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CASE REPORT

Surgical consideration in Hunter syndrome: a case of hydrocephalus and a case of epidural hematoma

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Abstract

Introduction Hunter syndrome (HS) is a rare X-linked lysosomal storage disorder which affects multiple organ systems. Surgical intervention and general anesthesia should be used with caution because of significant airway complications.

Case report Two HS patients underwent surgery with different prognosis are presented below. In the first case, symptoms of progressive disabilities on motor function, language, intelligence, and development last for 1 year in a 6-year-old boy; magnetic resonance imaging (MRI) showed severe hydrocephalus. Third ventriculostomy was performed in this patient to relieve the hydrocephalus. Unfortunately, this patient died postoperatively due to postsurgical tracheal collapse. In the second case, an 8-year-old girl was referred to our hospital with epidural hematoma because of a falling accident. Trephination surgery was performed under local anesthesia to remove the hematoma. Three days postsurgical, the patient was discharged uneventfully.

Conclusion General anesthesia in HS patients was associated with poor prognosis due to respiratory complications. Local anesthesia and less intensified treatment should be recommended.

Keywords Hunter syndrome · Surgery · Complications · Treatment

Introduction

Mucopolysaccharidoses (MPSs) are rare genetic disorders of glycosaminoglycan (GAG) metabolism [1]. Mucopolysaccharidosis type II (MPS II; Hunter syndrome) is an X-linked lysosomal storage disorder resulting from deficiency of lysosomal enzyme iduronate-2-sulfatase [2]. The main clinical features of Hunter syndrome (HS) patients are dysmorphic facial appearance, skeletal abnormalities, hepatosplenomegaly, cardiomyopathy, heart valve disease, respiratory problems, hearing impairment, and central nervous system involvement with mental retardation [3, 4]. Progressive airway obstruction is typical feature of respiratory complication and common cause of death [5]. Detailed information from two patients received neurosurgical operation are presented below. Surgical considerations are analyzed and discussed to achieve a better prognosis and to prevent the surgical complications in HS patients.

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Case reports

Case 1

A 6-year-old boy was admitted to our hospital due to progressive disabilities on motor function, language, intelligence, and development for the 1 year. Physical examination showed patient's features typical presentation of HS with coarse facial features, thick lips, macroglossia, protrusion of the abdomen, and short wide hands. Head MRI scan showed abnormal development of his brain accompanied with severe hydrocephalus and significant periventricular edema (Fig. 1a-b). Third ventriculostomy was performed through endoscopic procedure under general anesthesia. We administer 1 mg midazolam, 0.3 mg atrophine, and 40 mg methylprednisolone preoperatively. 0.06 mg fentanyl, 5 mg cisatracurium besilate, and 3 mg/kg/h propofol were used for induction of anesthesia. After intubation, 0.4 MAC sevoflurane, 3 mg/kg/h propofol,

and 0.1μg/kg/min remifentanil were used for maintenance of anesthesia. CPAP is used during surgery as patient's ventilation mode. Orotracheal tube was removed at the first postoperative day; however, patient developed respiratory difficulty and tracheostomy was performed later. Fiberbronchoscopy performed to adjust the tracheotomy tube revealed the collapsed trachea. Two days later, patient's blood saturated oxygen dropped suddenly. Patient deteriorated and eventually died because of respiratory dysfunction.

Case 2

An 8-year-old girl was transferred to our hospital due to a fall accident. Before admission, patient's family did not concern until she had nausea and vomiting, decrease appetite and lethargy. Physical examination showed typical appearances associated with HS (Fig. 1e). CT showed epidural hematoma on right parietal region with slight midline shift (Fig. 1c). Surgery was performed because of deterioration of the neurological function. To prevent respiratory complication, we performed trephination surgery under local anesthesia to remove the hematoma. Only part of hematoma was removed that can be reached through the burr hole (Fig. 1d); tube drainage was placed at the end of surgery. Tube drainage was removed and patient was discharge 3 days postsurgical with no complications.

Discussion

Hydrocephalus, myelopathy, and compressive neuropathy are the most common disorders of the CNS in patients with HS [6]. The etiology of hydrocephalus in MPS patient has not been defined yet, but the deposit of GAGs and the infiltration of PAS-positive leukocytes into the arachnoid villi [6–8] may cause poor CSF absorption and leading to communicating hydrocephalus. Another hypothesis suggests that leptomeningeal thickening hinders CSF absorption which lead to limitation on CSF outlets from the ventricle system causing obstructive hydrocephalus [9]. MRI is the best examination to determine the intracranial abnormality. It provide a great detail of the ventricle structure and their association with periventricular edema [10]. Ventriculoperitoneal shunt used to be the treatment modality, but recently, third ventriculostomy by endoscopic procedure is often performed in hydrocephalus case. Da Silva Neto et al. [10] reported a successfully treated hydrocephalus case in a patient with type VI MPS using endoscopic procedure. In our case, the patient had also undergone third ventriculostomy by endoscopic procedure. In contrast with Da Silva's report, respiratory complication eventually caused the death in this patient several days after operation.

Respiratory complication is the most common cause of death in HS patient [5]. It can be restrictive or obstructive and can involve any airway segment. GAGs that accumulate in the airway wall can lead to tracheal obstruction as well as intrinsic softening causing the airway collapse. Moreover, excessive secretions in the respiratory tract along with poor clearance are often in HS patient [11–13]. Enzymatic replacement therapy (ERT) is available since 2006 and has become the treatment modality for MPS [4, 14]. It has been reported to improve many somatic symptoms and signs of the disease [15–17]. However, no improvement was seen in respiratory function. What is interestingly is that patient in the literature reported by Da Silva Neto et al. has received ERT before while the patient in our first case did not received ERT. We suggested that the tracheal collapse might related with this ERT. Learnt from this experience, we suggest that operation should be delay in patient who did not have any ERT and

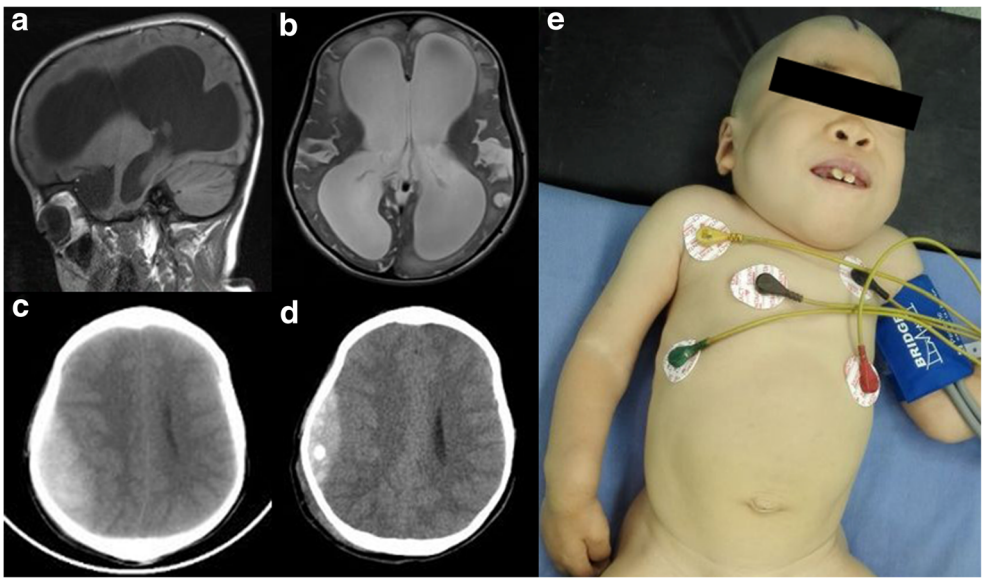

Figure 1 **a**, **b** Sagittal and axial MRI of the patient in the first case showed hydrocephalus with abnormal development of the brain, periventricular edema was significantly seen on T2WI. **c** Preoperative CT scan of the patient in the second case showed left side epidural hematoma. **d** Postoperative CT scan showed the hematoma was slightly reduced after surgery. **e** Features of HS patient in the second case showed short neck, depressed nasal bridge, thick lips, flexion contracture of fingers, and protuberant belly

operation can be performed after GAGs level decrease in a certain level.

Considering the tendency of tracheal collapse complication, we did not perform craniotomy to remove the epidural hematoma in the second patient. Under local anesthesia we performed trephination surgery. Although only small part of the hematoma was removed through the burr hole, the postoperative course was uneventful without leaving any neurological deficit. From this experience, we suggest that general anesthesia should be considered only when completely necessary and less aggressive procedure should be considered first in HS patients. However, in unconscious patient with severe brain herniation, craniotomy to remove the hematoma is still recommended. In this situation, a well-trained anesthesiologists who can use specific intubation/extubation techniques is highly recommended and a cardiothoracic surgeon is also recommended to perform any surgical intervention if tracheal collapse were occurred during or after operation [18–20].

Conclusion

General anesthesia is high risk and should be used with caution in MPS patient; it should be undertaken with great care as it is recommended in www.orpha.net and should be performed only when completely necessary. Local anesthesia and less intensified treatment were recommended to prevent postsurgical complications.

Compliance with ethical standards

Conflict of interest The authors declare that the study was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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