Anesthesiology
66:86-88, 1987

Anesthesia for Children with Jeune's Syndrome (asphyxiating thoracic dystrophy)

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During the last 2 years, we anesthetized seven children (aged 6 weeks to 10 yr) with Jeune's syndrome, or asphyxiating thoracic dystrophy. Such patients may have multiple organ failure or dysfunction.[1,2] The syndrome occurs in neonates who often have severe thoracic defects, and older infants and children who have end-stage renal disease. Both neonates and older children are represented in our patient group. Neonates with this syndrome, who previously often died, are having corrective thoracoplasty. We describe our experience with several anesthetics in patients with Jeune's syndrome.

Jeune's syndrome is inherited as an autosomal recessive disorder. Forty-five per cent of cases are reported in siblings and 7% in offspring of consanguineous unions. The syndrome is thought to be genetically heterogeneous because it occurs in both a neonatal form, with life-threatening thoracic deformity leading to death, and a form that develops in later childhood, manifested as renal dysfunction. One hundred and six cases have been reported in the literature. Fifty per cent of cases are diagnosed within the first month of life; the remaining patients are reported by 6 months of age. Thirty-nine per cent of patients die on the first day of life, 75% die by 6 months of age, and another 10% die by 1 yr of age; thereafter, the mortality is 4% per yr.[3-5]

The major feature of Jeune's syndrome in the neonate is deformity of the thoracic wall that prevents normal intercostal respiratory movement and causes respiratory insufficiency.[6] The most seriously affected patients have underlying pulmonary hypoplasia. Persistent pulmonary hypertension of the neonate as a result of decreased cross-sectional area of the pulmonary arterial vasculature has also been reported.[7] Renal failure from diffuse interstitial fibrosis with lymphoplastic infiltration and tubular changes including alternating dilation and atrophy may appear in late infancy or early childhood.[8,9] Hepatic fibrosis has been reported at this stage and later in survivors, although there is no evidence of bile stasis or involvement of the liver parenchyma or central veins. Finally, myocardial dysfunction can occur in older patients. Whether this is a result of long-term alveolar hypoventilation and hypoxemia or of intrinsic myocardial injury is not known.

We have cared for seven patients with this syndrome. Three patients with neonatal Jeune's syndrome underwent thoracoplasty and also required anesthesia for direct laryngoscopy, bronchoscopy, and tracheostomy. One of these infants subsequently required bilateral rib resection. Two infants remain ventilator-dependent at 29 months and 8 months of age, respectively. The other was weaned from ventilatory support by 18 months of age but died of *Haemophilus influenzae* meningitis and sepsis at 22 months of age. Four patients presented after infancy with end-stage renal disease. Operative procedures in these children included repeated insertion of dialysis catheters and renal transplantation. We describe the pathophysiology of this syndrome, and present three cases illustrating relevant issues in anesthetic management. Surgical procedures and the accompanying anesthetic technique for each of the seven patients are listed in table 1.

Report of Three Cases

Patient 1. This 10-week-old, 3.45-kg female infant was scheduled for a thoracoplasty to correct the thoracic defect of Jeune's syndrome (fig. 1). Arterial blood pressure was 127/60 mmHg and heart rate 160 beats/min. Preoperative laboratory findings included hemoglobin, 12.2 g/dl; hematocrit, 31.4%; *p*H, 7.33; Pa_{O_2}, 49 mmHg (fractional inspired O_2 concentration [$F_{I_{O_2}}$] = .21); and Pa_{CO_2}, 33 mmHg. Pulmonary status was assessed by measuring tidal volume with changes in thoracic and abdominal circumferences using inductance plethysmography (Respitrace®)[10],† and oxygenation with pulse oximetry (Nellcor®). At rest, rib cage and abdominal motion were synchronous, and oxyhemoglobin saturation was normal; when the patient was agitated, chest wall and abdominal motion became asynchronous and severe oxyhemoglobin desaturation occurred.

Anesthesia was induced and maintained with halothane, nitrous oxide, and oxygen *via* a preexisting tracheostomy supplemented by pancuronium 1 mg iv. Intraoperative blood loss was 20 ml. Lactated Ringer's solution, 70 ml, and D_{10}/0.2 NS, 100 ml were infused iv. The surgical procedure lasted 2.5 h. At 5 weeks postoperatively, forced vital capacity (FVC) and maximum expiratory flow at 25% of FVC ($\dot{V}_{max25}$) remained moderately decreased. (FVC had increased from 226 to 254 ml, while $\dot{V}_{max25}$ increased from 44 to 102 ml/s). This child, now 29 months of age, is at home and weighs 11.7 kg (10% for

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Key words: Anesthesia: pediatric, Jeune's syndrome.

† Motoyama EK: Deflation flow–volume curve: Pulmonary function testing in infants and children with artificial airways. Ped Pulmonol (In press).

TABLE 1. Surgical Procedures and Anesthetics in Seven Patients with Jeune's Syndrome

Patient No.	Age at Operation	Procedure	Anesthetic
1	7 weeks	Direct laryngoscopy, and bronchoscopy	Oxygen
	10 weeks	Thoracoplasty	Isoflurane, nitrous oxide, fentanyl, pancuronium
	3 months	Bronchoscopy, tracheostomy	Isoflurane, nitrous oxide, atracurium
	6 months	Removal of infected bone graft from chest	Isoflurane, nitrous oxide, gallamine
2	10 weeks	Thoracoplasty	Isoflurane, nitrous oxide, fentanyl, pancuronium
	11 weeks	Bronchoscopy, revise thoracoplasty	Isoflurane, nitrous oxide, fentanyl, pancuronium
	9 months	Revision of thoracoplasty	Halothane, nitrous oxide, pancuronium
	14 months	Right thoracoplasty	Isoflurane, nitrous oxide, fentanyl, pancuronium
3	4 yr	Renal transplant	Isoflurane, nitrous oxide, atracurium
4	10 yr	Renal transplant	Isoflurane, nitrous oxide, fentanyl, curare
5	8 months	Renal transplant	Isoflurane, nitrous oxide, atracurium
	8 months	Removal of transplanted kidney	Isoflurane, nitrous oxide, pancuronium
	8 months	Open-lung biopsy	Isoflurane, atracurium
	12 months	Bilateral myringotomies	Halothane, nitrous oxide
	13 months	Reposition of chronic peritoneal dialysis catheter, hernia repair	Isoflurane, nitrous oxide, atracurium
	28 months	Bilateral myringotomy	Halothane, nitrous oxide
	28 months	Renal transplant	Isoflurane, nitrous oxide, pancuronium
	32 months	Removal of transplanted kidney, placement of chronic peritoneal dialysis catheter	Halothane, nitrous oxide, atracurium
	35 months	Renal transplant	Halothane, nitrous oxide, atracurium
	36 months	Removal of transplanted kidney	Isoflurane, nitrous oxide, atracurium
6	4 yr	Placement of dialysis catheter	Halothane, nitrous oxide
	5 yr	Placement of dialysis catheter	Halothane, nitrous oxide, fentanyl
	5 yr	Placement of dialysis catheter	Halothane, nitrous oxide fentanyl
	5 yr	Renal transplant	Isoflurane, nitrous oxide, curare
	6 yr	Removal of dialysis catheter	Halothane, nitrous oxide
7	6 weeks	Thoracoplasty	Oxygen, pancuronium
	8 weeks	Bilateral rib resection	Fentanyl, pancuronium

age) but continues to require mechanical ventilation: intermittent mandatory ventilation (IMV) 10 breaths/min; spontaneous ventilation rate, 56 breaths/min; FI_{O_2}, 30%; peak inspiratory pressure (PIP), 28 mmHg; and continuous positive airway pressure (CPAP), 5 mmHg. *p*Ha was 7.40; Pa_{O_2}, 77 mmHg; Pa_{CO_2}, 39 mmHg; HCO_3, 25 mEq/l; base excess, +1 mEq/l; tc_{O_2}, 96 mmHg; and tc_{CO_2}, 41 mmHg. The hemoglobin was 13.1 g/dl and hematocrit 38.5%. FVC as measured with a deflation flow–volume curve technique[10,11] had increased to 79% of predicted for age. $\dot{V}_{max25}$ was markedly decreased, suggestive of lower airway obstructive disease. $\dot{V}_{max25}$/FVC improved after bronchodilator therapy.

Patient 2. This 10-week-old male infant was scheduled for thoracoplasty. His previous surgery included direct laryngoscopy, bronchoscopy, and tracheostomy. During mechanical ventilation (FI_{O_2}, 0.6; IMV, 38 breaths/min), pH_a was 7.33; Pa_{O_2}, 59 mmHg; Pa_{CO_2}, 47 mmHg; and PET_{CO_2}, 38 mmHg. Preoperative laboratory results included hemoglobin, 12 g/dl, and hematocrit, 38%. Pulmonary function was tested under general anesthesia immediately preoperatively during paralysis from pancuronium. FVC as measured by forced deflation was small (226 ml, 47% of the predicted 480 ml); $\dot{V}_{max25}$ and maximum expiratory flow at 25% of functional residual capacity ($\dot{V}_{max25}$/FRC) were also markedly reduced, indicating a moderate restrictive defect with lower airway obstruction. Anesthesia was induced with fentanyl and pancuronium iv and maintained with isoflurane and nitrous oxide. Surgery lasted 5 h, and the intraoperative course was uncomplicated. At 18 months of age the infant was weaned from the ventilator and had acceptable oxygenation and mild hypercarbia (Pa_{CO_2}, 48 mmHg) while breathing spontaneously *via* tracheostomy (FI_{O_2}, 0.35; CPAP, 2 mmHg). Three months later he died of H. influenza and sepsis.

Patient 3. This 15-kg, 4-yr-old female infant was scheduled for cadaveric kidney transplantation. Her medical history included mitral valve prolapse noted at birth. Although previously in congestive heart failure, the child was treated with digoxin 0.065 mg bid and was asymptomatic at the time of surgery. An episode of spontaneous bacterial peritonitis 4 days before surgery was treated with cephadine and cephalexin, and the child was afebrile immediately preoperatively. Roentgenogram of the chest showed the thoracic deformity of Jeune's syndrome, clear lung fields, normal cardiac size, and a small pleural effusion on the left.

Laboratory findings were: hemoglobin, 10.5 g/dl; hematocrit, 32.4%; serum sodium, 135 mg/dl; serum potassium, 3.7 mg/dl; blood urea nitrogen, 50 mg/dl; and creatinine, 5.7 mg/dl. The lungs were clear by auscultation, and the cardiac examination revealed no murmur or gallop. There was no intraabdominal organomegaly. Anesthesia was induced with thiopental 75 mg iv. Laryngospasm occurred, followed by cyanosis, hypotension (systolic arterial pressure, 50 mmHg), and bradycardia (heart rate, 64 beats/min). Positive-pressure ventilation *via* mask relieved the laryngospasm. Anesthesia was maintained with isoflurane, nitrous oxide, and paralysis from atracurium iv. Vital signs remained stable throughout the case. Early in the intraoperative period, bilateral wheezing was noted. A postsurgical intraoperative chest radiograph revealed a left-sided pneumothorax, which was treated by insertion of a chest tube. Neostigmine 1 mg and atropine 0.4 mg were given iv to antagonize the neuromuscular blockade, and the trachea

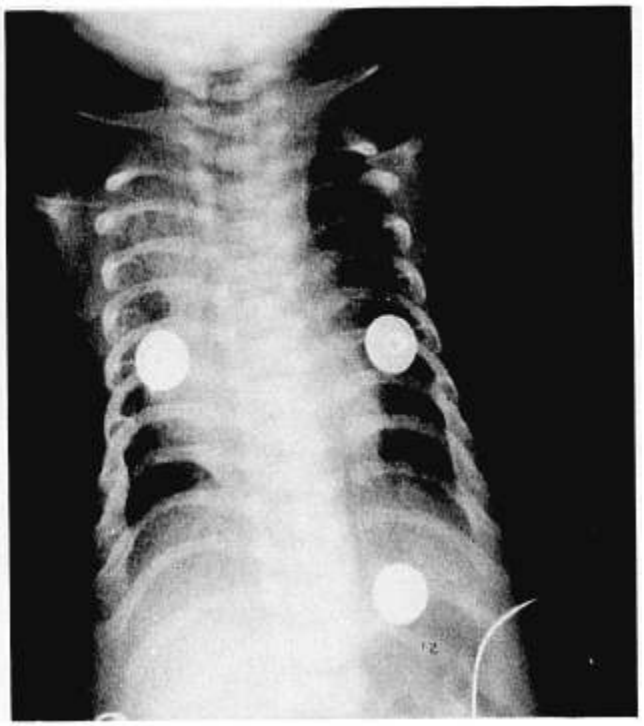

FIG. 1. Thoracic roentgenogram of patient no. 1. Abnormalities include high clavicles, narrowed thoracic cage with horizontal, short stubby ribs with a widened anterior portion, and right upper lobe atelectasis.

was extubated. She recovered without sequelae and was discharged home.

DISCUSSION

Our patients had the thoracic defects typical of Jeune's syndrome; the three neonates had more severe thoracic deformity. Both neonates and adolescents with this syndrome have decreased lung volumes. Pulmonary hypoplasia and persistent pulmonary hypertension are possible in the newborn. Even if normoxic at rest, these infants are prone to profound hemoglobin desaturation when agitated because of asynchronous rib and abdominal motion. During the intraoperative period, peak airway pressure should be maintained as low as possible to minimize both barotrauma and any adverse affect of increased airway pressure on the pulmonary arterial bed.

Infants who have thoracoplasty almost assuredly will need long-term ventilator support, because of an increased alveolar–arterial oxygen gradient and ineffective ventilation with persistent hypercarbia. Older children with Jeune's syndrome undergoing renal transplantation also have the typical thoracic deformity, although it is far less severe than in the neonate. In older patients pulmonary hypertension and cor pulmonale as a sequela of chronic hypoxemia may also occur. While the intraoperative pneumothorax that occurred in patient no. 3 most likely resulted from increased airway pressure due to coughing, laryngospasm, and our positive-pressure airway maneuver, decreased lung volumes may have contributed to this complication. In older patients without significant lung or cardiac disease the intraoperative anesthetic requirements for kidney transplantation are the same as for other patients undergoing this procedure.

In summary, the anesthetic management of a patient with Jeune's syndrome requires careful preoperative assessment of the pulmonary, cardiovascular, and renal function. The spectrum of the syndrome's pathophysiologic manifestations is rarely present simultaneously in an individual patient. Thus, the current physical examination, laboratory analysis, and surgical procedure should determine the anesthetic technique.

ADDENDUM

A deflation flow–volume curve is produced by manual inflation of the lung to total lung capacity followed by forced deflation by opening the endotracheal tube to a negative pressure reservoir. While the lungs are rapidly deflated, flow and volume signals are displayed on a X–Y storage oscilloscope. FVC and $\dot{V}_{max25}$ are obtained from the resultant maximum expiratory flow–volume curve. This is an approved clinical test method in our institution.

Pulmonary function tests were performed by Dr. E. Motoyama, Director of the Pulmonology Laboratory at the Children's Hospital of Pittsburgh, and Professor of Anesthesiology and Pediatrics, University of Pittsburgh School of Medicine. The author thanks Lisa Cohn for editorial assistance.

REFERENCES

1. Kaufman HJ, Kirkpatrick JA: Jeune's thoracic dysplasia: A spectrum of disorders? Birth Defects 10:101–117, 1974
2. Friedman JM, Kaplan HG, Hall JG: The Jeune syndrome (asphyxiating thoracic dystropy) in an adult. Am J Med 59:857–862, 1975
3. Tahernia AC, Stamps P: "Jeune syndrome" (asphyxiating thoracic dystrophy). Clin Pediatr 16:903–908, 1977
4. Schmidt R, Pajewshi M, Mundel G: Unusual features in familial asphyxiating thoracic dysphasia (Jeune's disease). Clin Genet 3:90–98, 1972
5. Canepa G, Saputo V, Gallina M, Manganini M, Monguzi W: La distrofia toracica di Jeune. Chir Organi Mov 64:11–49, 1978
6. Fruchter Z, Enachescu I: Asphyxiating thoracic dystrophy (Jeune syndrome). Radiological verification. Paediatrician 18:46–50, 1969
7. Borland LM: Persistent pulmonary hypertension of the newborn, Neonatal Anesthesia. Edited by Cook DR, Marcy JH. Pasadena, Appleton Davies (In press)
8. Shah KJ: Renal lesion in Jeune's syndrome. Br J Radiol 53:432–436, 1980
9. Bernstein J, Brough AJ, McAdams AJ: The renal lesion in syndromes in multiple congenital malformations. Birth Defects 10:35–43, 1974
10. Sackner JD, Nixon AJ, Davis B, Atkins N, Sachner MA: Noninvasive measurement of ventilation during exercise using a respiratory inductive plethysmograph. Am Rev Resp Dis 122: 867–871, 1980