

## PBL/Case based learning for Genetic Counselors

Title: Psychiatry in the era of Direct to Consumer (DTC) genetic testing

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Presentation: This case was designed to be presented in a 1 to 1.5 hour seminar.

### Objectives:

1. Understand some of the practical and psychosocial issues that can emerge in the context of direct to consumer genetic testing for psychiatric disorders
2. Provide a basic explanation of the etiology of mental illness in a manner suitable for patients/family members, and how to integrate direct to consumer genetic testing results into this explanation.
3. Understand and know how to address some of the psychosocial issues associated with genetic counselling for mental illness, and with receiving direct to consumer genetic testing results.

### USAGE NOTE:

Pages 1-10 are for participants to use in small groups, each facilitated by an individual who is using the facilitator guide appended to the end of this document. Pages are provided to groups one at a time, and each sheet should be fully worked through before the next page is presented by the facilitator.

### ADDITIONAL RESOURCES:

Peay and Austin. How to talk with families about genetics and psychiatric illness. W.W. Norton. 2011.

## Samantha Smith

You are a genetic counselor working in direct patient care; your next patient is Samantha Smith, a 42-year-old woman who has Bipolar Disorder (BD). She has been seen and treated by her family physician for about 20 years. The referral notes say that she has been treated with Lithium since being diagnosed with BD at 21. She is married and has one daughter (21), and has a very high-powered job at a local company, at which she has excelled despite having to take time off work during periods when she feels more depressed – the company is aware of her mental health problem and supportive of her (even writing articles about her in their company newsletter). But the referral notes indicate that she is frustrated and unsatisfied because poor mental health is affecting her work. Her BMI is 33, and she has poorly managed sleep apnea. The appointment has been made because she has had some genetic testing that the physician does not know how to interpret.

On arriving she tells you that she is thrilled to see you – an expert in genetics – because she hopes that you can help her understand what her test results mean because she is having a hard time making sense of it.

What is your immediate reaction to/how do you feel about her request and why?

What would you want to know from Samantha about the genetic testing?

Ask for Exhibit 1

Samantha says she got genetic testing because she wanted to give the information to her daughter because she too would soon develop the same condition. But now she is really confused by the test results she got – she was expecting that the results would tell her that she has a 100% chance to develop BD, because she has already been diagnosed with the disorder. But that’s not what the results say.

What questions do you need to ask?

You ask Samantha about what she means by saying her daughter will develop BD soon. She tells you that she herself developed BD at 21, and as her daughter just turned 21, she is certain to develop BD any day now, and this is something Samantha is extremely anxious about.

You ask Samantha what she understands about the causes of her BD.

She tells you that she knows BD is genetic, and this leads her to wonder aloud if somehow her sample or results got mixed up with someone else's, and she received the wrong ones. Alternatively, she wonders whether the test results mean that she does not in fact have BD, and whether – if this is the case - she can stop taking her medications? You tell her that those are really important topics that you can discuss with her, but first, you ask if there was anything else that she thinks might have played a role in the symptoms of her illness starting, and she answers with an unequivocal “no”. But you decide to explore possible environmental factors further, asking instead about what was going on for her around the time she was first diagnosed with BD. She becomes emotional and tells you a long list of highly stressful life events (death of a parent, personal physical injury in a car crash, best friend diagnosed with cancer, miscarriage) that occurred in rapid succession over a period of 6 months before her initial diagnosis.

You ask about family history of mental illness.

Samantha reports that aside from her mom who – she thinks - experienced one brief bout with depression, there was none. She now wonders out loud about that, and suggests in a half-joking manner that perhaps she is adopted, or her father is not biologically related to her, after all she must have inherited it from somewhere.....

What issues do you need to address?

What is the point of asking about family history here?

How could you use Exhibit 2 to help here?

You tell Samantha that the test result does **not** mean that they mixed up samples or that she doesn't have BD, and it doesn't mean that she should stop taking her medication. You tell her that her lack of family history of BD does not mean that she is adopted or that her father is not biologically related to her.

You tell her that it is far from a certainty that her daughter will develop BD. You start explaining for her that we don't typically inherit mental illness itself but we can inherit a vulnerability to mental illness.

You use the jar model (Exhibit 2) to illustrate for Samantha what we know about how BD is caused by a combination of genes and environment acting together. You tell Samantha that there are likely to be all sorts of different types of genetic variations that people can have that will make them more vulnerable to developing BD, and that this genetic test that she got only looked at one type of genetic difference. You talk about how everyone likely has some genetic vulnerability to mental illness, and that the reason no-one else in her family is affected is that they were lucky enough not to experience those life events that would fill the jar to the top.

Samantha smiles and nods and says she understands.

You are about to change gear, and ask her about how she is doing with her mood before moving to wrap up the appointment, but because you have a small uncomfortable feeling, you ask Samantha about the main thing that she took away from the discussion.

Samantha tells you that she understands that the test she had just didn't look at enough genes to provide the correct information that she does in fact have a 100% chance for developing BD (because she has it), and that her daughter will develop it soon, too.

What do you think happened here? And what do you do now?

You try again. You use new words to reiterate that BD is not caused by genes alone. You discuss how research shows that BD seems to be caused by both genes and environment acting together. You use the jar model to show Samantha how the stressful life events she experienced in the run-up to her diagnosis could have contributed environmental vulnerability factors to her jar. As you do so, you notice her face glazing over, and her colour draining.

What do you think could be going on? What do you do to address this?

You ask Samantha what is going on. She tells you that she is horrified to think that people know that she has BD now that she sort of understands that its not just genetic. She says that she would never have told anyone about her diagnosis had she known it was anything other than genetic in origin, for fear that people would blame her for her illness.

How do you feel? How do you respond?

You ask Samantha about where this idea comes from – she tells you that no, she has not heard anything from others that would make her think this is how they would react, but she believes it to be true.

You try to talk with her about how just because an illness is not entirely genetic does not mean that someone is culpable, and you tell Samantha gently but firmly that no-one could justifiably blame her for the stressful life events she described that preceded the onset of her symptoms.

She remains glazed and visibly upset, and blocks any further attempts to pursue this topic.

How would you proceed? What is the danger in finishing the session now? Is there any other strategy that you could try to use to try to show Samantha the positive side of the information you've given her?

How could you use Exhibit 3 to help here?



You talk with Samantha about the certainty she expressed that her daughter would develop BD too, and about how now that she knows that this condition arises as a result of genetics and environment acting together, this means that the chance for her daughter to develop BD is NOT 100%, and that there may be things that they can do to promote her daughter's mental health. She nods and seems to cheer a little.

You take the opportunity to talk with her about her own frustration with taking time off work for mental health reasons, and explore with her what she does to take care of her mental health. She tells you that beyond taking her Lithium every day, she doesn't feel that she does much. You talk with her about the recovery aspect of the jar model, and explain that if she accepts that there is more to the etiology of BD than genetics alone then perhaps there is more that could be done to help protect her mental health in addition to using psychotropic medications. You talk with her about cognitive behavioural therapy, and about how finding some better strategies for managing her sleep apnea could help her mood stability. You also discuss how the balance between exercising and eating regular healthy meals, along with feeling guilty about not being "perfect" in these regards can be tricky. Through all of this she says little. As you are now way over the time you had available for the appointment, you initiate closing the session, as she leaves your office she tells you, backwards over her shoulder, that had she and her husband known that the chance for children to be affected was <100%, they would have had more children, bursts into tears and dashes out.

How would you feel after an encounter like this? How would you manage it?

## Epilogue

One month later, Samantha is doing better psychiatrically than she has since her initial diagnosis with BD. She has a psychiatrist for the first time, who is trying some new medication strategies with her which already seem to be helping her more than the lithium did. She has bought into the need to address her sleep apnea as, on reflection, she was able to see for herself how her mood was most unstable after periods of very unsettled sleep. She is going for a walk every day, and is trying to do more to eat healthily – so far, she has lost 5kg. She and her daughter have just signed up for a mindfulness training series together.

What do you think might happen if someone attributes their BD entirely to environmental factors, rather than genetics?