PHYSICIAN RESOURCES MAPPED TO GENOMICS COMPETENCIES AND GAPS IDENTIFIED WITH CURRENT EDUCATIONAL RESOURCES AVAILABLE 06/04/14

Submitted resources from physician organization representatives were mapped to published competencies* by members of the Education Product Working Group. This summary provides a snapshot of the frequency of competencies currently being addressed by available G2C2 resources and can provide guidance when considering gaps in genomics education materials.

Method: For each major item below (i.e., 1-5) the total number of times resources were mapped to that item are listed (i.e., Family history was mapped to resources a total of 126 times). For each main subcategory, resources have been mapped to the main item (i.e., Family History, patient care was mapped to resources inclusive of all subcategories under patient care a total of 11 times). Then, if an individual item under the subcategory was mapped to individual resources, that number was also noted (i.e., 1A1 was mapped to individual resources 10 times).

Highlights:

1. The **most** frequently addressed EPA by currently available physician education resources is Genomic Testing (n= 217 all total).
2. The least frequently addressed EPA by currently available physician education resources is Microbial Genomic Information (n=8 all total)
3. There are key gap areas noted for all of the competencies even when other sub-items are addressed.
    a. For example Family History, Professionalism has few resources teaching to Patient Care 1A3, and 2 of the 3 subcategories 1E1, 1E3.
    b. Gaps are present in Genomic Testing Patient Care items 2A2, 2A3; Knowledge for Practice 2B4, 2B5; Professionalism 2E3; and Systems-Based Practice 2F3.
    c. Gaps are present in Patient Treatment, Practice-Based Learning and Improvement 3C2; and Interprofessional Collaboration 3G1.
    d. Gaps are present in Somatic Genomics, Systems-Based Practice 4F2.
    e. Gaps are present in Microbial Genomic Information-all items.

1. Family History (n=126 total)

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

1A. Patient Care (n=11 all inclusive)

- (10) 1A1. Conduct patient interview to assemble family history
- (3) 1A2. Use standard pedigree symbols in assembling family history

- (0) 1A3. Recognize patterns of Mendelian inheritance and calculate simple Mendelian risks; provide this information to patients and family members as appropriate
- (5) 1A4. Use empirical risk figures to provide appropriate information for complex (multifactorial) medical conditions
- (3) 1A5. Recognize that traits may cluster in families due to multifactorial rather than Mendelian patterns of inheritance1A6. Formulate an action plan to address relevant family history information
- (5) 1A6 Formulate an action plan to address relevant family history information

1B. Knowledge for Practice (n=10 all inclusive)

- (4) 1B1. Describe the basic patterns of Mendelian inheritance
- (2) 1B2. Explain the difference between Mendelian and multifactorial inheritance

1C. Practice-Based Learning and Improvement

- (5) 1C1. Incorporate family history information into health record

1D. Interpersonal and Communication Skills

- (13) 1D1. Explain and document findings from family history to patient, including implications for other family members

1E. Professionalism (n=7 all inclusive)

- (0) 1E1. Respect privacy of patient and family members in assembling and documenting family history
- (5) 1E2. Explain to patient relevant social and legal risks related to family history as well as relevant legal protections
- (0) 1E3. Recognize the potential of family history information to reveal unexpected family relationships such as consanguinity or misattributed paternity

1F. Systems-Based Practice (n=4 all inclusive)

- (5) 1F1. Focus family history on problems relevant to the individual patient's health
- (3) 1F2. Facilitate patient's desire to communicate relevant family history information among health providers and family members

1G. Interprofessional Collaboration

- (13) 1G1. Make appropriate referrals for specialty evaluation based on results of family history

1H. Personal and Professional Development (n=2 all inclusive)

- (6) 1H1. Identify sources of information on genetic disorders, such as OMIM (online Mendelian Inheritance in Man), and GeneReviews
- (10) 1H2. Maintain continuing medical education on matters of medical genetics

2. Genomic Testing (n= 217 all total)

EPA: Use genomic testing appropriately to guide patient management

2A. Patient Care (n=22 all inclusive)

- (18) 2A1. Discuss the indications for genomic testing – specifically the benefits, risks, and alternatives
- (1) 2A2. Explain the implications of placing genomic test results in the patient's medical record
- (1) 2A3. Discuss the possibility of incidental findings and how they will be handled
- (5) 2A4. 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
- (3) 2A5. Explain to the patient issues of costs and financial coverage of genomic testing
- (11) 2A6. Order, interpret, and communicate the results of appropriate genomic tests, within the physician's scope of practice
- (8) 2A7. Provide referral to an appropriate specialist for genomic testing of a condition outside the physician's scope of practice
- (6) 2A8. Respond to the results of an abnormal genetic screening test, such as newborn screening, including immediate management and appropriate referral

2B. Knowledge for Practice (n=12 all inclusive)

- (6) 2B1. Describe the major forms of genomic variability
- (7) 2B2. Explain how different genomic changes may result in different phenotypes
- (11) 2B3. Recognize that genomic tests require interpretation with respect to the patient's clinical status (e.g., pathogenic, likely pathogenic, benign)
- (1) 2B4. Explain the concepts of analytic validity, clinical validity, clinical utility as they relate to genomic testing
- (1) 2B5. Recognize that medically "non-actionable" genomic results can be useful to the patient and family (i.e., personal utility)

2C. Practice-Based Learning and Improvement (n=5 all inclusive)

- (8) 2C1. Incorporate genomic findings into the health record and patient-care plan
- (2) 2C2. Have a method for periodic review of 'new' genomic interpretation for clinical applications.

2D. Interpersonal and Communication Skills (n=4 all inclusive)

- (3) 2D1. Ensure that undergoing genomic testing is a joint decision of the patient and the physician
- (7) 2D2. Explain and document findings from genomic testing to patient, including implications for other family members
- (3) 2D3. Facilitate access to resources to enhance patient learning about the results of genomic testing

- (3) 2D4. Address the needs of the patient as an individual as well as the needs of family members

2E. Professionalism (n=9 all inclusive)

- (3) 2E1. Be aware of and comply with local and federal laws and regulations regarding use of genomic tests
- (2) 2E2. Be aware of and responsive to patients' concerns about genetic discrimination
- (1) 2E3. Respect patient's privacy and need to maintain confidentiality of genomic information

2F. Systems-Based Practice (n=6 all inclusive)

- (2) 2F1. Explain who could have access to a patient's genomic information
- (5) 2F2. Recognize the effects of the costs and coverage of genomic testing on utilization by patients
- (0) 2F3. Facilitate access of patients to relevant clinical studies or trials based on genomic testing

2G. Interprofessional Collaboration (n=6 all inclusive)

- (9) 2G1. Initiate responsible referrals to specialists or other health professionals
- (3) 2G2. Provide support to patients based on recommendations of specialists
- (11) 2G3. Maintain a dialog with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status

2H. Personal and Professional Development

- (12) 2H1. Engage in continuing education regarding advances in genomic medicine and changing indications for and interpretation of genomic testing

3. Patient Treatment Based on Genomic Results (n=200 all total)

EPA: Use genomic information to make treatment decisions

3A. Patient Care (n=17 all inclusive)

- (16) 3A.Identify medical conditions and drug responses that have a strong genetic component
- (6) 3A2. Recognize that variants affecting drug responses found in a patient may also have implications for other family members
- (9) 3A3. Discern the potential clinical impact of genetic variation on risk stratification and individualized treatment

3B. Knowledge for Practice (n=18 all inclusive)

- (6) 3B1. Appreciate the importance of genetic diversity of humans and the abundance of genetic variants in each individual genome
- (4) 3B2. Identify single-gene disorders that may be amenable to targeted pharmacological therapy
- (5) 3B3. Recognize that genomic test results may guide choice of therapy for multifactorial disorders
- (8) 3B4. Recognize that there is variability in the phenotypic expression of genetic variants and in response to therapy
- (8) 3B5. Recognize that the effects of some medications are strongly influenced by inherited or somatically acquired genetic variation

3C. Practice-Based Learning and Improvement (n=11 all inclusive)

- (4) 3C1.Use evidence-based recommendations of professional organizations and others in implementing knowledge gained from genetic discoveries to improve therapeutics
- (1) 3C2. Document and periodically reassess therapeutic decision making in the medical record of patients
- (2) 3C3. Incorporate a realistic assessment of personal genomic knowledge and skill in the selection and use of consultants and improve competencies in the wake of these interactions

3D. Interpersonal and Communication Skills (n=15 all inclusive)

- (6) 3D1. Discuss benefits, risks, and alternatives of various preventive and therapeutic approaches driven by genomic findings
- (3) 3D2. 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
- (3) 3D3. Discuss pharmacogenomics implications for future health

3E. Professionalism

- (12) 3E1. Respect and guard privacy of the patient and the family members

3F. Systems-Based Practice

- (12) 3F1. “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

3G. Interprofessional Collaboration (n=10 all inclusive)

- (1) 3G1. Recognize potential involvement of multiple organ systems in genetic disorders and therefore appreciate the need to seek appropriate consultation with experts in the field
- (2) 3G2. Make medical and genetic information available to other health-care professionals, upon obtaining proper consent, while keeping the patients’ interests as the primary priority

3H. Personal and Professional Development (n=5 all inclusive)

- (11) 3H1. Maintain the medical knowledge and clinical competence in genomics required for the provision of therapy
- (5) 3H2. 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

4. Somatic Genomics (n= 85 all total)

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

4A. Patient Care (n=2 all inclusive)

- (7) 4A1. Identify or facilitate identification of patients who may benefit from genomic testing of tissue
- (6) 4A2. 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
- (2) 4A3. Ensure that tissue biopsy procedures are coordinated to make certain that appropriate and sufficient material is obtained for testing
- (4) 4A4. Integrate genomic testing results into the patient-care plan

4B. Knowledge for Practice (n=5 all inclusive)

- (4) 4B1. Explain the concept of somatic genetic change
- (5) 4B2. Describe the role of genomic changes in the pathophysiology and treatment of cancer
- (5) 4B3. Explain how genomic testing can be used to guide choice of therapy and adjust drug dosage in patients with cancer

4C. Practice-Based Learning and Improvement

- (7) 4C1. 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

4D. Interpersonal and Communication Skills (n=3 all inclusive)

- (3) 4D1. 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
- (2) 4D2. Address any concerns the patient may have about test results
- (3) 4D3. Ensure that specialists involved in a patient's care are communicating with one another and with the patient
- (4) 4D4. Communicate to patients potential implications for his/her family

4E. Professionalism

- (3) 4E1. Ensure that the patient is aware of what will happen with any tissue samples obtained

4F. Systems-Based Practice (n=1 all inclusive)

- (4) 4F1. Maintain a dialog with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status
- (1) 4F2. Be prepared to refer patients to clinical trials designed to evaluate new approaches to cancer therapy

4G. Interprofessional Collaboration

- (6) 4G1. Make appropriate referrals to specialists and other health providers and support the patient in ongoing care

4H. Personal and Professional Development (n=9 all inclusive)

- (8) 4H1. Keep up-to-date with progress in the diagnosis and treatment of cancer and other tissue-based disorders

5. Microbial Genomic Information (n= 8 all total)

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

5A. Patient Care: (n=2 all inclusive)

- 5A1. Use genomic-based tests for infectious disease instead of classical strategies where appropriate (e.g., based on clinical validity and turn-around time)
- 5A2. 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
- 5A3. Interpret genomics-based tests for diagnosis, monitoring, and treatment of infectious disease

5B. Knowledge for Practice: (n=2 all inclusive)

- 5B1.Explain the core strategies for genomic testing for microbial disease
- 5B2. Describe how DNA or RNA sequence variations in the microbiome may predict response to therapy and clinical outcomes
- 5B3. Explain the potential reasons for false-positive and false-negative microbial genomic-based tests
- 5B4. Explain the importance of “normal” microbiome to health and disease

5C. Practice-Based Learning and Improvement: (n=0)

- 5C1. Monitor ongoing testing results and their implications for treatment and prognosis in chronic infection
- 5C2. Be aware of new genomic testing methods and their clinical applications and apply when appropriate
- 5C3. Maintain awareness of patterns of infection in your patient population and use genomic tests appropriate to these patterns

5D. Interpersonal and Communication Skills: (n=0)

- 5D1. Explain the results of microbial genomic testing to patients
- 5D2. Explain to patients and families results that signal a risk for contagion and take appropriate containment steps

5E. Professionalism: (n=0)

- 5E1. Provide guidance to patients on how to avoid transmission of microbial agents in the community
- 5E2. Appreciate the importance of genomic tests for public health and responsibilities of primary-care physicians in reporting results to the appropriate public health authorities

5F. Systems-Based Practice: (n=0)

- 5F1. Work with other health-care professionals to apply infection-control

measures when appropriate in both inpatient and outpatient settings

- 5F2. Reassure patients and health-care workers in those situations in which "infection control" is not indicated

5G. Interprofessional Collaboration: (n=2 all inclusive)

- 5G1. 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

- 5G2. Maintain a dialog with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status

- 5G3. Consult with infectious disease specialists as needed (e.g., to manage unusual or unexpected infection or infection that is highly resistant to treatment)

5H. Personal and Professional Development: (n=2 all inclusive)

- 5H1. Maintain up-to-date knowledge on genomic approaches to care for patients with microbial infection

*Competencies published in Genetics in Medicine advance online 24 April 2014

- Available at: http://www.g-2-c-2.org/start_search_map.php bottom of page select Physician: Framework for Physician Competencies