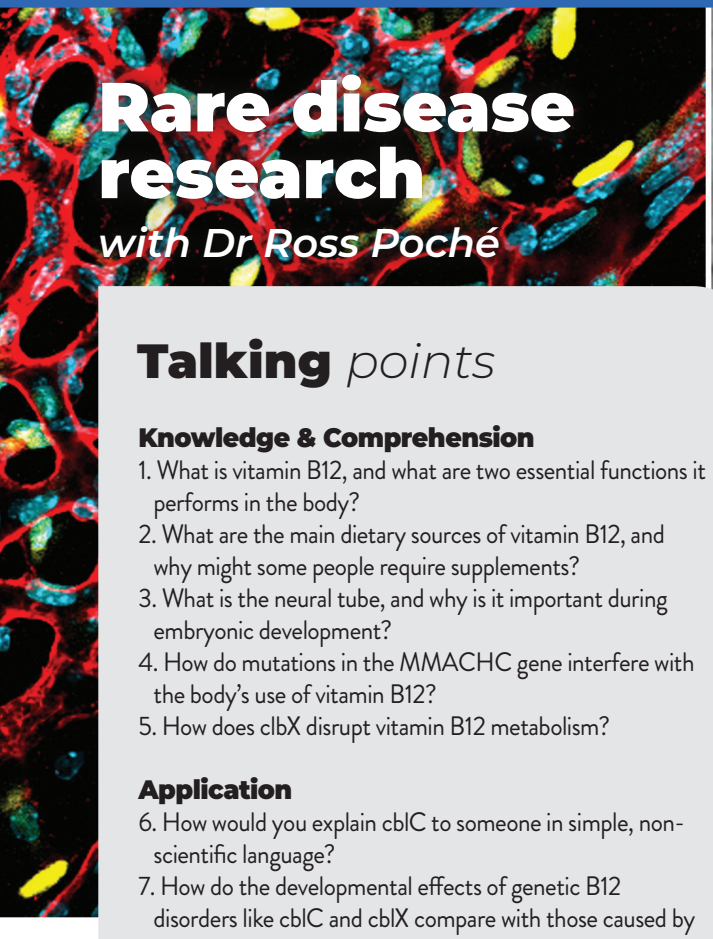

Rare disease research

with Dr Ross Poché

Talking *points*

Knowledge & Comprehension

1. What is vitamin B12, and what are two essential functions it performs in the body?
2. What are the main dietary sources of vitamin B12, and why might some people require supplements?
3. What is the neural tube, and why is it important during embryonic development?
4. How do mutations in the MMACHC gene interfere with the body's use of vitamin B12?
5. How does cblX disrupt vitamin B12 metabolism?

Application

6. How would you explain cblC to someone in simple, non-scientific language?
7. How do the developmental effects of genetic B12 disorders like cblC and cblX compare with those caused by simple dietary B12 deficiency? What implications might these differences have for treatment strategies during pregnancy?

Application

8. Why are mouse models often more useful than cell cultures for studying complex genetic disorders?
9. "Rare disease research not only addresses urgent medical needs for a small group of patients but also contributes to transformative discoveries with wide-reaching impacts," says Ross. In what ways can rare disease research lead to breakthroughs in common medical conditions?

Evaluation

10. "The best scientists aren't just skilled at doing science — they're also excellent communicators," says Ross. Why do you think it is important for good scientists to be skilled communicators? How good are your communication skills, and how could you develop them?

Creativity

11. Ross uses the analogy of workers in a city using power tools to explain how enzymes and coenzymes work together. Create your own analogy to explain this process.

Activity

Ross's research uses genetically modified mice to study rare conditions like cblC and cblX. These animal models provide vital insights into how certain genetic mutations affect vitamin B12 use and brain development. Some people argue that such research is necessary to develop treatments and save lives. Others believe that using animals in research is unethical, especially when alternatives such as advanced computer models and cell cultures are available.

1. Divide into two teams:
- Team A: Argue that using genetically modified animals in research is necessary and justified.
- Team B: Argue that the ethical concerns outweigh the benefits, and alternatives should be prioritised.

2. Do your research:
- Research real examples of medical breakthroughs that have come from animal studies.
- Research new tools and techniques that could potentially replace animal models.
- Consider the ethical concerns around animal suffering and discuss whether it can be justified.
- Investigate the laws, regulations and oversight in place to protect animals used in research.

3. Build your own arguments:
- Come up with at least three strong points to support your position.
- Make sure to include specific examples – such as real medical breakthroughs or ethical guidelines – to strengthen your case. It is also important to anticipate what the opposing team might argue and prepare thoughtful responses to challenge their points effectively.

4. Take part in the discussion:
- Each team should present their arguments, clearly explaining their position, then take turns responding to each other's points, challenging ideas and offering counterarguments. At the end of the debate, discuss with the classroom or your friends what you learned, whether any opinions changed, and why.

5. Reflect on the following questions:
- How should society decide which scientific goals are important enough to justify ethical trade-offs?
- What values should guide scientific research when it affects living creatures?
- How do personal beliefs and cultural perspectives influence opinions about scientific methods?
- What role should the public have in deciding how science is conducted, and how important is transparency in this process?

More *resources*

- Watch these Ted Talks about rare disease research: **youtube.com/watch?v=n0J1zCHURsQ** and **youtube.com/watch?v=CM7BZ9F72Kc**
- Baylor College of Medicine's Center for Educational Outreach has created a free digital science education platform containing interactive lessons, videos, and classroom resources for students and teachers: **bioedonline.org**
- Read this Futurum article about developmental biology: **futurumcareers.com/how-does-the-inner-ear-develop-into-a-sensitive-hearing-and-balance-organ**