

# Haematology

with Dr Jill M. Johnsen

## Talking points

### Knowledge

1. What is haemophilia?
2. What does the F9 gene do?

### Comprehension

3. What is the difference between the causes of haemophilia A and haemophilia B?
4. What is unique about the multiplex assay of variant effect (MAVE) used by Jill's team, compared to past MAVEs?

### Application

5. The Human Genome Project was a landmark 13-year international effort that produced the first sequence of the human genome, concluding in 2003. Nowadays, similar DNA sequencing tasks are done routinely and rapidly by scientists all over the world. What questions would you ask to learn more about how DNA sequencing has advanced so much in so little time?
6. How do you think Jill's approach to her research might be different if she did not have clinical experience of working with people with haemophilia?

### Analysis

7. Jill's team is only testing the effects of a single amino acid substitution on the functioning of the factor IX protein, but it is also possible to change two or more amino acids at the same time. Why do you think the team is not investigating such changes? Try to think of one practical and one biological reason.
8. How do you think other haematology researchers can use the library of variants generated by Jill's team within their research?

### Evaluation

9. A current issue within science is that we now have the tools to generate vast amounts of data, but fewer tools to process these data and find meaningful results. How do you think that technology, alongside the education of the next generation of scientists, can help address this issue? Use Jill's project to illustrate your points.

## Activities

### Individually

Design a poster that explores one of the two following topics:

- Haemophilia, including underlying genetic causes, the biomolecular mechanisms involved and symptoms
- DNA sequencing, including an overview of its history, its current power, and possible future advances

### In a pair

Science is all about collaboration between disciplines. Once you have completed your poster, pair up with a classmate who designed a poster on the other topic. Now, work together to use the information on both your posters, the article and extra online information to design a short presentation for younger students on how DNA sequencing is benefitting research into haemophilia.

For the poster and presentation, consider the following:

- Younger students will have less scientific/biological knowledge than you.
- Your aim is to communicate important and interesting points, without overwhelming your audience viewers with too much information
- Visual elements (such as illustrations and diagrams) will help you convey complex ideas.

### Reflection

Once you have finished your presentation, present to your class – or a class in younger year group, if you can. Afterwards, reflect on the process – both on the individual and collaborative creation. What are the advantages and disadvantages of each? Given the importance of collaboration in science, how do you think that collaborative processes could be made as effective as possible? What are the challenges of conveying scientific knowledge to others?

## More resources

- This factsheet from the Centers for Disease Control and Prevention explains what haemophilia is and how it is inherited: [www.cdc.gov/ncbddd/hemophilia/facts.html](http://www.cdc.gov/ncbddd/hemophilia/facts.html)
- This article from The DNA Universe takes a tour through the history of DNA sequencing and how it has advanced so dramatically since its inception: [the-dna-universe.com/2020/11/02/a-journey-through-the-history-of-dna-sequencing](http://the-dna-universe.com/2020/11/02/a-journey-through-the-history-of-dna-sequencing)
- This video from Precision Health explains the usefulness of genetic variant databases in medicine and beyond: [www.youtube.com/watch?v=zWSHTRkjUSE](https://www.youtube.com/watch?v=zWSHTRkjUSE)