

It's Not That Simple!

Genomic Research & The Consent Process

[First in a Series of ELSI Plays and Dramatic Vignettes](#)

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Authorship is equally shared for manuscript article and original play

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NARRATOR: As the play commences, we find Bobby, Amy, Sam, and Ellen Friedman in the Pediatric Genetic Clinic with Dr. Hardy. Bobby is a nineteen-year-old with an autosomal recessive genetic disorder. His degenerative symptoms manifested several years ago, and are of a similar nature to those beginning to affect his sister, Amy, age sixteen. Sam, their nine-year-old sibling, does not have the heritable condition. They reside with Ellen, their mom, who is an elementary school teacher. Their dad, Howie, lives in another town with his second wife and young son.

DR. HARDY: I don't think I have to tell you what you folks are all too aware of – both Bobby and Amy are clearly experiencing a decline in their health. Bobby more than Amy since his symptoms started first.....

MOM (interrupts): Isn't there anything new that could be done? I'm always seeing stories in the newspapers about all sorts of tests and treatments being developed for genetic diseases.

AMY: You'll find some way to cure us, won't you Dr. Hardy?

SAM: Why did I have to come? I'm not sick.

DR. HARDY: In fact, the reason I gathered you all here today is that I have some good news. We now have a better chance to understand what's causing your disorder -- and you can be part of a new study. That includes you too Sam.

SAM: How can I help them if I don't have their bad gene?

MOM: We'll do anything you say Dr. Hardy...as long as it's safe.

DR HARDY: There's little danger...just requires a simple blood draw. It's for a new type of analysis, called genome sequencing. We use this to help understand the causes of genetic disorders by looking at all the human genes we know about. I have consent forms for you all to sign. Take them home and read them before signing.

AMY: I know I want this test. I'll sign up for it right now! I don't need to read anything.

BOBBY: Right, another test that probably won't actually help us. And don't be so trusting Amy.

MOM: Don't be so cynical Bobby.

DR HARDY: Let me explain further. Doctors at several institutions are collaborating to further our understanding of genetic disease...and are especially interested in studying disorders with an adolescent-onset that are known to have increased prevalence in the Ashkenazi population. Hopefully we'll also be able to learn more about Bobby and Amy's specific heritable defect to help them.

MOM: So are you saying that this test is not just about my family but mostly about lots of Jewish teenagers with Jewish diseases?

SAM: But I don't have any disease.

DR HARDY: We will be looking at several groups, including healthy siblings. I have five consent forms here for all the members of your immediate family – it's highly preferable for dad to participate and have his specimen taken too.

MOM: Dr. Hardy.... it may be.... difficult.... to include Howie.

BOBBY: Dad hates doctor stuff, and anything to do with sick people. That's why he left us.

AMY: I bet he'll do it when he hears it can cure us.

NARRATOR: Bobby, Amy, Sam, and Mom have returned home from Dr. Hardy's office with an array of medical documents and consent forms.

MOM: After all these years with so few answers – finally, there's some really powerful genetic technology.

AMY: Let's start reading the consent forms already.

BOBBY: I wouldn't be so rushed if I were you or too optimistic. I've been going through all sorts of genetic tests longer than you Amy. Why do you think this is going to be so different and solve our problem?

AMY: Because Dr. Hardy's consent form says so right here:

“We will use new techniques to read all of the genetic information in your cells that might cause a health problem if it contained a mistake.”

See, Dr. Hardy's new test will find our genetic mistakes.

SAM: You mean YOUR genetic mistakes. Can I go to my friend's house now?

MOM: Wonder if they're doing all our DNA or just the part called Whole Exome. I read on the internet that exomes code for protein and some doctor back East just used the Whole Exome test that helped save a baby.

BOBBY: I bet they look at our exomes first; it's gotta be cheaper. Then...maybe they'll test all of our genes? All I know is it says: **“DNA tests can take years....”**

AMY: Oh, but we need to get our results sooner than that!

MOM: Just glancing at this, why are there so many warnings in this consent form?

SAM: Mom...Did you hear me? Can I walk over and play at Danny's?

MOM: OK, go play. I'll take care of your consent.

BOBBY: No wonder they need seven pages to explain things. They think we should be happy the whole world doesn't see our medical information. Listen: **“Researchers who have access to genetic information will take measures to maintain the confidentiality of your genetic information.”**

But how much of a measure, a ton or a drop in the bucket?

MOM: There's going to be lots of people reading about us when they bluntly say:
Even investigators at other participating institutions may see your information if they consult with other researchers"

BOBBY: So, other than the janitor, everyone will know about us? Wow, look at this paragraph,
"Breaches in confidentiality involving genetic information could impact future insurability, employability, or reproduction plans, or have a negative impact on family relationships."

AMY: What does that mean? Reproduction plans? Does that mean it hurts my chances of having a baby?

MOM: That's not something we need to be worried about now. But this part about hurting family relationships? What happens if my sisters discover that our family's genetic information is known to so many people? They'll hate me for that!

AMY: Why do you care what they think – remember, it's been years since you even spoke to one of your sisters! But do I have to worry that every boyfriend is going to know I have bad genes?

BOBBY: What about me? Is it going to hurt me from getting a job if I get better because of our screwed up DNA? Look at this....

"There may be a risk that genetic information obtained as a result of participation in research could be misused for discriminatory purposes."

AMY: Don't worry Bobby. See it says:

"However, state and federal laws provide some protections against genetic discrimination."

BOBBY: Here it even warns us: **“....The Federal Privacy Act allows release of some information from your medical record without your permission...”**

And they include **“law enforcement”** ... that’s the cops Amy!

AMY: Well, if you don’t get in trouble with the police, then there’s no problem.

MOM: I’m not worried about the police, I’m worried about what you and Amy have. We don’t want to sound ungrateful and risk losing our chance to get these tests. I don’t earn enough to pay for them, and I wouldn’t even know where else to go. Anyway, this may be our last hope.

BOBBY: But here they say they store our samples **“indefinitely”** that means everything about us goes into a federal databank. But why?...and forever?

AMY: No need to worry Bobby. Didn’t you read about everything being **“coded and locked up.”** The consent says all our information is **“password protected.”**

BOBBY: You have got to be kidding me. People hack into computer files all the time and crack codes.

AMY: They make sure to tell us how careful they are – Look:

“When results are reported in medical journals or at scientific meetings, study participants are not named and identified.”

MOM: But if you read on.... **“ However.... it may include your family history and other medical information. It is possible but unlikely that you and/or a family member could be recognized....”**

Grandma would be so upset if our family story was out there for others to know.

BOBBY: I wouldn't want the world to see that either. And what about Sam?

MOM: You know...I have another worry. This says we might get told what's wrong even if we don't want to know. Look,

“You will be given a choice to learn or not to learn the results of your genome sequencing....The only exception to opting out is if we find a result that has urgent importance to your health. We plan to share this type of result with you.”

[SAM RETURNS HOME]

SAM: Hi...I'm back! Danny had to do homework.

BOBBY: The doctors don't say anywhere in all these pages just how urgent or bad it has to be before they tell us bad news! Does **“urgent importance”** mean you're going to die in a week?

AMY: The form goes on to say, **“In the FUTURE, we may contact you to find out if you are interested in learning about your results or gene variants that are important to your health and/or the health of your relatives.”**

BOBBY: They say **“important”**but how will they decide what's really important enough to tell us?

AMY: So Mom, how are you going to check this off?

“___ I would like to be contacted about the diagnosis, possible treatment, or genetic causes of my (my child's) disorder.”OR

“___ Please do not contact me regarding the progress of this study or any specific gene change you found.”

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MOM: Why isn't there a checklist for us to say exactly which diseases we want to be told or which we really don't want to be told about?

BOBBY: But what if Amy wants to know everything and you don't, and it turns out both you guys have something like the BRCA gene you hear about so much? And what about Sam?

SAM: Don't worry about me. I'm the healthy kid in the family.

AMY: Of course I'd want to know everything---We'd all want to know.

MOM: Don't jump to any conclusions Amy...you can't speak for the whole family.

SAM: That's for sure.

MOM: Your dad and I are healthy. We obviously gave you something bad in our genes, but there's no need for us to know anything. And who knows if your dad will even show up to do his part of the test?

AMY: How could you not want to know what's in your genes? That's so old-fashioned. Well, I checked off that I want to know all the results!

BOBBY: So did I, because I wouldn't want the doctors to know all about me and my genes if I didn't know too.

MOM: I would not want any of us to know if something bad is going to happen way into the future, like Alzheimer's, or breast cancer. Why would we want to know ahead of time and worry?

SAM: I don't really need to hear this stuff. I'm going to do my homework.

AMY: The consent form also said something about our relatives learning results.

MOM: My relatives? I certainly wouldn't want my mother hearing about any results, and I wouldn't even know how to begin to tell my sister Rachel. As for my sister Sarah, it's none of her business. If the doctors find something...I'll just keep it a secret.

BOBBY: A secret? You're terrible at keeping secrets Mom.

AMY: That nice genetic counselor can always tell Aunt Rachel and Aunt Sarah.

MOM: They don't need to know anything. If my sisters are curious, they can get tested themselves.

AMY: Mom...Just sign the paper and keep it simple please.

MOM: ***It's Not That Simple!***

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