Clinical Challenge 4:

A 16-year-old girl was admitted via emergency department with abdominal pain, nausea, and vomiting. She is known to the gastroenterologists with a diagnosis of Functional Abdominal Pain. She had a normal upper GI endoscopy, ileo- colonoscopy, MR enterography and a video capsule endoscopy in the last 6 months. She has been prescribed peppermint oil capsules, which she takes erratically. She is also on oral contraceptive pills for menorrhagia. She had reported that the pain got worse in the past two days prior to the admission. She has reported history of diarrhoea without any mucus or blood and abdominal bloating in the last two days.

Examination did not show any pallor, jaundice or clubbing. Abdominal examination did not show any features of obstruction or peritonism. She was kept nil by mouth and started on IV fluids, IV omeprazole and IV morphine. Blood tests done at the time of admission showed normal full blood count, liver and renal function tests, amylase and normal CRP. Urine pregnancy test was negative. CT abdomen was organised by about 4 hours of admission because of severe abdominal pain, which showed massive swelling of the small bowel without any other abnormalities. The plan by the surgical team was to closely monitor and arrange a laparotomy if the pain did not improve. Abdominal pain improved in the next 24 hours and an US abdomen done on day 3 of admission shoed normal small bowel without any oedema.

What is the diagnosis?

Answer to Clinical Challenge 4

The correct answer is **Hereditary Angioedema (HAE Type III)**

Many congratulations to **Ed Giles, Arun Urs, Kornilia Nikaki and Dominic Dell Olio**, for getting the correct answer.

Self-remitting abdominal pain without clear organic aetiology that is recurrent and lasts >6 hours is a clinical criterion for HAE defined by the European working group (**Hereditary and acquired angioedema: problems and progress: proceedings of the third C1 esterase inhibitor deficiency workshop and beyond. *J Allergy Clin Immunol*. 2004;114:S51–S131**). This patient had experienced an episode of a facial angioedema some years ago. On further exploring, it was known that other family members had history of facial angioedema, partially associated with angiotensin converting enzyme inhibitor intake.

HAEs are subdivided into 3 types: Type I HAE is characterized by low levels and reduced functional tests of C1 esterase inhibitor, whereas type II HAE shows normal levels but reduced function of C1 esterase inhibitor. Type III HAE, which affects nearly exclusively females in an oestrogen-dependent fashion (precipitation or worsening of symptoms during pregnancy, treatment with oral contraceptives, or hormonal replacement therapy) shows normal levels as well as normal function of C1 esterase inhibitor. The oestrogen dependency

is explained by the fact that the expression of FXII is increased by oestrogens via oestrogen-responsive elements in the promoter region.

This patient, C3 and C4 levels as well as the level of C1 esterase inhibitor in functional testing were normal. Thus, the diagnosis was hereditary angioedema with normal C1 inhibitor levels. The patient improved spontaneously after symptomatic therapy within 2 days. Avoidance of oestrogen-containing oral contraceptives was recommended and no episodes were observed in the next several months. A clinical relapse after 6 months was again associated with the intake of an oral contraceptive.