16. Refsum’s disease

Eleanor Baldwin

Mr Andrews is a 25-year-old Caucasian male who was diagnosed with retinitis pigmentosa at the age of 14 years. His weight is stable at 80 kg, height 1.79 m and BMI 25 kg/m². His growth and development as a child were normal, but he has short third toes on both feet. His vision has recently been deteriorating more rapidly and he is complaining of itchy, scaly skin and mild ataxia. His plasma phytanic acid is currently 800 µmol/L. He has had genetic testing and been diagnosed with adult Refsum’s disease. He was referred for dietary advice.

- Describe how you would assess Mr Andrews using the ABCDE format.
- What is phytanic acid? Where is it found?
- What will be the effect on his symptoms of reducing its intake?
- What is the dietetic diagnosis?
- What is the dietetic intervention?
- Why is a regular meal pattern important for Mr Andrews? Why is it important that he does not fast or lose weight?
- Give five examples of foods that are high in phytanic acid and discuss suitable alternatives.
- Why should caffeine intake be reduced?
- How would you support him during an intercurrent illness?
- Describe how you would evaluate and monitor his progress. What outcome measures could you use?

Mr Andrews returned a month later and now weighs 75 kg. His symptoms have deteriorated despite following the diet; his plasma phytanic acid levels are now 1200 µmol/L.

- Why might Mr Andrew’s energy intake have deteriorated?
- What oral nutritional supplements would you recommend? Explain your rationale.
- If his symptoms rapidly deteriorate, how can they be relieved medically?

Mr Andrews continued to improve clinically and decided to take a holiday to the Far East. On his last day he developed gastroenteritis and travelled home avoiding food due to the nausea and vomiting; he managed to drink water. By the time he got home he had been fasting for over 40 hours.

- What would you estimate his phytanic acid level to be?
- What is your therapeutic goal?
• How would you try to achieve this therapeutic goal?