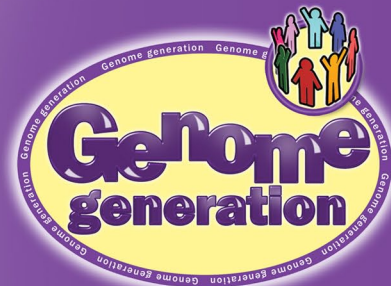


Genome generation

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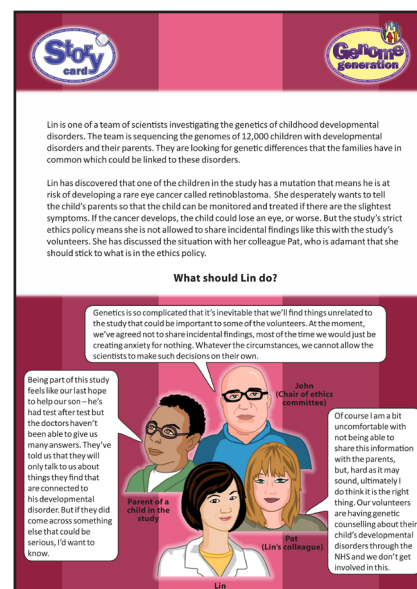
Should researchers share incidental findings?

This scenario is set in the present day.

Summary: This scenario is about a researcher (Lin) who is working on a study into childhood developmental disorders. She discovers that one of the children in the study has a mutation associated with the cancer retinoblastoma. The study has a strict policy not to share incidental findings with participants. The group has to decide whether Lin should tell the child's parents.

Initial question: Should Lin share the results with the participant's parents?

Key issues: *Unexpected or incidental findings; Access to data; Use of research data*



Complexity, sensitive issues & guidance on providing support

This scenario may be more suited to A-level students.

This scenario introduces groups to the issues of incidental findings in a research study, something that many people, particularly young people, will not have experienced. For this reason, this scenario may be more complex for some.

Some groups may have difficulty engaging with the discussion if they cannot imagine any reasons NOT to share the results with the parents. This might halt the conversation's progress. If so, remind them of some of the possible implications of sharing incidental findings for research studies, e.g. a significant proportion of money from the study will be taken away from the research to pay for genetic counselling, validation of results, clinical support.

Be aware that some people may be sensitive to the issues, if they have suffered from, or have relations with, retinoblastoma or developmental disorders. Be aware that some people can become more emotional and sensitive to issues that involve children.

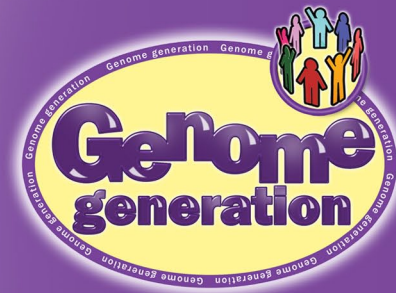
Some of the issues or questions that may be explored during discussions

Should incidental findings be shared with participants?

Should researchers share all incidental findings with the families of all study participants? There are a vast number of children involved in the study and numerous potential incidental findings. Which findings should be shared? Where do you draw the line?

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How significant is the risk of retinoblastoma?

With a condition like retinoblastoma the genetic mutation is more than a slight risk – it represents a high chance of developing the disease (90% of people with two mutant genes will develop the disease). Moreover, the condition presents itself in childhood.

Whose information is it?

If someone holds information about you or your child, should you have the right to that information? Should parents have the right to see all of the results? Are companies obliged to share their findings?

What are the financial and logistical implications of sharing findings?

The fact that there are so many participants (12,000 children) involved in the study – sharing all possible findings could make the study considerably more expensive to run. If the study chose to feedback all findings, it would have to provide appropriate genetic counselling and preparation, as well as ensuring that all results are validated before being fed back. The vast scope of this could mean that the original intended research might not go ahead.

Duty of care?

How would the researcher feel if she does not share the findings, but the child goes on to develop a condition that could have been prevented?

Further information – what they might need to know

Developmental disorders

It is estimated that between 30% and 40% of children with special needs do not have a specific diagnosis. Many developmental disorders are a result of duplications or deletions of particular parts of chromosomes, or an additional copy of an entire chromosome.

Disorders associated with chromosome 13

The *RB1* gene is located on chromosome 13 in a region called 13q14. Some cases of retinoblastoma are caused by the loss of regions of DNA in the q14 section of chromosome 13. Incidentally, deletions of this region can also cause developmental problems, such as intellectual disability, slow growth and certain facial features.

Ethics committees

Research organisations usually have their own ethics committees to evaluate and approve ethical safeguards for all types of research. In addition to this, the NHS's National Research Ethics Service oversees the ethics of any medical research to ensure that patients are properly protected. It works in line with a number of nationally and internationally derived guidelines.

Anonymous data and feedback

Scientists working on whole-genome sequencing studies usually deal with anonymous data to protect patient confidentiality. Access to the codes that allow individual patients to be linked to their data is heavily

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secured and accessible only to an authorised administrator. Scientists will communicate any findings to the administrator, who is then able to convey them to the patient's clinician. This will only happen if both the participant and the scientists have agreed that clinically relevant findings will be fed back.

Further reading

The following news story can be used in addition to the information cards to provide extra background information to help students understand the major issues raised in this scenario. The web link also includes a video that may also be of interest.

Do you want to know what lies in store?

<http://www.channel4.com/news/would-you-want-to-know-what-diseases-are-in-your-future>

Further information for students

Students can find out more information about retinoblastoma from the following websites:

NHS Choices

NHS Choices is produced by the National Health Service (NHS) and provides information on a range of different health issues. The page below provides information on retinoblastoma.

<http://www.nhs.uk/Conditions/retinoblastoma/Pages/Introduction.aspx>

Macmillan Cancer Support

Macmillan Cancer Support is a charity that provides information and support to people whose lives are affected by cancer.

<http://www.macmillan.org.uk/cancerinformation/cancertypes/childrenscancers/typesofchildrenscancers/retinoblastoma.aspx>

Cancer Research UK

Cancer Research UK is the world's leading cancer charity. It provides funding for research and provides information on cancer for patients, healthcare professionals and the public.

<http://cancerhelp.cancerresearchuk.org/about-cancer/cancer-questions/eye-cancer>

Follow up activity

Ask the students to create a diary entry (written or video) for Lin as she considers how she feels about the incidental findings and not being able to share the information with the child's parents.