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It is 2025 and Jill's 30th birthday is on the horizon. She has recently become a mother to baby Sophie and is keen to find out more about her family medical history. Jill never knew her father and her mother's knowledge of his side of the family is patchy. As a result Jill doesn't know much about her father's medical history. She therefore decides to get herself genotyped as an early birthday present. Her husband James is supportive of the decision, but her best friend Sara warns her against the idea, arguing that she may not be prepared for what she might find. Indeed, what Jill does not yet know is that she is at increased risk of developing schizophrenia.

The test will reveal a rare deletion in chromosome 3 that has been linked to schizophrenia, which she may also have passed to her daughter, Sophie. What's more, Jill is at an age where women with schizophrenia would typically start to show symptoms.

If you were Jill, would you want to know that you have an increased risk of developing schizophrenia?

Given that she has limited knowledge of her father's family history, Mrs Porter has indicated that she is keen to get herself genotyped. I would recommend that anyone who wishes to undertake such a test should initially undergo genetic counselling, to prepare them for any findings, such as an inherited predisposition to a disease.

I don't like it. You never know what might pop up during these tests. What if she has something really nasty? If it was me, I would rather not know!

Sara
(Jill's friend)

Jill's doctor

I think it's a brilliant idea! I know all about my parents' and grandparents' health issues, so I know what may be coming my way and I can change my lifestyle and diet to reduce my risk. Jill might have an issue that she needs to keep an eye on.

Jill

James
(Jill's husband)





With a condition like schizophrenia, is it better to find out now or to find out when symptoms occur?

What impact will the news have on the family, e.g. how might James react to the news?



Should baby Sophie (and any of her future siblings)
be genotyped? How might it
affect the way Sophie is raised?

What action can, or should, Jill take now that
she has received this news?



Who should have access to this information?
Jill's family, Jill's doctor? Insurance agencies?
Researchers? Potential employers?

Should anybody that does not know their family
history get themselves genotyped?



Most of the DNA in human cells is packaged into 46 chromosomes, in 23 pairs. Our genes are arranged along these chromosomes and carry the instructions to make molecules, such as proteins. To function properly, each cell depends on thousands of proteins doing the right job, in the right place, at the right time. Our complete set of DNA, containing all the instructions required to make us, is called a genome.



Gene mutations

A mutation is a permanent change in the DNA sequence. Mutations come in a number of different forms and can change the functioning of our genes. Some mutations may lead to a difference in the amount, or structure, of a protein produced by specific genes. Some mutations may switch genes on at the wrong time, preventing a protein from doing its job, or changing the way it does its job.



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What is schizophrenia?

Schizophrenia is a long-term mental illness. It can be devastating to families as it can cause dramatic changes to a person's behaviour. In extreme cases they may hear voices or have paranoid delusions. It can also cause severe depression; people with schizophrenia are 50 times more likely to commit suicide than people in the general population. Contrary to popular belief, people with schizophrenia do not have multiple personalities. Most people with schizophrenia do not show symptoms all of the time. Symptoms occur in "episodes" separated by symptom-free periods.

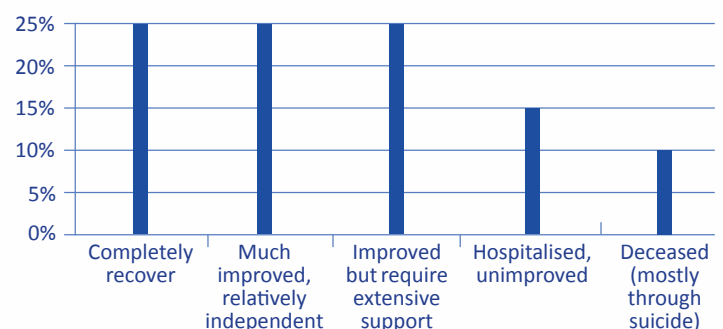


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Different experiences and levels of severity

Some people with schizophrenia have psychotic episodes lasting weeks or months but are able to recover and function normally between these episodes. Some people only experience one episode in a lifetime. At the other end of the spectrum, there are those who experience continuous symptoms.

10 year outcomes of people diagnosed with schizophrenia



Source: Surviving Schizophrenia



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A significant genetic risk

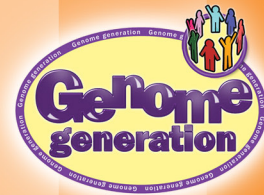
In most cases of schizophrenia, one individual gene is not responsible. However, a rare genetic deletion has been discovered on chromosome 3 of the human genome which shows a significant risk for schizophrenia. Schizophrenia affects less than 1% of the world's population but people who inherit this specific deletion are 17 times more likely to develop symptoms of schizophrenia than those with no deletion.



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Treatment of schizophrenia

As yet, there is no cure for schizophrenia but there are anti-psychotic medications which can be used to treat the condition. If medication for schizophrenia is discontinued, the chance of a relapse is approximately 80% within 2 years. With continued drug treatment, only about 40% of patients will have further episodes.

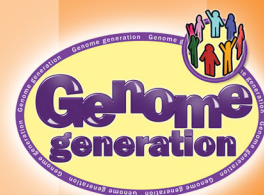


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Age of onset

Schizophrenia typically begins in early adulthood. Men tend to develop symptoms between 16-25 years, whereas women tend to develop symptoms later in life, often after the age of 30. It is very rare for people to develop schizophrenia under the age of 10 or over the age of 40.

There are some known environmental triggers for schizophrenia, in those who are susceptible to it. These include head injury, experiencing a stressful event and drug abuse.



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Detecting schizophrenia

The first signs of schizophrenia can be difficult to detect, e.g. becoming socially withdrawn and unresponsive, or experiencing changes in sleeping patterns. However, the earlier schizophrenia is diagnosed, the better. Early intervention and early use of new medications can lead to better medical outcomes for the individual.



Schizophrenia and employability

Compared to the average population, employment rates among people with schizophrenia are low. A study published in the British Journal of Psychiatry (2007) looked into the employment rates of people with schizophrenia in the UK, France and Germany. The study found that the proportion of people in employment was very low. Even among those who had fully recovered from the illness with only a single episode, the employment rate remained below 30%.



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Our DNA

Our bodies are made up of around 50 trillion cells. Most of these cells contain a person's genetic information within a long molecule called DNA (deoxyribonucleic acid). This DNA contains the genetic information that we inherit from our parents.

DNA is made up of specific sequences of letters or bases that encode instructions on how to make proteins. These bases [Adenine (A), Guanine (G), Cytosine (C) and, Thymine (T)] are lined up along two strands of DNA. The DNA is coiled into a double helix structure that resembles a twisted ladder.



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