Online Appendix 4: Knowledge items assessed during ELSI Genetics pre-post survey

Genetic content knowledge:

Sally was diagnosed with cystic fibrosis and shown to have both the delta F508 allele and a rare CFTR allele. Sally's mother tested positive for the one of the alleles (F508 allele). However, her father tested negative for both. What is the most likely reason that Sally's father tested negative for the two mutations?

a. Alternate paternity. Sally has different biologic father.
b. Laboratory error. This is a false negative result.
c. Maternal chromosome disomy in Sally.
d. Limited panel. The testing panel didn't include Sally's mutation.

Your patient has Klinefelter syndrome and is now on testosterone replacement. His mother wants his brother, who is 10 years old, to be tested. You know that Klinefelter syndrome almost always results from sporadic nondisjunction, and does not confer increased risk in other family members . Your best response is that her younger son:

a. Should be tested now, because effective treatment can begin as soon as the diagnosis is established.
b. Should not be tested, because he is too young to understand the implications of the disease.
c. Should not be tested, because he is not at increased risk.
d. Should not be tested, because of the stigma associated with the disorder.
e. Should be tested in a few years, because parents should plan how to help their children cope with infertility.

A patient's maternal triple screen results show a 1/95 risk of Down syndrome. Her regular physician is away, and you are cross-covering his clinic. You want to explain the results to her. Are the following statements are true or false?

"If 95 women like you had similar test results, one of those women would be carrying a baby with Down syndrome." ☐ True ☐ False

"If 100 women like you had similar test results, 95 of them would not be carrying babies with Down syndrome." ☐ True ☐ False

"The risk that the fetus has Down syndrome is about 1%." ☐ True ☐ False

"There is approximately a 99% chance that the fetus does not have Down syndrome." ☐ True ☐ False

Culture:

A 27-year-old woman from a small Asian village presents with venous thrombosis. She has no known risk factors. When you suggest testing for Factor V Leiden, she tells you to talk with her husband about her health decisions. When a female patient defers decision-making to her husband, the physician's approach should include:

Speaking only to the husband from that point onward. ☐ True ☐ False
Discussing with the patient the importance of individualism and self-determination. ☐ True ☐ False
Confirming the patient's wishes when the husband is out of the room. ☐ True ☐ False
Trying to talk with both the husband and the wife about decisions. ☐ True ☐ False
Respecting the patient's wishes. ☐ True ☐ False

Ethics:

Your patient's sister has been identified to have a BRCA1 mutation. Her family is pressuring your patient to be tested, because she may be at risk for breast and ovarian cancer. You are concerned about the extent to which your patient is able to consider what is right for herself, free of family and peer pressures. What is the term for this concept?

a. Voluntariness
b. Decision-making competence
c. Decision-making capacity
d. Assent

Advances in medical genetics will result in thousands of potential therapeutic targets to be identified over the next decade. If advances in genetic research lead to new treatments, equity in health care access will be an important social issue. Which of the following is the LEAST accepted principle of distributive justice?

a. To each person according to demonstrated need.
b. To each person according to their merit.
c. To each person according to social worth.
d. To each person an equal share.
e. To each person according to effort.

A pregnant woman had an amniocentesis. Chromosome analysis of the fetal cells confirmed that her fetus has Down syndrome. The parents are considering terminating the pregnancy. The disability community would raise which of the following ethical and social arguments about pregnancy termination in this case?

Health care resources should be used to develop treatments for genetic conditions, not to terminate pregnancies. ☐ True ☐ False

Aborting fetuses with genetic conditions encourages discriminatory attitudes toward people with genetic abnormalities. ☐ True ☐ False

Disabled children are best raised by disabled parents. ☐ True ☐ False

Health care resources should not be spent on the termination of pregnancy. ☐ True ☐ False

Many people with disabilities have a quality of life that is comparable with that of non-disabled people. ☐ True ☐ False

Pregnancy termination based on fetal characteristics, rather than other concerns (such as parental desire for a child), is wrong. ☐ True ☐ False

You are moonlighting in an outlying clinic. Your first visit was from a father who didn't want his daughter informed of her diagnosis of androgen insensitivity syndrome (46XY, with androgen resistance), who said, "It won't help her live her life." Your next visit was from a woman with inherited breast cancer, who didn't want her children informed about her gene mutation, saying, "My kids will make choices based on fear." Your third patient didn't want her father, who has Alzheimer disease, to know that her own genetic test results revealed a *PSEN1* mutation. "My dad will feel guilty that he passed on this gene to me." ***Paternalistic deception*** refers to the practice of withholding information from a person, when caregivers believe that disclosure would harm the person. Are the following statements about paternalistic deception true or false?

If no negative consequences are likely to arise, it is reasonable to withhold information. ☐ True ☐ False

Most women with androgen insensitivity syndrome wish they had not been told about their diagnosis. ☐ True ☐ False

Patients who discover that important medical conditions have been withheld usually feel isolated and betrayed. ☐ True ☐ False

Patients at risk for disease always have a right to know their medical condition. ☐ True ☐ False

If caregivers can be certain that patients will never discover their diagnosis, it is best that they not be told. ☐ True ☐ False

Legal issues

Your patient has a family history of hereditary nonpolyosis colon cancer. Your patient is worried that he may have the gene mutation as well, putting him at risk for colonic and extracolonic cancers. He is also concerned that a positive gene–test might affect his prospects for employment and life insurance. He decides to be tested, but he doesn't want the information put in the medical record. After the diagnosis of an inherited genetic disorder

Physicians should take special measures to ensure confidentiality of the medical record. ☐ True ☐ False

Physicians should let the patient know that the test result can't be kept out of their record. ☐ True ☐ False

Physicians should encourage genetic testing to identify family members at risk. ☐ True ☐ False

Patients should not to disclose their diagnosis, because they may be stigmatized. ☐ True ☐ False

Patients should not to disclose their diagnosis, because they may lose their life insurance. ☐ True ☐ False

Medical decision-making

The most important factor to consider when deciding whether to pursue genetic testing for a child at risk for genetic disease is:

a. The developmental level and age of the child.
b. Whether the medical management in childhood would be affected by the test result.
c. The severity of the suspected disease.
d. Whether insurance will cover the cost of the test.

Your patient has tested positive for a gene mutation that causes familial adenomatous polyposis, but she now refuses to have any further screening colonoscopies or a subtotal colectomy. "If I die, I die," she said. You know that the likelihood of developing colonic adenomas is 90% by age 40 in people with the mutation, and that most people with an APC gene mutation will develop colon cancer. Your patient is very upset. You wonder if she is depressed and lacking in decisional capacity. True or false? A patient with decisional capacity must:

a. Understand information relevant to the choice. ☐ True ☐ False
b. Be able to communicate their choice. ☐ True ☐ False
c. Have no concurrent psychiatric disorders. ☐ True ☐ False
d. Demonstrate a rationale for the choice. ☐ True ☐ False
e. Understand the implications of the choice. ☐ True ☐ False

Interaction with genetics professionals:

You have just identified a patient with probable hemochromatosis, based on serum ferritin and liver function tests. [note – these tests can provide highly suggestive results, but are not diagnostic]. Based on this family's pedigree, the disease seems to have an autosomal recessive inheritance pattern, consistent with HFE-associated hemochromatosis. Once you have established the diagnosis of HFE-associated hemochromatosis, which of the following is the best reason to refer the patient to a genetics professional?

a. To assure that treatment is appropriate.
b. To confirm that the patient's diagnosis is correct.
c. To discuss the timing of chromosome testing.
d. To discuss phenotype-genotype correlations in the patient.
e. To help identify family members at risk to be affected.

Informed consent

Informed consent prior to genetic testing is essential, because the implications of a positive genetic test result can be far-reaching for patients and families. However, under certain circumstances, a physician need not obtain informed consent from a patient. Which of the following is NOT a recognized exception to the duty to obtain an informed consent?

a. The patient declines a strongly recommended intervention.
b. The patient's condition constitutes a medical emergency.
c. The patient refuses to receive or to hear the information.
d. The patient may be seriously harmed by disclosure of the information.
e. The patient is unconscious and is likely to deteriorate without prompt intervention.

Your patient's father recently died from Huntington disease. Now, your patient wants to be tested so that he will know for certain whether he will develop the disease. You know that Huntington's is a trinucleotide repeat disorder that varies in age of onset and expression. It has no medical treatment, and although expression seems to be related to the number of trinucleotide repeats, this is not an absolute certainty. You are concerned that your patient might not understand the implications of testing, and that he can't give true informed consent. Are the following statements about informed consent true or false?

a. Informed consent can rarely be achieved, since most patients lack basic medical knowledge. ☐ True ☐ False
b. Use of medical jargon can interfere with informed consent. ☐ True ☐ False
c. Informed consent is a process, not an event. ☐ True ☐ False
d. Informed consent reflects respect for patient autonomy. ☐ True ☐ False
e. Informed consent is affected by both substance and method of information presentation. ☐ True ☐ False