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Framework for development of physician competencies in genomic medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics

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Completion of the Human Genome Project, in conjunction with dramatic reductions in the cost of DNA sequencing and advances in translational research, is gradually ushering genomic discoveries and technologies into the practice of medicine. The rapid pace of these advances is opening up a gap between the knowledge available about the clinical relevance of genomic information and the ability of clinicians to include such information in their medical practices. This educational gap threatens to be rate limiting to the clinical adoption of genomics in medicine. Solutions will require not only a better understanding of the clinical implications of genetic discoveries but also training in genomics at all levels of professional development, including for individuals in formal training and others who long ago completed such training. The National Human Genome Research Institute has convened the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) to develop and share best practices in the use of genomics in medicine. The ISCC has developed a framework for development of genomics practice competencies that may serve as a starting point for formulation of competencies for physicians in various medical disciplines.

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The promise of genomics to maintain health, facilitate diagnosis, and cure or mitigate disease is dependent on the skillful translation of genomic science into meaningful action at the bedside and in the clinic.[1,2] Surveys of both primary-care and specialist physicians, often by their professional societies, reveal unease, and even unwillingness, to use genomic data.[3,4] The use of genomics in caring for patients with certain cancers and for some pediatric patients is increasing in routine diagnosis and treatment, and this trend is likely to expand to other areas of medical practice in the coming years.[5,6]

Nearly half of practicing clinicians in the United States are more than 50 years of age; medical school and residency training for these physicians occurred before the completion of the Human Genome Project and the breakthrough advances in genomic medicine.[7] Current trainees are faced with a rate of progress in genomics that renders much of what they have learned out of date by the time they enter practice. Considering this rapid rate of change, substantial reductions in the cost of genome sequencing, and the increasing relevance of genomic information to the practice of medicine, the barriers to implementing genomic discoveries within medical practice have to be overcome. Moreover, misuse of genomics by untrained health-care providers may incur cost without advantage and may result in harm to patients based on inaccurate diagnosis or use of unnecessary or incorrect tests.

The National Human Genome Research Institute, together with 23 professional societies, 15 other institutes at the National Institutes of Health, and other organizations interested in physician education, developed the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) in the spring of 2013 (see **Supplementary Data S1** online). ISCC member organizations focus on physician training, starting with medical school matriculation and continuing through residency and fellowship, for active clinicians.[8] The ISCC seeks to "improve genomic literacy of physicians and other practitioners and to enhance the practice of genomic medicine through sharing of educational approaches and joint identification of educational needs." The ISCC developed four working groups: Genomic Medicine Competencies, Educational Products, Use Cases, and Specialty Boards (see **Supplementary Data S1** online).

The Genomic Medicine Competencies Working Group was charged with the development of a framework whereby

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competencies in genomic medicine could be identified for physicians working in various areas of practice. The specific applications of genomic medicine will differ for different medical disciplines, making it impossible to identify a single set of competencies that apply to all areas of practice. The Working Group instead chose to formulate a set of competencies that could be used as a starting point for professional societies and other relevant groups to develop a customized set of competencies for their areas of practice.

Medical educators have defined "entrustable professional activities" (EPAs) as "… those professional activities that together constitute the mass of critical elements that operationally define a profession."[9] The Competencies Working Group identified five EPAs that comprise a basic set of genomic skills:

- Family History: elicit, document, and act on relevant family history pertinent to the patient's clinical status;
- Genomic Testing: use genomic testing to guide patient management;
- Treatment Based on Genomic Results: use genomic information to make treatment decisions;
- Somatic Genomics: use genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes; and
- Microbial Genomic Information: use genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapeutics in infectious diseases.

The integrated core competencies for each EPA are based on the six core competencies used by the Accreditation Counsel for Graduate Medical Education as well as a recent proposed expansion to eight core competencies.[10] These core competencies guide residency training and are a commonly used structure to guide postgraduate medical education.

It is hoped that these EPAs and the embedded competencies will provide a starting point for development of more specific guidelines for individual medical disciplines. In some cases, particular EPAs may be unnecessary or may require modification, and in others, additional EPAs may need to be added. Specific competencies may also need to be modified according to the needs of specific groups. The ISCC Genomic Medicine Competencies Working Group may also make future revisions. This document, and future revisions will be posted on the Genetics/Genomics Competency Center website (http://www.g-2-c-2.org/start_search_map.php), and we hope that modifications made by other groups will be shared there as well. Future efforts will need to include development of educational materials to help physicians achieve these competencies, as well as expansion of the competencies to other types of health professionals. The process of developing a physician workforce that is prepared to implement genomic approaches in day-to-day medical practice will be a journey; this framework for identification of physician competencies in genomic medicine may facilitate various medical specialties in that effort.

FAMILY HISTORY

EPA: Elicit, document, and act on relevant family history pertinent to the patient's clinical status.

Patient care

- Conduct patient interview to assemble family history;
- Use standard pedigree symbols in assembling family history;
- Recognize patterns of Mendelian inheritance and calculate simple Mendelian risks; provide this information to patients and family members as appropriate;
- Use empirical risk figures to provide appropriate information for complex (multifactorial) medical conditions;
- Recognize that traits may cluster in families due to multifactorial rather than Mendelian patterns of inheritance; and
- Formulate an action plan to address relevant family history information.

Knowledge for practice

- Describe the basic patterns of Mendelian inheritance; and
- Explain the difference between Mendelian and multifactorial inheritance.

Practice-based learning and improvement

- Incorporate family history information into health record.

Interpersonal and communication skills

- Explain and document findings from family history to patient, including implications for other family members.

Professionalism

- Respect privacy of patient and family members in assembling and documenting family history;
- Explain to patient relevant social and legal risks related to family history as well as relevant legal protections; and
- Recognize the potential of family history information to reveal unexpected family relationships such as consanguinity or misattributed paternity.

Systems-based practice

- Focus family history on problems relevant to the individual patient's health; and
- Facilitate patient's desire to communicate relevant family history information among health providers and family members.

Interprofessional collaboration

- Make appropriate referrals for specialty evaluation based on results of family history.

Personal and professional development

- Identify sources of information on genetic disorders, such as OMIM (Online Mendelian Inheritance in Man), and GeneReviews; and
- Maintain continuing medical education on matters of medical genetics.

GENOMIC TESTING

EPA: Use genomic testing to guide patient management.

Patient care

- Discuss the indications for genomic testing—specifically the benefits, risks, and alternatives;
- Explain the implications of placing genomic test results in the patient's medical record;
- Discuss the possibility of incidental findings and how they will be handled;
- Discuss risks of having genomic testing done, e.g., psychological implications for the individual as well as the family, the potential for discrimination, and the potential effect on insurance coverage;
- Explain to the patient issues of costs and financial coverage of genomic testing;
- Order, interpret, and communicate the results of appropriate genomic tests, within the physician's scope of practice;
- Provide referral to an appropriate specialist for genomic testing of a condition outside the physician's scope of practice; and
- Respond to the results of an abnormal genetic screening test, such as newborn screening, including immediate management and appropriate referral.

Knowledge for practice

- Describe the major forms of genomic variability;
- Explain how different genomic changes may result in different phenotypes;
- Recognize that genomic tests require interpretation with respect to the patient's clinical status (e.g., pathogenic, likely pathogenic, and benign);
- Explain the concepts of analytic validity, clinical validity, and clinical utility as they relate to genomic testing; and
- Recognize that medically "nonactionable" genomic results can be useful to the patient and family (i.e., personal utility).

Practice-based learning and improvement

- Incorporate genomic findings into the health record and patient-care plan; and
- Have a method for periodic review of "new" genomic interpretation for clinical applications.

Interpersonal and communication skills

- Ensure that undergoing genomic testing is a joint decision of the patient and the physician;
- Explain and document findings from genomic testing to patient, including implications for other family members;
- Facilitate access to resources to enhance patient learning about the results of genomic testing; and
- Address the needs of the patient as an individual as well as the needs of family members.

Professionalism

- Be aware of and comply with local and federal laws and regulations regarding use of genomic tests;
- Be aware of and responsive to patient's concerns about genetic discrimination; and
- Respect patient's privacy and the need to maintain confidentiality of genomic information.

Systems-based practice

- Explain who could have access to a patient's genomic information;
- Recognize the effects of the costs and coverage of genomic testing on utilization by patients; and
- Facilitate access of patients to relevant clinical studies or trials based on genomic testing.

Interprofessional collaboration

- Initiate responsible referrals to specialists or other health professionals;
- Provide support to patients based on recommendations of specialists; and
- Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status.

Personal and professional development

- Engage in continuing education regarding advances in genomic medicine and changing indications for and interpretation of genomic testing.

PATIENT TREATMENT BASED ON GENOMIC RESULTS

EPA: Use genomic information to make treatment decisions.

Patient care

- Identify medical conditions and drug responses that have a strong genetic component;
- Recognize that variants affecting drug responses found in a patient may also have implications for other family members; and
- Discern the potential clinical impact of genetic variation on risk stratification and individualized treatment.

Knowledge for practice

- Appreciate the importance of genetic diversity in humans and the abundance of genetic variants in each individual genome;
- Identify single-gene disorders that may be amenable to targeted pharmacological therapy;
- Recognize that genomic test results may guide choice of therapy for multifactorial disorders;
- Recognize that there is variability in the phenotypic expression of genetic variants and in response to therapy; and
- Recognize that the effects of some medications are strongly influenced by inherited or somatically acquired genetic variation.

Practice-based learning and improvement

- Use evidence-based recommendations of professional organizations and others in implementing knowledge gained from genetic discoveries to improve therapeutics;
- Document and periodically reassess therapeutic decision making in the medical record of patients; and
- Incorporate a realistic assessment of personal genomic knowledge and skill into the selection and use of consultants and improve competencies in the wake of these interactions.

Interpersonal and communication skills

- Discuss benefits, risks, and alternatives of various preventive and therapeutic approaches driven by genomic findings;
- Communicate clearly with other medical professionals involved in the care of the patient about the therapeutic implications of the genetic information garnered about the patient; and
- Discuss pharmacogenomics implications for future health.

Professionalism

- Respect and guard the privacy of the patient and the family members.

Systems-based practice

- "Treat the patient who has the disease," i.e., be aware of the patient's needs as an individual who also has a genetic disease or pharmacogenomic variation.

Interprofessional collaboration

- Recognize potential involvement of multiple organ systems in genetic disorders and therefore appreciate the need to seek appropriate consultation with experts in the field; and
- Make medical and genetic information available to other health-care professionals, upon obtaining proper consent, while keeping the patient's interests as the primary priority.

Personal and professional development

- Maintain the medical knowledge and clinical competence in genomics required for the provision of therapy; and
- Be familiar with the available databases and resources relevant to genetic variation, including ongoing clinical trials involving patients with genetic disorders, pharmacogenomics, and patient-oriented Internet resources from reliable organizations.

SOMATIC GENOMICS

EPA: Use genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes.

Patient care

- Identify or facilitate identification of patients who may benefit from genomic testing of tissue;
- Explain the benefits and limitations of somatic genomic testing to the patient, including implications regarding treatment of the condition and clarification of his/her prognosis;
- Ensure that tissue biopsy procedures are coordinated to make certain that appropriate and sufficient material is obtained for testing; and
- Integrate genomic testing results into the patient-care plan.

Knowledge for practice

- Explain the concept of somatic genetic change;
- Describe the role of genomic changes in the pathophysiology and treatment of cancer; and
- Explain how genomic testing can be used to guide choice of therapy and adjust drug dosage in patients with cancer.

Practice-based learning and improvement

- Maintain an awareness of and follow evidence-based guidelines and other professional resources regarding somatic genetic disorders appropriate to the physician's scope of practice.

Interpersonal and communication skills

- Communicate to the patient the importance of genomic testing of his/her tissue sample, including potential implications for treatment and prognosis, and the limitations of genomic testing;
- Address any concerns the patient may have about test results;
- Ensure that specialists involved in a patient's care are communicating with one another and with the patient; and
- Communicate to patients potential implications for his/her family.

Professionalism

- Ensure that the patient is aware of what will happen with any tissue samples obtained.

Systems-based practice

- Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status; and
- Be prepared to refer patients to clinical trials designed to evaluate new approaches to cancer therapy.

Interprofessional collaboration

- Make appropriate referrals to specialists and other health providers and support the patient in ongoing care.

Personal and professional development

- Keep up to date with progress in the diagnosis and treatment of cancer and other tissue-based disorders.

MICROBIAL GENOMIC INFORMATION

EPA: Use genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapeutics in infectious diseases.

Patient care

- Use genomics-based tests for infectious disease instead of classic strategies where appropriate (e.g., based on clinical validity and turnaround time);
- Appreciate the sensitivity and specificity of genomics-based tests for diagnosis of infectious disease based on the clinical presentation, suspected pathogen type, and testing method; and
- Interpret genomics-based tests for diagnosis, monitoring, and treatment of infectious disease.

Knowledge for practice

- Explain the core strategies for genomic testing for microbial disease;
- Describe how DNA or RNA sequence variations in the microbiome may predict response to therapy and clinical outcomes;
- Explain the potential reasons for false-positive and false-negative microbial genomics-based tests; and
- Explain the importance of "normal" microbiome to health and disease.

Practice-based learning and improvement

- Monitor ongoing testing results and their implications for treatment and prognosis in chronic infection;
- Be aware of new genomic testing methods and their clinical applications and apply when appropriate; and
- Maintain awareness of patterns of infection in your patient population and use genomic tests appropriate to these patterns.

Interpersonal and communication skills

- Explain the results of microbial genomic testing to patients; and
- Explain to patients and families results that signal a risk for contagion and take appropriate containment steps.

Professionalism

- Provide guidance to patients on how to avoid transmission of microbial agents in the community; and
- Appreciate the importance of genomic tests for public health and responsibilities of primary-care physicians in reporting results to the appropriate public health authorities.

Systems-based practice

- Work with other health-care professionals to apply infection-control measures when appropriate in both inpatient and outpatient settings; and
- Reassure patients and health-care workers in those situations in which "infection control" is not indicated.

Interprofessional collaboration

- Identify appropriate specialists and public health officials who need to be included in the care of the patient with

infectious disease and function as a member of the care team;
- Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status; and
- Consult with infectious disease specialists as needed (e.g., to manage unusual or unexpected infection or infection that is highly resistant to treatment).

Personal and professional development

- Maintain up-to-date knowledge on genomic approaches to care for patients with microbial infection.

SUPPLEMENTARY MATERIAL

Supplementary material is linked to the online version of the paper at http://www.nature.com/gim

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DISCLOSURE

The authors declare no conflict of interest.

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