Supplement A.

Mayo Clinic TRUST Survey

Qa. How much have you heard or read about genetic testing?

1. A great deal
2. Something, but not very much
3. Nothing at all

Qb. We are interested in learning which relatives you now have, as this may affect how people think about genetic testing. Indicate those <u>biological relations</u> that are currently living:

	Yes	No
Parent(s)		
Grandparent(s)		
Sister(s) or brother(s)		
Child or children		
Aunts or Uncles		
Nieces or nephews		
Cousins		

The following survey is sponsored by the Mayo Clinic and is about health and genetics. Before you begin, you will watch a short slideshow about what you could learn about your health from genetics. We will ask you to imagine that you had genetic testing and were being given choices about what sorts of results you would want to know about and what sorts of results you might NOT want to know about.

We will also tell you about different medical situations and ask you to think about those situations. However, some people may find it causes stress or anxiety or discomfort to think about serious medical problems. If you experience these feelings, you can stop the survey at any time.

The researchers are interested in learning the kind of information different people need or want in order to make decisions for themselves regarding possible genetic testing information.

This is just a survey of your opinions. We will not do actual genetic testing. Additionally, there are no wrong answers. All answers you provide will be confidential.

Click next to view and hear the short video.

Video 1 Narrative:

Slide 1: *You already know some things about genetics, even if you never studied it in school*
Slide 2: *Maybe you got your mother's eyes…..*
Slide 3: *Or your fathers hands…*
Slide 4: *These traits are influenced by the genetic information given to you by your parents*
Slide 5: *Parents pass half of their genetic building blocks to each child. The building blocks are called genes.*
Slide 6: *Genes can be passed from generation to generation.*
So, some traits or conditions can be seen as "running in families"
Slide 7: *All genes are made of letters (A, T, C, and G). Some genes have changes, such as extra, missing or different letters from what is expected. Genetic testing can detect these changes. Most changes are not harmful. But some changes can cause the gene not to work, and cause risk for disease.*
Slide 8: *About 20 years ago doctors were first able to order blood tests for genetic changes that might increase the chances for some medical conditions. But the testing was slow and very expensive*
Slide 9: *Today, scientists can read all your genes at once. There are 23,000 total genes. As of today about 4000 genes are understood well enough to connect to risks for particular medical conditions.*
Slide 10: *So if you could test all your genes, would you like to know all the conditions that you may have a higher chance of getting?*
Slide 11: *[First Panel] I want to know as much as possible about what medical conditions I might get. [Second panel] I don't think I want to know and worry about what might happen in the future.*
Slide 12: *[First panel] Bring it on– what is the point of staying in the dark if it is already in my genes? [Second panel] Would hearing about future risks make me feel bad?*
Slide 13: [*First panel]: If there is something I can do to stay healthy, I am going to do it. [Second panel]: Even if there is no prevention I can still plan for the future better if I know what might happen*
Slide 14: *[First panel]: I think my health care provider is in the best position to decide what I need to know. [Second panel] I want to be the one to decide about what information I get*
Slide 15: *Can health care providers give enough information to people to help them make good choices about genetic testing results? How can thousands of genetic conditions be discussed?*
Slide 16: *To cover the possibilities we have grouped genetic information into different categories. If you were having genetic testing, which groups of conditions would you want to know about? Through this survey we hope to improve how people learn about and make choices for getting genetic testing results.*

Q1. Imagine you were going to have testing done on all your genes and that it was possible to get all the results from your doctor or healthcare provider. There could be information about many different kinds of medical conditions available to you. For the sake of this survey, imagine that testing would be free and that results would not affect your healthcare insurance.

Here are some reasons some people might want their genetic testing results. Please indicate how important each reason is for you.

	Not at all Important	Slightly Important	Moderately Important	Very Important	Extremely Important
For knowing about my own health					
To know more about immediate health risks for my children					
To know more about health risks for my family members other than my children (parents, brothers, sisters, cousins etc.)					
For family planning					
I am just curious					
To understand health problems that seem to run in my family					

Video 2 Narrative

Slide 1: *So, Doc, is it bad?*
Slide 2: *Well, that depends on what you consider bad*
Slide 3: *Anything less than perfect health can be considered a problem– but how big a problem? How medical conditions affect us could depend on how old we are, what we do and what is important to us. . What may seem horrible to one person may be rather minor to another.*
Slide 4: *We have tried to organize all genetic conditions by how severe, or how serious, they are and by how actionable they are. By actionable we mean how much someone can do something about it. For severity we have grouped conditions into mild moderate and severe by how much the condition puts your health at risk, if not treated or if it is not treatable. Again, actionability refers to the ability to treat or prevent a condition.*
Slide 5: *Let's look at some examples. Low Severity conditions include genetic changes that will not really change how long you live or will not make a big change with your quality of life. Color blindness, or not being able to see red and green colors, would be considered as low severity. A scaly skin condition, or when the skin is flaky, is also considered low severity.*
Slide 6: *High severity conditions can have a big change on how long a person can live or how they are able to function in daily life. Examples might be getting a genetic form of a disease like Alzheimer's disease that leads to more and more problems with thinking and memory in middle age and can lead to death 10-20 years later…or a toddler who loses all of his or her muscle strength and dies before age 5.*
Slide 7: *Most genetic conditions would fall in the middle group, that of moderate severity.*

Q2. What seems bad to one person is perhaps not so bad to a different person. There are different reasons a person may think a medical condition is severe or not severe. Please indicate how important each condition is to you…. in deciding if something is severe.

	Not at all Important	Slightly Important	Moderately Important	Very Important	Extremely Important
May cause loss of ability to see or hear					
May cause loss of ability to walk or use your body normally					
May cause loss of brain function					
May cause loss of ability to have children					
Has serious financial consequences					
Caused difficulties for those closest to you					
Could be passed to your children					
Leads to your death					

Q3. When deciding which genetic testing results you would want to know about, how useful would it be for those medical conditions to be classified by how "severe" that condition is?

1. Extremely
2. Very
3. Slightly
4. Not at all

Next, we are interested in learning more about how the ***chances*** of a bad event changes people's thoughts about severity.

Q4. If a gene mutation can cause sudden death but this happens to only 1% (1 out of 100) of the people who have the gene mutation, would you consider this medical condition to be…?

1. Mild
2. Moderate
3. Severe

Q5. If a gene mutation can cause sudden death but this happens to only 5% (5 out of 100) of the people who have the gene mutation, would you consider this medical condition to be…?

1. Mild
2. Moderate
3. Severe

Q6. If a gene mutation can cause sudden death but this happens to only 10% (10 out of 100) of the people who have the gene mutation, would you consider this medical condition to be…?

1. Mild
2. Moderate
3. Severe

Video 3 Narrative

Slide 8: *So Doc, is it bad?*
Slide 9: *In thinking about medical conditions, information on how treatable or preventable the conditions are may play into whether or not people want to know of their risks for those conditions. " Actionable" is a word used to describe whether or not actions can be taken that will help prevent the condition or can treat the condition. For all illnesses, people may be able to have counseling, pain control, glasses or hearing aids, special education, or use mobility devices like wheel chairs and braces. This type of care (called supportive care) is not part of how we use the term actionability.*
Slide 10: *If a medical condition is actionable, there are medical treatments that can protect a person's health or return a person to ideal health.*
We have scored genetic disorders for how actionable they are on an A-B-C-D scale.
Slide 11: *Those in Actionabilty group A (treatable/preventable) can be mostly cured with medical treatment. An example might be a very serious condition in which a child's brain does not develop and leads to death. Treatment with a nutritional supplement (when something is added to what the child eats) leads to a healthy normal life.*
Slide 12: *For Group B Actionability (mostly treatable/preventable), medical treatments or prevention are very helpful but do not cancel out all of the issues or risks. For example, a change in a particular gene increases risks for cancer. Regular checking for early cancers, will usually, but not always, prevent cancer deaths.*
Slide 13: *In Actionability Group C (NOT mostly treatable/preventable), Some medical management is possible but the most serious parts of the disease are not prevented or made better. For example, changes in a gene causes on-going weakness that gets worse over time and leads to the inability to walk. Other symptoms of this condition such as problems with the eyes and diabetes (sugar problems) can be treated with surgery and medications.*
Slide 14: *For Actionability Group D (NOT at all treatable/preventable) No specific medical management changes the outcome of this condition. For example:*
Males with a certain gene mutation have lifelong moderate mental disability, or a gene change affecting bone growth causes some people to be very short, or a gene causes loss of vision and blindness by middle age. Remember the conditions can be mild, moderate or severe. Actionabilty refers only to the ability to treat or prevent.

Q7. When deciding which genetic testing results you would want to know about, how useful would it be for those medical conditions to be classified by how "actionable" that condition is?

1. Extremely
2. Very
3. Slightly
4. Not at all

Q8.Do you understand how we explain the actionability scale described in the slides? ***Actionability scale shown.***

1. No
2. Yes
3. Not Sure

Q9. How well did you understand the medical conditions discussed in the slides you just viewed?

1. I did not understand the medical conditions at all
2. I only understood the medical conditions a bit
3. I understood the medical conditions well
4. Not sure

Please read the information below:

When we learn about our own genes, we are also learning about things that may be present in our relatives. We have half of our genes in common with our parents, our children and our brothers and sisters. For second-degree relatives (grandparents, grandchildren, aunts and uncles), we have a quarter of our genes in common.

We have two copies of most genes, one inherited from each parent. For some genetic conditions, having one gene change means you are at risk for that medical condition; for other genetic conditions, having only one gene change means you are not at much risk yourself but could pass this on to your children, who could end up with the condition if their other parent also passed them a gene change.

So, if you received all the results of your genetic analysis it could have gene changes that pose no risk for yourself but might be "carried" silently, only to show up in your children or other blood relatives if a certain combination of genes came together. If you are a carrier, then likely one of your parents is a carrier and your children, brothers and sisters and more distant relatives also may be carriers.

Q10. Some genetic testing labs allow people to say if they want to know only about genetic changes that increase their chance of getting the genetic condition or genetic changes that don't carry much risk for that person as they are just "carriers". Do you think the information you just read should be explained in more detail to people who need to make decisions about learning about what genetic test results they want?

1. No, this amount of information is enough
2. Yes, more information is needed to understand what choices I might have between genetic changes for conditions I might get and genetic changes for conditions I might just be carrying silently
3. Not sure

Imagine now that you are the person making a decision about what results to learn from the testing of all your genes. Put a check in any of the boxes for the conditions you would want to know about. For example, if you would want to know only about high severity conditions if completely curable/preventable, you would put check just under column A; if you would want to know everything possible, you would put check in all columns.

Image shown.

Remember, you are thinking about genes that you might carry that could also be carried by your blood relatives including your children. So even if you obviously don't have this condition, would you want to know of the increased risk to other family members?

Q11. Considering both the severity of consequences and how responsive some medical conditions are to lifestyle changes and treatment, which types of conditions would you want genetic testing to identify? *(select all that apply)*

	Curable or preventable	**Mostly curable or preventable**	**Mostly NOT curable or preventable**	**NOT curable or preventable**
Low Severity				
High Severity				

Now let's look more closely at medical conditions that are in the middle: those of *moderate severity*. Most medical conditions probably fall into this category.

Image shown.

And remember, we are asking about gene mutations that you might carry silently but that could affect your blood relatives. Is it useful to you to know what body part or function is affected? Please indicate those results you would choose to learn about if given such a choice.

Q12. For conditions of <u>moderate severity</u>, which types of outcomes would you want to know about, related to your...? *(select all that apply)*

	Curable or preventable	**Mostly curable or preventable**	**Mostly NOT curable or preventable**	**NOT curable or preventable**
Eyes				
Seizures (sometimes called epilepsy, convulsions, or fits)				
Balance, coordination, or strength				
Bones: deformity, early bone loss, broken bones				

Heart diseases				
Kidney diseases (makes urine to remove body waste)				
Digestive diseases: disorders of esophagus, stomach, intestines, liver, gallbladder, pancreas				
Problems with fertility (being able to get pregnant or get someone pregnant)				
Immune system problems: getting a lot of infections or autoimmune disorders				
Increased risks for cancers in any part of body				

Q13. When thinking about genetic testing, do you believe it would be useful to select types of results by the body part(s) impacted?

1. Extremely useful
2. Somewhat useful
3. Somewhat NOT useful
4. NOT at all useful

In this section we describe some health problems caused by changes in the genes, or genetic mutations. We want to learn how much agreement there is on the severity and actionability of certain medical conditions. <u>We also ask you if you personally think you would want to know about this.</u> There are no right or wrong answers and… Remember, you are thinking about genes that you might carry that could also be carried by your blood relatives including your children. So even if you obviously don't have this condition, would you want to know of the increased risk to other family members?

*For **Severity,** consider how much a particular condition changes a person's ability to lead a typical full life. You will decide if it is mild, moderate, or severe.*

*For **Actionability**, think, "Is there something I can do about it?"*

Imagine a genetic condition in which a person starts to lose eyesight as a child. By age 40, one could be completely blind. Their health is fine in all other ways. There is no prevention or cure for this condition and their vision cannot be corrected.

S1Q1. How severe is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S1Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S1Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change would cause a child to be born blind and no treatment can bring back the child's vision.

S2Q1. How severe is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S2Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S2Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a medical condition where beating of the heart is not regular or beating can stop completely. This problem can be treated with medicine or maybe a pacemaker. If you don't get treatment, and even in some people who take medicine, your heart could stop beating, causing death.

S3Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S3Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S3Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S3Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a medical condition in which the kidneys stop working after a number of years. (As a reminder, kidneys clean waste products from blood.) If not treated, kidney failure leads to death. Medications may slow down how quickly the kidneys stop working, but cannot stop it entirely. Kidney failure can be treated with dialysis, in which the blood is cleaned by filtering the blood for several hours several times a week, or with transplantation of a kidney, which is not always readily available.

S4Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S4Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S4Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S4Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

There is a gene change that would cause you to begin, around middle age, to lose your ability to remember things and your personality changes. This worsens over the next 10-20 years until you have severe thinking and memory problems (dementia) and death. There is no medication known to prevent or treat this medical condition.

S5Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S5Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S5Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to be born with moderate mental retardation/impairment, and the need to live in a home where someone can help take care of them. The person with this condition cannot work, read or drive. There is no known prevention or treatment for this medical condition.

S6Q1. How <u>severe</u> is this condition?

1. Low/mild severity

2. Moderate severity
3. High severity

S6Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S6Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to get a lot of infections in the lungs. Breathing gets harder over time and there are problems with food digestion, requiring regular use of nutritional supplements and other medicines (enzymes). Men with this medical condition are infertile. Medical treatment can help people with this medical condition to live to be around 40 years old. If a person with this medical condition had a lung transplant, they might live longer.

S7Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S7Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S7Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S7Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely

5. Extremely likely

A gene change causes a person to have problems with their heart muscle that gets worse over time, making them feel weak and tired and they might even die. Medications can improve the weakness and tiredness but some people will eventually need a heart transplant to live.

S8Q1. How severe is this condition?
1. Low/mild severity
2. Moderate severity
3. High severity

S8Q2. How actionable is this condition?
1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S8Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?
1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S8Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?
1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

There is a gene change that causes Type 2 diabetes (problems with blood sugar) as a young adult. Diabetes can be treated with medicines, sometimes insulin shots. Good control of blood sugar can help prevent problems with eyes, kidneys, heart, and circulation that may develop in those with diabetes after many years.

S9Q1. How severe is this condition?
1. Low/mild severity
2. Moderate severity
3. High severity

S9Q2. How actionable is this condition?
1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S9Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S9Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change increases the chance of getting breast cancer and ovarian cancer. Screening for breast cancer more often and using newer techniques can help find breast cancers at earlier, more curable stages. Removing the breasts with surgery greatly reduces the chances that a breast cancer will develop. There are no good ways to screen for early ovarian cancer but taking out the ovaries (with surgery) by age 40 greatly reduces the chances that ovarian cancer will develop.

S10Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S10Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S10Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S10Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A person is born without any pigment in their skin or hair (they are albino, their skin is very white in color). This also affects their eye sight which cannot be corrected to all the way back to normal vision but people can wear glasses or get treatment that allows them to see well enough to do most jobs. A person can get easily sunburned and there is no correction for the loss of pigment in the skin. If a person has a lot of sunburns, this can lead to skin cancer.

S11Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S11Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S11Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S11Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A person has frequent epileptic seizures (also called convulsions or "fits"). With medicines, these can be controlled fairly well with only rare seizures happening. The person is otherwise healthy.

S12Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S12Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S12Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S12Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A child is normal at birth but by age 1 is not developing normally, develops seizures, loss of eye sight and despite good medical care, dies around age 3 years.

S13Q1. How severe is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S13Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S13Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to have problems with control of their legs when they become an adult. This makes it harder and harder to walk. They may need to get around with a wheel chair by middle age. Medications can relax muscles but do not change that the person cannot walk.

S14Q1. How severe is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S14Q2. How actionable is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S14Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S14Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to have a condition affecting the bones of the body (skeleton), leading to very short stature (being about three and a half feet tall as an adult). This person may also have sore joints from having joints that are not shaped correctly. Joints can be replaced by surgery. The person is otherwise healthy.

S15Q1. How severe is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S15Q2. How actionable is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S15Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S15Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely

3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to be born with profound deafness that cannot be corrected with hearing aids. They can learn sign language and attend schools for the deaf. They can have a cochlear implant which is an electrical device put into the ear. The device has a wire that the person wears on his or her head. This treatment works well enough that they can learn to speak and communicate by talking.

S16Q1. How <u>severe</u> is this condition?
1. Low/mild severity
2. Moderate severity
3. High severity

S16Q2. How <u>actionable</u> is this condition?
1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S16Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?
1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S16Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?
1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

A gene change causes a person to have an immune system that does not work, so they cannot fight off germs. They get frequent and severe infections. These can be treated with medicines or prevented with vaccines, but most people with this medical condition will die in childhood of infections. Death in childhood might be avoided if a bone marrow transplant is done. About 10% (10 out of 100) of children who have a bone marrow transplant will die of problems from the transplant.

S17Q1. How <u>severe</u> is this condition?
1. Low/mild severity
2. Moderate severity
3. High severity

S17Q2. How actionable is this condition?

S17Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S17Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
6. Slightly likely
7. Moderately likely
8. Very likely
9. Extremely likely

A gene change causes a child to be born with a head and lips that are not shaped correctly. They are otherwise healthy and development is normal. Surgery can help the head grow in a more normal shape and can repair the lip, but some visible differences will remain.

S18Q1. How <u>severe</u> is this condition?

1. Low/mild severity
2. Moderate severity
3. High severity

S18Q2. How <u>actionable</u> is this condition?

1. Treatable or preventable
2. Mostly treatable or preventable
3. Mostly NOT treatable or preventable
4. Not at all treatable or preventable

S18Q3. How likely is it that you would choose to learn now if your genetic test showed a result that increases the chance of this medical condition occurring in or being carried by you or your family?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

S18Q4. Imagine that your circumstances change, how likely is it that you might change your mind about learning a genetic result like this in the future?

1. Not at all likely
2. Slightly likely
3. Moderately likely
4. Very likely
5. Extremely likely

F1. Now that you have reviewed and scored several complex medical conditions in the previous examples, how helpful do you think it is to try to group these medical conditions by severity when thinking about the types of genetic tests you might want to learn about?

1. Not at all helpful
2. Slightly helpful
3. Moderately helpful
4. Very helpful
5. Extremely helpful

F2. How helpful do you think it is try to group these medical conditions by actionability when thinking about the types of genetic tests you might want to learn about?

1. Not at all helpful
2. Slightly helpful
3. Moderately helpful
4. Very helpful
5. Extremely helpful

F3. It would be possible to skip talking about how severe a disease is and just focus on how treatable/preventable it is. Would this be better than trying to break it down by how severe the conditions are?

1. No
2. Yes
3. Not Sure

F4. How certain are you that you can make a choice that is right for you about whether to learn genetic test results for different conditions? ***(Place your cursor on the ruler and select a position that best fits with how confident you feel)***

F5. How confident are you in your ability to…

	Not at all confident	Slightly confident	Moderately confident	Very confident	Extremely confident
Make decisions on how to categorize or group genetic information?					
Think about actionability (treatability/preventability) when making decisions about genetic information?					
Think about actionability and severity together when making decisions about genetic information?					
Make a choice that is right for you about whether or not to learn genetic test results for different conditions					

F6. Some experts say that it should be a policy to give back medically actionable genetic findings to patients whether or not they want to know the information.

Example: A committee of genetic experts recommends that if changes in a particular gene are found that are believed to increase the chances of getting cancer then this finding should be reported back to all patients regardless of whether this was requested or wanted, because it may be medically useful to that patient.

Do you like the idea of using a list prepared by experts for deciding what you should be told?

1. No
2. Yes
3. Not Sure

F7. Please indicate how much you agree or disagree with each of the following statements about receiving genetic test results.

	1 - Strongly Disagree	**2**	**3**	**4**	**5**	**6**	**7 - Strongly Agree**
For me, receiving genetic test results would be beneficial.							
For me, receiving genetic test results would be harmful.							
For me, receiving genetic test results would be worthwhile.							
For me, receiving genetic test results would be important.							
For me, receiving genetic test results would cause anxiety.							
For me, receiving genetic test results would be reassuring.							

And, finally, to better understand the information you and others provide to this survey, we would like to ask you a few questions about yourself.

D1. Which description best represents your personal situation. *(Please do not include non-biological children)?*

1. I don't currently have children, and may have children later.
2. I don't currently have children, and I don't intend to have children later.
3. I have children, and could have more children.
4. I have children, but do not intend to have any more children.

D2. In your opinion, compared to other people of your age, race, and gender, what do you think your chances are of getting cancer in your lifetime? Are your chances of getting cancer…

1. Much Lower
2. A little lower
3. About the same
4. A little higher
5. Much higher
6. I have already had cancer. Please tell us what type of cancer you had *(please specify)*

D3. In your opinion, compared to other people of your age, race, and gender, what do you think your chances are of having heart disease in your lifetime? Are your chances of having heart disease…

1. Much lower
2. A little lower
3. About the same
4. A little higher
5. Much higher
6. I have already had heart disease

D4. What is your experience with genetic testing (please select all that apply)?

1. I have no experience with genetic testing
2. I have had clinical genetic testing for cancer
3. I (or my partner) had prenatal testing during pregnancy
4. Someone in my family has had genetic testing for cancer
5. I have had genetic testing done through a personal genomics website like 23andMe or Navigenics
7. Other *(please specify)*

D5. How would you describe your current state of health?

1. Poor
2. Fair
3. Good
4. Great

D6. What is your biggest health concern?

D7. How important is it to you to live a healthy lifestyle?

1. Not at all important
2. A little important

3. Somewhat important
4. Very important

D8. Are you male or female?
1. Male
2. Female

D9. What is your marital status?
1. Married, living with spouse
2. Separated
3. Divorced
4. Widowed
5. Single, never married
6. Domestic partnership

D10. What is the highest level of education you have completed?
1. Did not graduate from high school
2. High school graduate
3. Some college, but no degree (yet)
4. 2-year college degree
5. 4-year college degree
6. Postgraduate degree (MA, MBA, MD, JD, PhD, etc.)

D11. In what year were you born?

D12. What racial or ethnic group best describes you?
1. White
2. Black or African-American
3. Hispanic or Latino
4. Asian or Asian-American
5. Native American
6. Middle Eastern
7. Mixed Race
8. Other

D13. What is your zip code?

Supplement B.

Eighteen prototypical medical conditions presented to participants.

Story 1: Adult blindness. Imagine a genetic condition in which a person starts to lose eyesight as a child. By age 40, one could be completely blind. Their health is fine in all other ways. There is no prevention or cure for this condition and their vision cannot be corrected.

Story 2: Congenital Blindness. A gene change would cause a child to be born blind and no treatment can bring back the child's vision.

Story 3: Cardiac Conduction Disorder. A gene change causes a medical condition where beating of the heart is not regular or beating can stop completely. This problem can be treated with medicine or maybe a pacemaker. If you don't get treatment, and even in some people who take medicine, your heart could stop beating, causing death.

Story 4: Renal Failure. A gene change causes a medical condition in which the kidneys stop working after a number of years. (As a reminder, kidneys clean waste products from blood.) If not treated, kidney failure leads to death. Medications may slow down how quickly the kidneys stop working, but cannot stop it entirely. Kidney failure can be treated with dialysis, in which the blood is cleaned by filtering the blood for several hours several times a week, or with transplantation of a kidney, which is not always readily available.

Story 5: Dementia. There is a gene change that would cause you to begin, around middle age, to lose your ability to remember things and your personality changes. This worsens over the next 10-20 years until you have severe thinking and memory problems (dementia) and death. There is no medication known to prevent or treat this medical condition.

Story 6: Mental Subnormality. A gene change causes a person to be born with moderate mental retardation/impairment, and the need to live in a home where someone can help take care of them. The person with this condition cannot work, read or drive. There is no known prevention or treatment for this medical condition.

Story 7: Cystic Fibrosis. A gene change causes a person to get a lot of infections in the lungs. Breathing gets harder over time and there are problems with food digestion, requiring regular use of nutritional supplements and other medicines (enzymes). Men with this medical condition are infertile. Medical treatment can help people with this medical condition to live to be around 40 years old. If a person with this medical condition had a lung transplant, they might live longer.

Story 8: Heart Failure. A gene change causes a person to have problems with their heart muscle that gets worse over time, making them feel weak and tired and they might even die. Medications can improve the weakness and tiredness but some people will eventually need a heart transplant to live.

Story 9: Type II Diabetes. There is a gene change that causes Type 2 diabetes (problems with blood sugar) as a young adult. Diabetes can be treated with medicines, sometimes insulin shots. Good control of blood sugar can help prevent problems with eyes, kidneys, heart, and circulation that may develop in those with diabetes after many years.

Story 10: Breast/ovarian cancer. A gene change increases the chance of getting breast cancer and ovarian cancer. Screening for breast cancer more often and using newer techniques can help find breast cancers at earlier, more curable stages. Removing the breasts with surgery greatly reduces the chances that a breast cancer will develop. There are no good ways to screen for early ovarian cancer but taking out the ovaries (with surgery) by age 40 greatly reduces the chances that ovarian cancer will develop.

Story 11: Albinism. A person is born without any pigment in their skin or hair (they are albino, their skin is very white in color). This also affects their eye sight which cannot be corrected to all the way back to normal vision but people can wear glasses or get treatment that allows them to see well enough to do most jobs. A person can get easily sunburned and there is no correction for the loss of pigment in the skin. If a person has a lot of sunburns, this can lead to skin cancer

Story12: Epilepsy. A person has frequent epileptic seizures (also called convulsions or "fits"). With medicines, these can be controlled fairly well with only rare seizures happening. The person is otherwise healthy.

Story13: Childhood neurodegenerative. A child is normal at birth but by age 1 is not developing normally, develops seizures, loss of eye sight and despite good medical care, dies around age 3 years.

Story 14: Mobility loss. A gene change causes a person to have problems with control of their legs when they become an adult. This makes it harder and harder to walk. They may need to get around with a wheel chair by middle age. Medications can relax muscles but do not change that the person cannot walk.

Story 15: Dwarfism. A gene change causes a person to have a condition affecting the bones of the body (skeleton), leading to very short stature (being about three and a half feet tall as an

adult). This person may also have sore joints from having joints that are not shaped correctly. Joints can be replaced by surgery. The person is otherwise healthy.

Story 16: Congenital Deafness. A gene change causes a person to be born with profound deafness that cannot be corrected with hearing aids. They can learn sign language and attend schools for the deaf. They can have a cochlear implant which is an electrical device put into the ear. The device has a wire that the person wears on his or her head. This treatment works well enough that they can learn to speak and communicate by talking.

Story 17: Immune Deficiency. A gene change causes a person to have an immune system that does not work, so they cannot fight off germs. They get frequent and severe infections. These can be treated with medicines or prevented with vaccines, but most people with this medical condition will die in childhood of infections. Death in childhood might be avoided if a bone marrow transplant is done. About 10% (10 out of 100) of children who have a bone marrow transplant will die of problems from the transplant.

Story 18: Craniofacial Anomaly. A gene change causes a child to be born with a head and lips that are not shaped correctly. They are otherwise healthy and development is normal. Surgery can help the head grow in a more normal shape and can repair the lip, but some visible differences will remain.

Supplementary Table C. Percent of respondents who reported they would want to know about genetic results of moderately severe conditions, according to actionability groups.[a]

Body part affected	Level of actionability			
	Curable/Preventable	Mostly curable/preventable	Mostly not curable/preventable	NOT curable/preventable
Eyes/Vision	63.3	72.4	47.4	44.6
Seizures	62.8	65.9	51.2	43.8
Balance/Coordination	61.9	68.7	49.5	41.2
Bones	62.2	66.6	53.7	45.5
Heart disease	67.1	68.6	50.1	45.0
Kidney	66.8	69.3	49.7	44.5
Digestive	63.4	69.9	50.4	43.4
Fertility	57.3	55.2	44.9	38.9
Immune system	64.9	67.8	51.4	44.0
Cancer	65.9	65.6	52.7	44.4

[a]This exercise explored whether the body part or disease type affected these choices.

Supplementary Table D. Proportion of respondents who indicated they wanted to learn of their genetic test results on each of the nine scenarios they scored.[a]

Scenario number	Total number who want to know[b]	
	Frequency	Percent
0	143	15.9
1	60	6.7
2	61	6.8
3	55	6.1
4	57	6.3
5	52	5.8
6	49	5.4
7	64	7.1
8	83	9.2
9	274	30.4

[a]For example, 60 individuals (6.7%) indicated they would want to learn results on just 1 of the 9 scenarios they scored.

[b]"Want to know" defined as very to extremely likely respondent would want genetic test.