

Letters

CEC SYNDROME –A RARE MANIFESTATION OF COELIAC DISEASE

Editor,

We report a case of incomplete CEC syndrome, a rare condition comprising coeliac disease and bilateral occipital calcification. There are no previous published reports of CEC syndrome in Northern Ireland.

Case Report: A thirty-six year old lady presented following two episodes of facial numbness of ten minutes duration, one of which was associated with blurred vision. There was a three-kilogram weight loss over six months and there was no significant past medical history. Clinical examination was unremarkable.

Routine blood investigations revealed a mildly decreased folate level and a CT scan of the brain revealed cortical-based bilateral serpiginous calcification. An MRI scan showed no evidence of Sturge-Weber syndrome, a prominent cause of cerebral calcification, which is characterised by prominent deep cerebral veins, focal atrophy, ipsilateral choroid plexus enlargement and enhancement at the sites of calcification. On further investigation coeliac disease was confirmed by a raised serum anti-transglutaminase and anti-endomysial antibodies, and villous atrophy and intra-epithelial lymphocytes on duodenal biopsy. A gluten-free diet was commenced and there were no subsequent similar symptoms. A diagnosis of incomplete CEC syndrome was reached on the basis of radiological and clinical findings

Discussion: CEC syndrome is a rare condition characterized by coeliac disease (C), epilepsy (E) and cerebral calcification (C) that can present in any age group.

C: Coeliac disease is an inflammatory disease of the upper small intestine resulting from gluten ingestion in genetically susceptible individuals. Classic signs are related to the gastrointestinal tract. Extra intestinal manifestations include dermatitis herpetiformis, anaemia, infertility, metabolic bone diseases, coagulopathy, psychiatric syndromes and neurological disorders¹. Neurological complications are estimated to occur in six to ten percent of patients with coeliac disease².

E: In contrast to incomplete CEC syndrome, a diagnosis of complete CEC syndrome includes a history of seizures. Seizures associated with CEC syndrome are usually occipital in origin and of focal or complex partial type. They may present as paroxysmal visual manifestations such as blurred vision, loss of focus, visualised coloured dots, and brief stereotyped complex visual hallucinations³. The seizures associated with this type of calcification can be difficult to treat. However, in some of the cases in the literature, seizure control was improved after institution of a gluten-free diet with folic acid supplements⁴, which also seems to have occurred in our case as evidenced by no recurrence of symptoms for a year.

C: Cerebral calcification in CEC syndrome is classically

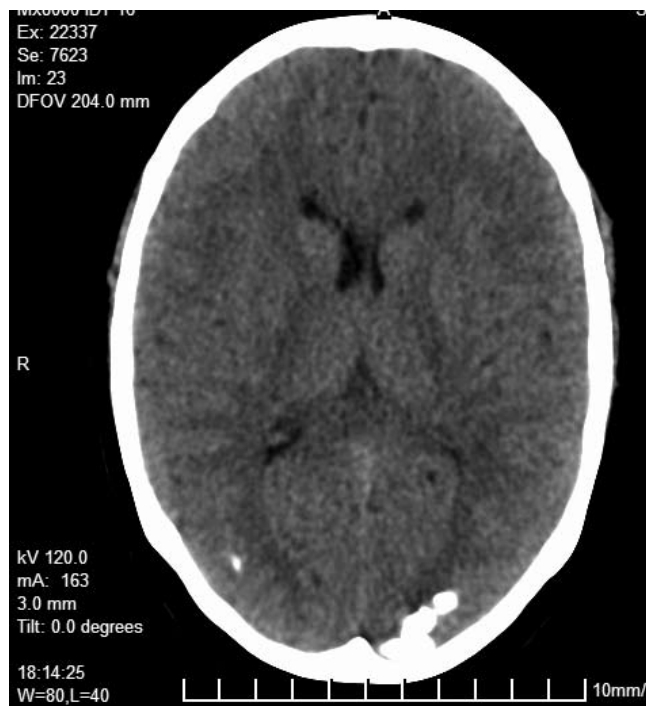


Fig. CT Scan showing bilateral calcification in occipital region in a patient with coeliac disease.

represented radiologically by bilateral cortico-subcortical occipital calcifications without contrast enhancement and brain atrophy. Reduced central nervous system folate levels secondary to folate malabsorption is a suggested mechanism of calcification in CEC syndrome⁵ as folate deficiency is seen in all major causes of cerebral calcification: Sturge-Weber syndrome, intrathecal methotrexate administration, central nervous system irradiation in childhood leukaemia, congenital folate malabsorption, coeliac disease and some atypical forms of other phakomatoses⁵.

Conclusions: Although CEC syndrome is very rare, it should be considered in individuals with coeliac disease presenting with neurological symptoms.

The authors have no conflict of interest.

*Arun P Sunnikutty, Senior House Officer, General Medicine

Janet Harding, Consultant Physician, with an Interest in Diabetology and Endocrinology

James C Nelson, Foundation Year 2, General Medicine

abiarun@hotmail.com

REFERENCES:

1. Wills AJ, Unsworth DJ. The neurology of gluten sensitivity: separating the wheat from the chaff. *Curr Opin Neurol* 2002;**15**(5):519-23.
2. Lagerqvist C, Ivarsson A, Juto P, Persson LA, Hernell O. Screening for adult coeliac disease - which serological marker(s) to use? *J Intern Med* 2001;**250**(3):241-8.
3. Taylor I, Scheffer IE, Berkovic SF. Occipital epilepsies: identification of specific and newly recognized syndromes. *Brain* 2003;**126**(Pt 4):753-69.
4. McIntosh S, Fischer D, Rothman SG, Rosenfield N, Lobel JS, O'Brien RT. Intracranial calcifications in childhood leukemia. An association with systemic chemotherapy. *J Pediatr* 1977;**91**(6):909-13.
5. Gobbi G, Bouquet F, Greco L, Lambertini A, Tassinari CA, Ventura

A, *et al.* Coeliac disease, epilepsy and cerebral calcifications. The Italian Working Group on Coeliac Disease and Epilepsy. *Lancet* 1992;**340**(8817):439–43.

HUMAN RIGHTS AND PRESUMED CONSENT FOR ORGAN DONATION IN THE UK

Editor,

Following the recent commentary on the importance of body donation for anatomical examination and teaching in Northern Ireland¹, the issue of organ donation has also received a fresh impetus. Organ donation is a fundamental concept in medical treatment. A recent survey on organ donation has indicated that approximately 90% of the UK population is in favour of organ donation. However, out of those, only 24% has signed the Organ Donation Register.

Over the past year, opinion in the UK among the public, politicians and the media, has shifted towards presumed consent whereby making donation the default position, from which everybody would retain the right to opt out during their lifetime. Recently, the Prime Minister Gordon Brown has pledged his support for such a system. A public opinion poll taken in October 2007 showed that 64% of respondents were in favour of a soft system of presumed consent, compared with 59% in 2004 (UK Transplant Records 2006-2007)².

The concept of presumed consent in organ donation is not new and has been the subject of a considerable debate among medical ethicists in the 1990s. There are basically three major schools of thoughts differing in their response to the idea of presumed consent

1. Is presumed consent the answer to organ shortages? Yes.

A leading view in this group is that of Veronica English (2007)³ a deputy head of medical ethics, British Medical Association, London. English argues that assuming people want to donate unless there is evidence to the contrary evidence will increase availability of donated organs. According to English, the new system would work when a person is identified as a potential donor, doctors must check the opt-out register. If the person has not opted out, the relatives are informed of this and, as an added safeguard, are asked if they are aware if the person has any unregistered objection. If the answer is no, the relatives are informed of the intention to proceed with donation.

2. Is presumed consent the answer to organ shortages? No.

A leading view in this group was initiated by the bioethicist Linda Wright (2007)⁴ at the University of Toronto. Wright's argument is based on the fact that presumed consent is hard to evaluate as it is implemented in different ways in different contexts, with different results. Wright compared two countries and found that the rate of donation in France in 2005 was 22.2 donors per million population while in Spain it was 35.1 per million. Both countries operate presumed consent and routinely ask families for their consent to donation, yet their organ donation rates vary greatly.

3. Is there any human rights influence on either of these arguments? Yes and No.

Under the ECHR, Article 8-the right to respect private and family life- would be violated where a person's organs

could be removed, after death, without consent having been obtained during their lifetime. On the human rights side of the argument (although, none of the Articles in either ECHR or Human Rights Act 1998 contain any provision to health care) organ donation with informed consent does satisfy Article 8. However, presumed consent *per se* could violate the right to respect the private life and diminish the support for organ donation.

The author has no conflict of interest.

*Jalal M Cartwright-Shamoon, *Researcher in Medical Law*

School of Law, University of Ulster, Jordanstown campus, Shore Road, Newtownabbey, Co. Antrim BT37 0QB. United Kingdom.

cartwright_shamoon-j@ulster.ac.uk

REFERENCES:

1. Taylor SJ, Wilson DJ. The Human Tissue Act (2004), anatomical examination and the importance of body donation in Northern Ireland. *Ulster Med J* 2007;**76**(3):124-6.
2. NHS Blood and Transplant. UK Transplant. Potential donor audit. Summary report for the 24 month period 1 April 2005 - 31 March 2007. Available from: http://www.uktransplant.org.uk/ukt/statistics/potential_donor_audit/pdf/pda_summary_report_2005-2007.pdf [Last accessed May 2008].
3. English V. Is presumed consent the answer to organ shortages? Yes. *BMJ* 2007;**334**(7603):1088.
4. Wright L. Is presumed consent the answer to organ shortages? No. *BMJ* 2007;**334**(7603):1089.

MILIARY TUBERCULOSIS CAUSING MULTIPLE INTESTINAL PERFORATIONS IN AN IMMIGRANT WORKER

Editor,

The incidence of tuberculosis (TB) in Northern Ireland is increasing¹. We present an uncommon case of perforated intestinal TB in an immigrant patient. The clinical presentation and endoscopic findings suggested inflammatory bowel disease (IBD). Subsequent multiple perforations necessitated emergency intestinal resection. With an increasing immigrant population, intestinal TB should be considered in such patients presenting with intestinal symptoms and signs.

Case Report: A 46-year old Polish immigrant presented with weight loss, abdominal pain and bloody diarrhoea. He appeared cachectic and had right iliac fossa tenderness. Colonoscopy revealed segmental ulceration with caecal involvement (Fig. 1). Given the distribution, Crohn's disease was suspected. However colonoscopic biopsies demonstrated caseating granulomatous inflammation and acid-fast bacilli.

Further examination revealed cervical lymphadenopathy and bilateral chest crepitations. Chest radiography showed bilateral infiltrates (Fig. 2). Identification of acid-fast bacilli in sputum and isolation of mycobacterium tuberculosis confirmed pulmonary TB.

After commencing anti-tuberculous treatment, the patient developed an acute abdomen. CT scanning demonstrated a pelvic collection, with free intra-peritoneal fluid.

Emergency laparotomy revealed generalised peritonitis due

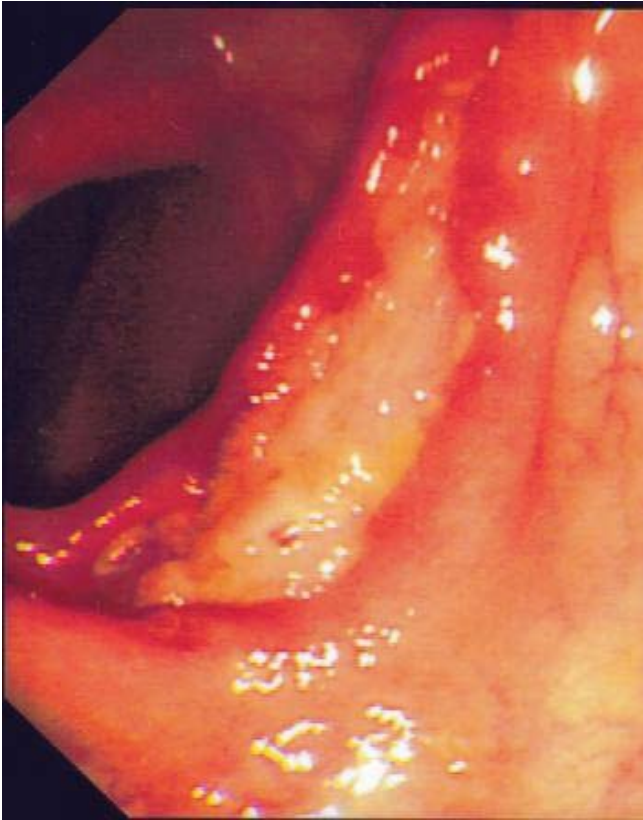


Fig 1. Caecal tuberculous ulcer.

to multiple ileal perforations. Apart from a short segment of proximal jejunum the entire small bowel and caecum were grossly diseased. An extensive enterectomy and caecal resection was performed, with a high jejunostomy and mucous fistula fashioned.

Histopathology revealed marked small bowel and caecal ulceration. Extensive caseating granulomatous inflammation (Fig. 3) and acid-fast bacilli confirmed intestinal TB.

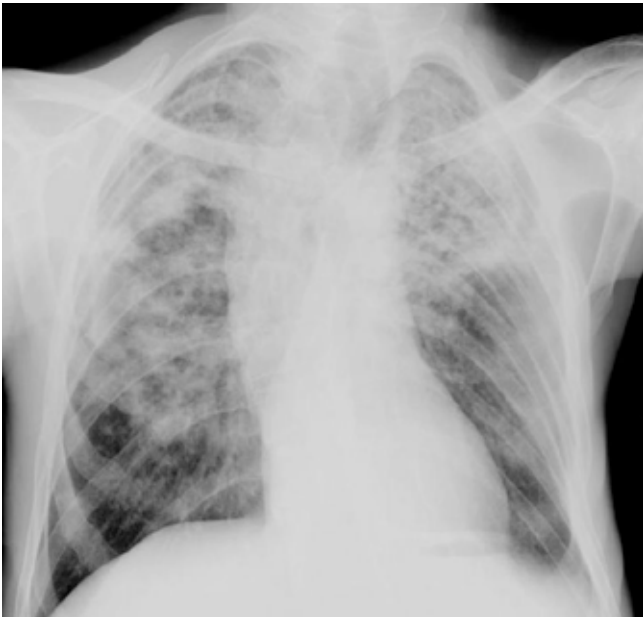


Fig 2. Bilateral upper and mid-zone infiltrates consistent with active pulmonary tuberculosis.

Discussion: In recent years the incidence of intestinal TB in developed countries has increased. In the UK higher rates have been identified in non-UK born individuals. This reflects increasing levels of disease in areas from which migrants are coming to the UK and increasing numbers arriving from high incidence areas¹.

In Northern Ireland the incidence of TB is rising, with a notification rate of 4.7/100,000 in 2004 (compared with 24.6/100,000 in Poland). Furthermore, the proportion of non-UK born cases of TB in Northern Ireland rose to 37% of those reported in 2006².

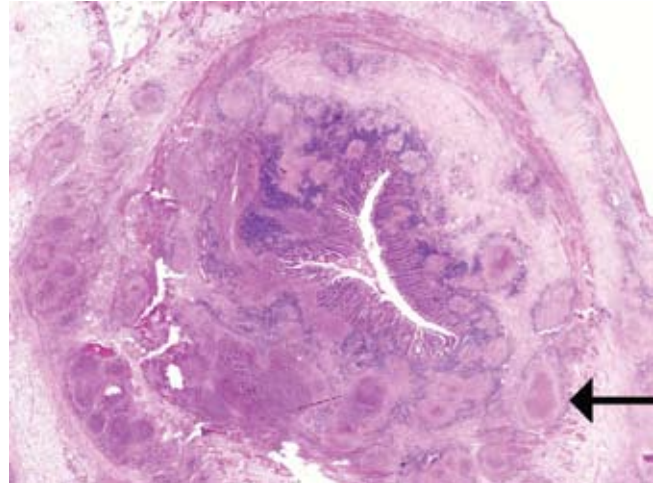


Fig 3. Photomicrograph of appendix demonstrating multiple trans-mural caseating (arrow) and non-caseating granulomata. (Haematoxylin and eosin, low power x1).

Intestinal TB presents a diagnostic challenge. Patients can present with abdominal pain, diarrhoea and weight loss, mimicking IBD³. The ileo-caecal region is the most frequent site of intestinal TB (similar to classical Crohn's). Colonic mucosal ulceration is often segmental and may be indistinguishable from Crohn's disease endoscopically. Colonoscopy is valuable in aiding the histopathological diagnosis of ileo-caecal disease⁴. The presence of caseating granulomata differentiates intestinal TB⁵. Identification of acid-fast bacilli together with isolation of mycobacteria confirms the diagnosis.

Intestinal TB is primarily managed with anti-tuberculous agents. Surgical intervention is reserved for complications including perforation which is an uncommon but serious complication with high mortality rates. Perforations may be solitary or multiple and surgical resection is required.

With an increasing incidence of TB and a rising immigrant population this case demonstrates the importance of considering intestinal TB in patients, particularly non-UK born, who present with symptoms suggestive of IBD. The role of endoscopic biopsy in differentiating intestinal TB from Crohn's disease is highlighted. This case also underlines the importance of recognition of perforated intestinal TB and the role of timely surgery.

The authors have no conflict of interest.

*Kevin McElvanna, *SHO General Surgery*

Roderick T Skelly, *SpR General Surgery*

Ciaran O'Neill, *SpR Histopathology*

Gary M Spence, *Consultant General Surgeon*

Department of General Surgery, Ulster Hospital, Dundonald, United Kingdom.

kevinmcelvanna@doctors.org.uk

REFERENCES

1. Tuberculosis Section. Health Protection Agency Centre. Focus on Tuberculosis. Annual surveillance report 2006 – England, Wales and Northern Ireland. London: Health Protection Agency Centre for Infections. November 2006. Available from: http://www.hpa.org.uk/web/HPAwebFile/HPAweb_C/1204100456946 [Last accessed May 2008].
2. Kennedy HE. Surveillance of tuberculosis in Northern Ireland 2004. Northern Ireland Communicable Disease Surveillance Centre; 2006. Available from: <http://www.cdscni.org.uk/publications/AnnualReports/pdf/TBReport2004.pdf>
3. Sibartie V, Kirwan WO, O'Mahony S, Stack W, Shanahan F. Intestinal tuberculosis mimicking Crohn's disease: lessons relearned in a new era. *Eur J Gastroenterol Hepatol* 2007;**19**(4):347-9.
4. Pulimood AB, Peter S, Ramakrishna B, Chacko A, Jeyamani R, Jeyaseelan L, *et al*. Segmental colonoscopic biopsies in the differentiation of ileocolic tuberculosis from Crohn's disease. *J Gastroenterol Hepatol* 2005;**20**(5):688-96.
5. Kirsch R, Pentecost M, Hall P de M, Epstein DP, Watermeyer G, Friederich PW. Role of colonoscopic biopsy in distinguishing between Crohn's disease and intestinal tuberculosis. *J Clin Pathol* 2006;**59**(8):840-4.

Letters to the Editor are welcomed on new scientific advances and should be no more than 500 words and contain up to 5 references and one figure and / or table.

Correspondence on articles on this issue of the journal can be sent by email as an attached word file, or by post on CD to the editorial office, and should be less than 300 words and a maximum of three references and one table or figure.