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lesson of the month (1)

Diarrhoea in a family from Pakistan

Lactose intolerance, the inability to metabolise lactose in milk and dairy products is due to deficiency of lactase in the gastrointestinal (GI) tract resulting in multiple GI and systemic symptoms. Diagnosis requires a degree of clinical suspicion.

Lesson

A father and his twin daughters were referred for gastrointestinal (GI) symptoms including abdominal pain and diarrhoea. The father's symptoms were mainly abdominal pain and diarrhoea occurring a few minutes after ingestion of dairy products but also wheat, most notable after consuming pizza. His problems started after the death of his first wife 10 years prior to the referral. The first of the twins complained about nausea and vomiting, mainly after drinking milk, with occasional watery stools but without increase in stool frequency. The other daughter had vomiting as a baby on weaning to cow's milk with persisting nausea but improving vomiting. She then developed diarrhoea after milk and abdominal pain after meals, in particular pasta and spicy foods. Defecation improved this daughter's symptoms. The family originates from Pakistan, the parents were blood related and the twins and two other healthy children were born in the UK. The father has a sister with similar problems. Physical examination, routine blood tests and tissue transglutaminase of the three patients were normal. An oral lactose challenge revealed no rise in glucose levels after lactose ingestion. Upper GI endoscopies with duodenal biopsies showed unremarkable histology consistent with the absence of coeliac disease in the three patients and the snap frozen samples were sent for disaccharidase measurements. All three patients had complete lactase deficiency with sucrase and isomaltase not being affected confirming selective lactase deficiency.

Discussion

Disaccharide lactose is only found in dairy products. In lactose intolerance, lactase, which cleaves lactose into the monosaccharide glucose and galactose, is absent in the intestinal tract. About 75% of adults worldwide show decreased lactase activity with a prevalence from 5% in northern Europe to more than 90% in African and Asian countries and Peru.[1–2] Humans, with a specific allele of the lactase promoter maintain lactase production throughout life. This adaptation to milk consumption is found in northern European and east African populations allowing ingestion of high doses of lactose.[3] In the absence of the disaccharidase, uncleaved lactose passes unchanged through the bowel leading to osmotic diarrhoea and bacterial fermentation. Resulting symptoms include cramps, bloating and flatulence. Systemic symptoms, including headaches, light headedness, loss of concentration and difficulty with short-term memory, tiredness, muscle and joint pains are described.[4] Primary lactose intolerance is unmasked on weaning, as illustrated in one twin.[5] Secondary lactose intolerance results from GI diseases including coeliac disease, parasitoses such as *Giardia* or rotavirus and is associated with reduction of other disacharidases.[6–7] A high level of suspicion is important and pointers are in the history in a patient who has migrated from a non-milk consuming to a milk consuming country. However, in the same populations, chronic GI infections are also common. For diagnostic tests see Box 1. In societies with little dairy exposure, lactose intolerance often does not require specific treatment. However, migration to largely lactose-tolerant societies can lead to troublesome symptoms. Treatment is by lactose avoidance, with substitution of nutrients found in dairy, including calcium, or enzyme substitution. Many patients learn through trial and error how much lactose they can handle with the tolerance of certain cheeses being an example. Special attention should be paid to processed food containing dairy, including bread, cake mixes, soft drinks, meat and lagers. Water-soluble lactose is not found in the fat portion of milk but is in higher concentrations in 'fat-reduced' or 'fat-free' products. Additionally, low-fat dairy foods often have various dairy derivatives including lactose added to enhance sweetness. Lactose-free milk substitutes include soy, rice or almond milk and they are often fortified with supplementary calcium, potassium and vitamins. Symptomatic patients do not often require dietary advice. When lactose avoidance is impossible or difficult, enzymatic lactase supplements are available.[8]

Box 1. Diagnostic tests for lactase deficiency.

- Stool acidity on weaning as a pointer in small infants
- Oral lactose challenge with absent increment of blood or breath metabolite
- Intestinal biopsy and presence of other disaccharidases but selective absence of lactase[5]
- Histology with absence of lactase mRNA
- Polymorphisms on DNA analysis is now possible[4]

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lesson of the month (2)

Acute aortic dissection with a high D-dimer and pleuritic chest pain in an airline passenger

D-dimer can be significantly elevated in acute aortic dissection and poses a diagnostic challenge in someone with pleuritic chest pain occurring after a flight. Electrocardiogram abnormalities in isolated acute aortic dissection may mimic other acute cardiovascular conditions.

Lesson

A 68-year-old previously fit Caucasian man was admitted from Heathrow airport complaining of chest pain. He was in transit, having flown from France, and was lifting his cabin baggage on his connecting flight when he complained of pain in his left temple. The pain moved to the left side of his face and then to the centre of his chest. The chest pain was sharp, pleuritic and severe associated with shortness of breath, sweatiness, nausea and the patient feeling hot. It eased with glyceryl trinitrate spray provided by the London Ambulance Service (LAS) but worsened after he had reached the emergency department (ED). Past medical history included diet-controlled hyperlipidaemia and mild hypertension, treated with atenolol.

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The patient was tired and pale with cool peripheries. Blood pressure was 99/62 mmHg, oxygen saturation 96% on room air and pulse 58 beats per minute. Pulses and blood pressure were equal between the arms. First and second heart sounds were audible along with a soft systolic murmur in the aortic area. Mild bibasal crepitations were heard in the chest. The abdomen was soft and non-tender with no organomegaly detected. No neurological deficit was found.

Arterial blood gas (ABG) sampling while breathing room air showed a (normal ranges in brackets) pH of 7.39 (7.35–7.45), pCO_2 of 4.31 KPa (4.67–6.40), pO_2 of 9.91 KPa (11.10–14.40), HCO_3- of 21.2 mmol/l, lactate of 3.2 mmol/l (0.5–1.6) and base excess of −3.8. A chest radiogram (CXR) was unremarkable. Electrocardiogram (ECG) showed 0.5–1 mm concave ST segment elevation in leads II, aVF and V2–6 (Fig 1), mimicking the ECG appearance of pericarditis.

Given the chest pain and ECG changes acute coronary syndrome (ACS) was diagnosed by the ED and 300 mg of clopidogrel and clexane (1 mg/kg body weight twice daily) were administered. The patient had already been given 300 mg aspirin by the LAS.

At the time of review by the acute medical team the D-dimer result was 3,520 μg/l (0–275). Five hour troponin I was also mildly positive at 0.10 μg/l (0–0.04). Full blood count and urea and electrolyte levels were within normal range. At this point the differentials included pulmonary embolus (PE) (given the history of the flight, pleuritic chest pain, slightly elevated troponin and significantly