Ehlers–Danlos syndrome type IV in a child admitted in emergency with peritonitis

Editor—The diagnosis of Ehlers–Danlos syndrome (EDS) of type IV is rare in children. A 7-yr-old child was admitted to our hospital with septic shock and peritonitis. Emergency laparotomy found spontaneous perforation of the colorectal junction with bleeding and serious disruption on manipulation. Several biopsies were taken. The child’s progress was marked by a thrombosis in the inferior vena cava inferior on a central venous catheter and reappearance of peritonitis 10 days later. Laparotomy revealed large amounts of intraperitoneal blood with multiple haematomas along the colon and diffuse bleeding. Haemorrhage was treated with plasma and platelets. EDS type IV was suspected and genetic assessment was sought. The diagnosis was confirmed by colonic biopsies, molecular biology which showed a heterozygous mutation of the COL3A1 gene, and fibroblastic culture of the skin. The family investigation was negative. One year later, the child has had a total colectomy with ileorectal anastomosis.

The incidence of EDS is 1:5000 births and type IV represents 3–5% of the cases. It is a genetic abnormality affecting the collagen metabolism, of autosomal dominant trait, although a significant percentage of mutations are sporadic as in this case. Only 14 cases of spontaneous colonic perforation have been reported in the literature, but only two in children <10 yr of age. Skin and joint manifestations are less prominent than in other types of EDS, and type IV presents with complications such as arterial, visceral, or colonic rupture, as in our patient.

In retrospect, this child had several major criteria of EDS: history of spontaneous colonic perforation, easy bruising, characteristic face, keloid on the chin and forehead, and translucent dry skin with venous prominence at thoracic area, thin fingers, and joint hyper mobility. The coagulation disorders associated with the type IV EDS are related to platelet abnormalities, or deficiency in factors VIII, XI, or XIII but tests of haemostasis are often normal. The major abnormality of coagulation in EDS type IV is decreased vascular wall reactivity which prevents slowing of the microcirculation and the accumulation of platelets needed for the formation of thrombus and haemostasis. There are no reports of thrombosis associated with EDS in the literature, but those observed in our patient may be related to the presence of the central venous catheter.

The median survival with EDS is around 48 yr because of vascular or gastrointestinal complications. Perioperative management should avoid triggers, such as raised arterial pressure, that can lead to vascular or gastrointestinal perforations. The long-term administration of β-adrenergic blockers has been proposed. The use of non-steroidal anti-inflammatory agents may increase the risk of bleeding. Any invasive surgical procedures must be performed with great care. The surgeons chose a total colectomy because recurrent perforation is frequent in patients treated with resection and diversion.

The association of ‘spontaneous’ colonic perforation, haemostatic disorders with tendency to easy bruise should raise a suspicion of a vascular type EDS. Early diagnosis will facilitate and improve the perioperative management.
Bispectral index may not reflect the depth of anaesthesia in a patient with glycogen storage disease type I

Editor—A 17-yr-old boy with glycogen storage disease (GSD) type I was undergoing resection of multiple hepatic adenomas under general anaesthesia. Before operation, the blood glucose was kept at 4–5 mmol litre$^{-1}$. After the induction of anaesthesia and tracheal intubation, continuous inhalation of sevoflurane and i.v. infusion of remifentanil were started. Rocuronium 0.2 mg kg$^{-1}$ was given i.v. every 30–40 min. Arterial and central venous cannulas were placed. Bispectral index (BIS) was used to measure the anaesthesia depth. Blood glucose level and arterial gas analysis were done every 30 min. Dextrose 10% and sodium bicarbonate 5% were given intraoperatively guided by the blood glucose level and the acid-base status, respectively. After induction, the inhaled concentration of sevoflurane and infusion speed of remifentanil were adjusted to keep the BIS score between 40 and 60. During the first 30 min, the heart rate and arterial pressure were normal. After skin incision, the heart rate and arterial pressure increased gradually, whereas the BIS score remained between 40 and 60. Blood glucose level remained below 5 mmol litre$^{-1}$ with a lowest reading of 3.5 mmol litre$^{-1}$. The rate of infusion of dextrose 10% was increased and the BIS score increased to 70 with the normalization of blood glucose level. To increase the anaesthesia depth, we increased the inhaled concentration of sevoflurane and the infusion rate of remifentanil. The heart rate and arterial pressure returned to normal and the BIS score decreased to 45. After operation, the patient was discharged home on day 10.

GSD type I is caused by deficiency of glucose-6-phosphatase activity which results in an inability to form glucose during periods of fasting which could cause intraoperative hypoglycaemia and lactic acidosis. The brain is dependent upon a continuous supply of glucose. Thus, keeping the blood glucose level normal is important for the brain function. Recording EEG during insulin-induced hypoglycaemia in 14 young adults, Glass and colleagues reported a decrease in alpha-activity and an increase in delta- and theta-activities, which would be similar to the findings in patients given general anaesthesia. BIS is derived from pooled data of EEG changes in patients given different anaesthetics. In our case, the patient’s BIS score fluctuated between 40 and 60 after skin incision. It appeared that the depth of anaesthesia was sufficient, but the heart rate and arterial pressure increased gradually. At the same time, hypoglycaemia developed. When the blood glucose level was returned to normal, BIS score increased followed by the changes in heart rate and arterial pressure. In view of the known EEG changes associated with hypoglycaemia state as described above, the decrease in BIS during anaesthesia may have been due to hypoglycaemia and not necessarily to deepening of anaesthesia. It is important to remember that BIS score might not reflect the depth of anaesthesia in patients with GSD type I who are prone to develop hypoglycaemia.

X. Yu
Y. Huang*
J. Du
Beijing, China

*E-mail: punchhyyg@yahoo.com.cn


doi:10.1093/bja/aep250