

Ventricular Septal Defect Closure in a Neonate with Osteogenesis Imperfecta

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A male patient weighing 2.5 kg was admitted for respiratory difficulty, and a large ventricular septal defect (VSD) was diagnosed. During care, sudden right leg swelling with a femur shaft fracture occurred. The patient's father had a history of recurrent lower extremity fractures; thus, osteogenesis imperfecta was considered. The patient's respiratory difficulty became aggravated, and VSD repair in the neonatal period was therefore performed with gentle sternal traction and great vessel manipulation under total intravenous anesthesia to prevent malignant hyperthermia. The patient was discharged without notable problems, except minor wound dehiscence. Outpatient genetic testing revealed that the patient had a *COL1A1/COL1A2* mutation.

Key words: 1. Osteogenesis imperfecta
2. Malignant hyperthermia
3. Cardiac surgery

Case report

A male patient weighing 2.5 kg was admitted for decreased activity and respiratory difficulty with bradycardia after birth. Mechanical ventilation was initiated, and a large ventricular septal defect (VSD), atrial septal defect (ASD), and patent ductus arteriosus were detected on echocardiography. On hospital day (HD) 3, during care in the intensive care unit (ICU), right leg swelling suddenly developed, and a right femur shaft fracture without any traumatic history was identified (Fig. 1). The patient's father had a history of recurrent lower extremity fractures; thus, osteogenesis imperfecta (OI) was considered. The patient did not show blue sclera and had no hearing loss.

His respiratory difficulty gradually increased after extubation on HD 3. We therefore recommended sur-

gical correction of his heart anomaly and a genetic mutation test. At first, his parents refused these recommendations due to economic problems. However, with the parents' consent, we decided on VSD closure because the patient's respiratory difficulty became aggravated. OI could not be ruled out, meaning that several factors had to be considered preoperatively in terms of an appropriate surgical technique. We chose total intravenous anesthesia (TIVA) to prevent malignant hyperthermia (MH) after the operation. Thus, propofol-, remifentanyl-, and rocuronium-based TIVA was applied under bispectral index monitoring. We performed very gentle intubation, and we opened the patient's sternum with electrocautery and widened it very slowly to prevent rib or costal cartilage fracture. We focused on extensive hemostasis during the operation, due to the

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Fig. 1. Right femur shaft fracture without any traumatic history.

patient's bleeding tendency. Intraoperatively, the sternum was relatively hard, but a bleeding tendency was noted when we grasped the aorta with forceps. However, the muscular power of the ventricle was also relatively good. The VSD and ASD were closed without problems, and there was no evidence of rib fracture after the operation on a chest X-ray.

After the operation, we carefully monitored the patient's body temperature during postoperative ICU care. Since no evidence of MH was detected, we performed extubation on postoperative day 2, and the patient had an uneventful postoperative recovery. In the outpatient department, a genetic analysis was conducted to determine whether a *COL1A1/COL1A2* mutation was present, and the analysis detected *COL1A1* c.1471G>A, p.Gly491Ser (heterozygous, autosomal dominant). Wound dehiscence without any infectious signs occurred after discharge. The patient was therefore readmitted and the wound was re-sutured.

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all patients, parents, or guardians included in the study.

Discussion

OI involves impaired type I collagen synthesis and

shows an autosomal dominant hereditary pattern. Thus, OI patients have a greater susceptibility to bone fracture, brittle teeth, blue sclera, and hearing loss [1]. Cardiovascular involvement is rare, and the severity of skeletal involvement does not predict the extent of cardiovascular involvement [2].

Cardiovascular surgery in OI patients has been associated with increased morbidity and mortality due to their tissue fragility, a bleeding tendency, and delayed wound healing [3,4]. In addition, MH, which is a life-threatening clinical syndrome of hypermetabolism involving the skeletal muscle after surgery, showed an association with OI during surgery [5]. MH induces heat production, leads to disseminated intravascular coagulation, and can cause multiple organ failure. MH occurs mainly during anesthesia or postoperative ICU management. Since it has been reported that propofol-based TIVA can decrease the incidence of MH by blocking the triggering factors, such as inhaled anesthetics or succinylcholine during anesthesia [6], we applied TIVA despite concerns regarding intraoperative awareness and a slow response to anesthetic depth in pediatric patients [7]. We took several surgical measures to prevent costal cartilage or rib fractures. We made a wide surgical incision and minimized the opening of the surgical field to the extent possible, to decrease the stress placed on the rib cage. Furthermore, we applied very gentle tissue handling and dissection around the aorta. However, the ventricular muscle strength was similar to that of other VSD patients during the operation. Delayed wound healing is another characteristic problem in patients with OI. In our patient, delayed wound dehiscence occurred without any signs of infection, and wound dehiscence was resolved, with no problems after re-closure.

Cardiac involvement in OI has been rarely reported. Furthermore, few cases have been reported of congenital heart disease in individuals with OI. A previous report described VSD closure in a 6-year-old with OI, which is youngest age for which such an operation has been reported [8]. This is the first report of congenital heart surgery in a neonate with OI, and it demonstrates that open heart surgery can be performed safely in neonates with OI.

Conflict of interest

No potential conflict of interest relevant to this ar-

ticle was reported.

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