Extensive jejunal diverticulosis in a family, a matter of inheritance?

**ABSTRACT**

Diverticulosis of the jejunum is a rare finding (0.06 to 1.3%). Possible complications are bacterial overgrowth, malabsorption, bleeding, mechanical obstruction, volvulus and perforation. At present only one case report on familial jejunal diverticulosis has been published. We describe three patients with jejunal diverticulosis within one family, which might suggest inheritance.

**CASE REPORT**

The family consists of a mother and four children; no information is available about the father (figure 1).

Patient A: The mother died at the age of 83 due to infection and rapid dehydration. She had documented colonic cancer for which she was operated at the age of 50. She suffered from celiac sprue and chronic anaemia. Autopsy revealed extensive jejunal diverticulosis.

A son had a myocardial infarction at the age of 50 and colonic cancer was diagnosed at the age of 60. He shows no symptoms related to jejunal diverticulosis. Another son died of a myocardial infarction, no other data are available.

Patient B: A son is currently being treated intermittently for bacterial overgrowth due to jejunal diverticulosis diagnosed with enteroclysis.

Patient C: A daughter came to our hospital with complaints of cramp-like abdominal pain, nausea, vomitus, flatulence and weight loss. She had lost over 25 kg in two years time, and her BMI had dropped to 18 kg/m². Gastroduodenoscopy showed no abnormalities, biopsies excluded coeliac sprue. H₂-breathing tests were positive for lactose and glucose, suggesting bacterial overgrowth. Enteroclysis demonstrated jejunal diverticula (figure 2). She was treated with doxycycline for bacterial overgrowth due to jejunal diverticulosis. This alleviated most of her symptoms and she gained 13 kg in weight. She kept having episodes of abdominal pain and vomiting. She underwent a diagnostic laparoscopy with resection of a diverticulum, which was probably the cause of these transient symptoms related to ileus. Laparoscopy revealed jejunal diverticula involving about half of the small bowel.

**Figure 1.** Pedigree chart compatible with an autosomal dominant trait

**Figure 2.** Enteroclysis of patient C demonstrating jejunal diverticulosis
One of the diverticula was stapled and histology showed pneumatosis intestinalis probably due to her severe form of bacterial overgrowth or as a result of her motility disorder. She was treated over a long period of time with a lactose-free diet, cobalamin injections every three months and doxycycline. A surgical treatment is not feasible as it would involve a small bowel resection extending over 50% of the total small bowel length.

As was calculated in the article by Sternberg et al., simple calculation shows a chance of \(1.28 \times 10^{-3}\) that three people in a family of five have jejunal diverticulosis coincidentally:

\[(p)^3 \times (1-p)^2 \times (4!/2!x2!); p=0.013 \text{ (the highest prevalence mentioned)}\]

Although it is statistically only partially correct to use prevalence as a P value, it is a very strong indication that coincidence is highly unlikely.

**CONCLUSION**

We present a family with jejunal diverticulosis in the absence of apparent connective tissue disease. The inheritance pattern is compatible with an autosomal dominant inheritance trait, which clearly infers a genetic cause. As such it is the second family to be described.

**REFERENCES**


**ANSWER TO PHOTO QUIZ (ON PAGE 153)**

**SINE WAVES**

ECG findings with hyperkalaemia are well described in the literature. When the potassium level is between 5.5 and 6.5 mEq/l, large-amplitude T waves may be seen. PR prolongation, P-wave flattening or disappearance, QRS-complex widening, and conduction blocks with escape beats are typical findings, which may be associated with a potassium level of 6.5 to 8.0 mEq/l. Sine-wave appearance is an ominous ECG finding which precedes ventricular fibrillation. Usually sine waves correlate with a potassium level >8.0 mEq/l. It should be emphasised that hyperkalaemia may present with minimal ECG changes.

**REFERENCE**