

# CLINICAL GENOMICS

## WITH PROFESSOR JAMES LUPSKI

### Talking points

#### KNOWLEDGE & COMPREHENSION

1. How many base pairs are present in a diploid human genome?
2. What is the haploid reference human genome (HRHG), and what is it used for?
3. What is the difference between a haploid and diploid genome?
4. What are the three main types of genetic mutation? What happens to the DNA in each?

#### APPLICATION

5. Whole genome sequencing has many applications in medicine and genetics. Which other areas of science might benefit from this technique?
6. What questions would you want to ask James about his research and career?

#### ANALYSIS

7. How have James's personal experiences influenced his research?
8. Personal genome sequencing is currently still expensive. What are the ethical implications of this for people with genetic conditions?

#### SYNTHESIS

9. Whole genome sequencing is a powerful tool with the potential to improve many people's lives. If it was available to everyone, how might society be impacted?

#### EVALUATION

10. Despite the genetic variation resulting from sexual reproduction and mutations, only 0.1% of your genome is unique to you. As humans, we share 99.9% of our genome with each other. How does this make you feel? How might society be different if more people were aware of our similarities?
11. If the opportunity arose, would you have your personal genome sequenced? Why, or why not?

### Activity

#### The ethics of genomics

While James's genome was sequenced for the advancement of science, many people now choose to have their personal genome sequenced. In these cases, who should have access to these data?

Imagine that Mr X has had his genome sequenced and wants to keep his genetic information private. However, several people want to access these data. Build a case for each of the following, as to why they should be allowed to access Mr X's genetic information:

- Mr X's employer
- Mr X's life-insurance broker
- Mr X's child
- A scientist researching genetic diseases

In groups, discuss your different cases and decide who (if anyone) should be given access to Mr X's genetic information. Which arguments do you find the most and least compelling? What would the implications be if each person had access to Mr X's genetic data?

### More resources

- Learn more about James's research: [www.lupskilaboratory.org/home](http://www.lupskilaboratory.org/home)
- Learn more about the research conducted in James's department at Baylor College of Medicine: [www.bcm.edu/departments/molecular-and-human-genetics](http://www.bcm.edu/departments/molecular-and-human-genetics)
- Learn more about the Human Genome Project ([www.genome.gov/human-genome-project](http://www.genome.gov/human-genome-project)) and explore educational resources about genomics ([www.genome.gov/About-Genomics/Educational-Resources](http://www.genome.gov/About-Genomics/Educational-Resources))
- Visit James's Futurum webpage to read his article in Spanish and listen to his podcast: [www.futurumcareers.com/the-human-story-behind-human-genome-sequencing](http://www.futurumcareers.com/the-human-story-behind-human-genome-sequencing)