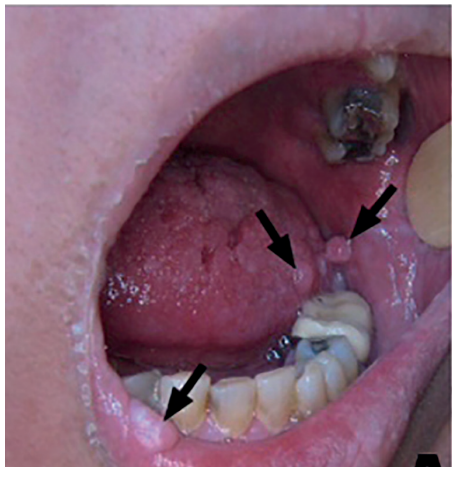
Cowden syndrome

Introduction

A 43-year-old woman with epigastric pain and mouth nodules on physical exam underwent endoscopy which revealed nodularity of the oesophageal mucosa over its entire length, bulb and the second part of duodenum. A diagnosis of Cowden syndrome was considered likely and a molecular study showed that she had a phosphate and tensin homologue (PTEN) gene mutation.

Figure 1. Nodules in the mouth.



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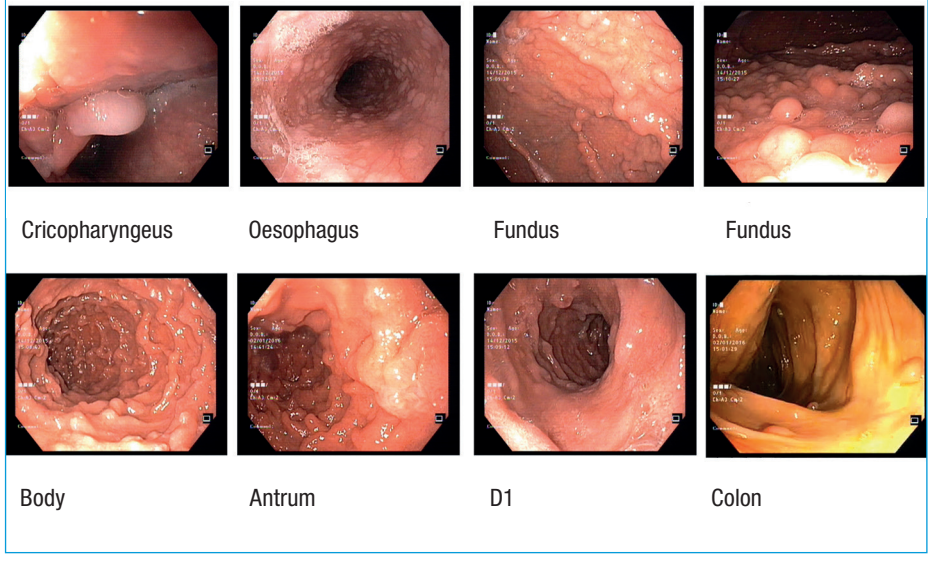
Discussion

Cowden syndrome is an autosomal dominant hamartoma/neoplasia syndrome which affects all three germ cell layers but most commonly arises from the ectodermal and endodermal elements (Blumenthal and Dennis, 2008). Almost all patients (90–100%) have mucocutaneous lesions that include trichilemmomas, acral keratoses and oral papillomas (O'Hare et al, 1997). Breast lesions affect the majority of female patients and include fibroadenomas, fibrocystic disease and adenocarcinomas (25–50%).

Thyroid abnormalities, such as multinodular goitre and follicular adenoma, are found in one half to two thirds of patients with Cowden syndrome. Thyroid carcinoma occurs in 3–10% of patients. Macrocephaly, cerebellar gangliocytoma and genitourinary malformations are

Figure 2. Colonoscopy revealed multiple polyps.



CASE REPORT

A 43-year-old woman presented with a 1-year history of epigastric pain and constipation. Her mother had had breast cancer and the patient had a son with intellectual disabilities, who had a cerebellar tumour diagnosed as dysplastic gangliocytoma. On physical examination, there were a few nodules in the mouth which had been there for 2 months (*Figure 1*). There were a few acral keratoses and thyroid nodules. The nodules were biopsied and shown to be simple cysts.

The patient underwent endoscopy. This showed a single nodule on the epiglottis, nodularity of the oesophageal mucosa along the entire length, coarse nodularity in the stomach from the cardia and fundus to the antrum, and nodularity of the bulb and second part of the duodenum. Colonoscopy revealed multiple polyps of different sizes (*Figure 2*) (*Video 1* available on www.bjhm.co.uk).

Biopsy of the upper gastrointestinal endoscopy samples showed papilloma, and the colon polyps were reported as hamartomatous polyps. With a clinical suspicion of Cowden syndrome a molecular study was done which showed germline mutations in the tumour suppressor, phosphate and tensin homologue deleted on chromosome ten (phosphate and tensin homologue gene mutation; PTEN) which is the prototype of the PTEN hamartoma tumour syndrome.

also frequent components of Cowden syndrome. Patients with Cowden syndrome have a higher risk of developing endometrial carcinoma or renal cell carcinoma. Benign tumours can cause significant morbidity but 40% of patients experience a malignant primary tumour. In this regard, annual or biannual follow-up visits with multidisciplinary care are recommended. BJHM

Blumenthal GM, Dennis PA. PTEN hamartoma tumor syndromes. Eur J Hum Genet. 2008 Nov;16(11):1289–1300. https://doi.org/10.1038/ejhg.2008.162

O'Hare AM, Cooper PH, Parlette HL 3rd. Trichilemmomal carcinoma in a patient with Cowdens disease (multiple hamartoma syndrome). J Am Acad Dermatol. 1997 Jun;36(6):1021–1023. https://doi.org/10.1016/S0190-9622(97)80295-2

LEARNING POINTS

- Cowden syndrome is an autosomal dominant hamartoma.
- Almost all patients with Cowden syndrome have mucocutaneous lesions and are at increasing risk of cancer.
- Patients with Cowden syndrome should have annual or biannual follow up.

In-hospital newborn falls: should all neonates undergo neurological imaging?

In-hospital newborn falls, although not uncommon, are under-recognized and often considered to be trivial. Skull fractures and intracranial haemorrhages can occur in the absence of clinical signs (Ruddick et al, 2010).

Maternal risk factors associated with in-hospital newborn falls include delivery by caesarean section, general or spinal anaesthesia, severe obesity, pre-existing health conditions (e.g. diabetes, epilepsy), opiate analgesics, and low haemoglobin level (<105 g/litre) (Helsley et al, 2010).

A 1-day-old neonate, delivered by caesarean section, whose mother was on opiate analgesics, slipped from the mother's arms to the floor when the mother fell asleep in a chair on the postnatal ward. Clinical examination was normal. Computed tomography scan (*Figures 1a,b*) showed a small acute traumatic subarachnoid haemorrhage overlying the right occipital lobe and a subgaleal haematoma overlying the left parietal bone. There was no skull fracture.

The paediatric neurosurgical team recommended conservative management. Medical review the next day detected swelling over the left occipitoparietal area measuring 4x4 cm (*Figure 2*). Developmental progress and head circumference were normal at clinic review 6 weeks later.

The authors suggest that early neurological imaging be performed in all unwitnessed in-hospital newborn falls and in witnessed falls where the mechanism is considered to be serious, even in the absence of initial clinical signs. BJHM

Helsley L, McDonald JV, Stewart VT (2010) Addressing in-hospital falls of newborn infants. *Jt Comm J Qual Patient Saf* **36**(7): 327–AP3. https://doi.org/10.1016/S1553-7250(10)36049-1

Ruddick C, Platt MW, Lazaro C (2010) Head trauma outcomes of verifiable falls in newborn babies. *Arch Dis Child Fetal Neonatal Ed* **95**(2): F144–F145. https://doi.org/10.1136/adc.2008.143131

Figure 2. Swelling of the scalp (arrow) appeared 24 hours later at the suspected point of impact.

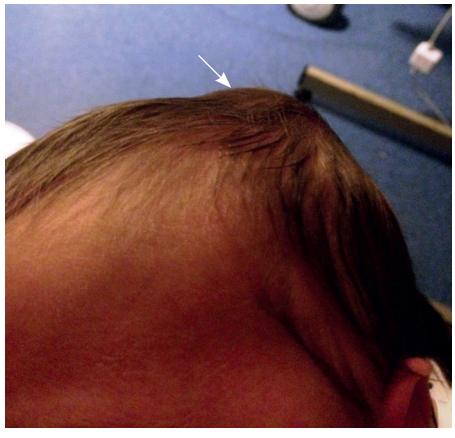
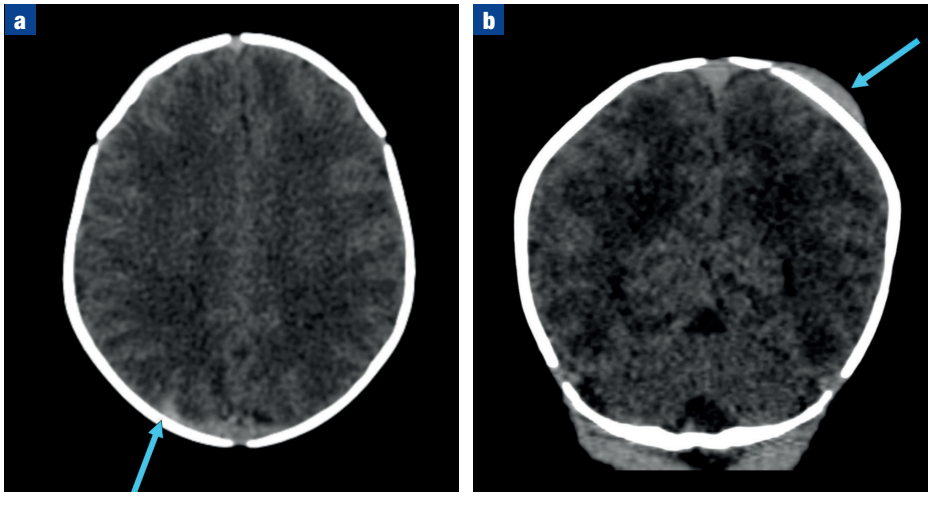


Figure 1. **a.** Axial image labelling the right occipital subarachnoid haemorrhage. **b.** Coronal view showing the left parietal haematoma.



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