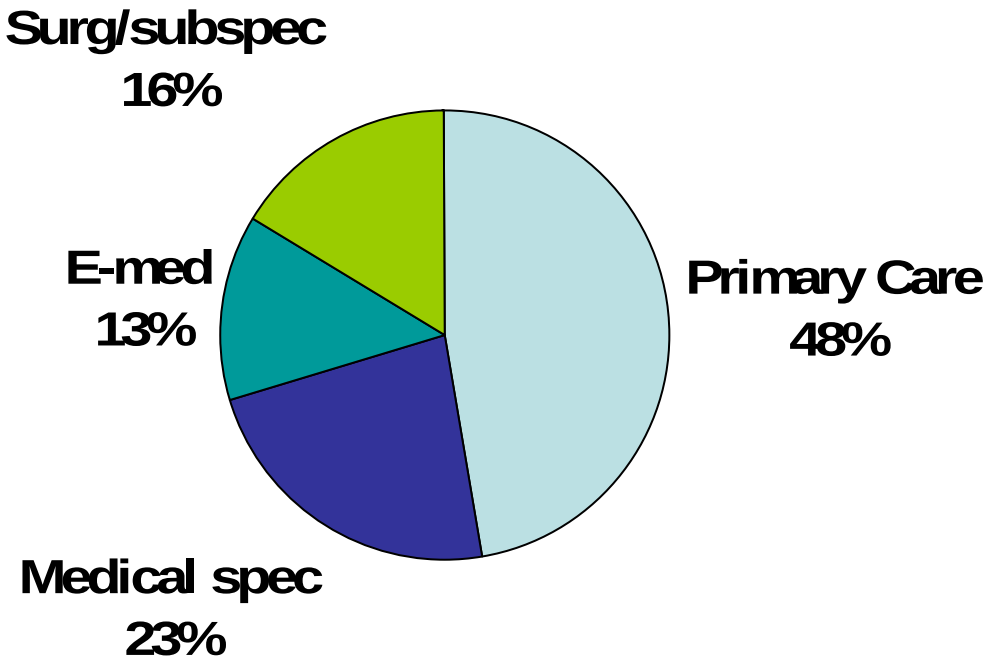
How knowledgeable do you feel that your supervising physician is regarding genetics (if you have more than one supervising physician, estimate the average knowledge)?



How accessible are genetic service providers in your clinical setting?



Sub-analyses by broad specialties



Sub-group family history

- Gathering family history - less frequently by surgical/subspecialties (76% v 90+%)
- Using family history - primary care (73%) and medical specialties (79%) higher than ER (57%) or surgery (35%)
- PAs out of school longer more frequently gathered and used family history
  - 0-4 years always the lowest (64% and 55%, respectively)

Sub-group referrals

- Referral to genetics expert – “Never” most common across the board
- Referral to non-geneticist – most “frequently” by ED PAs
- Two-thirds have never referred to a genetic specialist – those having a geneticist in the “same location” refer more often

Sub-group perceived supervising MD knowledge

- Attendings' knowledge – perceived highest knowledge in medical specialties (54%), lowest knowledge in emergency medicine (14%)

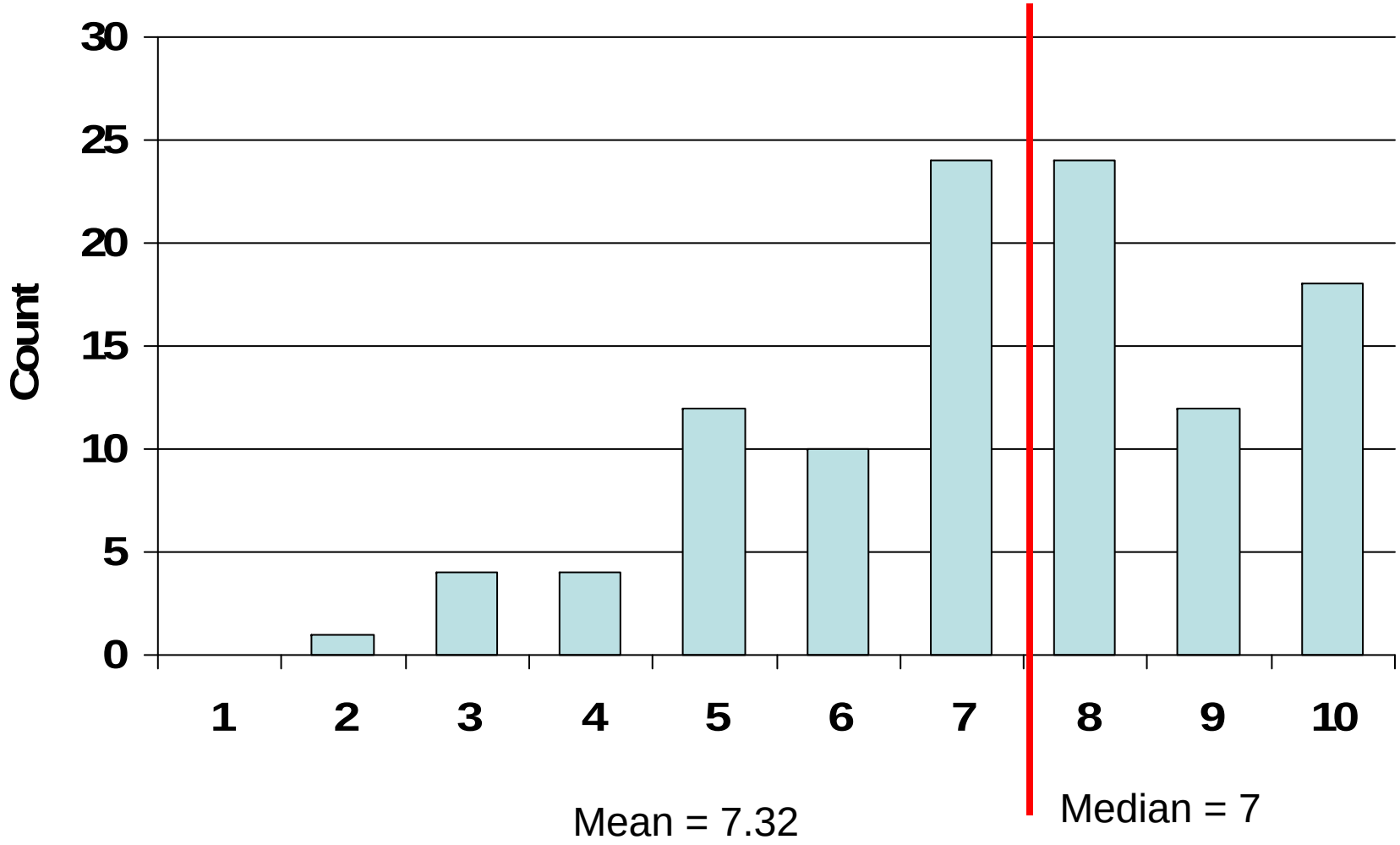
Learning about genetics

NCHPEG

CME

Family history tools

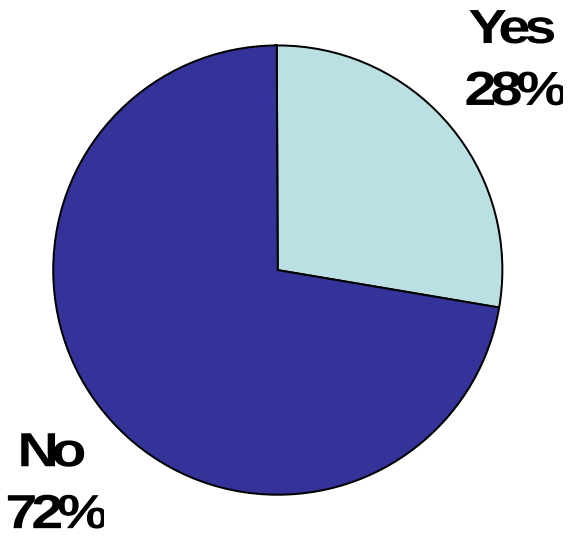
On a scale of 1-10 how important is genetics in the education of physician assistants?



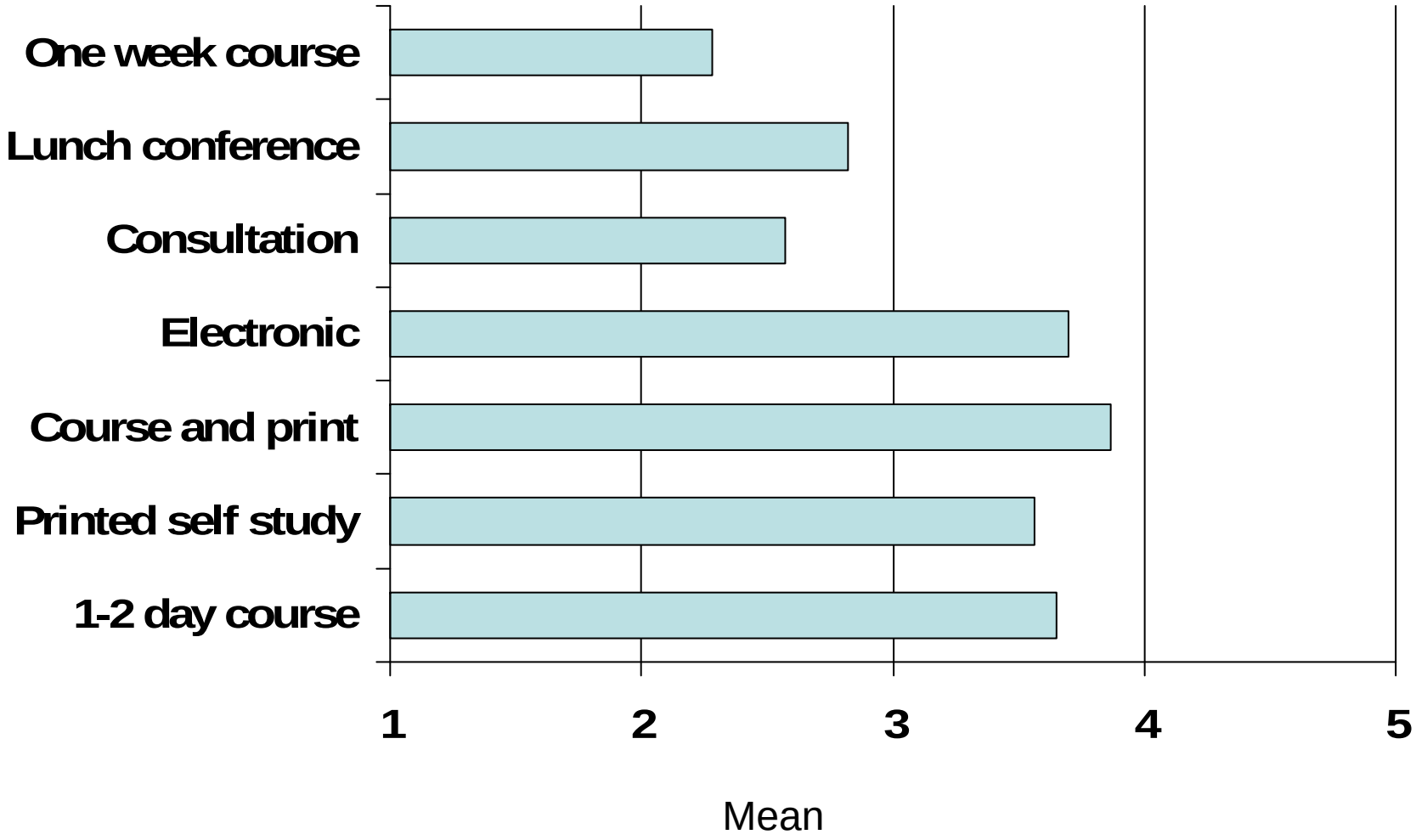
Importance related years since graduation

- 0-4 years 6.6
- 5-9 years 7.1
- 10-19 years 7.5
- 20-35 years 7.5

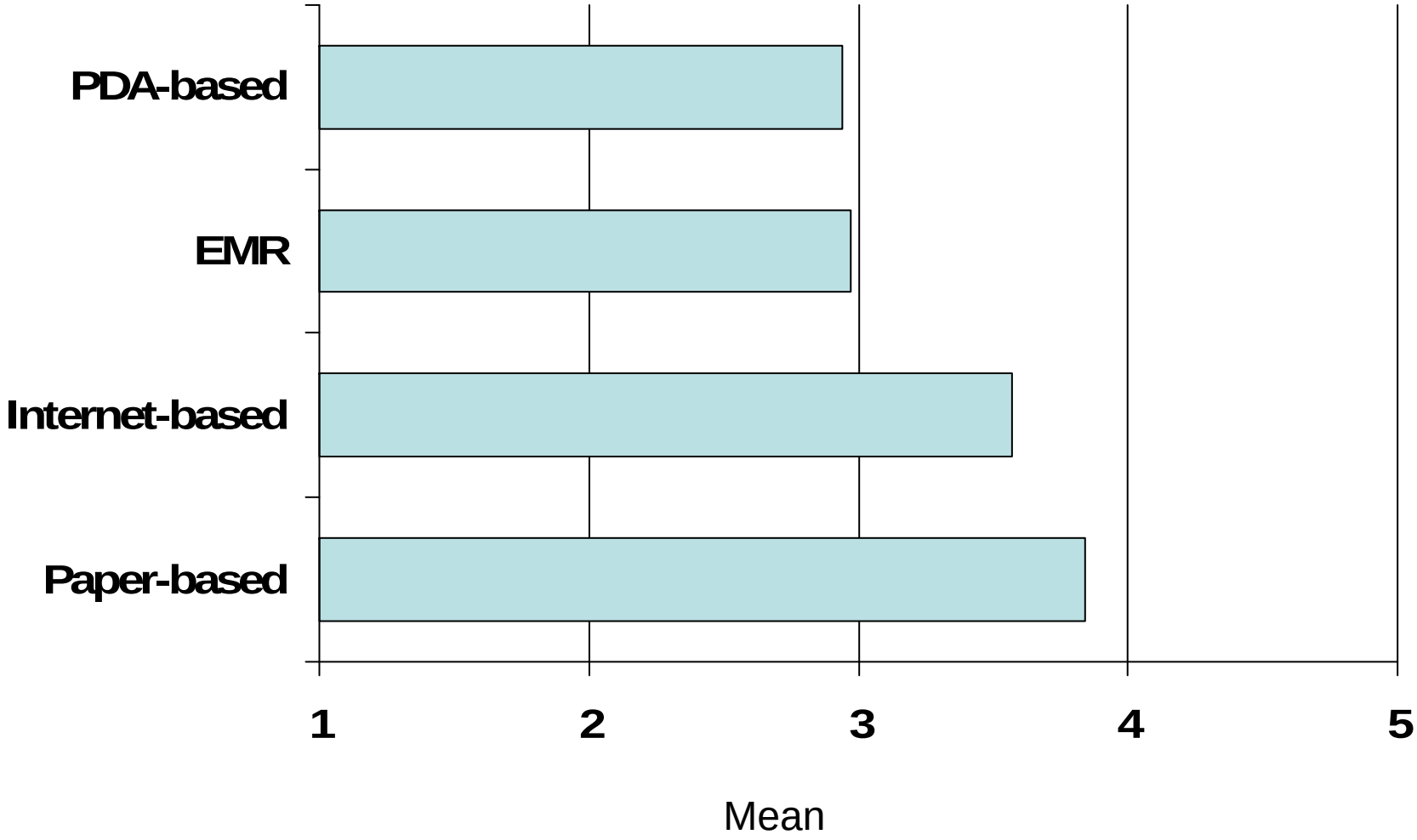
Have you heard of the National Coalition for Health Professional Education in Genetics?



On a scale of 1 to 5, with 1 = not at all helpful and 5 = most helpful, please rate the usefulness of the following formats for learning about genetics.



On a scale of 1 to 5, with 1 = not at all helpful and 5 = most helpful, please rate the usefulness of the following types of clinical tools to incorporate genetics into your clinical practice.



Summary

- This is a subset of a subset of a subset of a subset of PAs, but we learned...
- Most of these PAs gather and use family history frequently
- Few of these PAs refer patients to genetic specialists
- Specialty, geography and experience influence genetic knowledge

Summary

- Most of these PAs believe genetics is important
- Most are not aware of NCHPEG
- PAs are open to a variety of educational media as long as they are print, electronic or short in-person conferences
- Paper is still king for tools in practice

Final thoughts

- If we could pick a handful of questions on genetics for next year’s conference survey what would you pick?

- Thanks again to Murugu!

Translation of genomic discoveries to primary care – A role for the PA?

Greg Feero, M.D., Ph.D.
Jean Jenkins, R.N., Ph.D.
Michael Rackover, M.S., PA-C

Sept. 19, 2007

Translating Genomics…

- Genomic discoveries relevant to common disease diagnosis and management are coming at an increasing rate.
- Basic discoveries are leading to the development of clinical applications.
- Ergo, improved healthcare is around the corner!

Translating Genomics…

- Genomic discoveries relevant to common disease diagnosis and management are coming at an increasing rate.
- Basic discoveries are leading to the development of clinical applications.

Mind the gap!

- Ergo, improved healthcare is around the corner!

Translating Genomics…

Filling the gap

- Does the application address a clinical need?
- Does the application meet a clinical need?
- Is the application acceptable to patients?
- Is the application acceptable to health care providers?
- Is the application acceptable to insurers?
- Is the application acceptable to society?
- How are patients best educated about the application?
- How are providers best educated about the application?

Who will fill the gap?

Bench Scientists

↓

Clinical Scientists

↓

Clinicians

↓

Patients

Multiplex ClinSeq

PUHGV

NATIONAL HUMAN GENOME RESEARCH INSTITUTE Division of Intramural Research

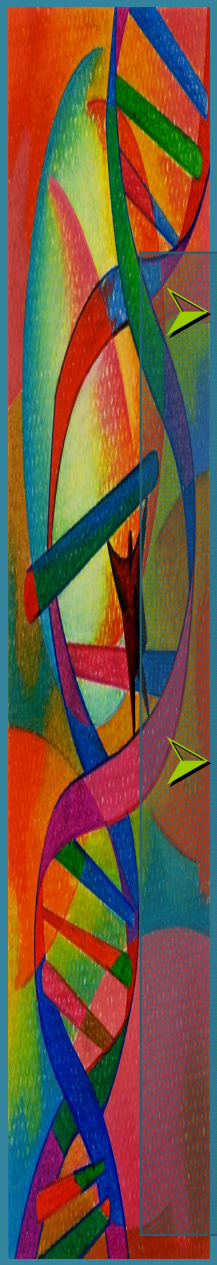
Multiplex Genetic Susceptibility Testing:

A prototype for applied research to inform personalized medicine

Colleen M. McBride, PhD. & Larry Brody, Ph.D.

Research Partners:

National Human Genome Research Institute
Henry Ford Health System
Group Health Cooperative
Cancer Research Network (NCI)



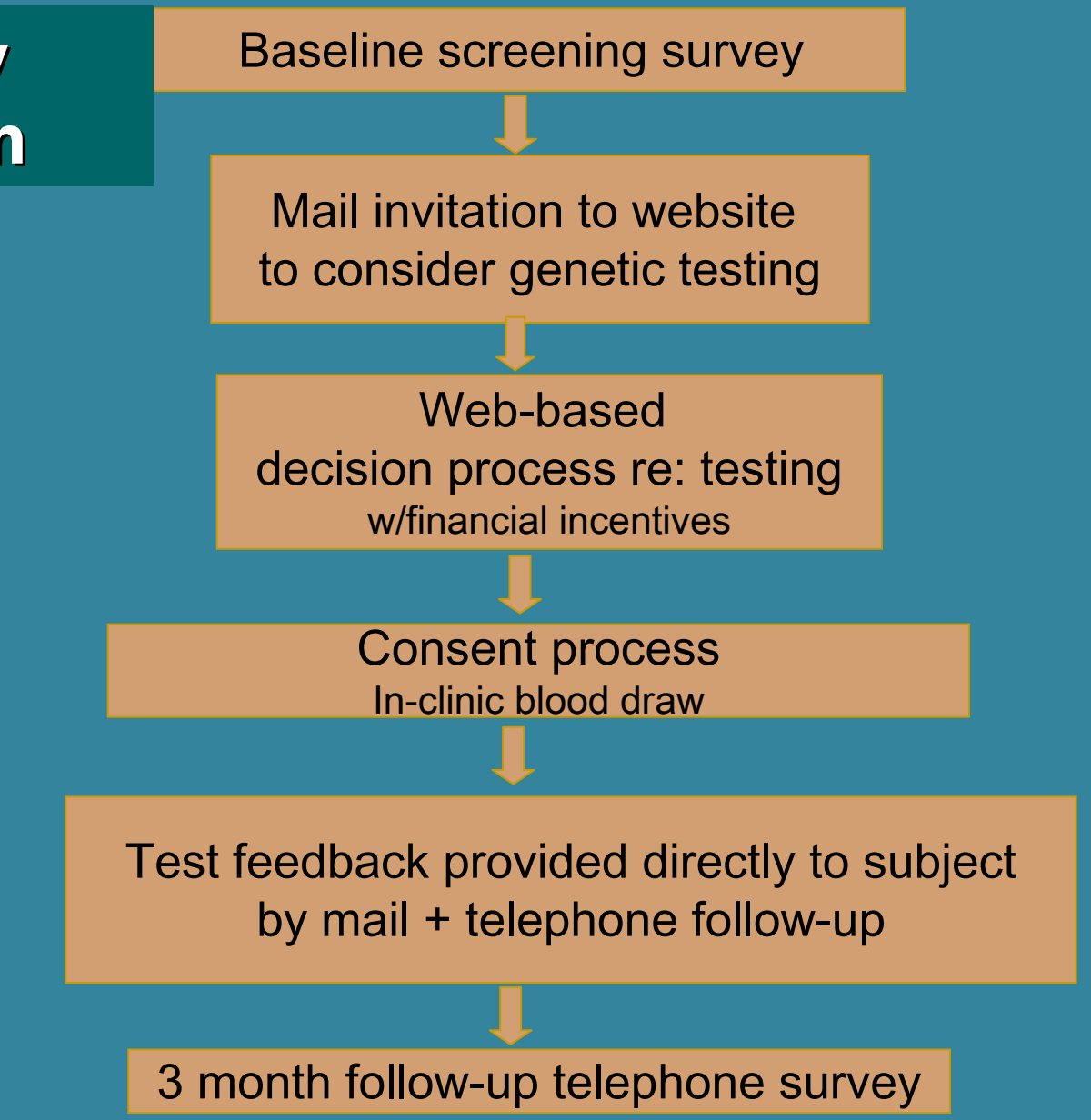
Multiplex Project Aims

- **To develop a prototype for multiplex genetic susceptibility testing**
  - Multiple markers of susceptibility for multiple diseases
  - Provide risk feedback to target populations
- **To create an infrastructure to facilitate public health research**
  - Decide upon “standard of care” for consent, feedback & support services
  - Identify optimal study population(s) & recruitment approach

Clinic-based population

- **Cancer Research Network (NCI-funded)**
  - Full complement of preventive services
  - Patient bases geographically distributed with racial-ethnic & SES diversity
  - Henry Ford Health System clinical recruitment site
  - Group Health Cooperative (HMO Research Network), Survey coordination
- **Sample size: 5000+ touched ~ 1000 tested**
- **Healthy adults**
  - -- Ages 25-40
  - -- Without diseases included on test batter

Study Design



ClinSeq: A translational research project in clinical genomics

Medical & Statistical Genetics

NHLBI

ClinSeq

NIH Clinical Center

NIH Intramural Sequencing Center

Specific Aims

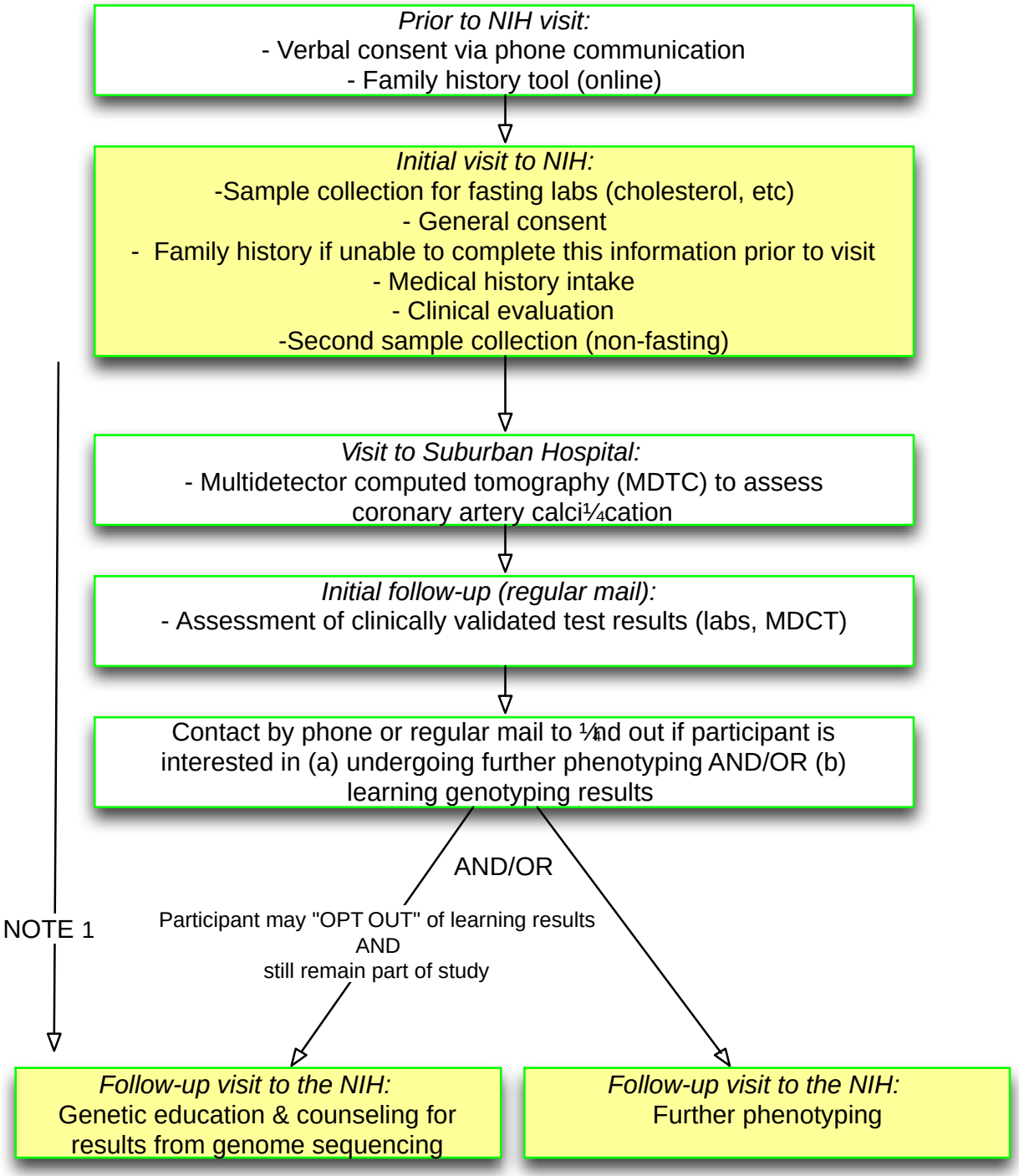
1. **Develop a robust infrastructure for the generation and use of LSMS in a clinical research setting**
2. **Use LSMS data to develop novel approaches to clinical biomedical research**
3. **To understand how to interact with subjects re LSMS**

Approach

- **Phenotype 1,000 subjects**
- **Sequence 200-400 candidate genes**
- **Follow-up studies**
- **Interpret variants and validate *some***
- **Return results**

Clinical evaluation

- Family history (semiautomated)
- Medical history (form-driven)
- Blood pressure
- Coronary calcium score (MDCT)
- Echo/electro-cardiography
- Clinical & research bloods


Prior to NIH visit:
- Verbal consent via phone communication
- Family history tool (online)
Initial visit to NIH:
-Sample collection for fasting labs (cholesterol, etc)
- General consent
- Family history if unable to complete this information prior to visit
- Medical history intake
- Clinical evaluation
-Second sample collection (non-fasting)
Visit to Suburban Hospital:
- Multidetector computed tomography (MDTC) to assess coronary artery calci¼cation
Initial follow-up (regular mail):
- Assessment of clinically validated test results (labs, MDCT)
Contact by phone or regular mail to ¼nd out if participant is interested in (a) undergoing further phenotyping AND/OR (b) learning genotyping results
AND/OR
Participant may "OPT OUT" of learning results
AND
still remain part of study
NOTE 1
Follow-up visit to the NIH:
Genetic education & counseling for results from genome sequencing
Follow-up visit to the NIH:
Further phenotyping

Health Professionals' Understanding of Human Genetic Variation Study

Vence Bonham, JD
Associate Investigator
Social and Behavioral Research Branch
Principal Investigator

Project Aim

To investigate health professionals' ***knowledge*** of human genetic variation, ***beliefs*** about biological and genetic differences based upon their patients' race and ethnicity and its ***use*** in clinical practice.

Health Professionals' Genetics Education Needs Exploration (HP GENE) Survey

National Human Genome Research Institute

National Institutes of Health

0% 100%

7. Random mutations cause all of the genetic variation in the human genome.

true	false	scientific evidence inconclusive	don't know
○	○	○	○

8. The variation in the human genome includes both disease causing gene variants and variants that have no effect on health and disease.

true	false	scientific evidence inconclusive	don't know
○	○	○	○

STUDY DESIGN

Phase I Qualitative Study

Dimensional analysis and qualitative content analysis were used to explore physicians' perceptions of and experiences with racial factors in clinical decision-making, determining the racial background of a patient, and perceptions of the race-related causes of health differences.

Phase II Scale Development

Focus groups were used to assist in question development. The process of scale development occurred in an iterative fashion. Thirty-two cognitive interviews with physicians were used to refine the instrument and scale. Two panels of experts, geneticists with expertise in human genetic variation and social scientists with expertise in survey methodology provided input.

Phase III National Physician Survey

A pilot survey of 400 physicians will be conducted fall 2007 to examine psychometrics of the scale. The scale will be revised based upon the findings. In 2008 a National Survey of 3000 Primary Care Physicians will be conducted using the final HGVB scale.

Phase IV National Physician Assistants Survey ????

Thanks to:

- Colleen McBride Ph.D., DIR, NHGRI
  Multiplex
- Les Biesecker, M.D., DIR, NHGRI
  ClinSeq
- Vence Bonham, J.D., ECIB, NHGRI
  PUHGV

Possible discussion topics:

- To what extent will these sorts of research questions interest the PA community?
- What unique perspectives could the PA community bring to this type of research?
- To what extent do PA training centers participate in research? Independent? Part of a larger academic center?
- Do PA's have a research society? NAPCRG? How to engage PA's with interest?

Possible discussion topics:

- What other factors need to be considered to facilitate the translation of genomic discoveries to primary care?