PMI Public Meeting

Case Studies

Disclaimer: These case studies are meant to be forward thinking examples to help the FDA receive input from the patient and medical professional communities regarding the return of genetic test results. These are not meant to represent tests currently available or the scientific knowledge currently available.

Case Study 1 - Well Patient Tests/Predictive tests

John is a 30 year old Latino with a high school education. He is concerned that his grandfather was recently diagnosed with Alzheimer's disease at age 85 and read about a new test that can tell him if he's likely to develop the condition.

A: You are John. Consider the following:
- You have a mutation that, based on published data, gives you a 90% chance of developing early onset Alzheimer's.
- You have a mutation that, based on published data, gives you a 35% chance of developing Alzheimer's.
- You have a mutation that has contradictory evidence suggesting you are anywhere between 3 and 30% more likely to develop Alzheimer's than average.
- There is limited evidence regarding the clinical effect of this mutation
- You do not have any mutations that we know are connected to developing Alzheimer's disease.

In which of these cases would you want to know if you carried the mutation?
Would you want to receive this information directly from a laboratory, or through your physician?
What would you do with this information?
What if there was some evidence about the mutation and the chance of developing Alzheimer's, but not enough to be sure of the link?
What type of information would you need to understand your risk in each of these cases?

B: You are John's primary care doctor. What information would you like? How would you like it presented? What would you do with it?
- John has a mutation that, based on published data, gives him a 90% chance of developing early onset Alzheimer's.
- John has a mutation that, based on published data, gives him a 90% chance of developing early onset Alzheimer's but you didn't order the test for that mutation. Do you still want the result? What will you do with it?
- John has a mutation that, based on published data, gives him a 35% chance of developing Alzheimer's.
- John has a mutation that has contradictory evidence suggesting he is anywhere between 3 and 30% more likely to develop Alzheimer's than average.
- There is limited evidence regarding the clinical effect of this mutation
- He does not have any mutations that we know are connected to developing Alzheimer's disease.

Case Study 2 - Well Patient Tests/Predictive tests

Zoe is a 34 year old Caucasian unmarried woman who does not yet have, but wants, children. Her paternal aunt died of breast cancer at the age of 52. No one in her family that she knows of is of Ashkenazi Jewish descent.

A: You are Zoe. Consider the following:
- Do you want to know if you have a BRCA mutation that increases your likelihood of developing breast and ovarian cancer?
- Do you want to know if you may have other genetic mutations that could increase your likelihood of other cancers?
- What percentage increase in risk would cause you to consider increased surveillance or prophylactic surgery to remove your breasts and/or ovaries?
- How would you want to receive this information – from a laboratory or your physician? Or another source?
- Would major life decisions and estate planning be changed based on the results of your test?

B: You are Zoe's gynecologist.
- As you considering ordering these tests, do you consider that Zoe does not have a strong family history of cancer? Do you consider her ethnicity and the population for which the test is validated?
- Do you want the results of the test? Do you want do know if there are variants of unknown significance?
- What other information would you like?
- Would you refer your patient to a specialist? What type?
- How would you like this information presented?
- What would you do with it?

Case Study 3 – Oncology Tests

Carole is a 63 year old college educated woman who has a family history of cancer but no known pattern of specific cancers. Her family is of Middle Eastern and Asian heritage. She is diagnosed with lung cancer and has her tumor's genome sequenced.

A: You are Carole. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- Your lung cancer has a mutation for an FDA-approved companion therapy for lung cancers
- Your lung cancer has a mutation for a an FDA-approved companion therapy for breast cancers
- Your lung cancer has a mutation that may be connected to higher response rates in prostate cancers
- Your lung cancer has multiple mutations that may suggest different courses of therapies

B: You are Carole's oncologist. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- Her lung cancer has a mutation for an FDA-approved companion therapy for lung cancers
- Her lung cancer has a mutation for a an FDA-approved companion therapy for breast cancers
- Her lung cancer has a mutation that may be connected to higher response rates in prostate cancers

- Her lung cancer has multiple mutations that may suggest different courses of therapies

Case Study 4 – Pharmacogenomic Tests

Leslie is a 70 year old African-American woman with a number of chronic health conditions being managed by multiple medications. She recently learned about a gene that is involved in the metabolism of drugs and may help predict the right dosage of many different types of medication. She asks her primary care doctor to order the test for her and she finds out that she is a "slow metabolizer".

A: You are Leslie. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- The pharmacogenomics test that your doctor orders has been cleared to help find the right dosage of several medications – including some of the ones you are taking.
- The test suggests you should change the dosage of some of your medications, though your conditions are being well managed at your current dosages.
- You are taking medications that the test currently does not predict the correct dosing for. Would you lower the doses of those drugs too?

B: You are Leslie's primary care provider. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- The pharmacogenomics test that you order has been cleared to help find the right dosage of several medications – including some of the ones Leslie is taking.
- The test suggests Leslie should be on lower doses of some of her medications, though her conditions are being well managed at the current dosages.
- Leslie are taking medications that the current test does not predict the correct dosing for. Would you lower the doses of those drugs too?

Case Study 5 - Chronic Disease Tests

Doug is 22 year old Caucasian with a history of depression and schizophrenia that are moderately well-controlled with drugs and therapy. His aunt tells him about a test his doctor can order to help him find the perfect drugs for his conditions by sending in a simple cheek swab.

A: You are Doug. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- The data guiding the treatment recommendations is not well-developed
- The treatment recommendations are provided as strongly recommended for a number of different options
- The treatment recommendations from the test conflict with your current regimen

B: You are Doug's psychiatrist. What information would you like? How would you like it presented? What would you do with it? Consider the following:
- The data guiding the treatment recommendations is not well-developed
- The treatment recommendations are provided as strongly recommended for a number of different options
- The treatment recommendations from the test conflict with his current regimen