Primary amenorrhoea due to a rare cause: epidermolysis bullosa causing haematometra

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SUMMARY

A 27-year-old woman with junctional epidermolysis bullosa (EB) presented with primary amenorrhoea and massive haematometra. Considering her severe physical handicap and anticipated problems in optimising menstrual hygiene, the patient and her parents opted for hysterectomy which was performed uneventfully. The patient was recovering well postoperatively until the eighth postoperative day when she developed acute dilation of stomach and unfortunately died on day 17.

BACKGROUND

Epidermolysis bullosa (EB), a mechanobullous disease characterised by increased skin fragility, is rarely encountered in gynaecological practice. When patients do present, they can be managed surgically if precautions are taken to avoid bullae formation, as evident by the uneventful immediate postoperative course of this patient. Unfortunately, the patient died due to an uncommon complication with high death rate. We feel that the case should be reported so as to help in future diagnosis of this extremely rare condition and to provide guidance on management.

CASE PRESENTATION

A 27-year-old woman with multiple healed scars all over body presented to emergency complaining of acute abdominal pain. She gave a history of primary amenorrhoea. The patient was non-ambulatory due to stunted growth and musculo-skeletal deformities that had been present since childhood and were progressively worsening. There was history of similar deformities in two other siblings who expired in infancy. She had three healthy living siblings. There was no history of consanguinity.

The patient was conscious and well oriented. Her body mass index was 17.36 kg/m². She had limited jaw opening with ill formed teeth and poor orodental hygiene. All digits were scarred, with dysplastic nails and pseudosyndactyly (figures 1 and 2). Secondary sexual characters were juvenile with sparse pubic and axillary hair. Breast development was Tanner stage II. Abdominal examination revealed a smooth, well-defined, cystic, non-tender mass arising from the pelvis corresponding to a 28-week gravid uterus (figures 3). There was no hepatosplenomegaly or ascites. Erosions and bullae were present on intercrural folds. Clitoris was normal though labia majora and minora were underdeveloped. Vaginal examination revealed a 3–4 mm deep blind pouch. On rectal examination, lower limit of the abdominal mass was palpable two inches above anal verge.

INVESTIGATIONS

The patient's haemoglobin on admission was 3 gm% with peripheral smear suggestive of severe dimorphic anaemia. The platelet count and coagulation profile were within normal limits. Cervical spine x-ray revealed fused C2 C3 vertebrae. Ultrasound of the abdomen showed grossly distended uterus and bilateral hydronephrosis. MRI reported vaginal atresia/agenesis with haematometra with bilateral hydronephrosis. Skin biopsy (figure 4) confirmed the diagnosis of junctional EB on both histopathology and electron microscopy showing subepidermal acellular bullae which are characteristic of junctional EB.

DIFFERENTIAL DIAGNOSIS

Thus, the diagnosis of junctional EB leading to vaginal agglutination and subsequent development of haematometra was made.

TREATMENT

Once the diagnosis was established, haematometra drainage was planned. The decision regarding a vaginal versus abdominal approach could not be resolved even after extensive literature search, there being only one case report of acquired haematometra in a patient with EB.¹ Blood transfusion was given to bring up the haemoglobin. Despite extensive counselling, the patient and her parents requested for hysterectomy considering the severe physical handicap and anticipated problems in optimising menstrual hygiene. After a detailed discussion about the advantages and disadvantages of surgery, an informed consent for hysterectomy was taken from the patient and her parents.

Examination under anaesthesia was conducted which confirmed the previous findings. Laparotomy revealed an enlarged uterus and 2.5 litres of old collected blood was drained (figures 5 and 6). Bilateral tubes and ovaries were normal. Hysterectomy was performed. The anatomic distinction between the cervix, vagina and uterus was lost due to gross distension. As the vaginal edge was not identifiable, vaginal dissection was also planned in order to ensure external communication to avoid the possibility of subsequent mucocolpos formation. On dissection from below, about 5 ml of pus was drained which confirmed the acquired nature of haematometra. Both Gram staining and culture sensitivity of pus were negative.

During the surgery, special precautions were taken to prevent bullae formation including gentle handling of the patient, padding of the pressure points, use of steroid cream before applying ECG electrodes and use of gauze/bandage in place of adhesive tapes. The surgery was performed uneventfully.

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Figure 1 Distal extremity contractures with pseudosyndactyly.

OUTCOME AND FOLLOW-UP

The patient was given antibiotics and anti-inflammatory drugs as well as epidural analgesia for 2 days postoperatively. The first postoperative week was uneventful wherein the patient was afebrile, was allowed orally on the third day and passed stools.

On eighth postoperative day, she developed acute gross abdominal distension and vomiting. An abdominal x-ray showed diffuse gastric distension with air fluid levels. Diagnosis of acute dilation of stomach was made. The patient was shifted to the intensive care unit and managed conservatively. However, she developed aspiration pneumonitis with high-grade fever on postoperative day 10, which did not respond to intravenous antibiotics and supportive therapy. The patient expired on postoperative day 17.

DISCUSSION

Epidermolysis Bullosa is a mechanobullous disease characterised by increased skin fragility and blistering of skin and mucosae following even mild mechanical trauma or increase in temperature. Its prevalence as per the National EB Registry Project of the USA is 8.22 per 10 000 000 population.²

Based on pathological findings and type of inheritance, it is divided into three types: EB simplex, junctional EB and EB dystrophica. Junctional EB has an autosomal recessive inheritance and is further subdivided into Herlitz and non-Herlitz types. The former is characterised by blistering and erosions at or soon after birth and results in death in infancy. The non-Herlitz type

Figure 2 Scarred upper limb digits with dysplastic nails.

Figure 3 Skin biopsy showing separation at level of lamina lucida.

follows a more indolent course and is characterised by severe teeth and nail involvement apart from generalised skin fragility and blistering. Lesions typically heal with atrophic scarring. These patients usually survive into adulthood. Our patient had the non-Herlitz type of junctional EB.

The diagnosis of EB requires thorough history, examination and an awareness of the disease. Skin biopsy and electron microscopy confirm the diagnosis. In adults, the timing of onset of blistering allows inherited EB to be differentiated from acquired immunobullous disorders.

There is no specific treatment available for EB. Management is essentially conservative and includes avoidance of provoking factors for blistering. The major challenge in patients who reach adulthood is management of chronic disability.

Genital involvement in EB is rare though perianal and vulvar scarring has been reported.^{3 4} To the best of our knowledge, this is the first reported case of junctional EB presenting with

Figure 4 Abdominopelvic mass corresponding to the size of 28 week gravid uterus.

Figure 5 Intraoperative picture showing distended uterus.

gynaecological involvement. Only one previous case of vaginal agglutination following tampon use leading to haematometra and secondary amenorrhoea has been reported in a patient of dystrophic EB.¹

This patient posed a challenge both in establishing a diagnosis as well as in deciding management. The standard treatment for primary amenorrhoea and haematometra is drainage and correction of the defect causing obstruction to the menstrual flow which may vary from a simple cruciate incision for intact hymen to complex vaginal reconstructive surgery. This patient

Figure 6 Old collected blood in the uterine cavity.

would have required surgical intervention for haematometra drainage though the extent of surgery might be debatable. Unfortunately, the outcome in this patient was unfavourable due to development of a rare postoperative complication which has a high mortality rate.⁵ However, we feel that this case should be reported in view of the rare association of EB with haematometra and especially due to paucity of literature to guide management of such a patient. These patients can be operated upon if precautions are taken to avoid bullae formation, as evident by the uneventful immediate postoperative course of this patient.

Learning points

- ▶ Although rare, epidermolysis bullosa may present with gynaecological complaints in young adult women. Thus, rare causes of primary amenorrhoea and haematometra should be considered.
- ▶ These patients require multidisciplinary care with gynaecologist, dermatologist, geneticist, physiotherapist, anaesthetist and medical social worker.
- ▶ There are no guidelines for management of this rare condition; hence management should be guided by whatever limited evidence is available and decisions be taken jointly by the patient, her guardians, caretakers and the physicians involved in patient care.

Competing interests None.

Patient consent Obtained.

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