PBL/Case based learning for Genetic Counselors

Title:	Psychiatry in the era of Direct to Consumer (DTC) genetic testing
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Version:	6 (March 2023)
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

Case author: Jehannine.austin@ubc.ca

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 and 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 dosen'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?

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?

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Facilitator guide

The following pages are for the use of the group facilitator only.

The facilitator's job is to guide participants through the case, ensuring that the participants' time is spent discussing topics that address the objectives of the case.

You do not need to be a subject matter expert to facilitate a group discussion, but familiarity with the content of the facilitator guide before starting is highly recommended.

The facilitator should encourage discussion amongst group members and should step in to redirect only when necessary. The notes outlined in these pages should help you to achieve that. The case could be completed in 1 hour, but the timing is very tight – more time is better - 1.5 hours is more optimal. If you are aiming to do this in one hour – paying close attention to the time allocated in the facilitator guide for each sheet is critical!

The facilitator should hand out the pages for participants one at a time, with each new page only presented once the group has addressed all of the relevant questions from the previous page. The facilitator should ask a member of the group to read aloud each new page that they present.

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.

READING THIS PAGE ALOUD, LOOKING AT EXHIBIT 1, AND ANSWERING THE QUESTIONS ON THIS PAGE should take 10 minutes (1 min to read, 4 mins discussing feelings, 2.5 mins each discussing sleep apnea and genetic testing).

PRESENT EXHIBIT 1: 23 and me genetic test results for BD.

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

Explore how the residents feel about addressing genetic testing for psychiatric disorders and where this comes from.

Participants may express the desire to go away and research the background for the test that Samantha had. You can inform them that this kind of testing is easy to access. People can upload their raw genetic test data from companies like 23andMe or Ancestry.com into any one of a variety of 3rd party websites that will produce data like this. Don't let this conversation continue too long! If needed, prompt them to step back and think about this from the perspective of the situation at hand – help them if needed to say that we know that psychiatric disorders are complex conditions that arise as a result of genes and environment acting together. PRSs for psychiatric disorders including BD account for less than 10% of the overall liability for the condition.

In sum, this means that the test result that she received means little from a clinical perspective.

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

If it doesn't happen spontaneously, draw them towards acknowledging that it would be important to know what her motivations were for initiating genetic testing in the first place.

GIVE THEM THE NEXT PAGE OF THE CASE!

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.

READING THIS PAGE ALOUD, AND ANSWERING THE QUESTION ON THIS PAGE should take 10 minutes (1 min to read, 9 mins discussing question).

What questions do you need to ask?

- If it doesn't happen spontaneously, help the participants identify that they need to understand:
- 1) What she means by saying that her daughter will soon develop BD
- 2) What she understands about the causes of BD and get them to think about how they would elicit this. What would they do if they ask directly and she says she doesn't know? What if they ask, and her response is: "Just genetics"? Would they go any further? Try to get them to suggest asking something like: "what was going on for you around the time you first got sick?" this is a really good way to get at some of the environmental factors that may have contributed.
- 3) Whether there is any family history of mental illness. Again get the participants to think about *how* they would ask about family history of mental illness. It is not sufficient just to ask "Does anyone have mental illness?" You need to ask multiple different ways because people think about it differently. If they don't come up with these example questions themselves, offer them as possibilities: "Did anyone in the family receive a diagnosis of something like (GIVE EXAMPLES) depression, anxiety, OCD?", "Did anyone seem to have dramatic mood swings?", "Was anyone ever hospitalized for mental health reasons?", "Did anyone have a nervous breakdown?", "Did anyone attempt suicide?"
- When they are done with discussing how they would ask, you can provide the family history information. Try to keep this quick. Simply tell them that there is no family history of mental illness aside from a single episode of postpartum depression in Samantha's mom. If for some reason they desperately need more info (please don't encourage this, it detracts from the point), give them the following:
- She and her husband (44) have one child only (21). Her husband has no mental health problems and no family history of mental health problems among his 4 sisters and aging parents, neither of whom have siblings. All his grandparents died in their 80s, his maternal grandmother had a "nervous breakdown" after delivering his mom, but aside from that there are no mental health problems.

Samantha has one brother (36) who has two young daughters (2 and 4), her parents have no experience with mental health problems aside from a brief bout of depression for Samantha's mom after delivering Samantha's brother. Samantha's parents each have one brother, no mental health problems amongst their kids or the grandparents. 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.....

READING THIS PAGE ALOUD, LOOKING AT EXIBIT 2, AND ANSWERING THE QUESTIONS ON THIS PAGE should take 10 minutes (1 min to read, 9 mins discussing question).

What issues do you need to address?

- Get the participants to identify that they need to address the following issues, they should refer back to the previous page:
- 1) That the test results do not mean that she does not have BD, and that this does not mean that she should stop her lithium.
- 2) Her lack of biological family history of BD does not mean that she is adopted/there is non-paternity/that they have mixed up the test results.
- 3) It is not a certainty that her daughter will develop BD the specific number

doesn't matter at this point - the key/fundamental issue is that it is not a certainty.

What is the point of asking about family history here?

Get the participants to say that

- 1) it is important to understand her experience of psychiatric illness in her family, and that if there were lots of other family members who were affected, it may have helped to explain why she feels that BD is genetic. The fact that there is no family history is interesting given her existing explanation of cause.
- 2) The family history can be used as a tool to have an individualized discussion based on her own personal and family history about how genes and environment contribute together to the development of these conditions.
- 3) It allows you to derive recurrence risks if necessary. If the participants want to talk about what the actual numbers are, explore with them whether numbers and risk estimates are of the highest importance here. If they don't get to it themselves, suggest that this is NOT the most important aspect of this particular appointment. It may help her to be told that the chance for her daughter to develop BD is not 100%, but perhaps that's all she needs to know, and the specific figure doesn't matter. Remind participants that actually, not all patients will actually want to know specific numbers regarding chances. But as you have spoken with Samantha, it has become clear that there are other things going on here that need to be dealt with, and that it is important for her to understand more than just the number to address these other issues. If they still want to talk about recurrence risk, point out that this is not one of the questions they have been asked to address at this point. But if it is desperately important and they cant move on, the chance for the daughter to develop BD would be around 15% based on her family history. Tell them that we can discuss this at the end.

How could you use Exhibit 2 to help here?

PRESENT EXHIBIT 2

Get the participants to talk through how its genes and environment acting together, and how that relates to the genetic test that she had, her lack of family history. Get them to realize (tell them if necessary) that its very common for there to be no family history amongst people with psychiatric disorders. Get participants to discuss how that is compatible with the jar model explanation of the etiology of mental illness – if they don't get there themselves, tell them that just because no other family members have been affected, this doesn't mean that they had no genetic vulnerability to mental illness. It looks like (from the research evidence) it is likely that we all have some, we just vary individually in how much. So, all it means if there is no-one else in the family who has experienced psychiatric problems, is that they were lucky enough to not have their jars fill all the way to the top. You tell Samantha that the test result does **not** mean that they mixed up results 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.

READING THIS PAGE ALOUD, AND ANSWERING THE QUESTION ON THIS PAGE should take 6 minutes (1 min to read, 5 mins discussing question).

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

- Maybe she didn't understand (you were not clear enough) or maybe her understanding of the cause as genetic is very important for her psychologically for some reason. Get them to try to figure out why that might be (e.g. to her, it means that she is not culpable). But the problems with this belief are that she might be doing little to protect her own mental health (remind them that her mental health is currently poor, time off work etc), that she is so anxious about her daughter. So there would be some benefits to shifting this belief, particularly if you can help her to see that environmental vulnerability does not necessitate culpability.
- Get the participants to say that they would try again using different words saying the key messages explicitly - and using examples of things she has shared with you to illustrate points.

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.

READING THIS PAGE ALOUD, AND ANSWERING THE QUESTION ON THIS PAGE should take 3 minutes.

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

She could be bored and nauseous for unrelated reasons, it could be that she doesn't understand and is feeling overwhelmed with it, or she could be horrified because of the reasons discussed on the last page. Get the participants to indicate that they would just ask her what is going on! 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.

READING THIS PAGE ALOUD, AND ANSWERING THE QUESTION ON THIS PAGE should take 6 minutes.

How do you feel? How do you respond?

The participants are likely feeling anxious and possibly guilty that they have removed a belief system for someone in such a way as to leave her feeling vulnerable. They may be unsure what to do now and worried that they have done harm. Remind them that we thought about this before doing it, and there were reasons for doing so – poor mental health, concern for her daughter etc.

Get the participants to say that they would explore where this belief comes from and to explain to Samantha that the things that happened to her were no-ones fault, certainly not hers.

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 and 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.

READING THIS PAGE ALOUD, LOOKING AT EXHIBIT 3 AND ANSWERING THE QUESTIONS ON THIS PAGE should take 8 minutes (1 min to read, 7 mins discussing question).

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?

Get the participants to think about whether they would finish or continue the session. The danger is that you've essentially opened a can of worms for her, its not really right not to make a decent effort to try to help her realize the benefits of this pain that were the reason for getting into this in the first place. Ask the participants to imagine that they do have the flexibility of time to continue. They could talk with her about the implications of BD not being entirely genetic for her daughter, and how that might affect her positively: relief that its not a done deal that her daughter will have BD, things she can do to protect her own and her daughter's mental health

How could you use Exhibit 3 to help here?

PRESENT EXIBIT 3 and get participants to think about helping her see that there are strategies she could use to protect her own mental health and that of her daughter, so that she has to take less time of work for being depressed – this is something she would value. 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 dosen'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.

READING THIS PAGE ALOUD, AND ANSWERING THE QUESTION ON THIS PAGE should take 6 minutes.

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

Get the participants to talk about peer supervision for self-care, plans for follow up with the patient etc.

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?

Talk about how people can feel huge guilt if they feel their illness is entirely their own fault, and how if people think their illness is entirely attributable to environmental factors, then taking medication is not logical – they just need to try harder. They key is helping people to see that it is BOTH, while empowering them to feel that have some control, but not ultimate culpable responsibility.

Exhibit 1



Exhibit 2: Explaining development of psychiatric illness





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Exhibit 2 cont'd: Explaining development of psychiatric illness

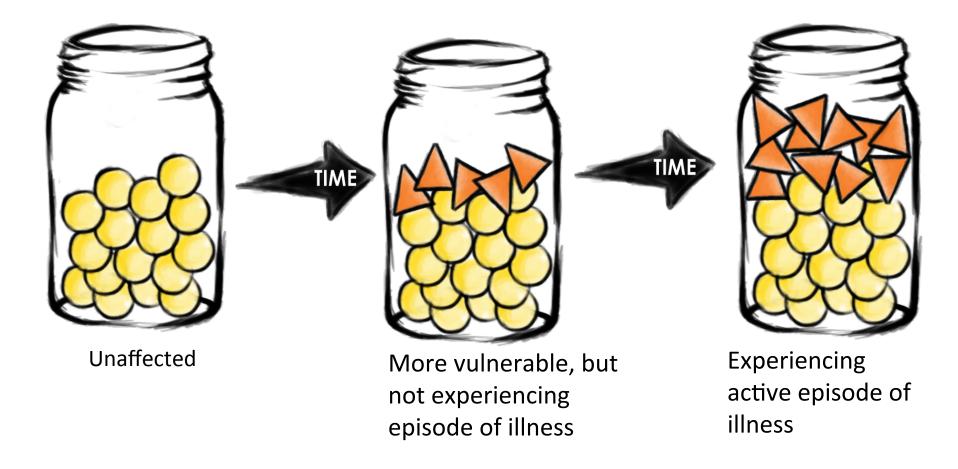
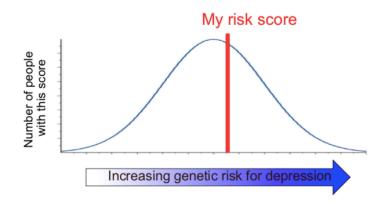




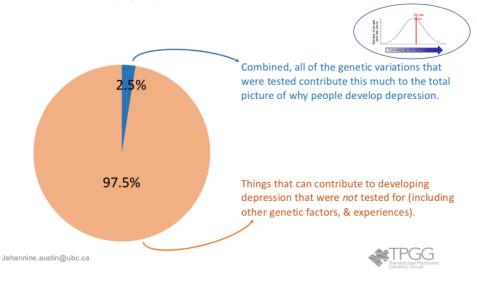
Exhibit 2 continued

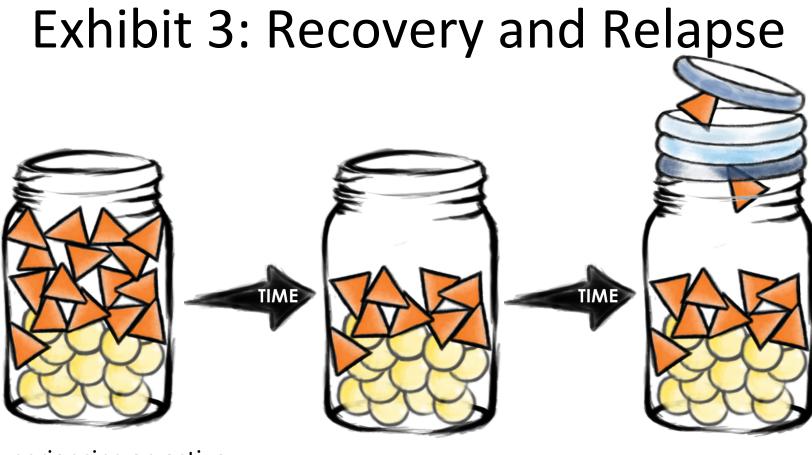
My genetic risk score for depression



This is how your genetic risk - according to the variations tested - compares to others' risk from testing the same set of variations

Pie chart representing the information that was provided by your genetic test in relation to *all* of the factors (including genetic & environmental) that can contribute to developing depression



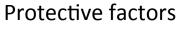


Experiencing an active episode of illness



Vulnerable, but not actively ill

Less likely to relapse



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