

# ISCC Competencies Working Group – EPAs (FINAL PRODUCT)

## 1. Family History

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

### 1A. Patient Care

- 1A1. Conduct patient interview to assemble family history
- 1A2. Use standard pedigree symbols in assembling family history
- 1A3. Recognize patterns of Mendelian inheritance and calculate simple Mendelian risks; provide this information to patients and family members as appropriate
- 1A4. Use empirical risk figures to provide appropriate information for complex (multifactorial) medical conditions
- 1A5. Recognize that traits may cluster in families due to multifactorial rather than Mendelian patterns of inheritance
- 1A6. Formulate an action plan to address relevant family history information

### 1B. Knowledge for Practice

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

### 1C. Practice-Based Learning and Improvement

- 1C1. Incorporate family history information into health record

### 1D. Interpersonal and Communication Skills

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

### 1E. Professionalism

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

### 1F. Systems-Based Practice

- 1F1. Focus family history on problems relevant to the individual patient's health

- 1F2. Facilitate patient's desire to communicate relevant family history information among health providers and family members

**1G. Interprofessional Collaboration**

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

**1H. Personal and Professional Development**

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

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## 2. Genomic Testing

**EPA:** Use genomic testing appropriately to guide patient management

### **2A. Patient Care**

- 2A1. Discuss the indications for genomic testing – specifically the benefits, risks, and alternatives
- 2A2. Explain the implications of placing genomic test results in the patient’s medical record
- 2A3. Discuss the possibility of incidental findings and how they will be handled
- 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
- 2A5. Explain to the patient issues of costs and financial coverage of genomic testing
- 2A6. Order, interpret, and communicate the results of appropriate genomic tests, within the physician’s scope of practice
- 2A7. Provide referral to an appropriate specialist for genomic testing of a condition outside the physician’s scope of practice
- 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**

- 2B1. Describe the major forms of genomic variability
- 2B2. Explain how different genomic changes may result in different phenotypes
- 2B3. Recognize that genomic tests require interpretation with respect to the patient’s clinical status (e.g., pathogenic, likely pathogenic, benign)
- 2B4. Explain the concepts of analytic validity, clinical validity, clinical utility as they relate to genomic testing
- 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**

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

### **2D. Interpersonal and Communication Skills**

- 2D1. Ensure that undergoing genomic testing is a joint decision of the patient and the physician
- 2D2. Explain and document findings from genomic testing to patient, including implications for other family members

- 2D3. Facilitate access to resources to enhance patient learning about the results of genomic testing
- 2D4. Address the needs of the patient as an individual as well as the needs of family members

## **2E. Professionalism**

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

## **2F. Systems-Based Practice**

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

## **2G. Interprofessional Collaboration**

- 2G1. Initiate responsible referrals to specialists or other health professionals
- 2G2. Provide support to patients based on recommendations of specialists
- 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**

- 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

**EPA:** Use genomic information to make treatment decisions

#### **3A. Patient Care**

- 3A.1. Identify medical conditions and drug responses that have a strong genetic component
- 3A.2. Recognize that variants affecting drug responses found in a patient may also have implications for other family members
- 3A.3. Discern the potential clinical impact of genetic variation on risk stratification and individualized treatment

#### **3B. Knowledge for Practice**

- 3B.1. Appreciate the importance of genetic diversity of humans and the abundance of genetic variants in each individual genome
- 3B.2. Identify single-gene disorders that may be amenable to targeted pharmacological therapy
- 3B.3. Recognize that genomic test results may guide choice of therapy for multifactorial disorders
- 3B.4. Recognize that there is variability in the phenotypic expression of genetic variants and in response to therapy
- 3B.5. Recognize that the effects of some medications are strongly influenced by inherited or somatically acquired genetic variation

#### **3C. Practice-Based Learning and Improvement**

- 3C.1. Use evidence-based recommendations of professional organizations and others in implementing knowledge gained from genetic discoveries to improve therapeutics
- 3C.2. Document and periodically reassess therapeutic decision making in the medical record of patients
- 3C.3. 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**

- 3D.1. Discuss benefits, risks, and alternatives of various preventive and therapeutic approaches driven by genomic findings
- 3D.2. 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
- 3D.3. Discuss pharmacogenomics implications for future health

#### **3E. Professionalism**

- 3E.1. Respect and guard privacy of the patient and the family members

### **3F. Systems–Based Practice**

- 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**

- 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
- 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**

- 3H1. Maintain the medical knowledge and clinical competence in genomics required for the provision of therapy
- 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

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

### 4A. Patient Care

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

### 4B. Knowledge for Practice

- 4B1. Explain the concept of somatic genetic change
- 4B2. Describe the role of genomic changes in the pathophysiology and treatment of cancer
- 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

- 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

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

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

### 4F. Systems-Based Practice

- 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

- 4F2. Be prepared to refer patients to clinical trials designed to evaluate new approaches to cancer therapy

#### **4G. Interprofessional Collaboration**

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

#### **4H. Personal and Professional Development**

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

## 5. 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

### 5A. Patient Care:

- 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:

- 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:

- 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:

- 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:

- 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:**

- 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:**

- 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:**

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

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