Chapter 28
Bone and Connective Tissue Disorders

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An 11-year-old boy with achondroplasia presents for Ilizarov leg lengthening. His admission VS are HR 92 bpm, BP 110/80 mmHg, and RR 16/min. Hct 34. No prior history of surgery.

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Preoperative Evaluation

Questions

1. The patient has a large head. Is this normal? Why/why not? What might it signify? How could you evaluate the presence or absence of hydrocephalus? Is this important? Why/why not? How would it alter your anesthetic management if he had normal pressure hydrocephalus? What if this patient had progressive difficulty walking; what would this suggest? How would you evaluate it? How could this be distinguished from his short-limb difficulties?

2. What are the anesthetic implications of his bony abnormalities? Is this more likely to occur cranially or caudally? Is the spinal defect only a mesodermal defect, or does it involve neuroectoderm as well? The parents want to find out about a spinal; what would you tell them? Is this likely to be easy in this patient (aside from psychological issues)? What level would you choose to perform the block?

3. Does this patient need soft tissue films of his neck? Why/why not? An airway MRI? C spine films? Why/why not? How will it affect your anesthetic management?

Preoperative Evaluation

Answers

1. Yes, a large head is normal in achondroplasia patients. While macrocephaly is typical of the syndrome, there is a higher incidence of hydrocephalus due to foramen magnum stenosis. Various imaging modalities will show dilated ventricles. If found, foramen magnum stenosis predisposes to brainstem compression and respiratory arrest. The absence of hydrocephalus or normal pressure hydrocephalus does not rule out the presence of foramen magnum stenosis, and the risk of brainstem compression remains unchanged. This may be due to spinal cord compression from spinal canal stenosis. Imaging of the thoracic and lumbar spine would help with a more specific diagnosis. Difficulty walking owing to short limbs is not associated with neurological symptoms. However, spinal cord compression is associated with lower extremity changes in sensation and motor function. Bladder and rectal sphincter dysfunction may also occur with thoracolumbar stenosis.

2. Failure of bone growth in this syndrome may result in stenosis of the foramen magnum and spinal canal and vertebral malalignments, most likely in the lumbar spine. Embryologically, the problem is primarily a defect of mesoderm, i.e., the failure of the cartilage to form bone. Lumbar spinal anesthesia has been performed safely in patients with spinal stenosis and vertebral malalignment but is associated with a potentially higher incidence of direct neural injury by the block needle or as a result of epidural bleeding. Therefore, it is advisable to perform spinal anesthesia if it offers distinct advantages over general anesthesia. In this particular patient, there is no obvious benefit of spinal anesthesia over general anesthesia. It is likely to be difficult due to the lordotic deformity and vertebral malalignment. I would choose a site that is not involved with lordosis, kyphosis, stenosis, or vertebral malalignment after reviewing the imaging studies and documenting any preprocedural neurological deficit. L5–S1 may be safe because the deformities are usually located above this level. Nevertheless, the risk of cord compression is possible with the use of a large volume of local anesthetic and epidural venous bleeding.

3. This patient does not need soft tissue films of the neck. Achondroplasia is a disease of underdevelopment of bone. Midface hypoplasia is expected with this disorder. MRI of the airways can be helpful to determine the extent of airway involvement. As a disorder of bone underdevelopment, achondroplasia can be associated with dysgenesis of the odontoid process, resulting in atlanto-axial instability. The presence of atlanto-axial instability requires protection of the spinal cord from compression by stabilization of the spine in extension during tracheal intubation maneuvers and throughout the perioperative period. Rigid videolaryngoscopy, now readily available, will likely result in a better view of the larynx and glottis without significant manipulation of the head and neck. It is reasonable to anticipate a difficult intubation due to midface hypoplasia, micrognathia, or macrognathia and prepare the patient and the equipment accordingly.

Intraoperative Course

Questions

1. What kind of monitoring (aside from routine) would you choose for this case? Why/why not? Does this patient need an arterial line? Why/why not? A CVP? A precordial Doppler? Why/why not?

2. Your colleague walks by the room, takes a look in as you are placing monitors, and suggests that you consider a rapid sequence induction because the patient is a preteen, and they all have full stomachs. What do you think? How would you decide which approach to use? Would you use succinylcholine for a rapid sequence induction? Why/why not? Any association with malignant hyperthermia? The patient can extend his neck through an arc of 15–20°. Does this influence your choice of anesthetic induction techniques? How will you do an awake intubation? Would a light wand be just as good as a fiber-optic bronchoscope? Would an anterior commissure scope work just as well for this problem? What are the advantages and disadvantages of each in patients with odontoid hypoplasia and decreased neck extension?

Intraoperative Course

Answers

1. I would monitor the patient with standard noninvasive monitoring. I would not use an arterial line or a CVP catheter for this low-risk procedure. I would, however, use a precordial Doppler because of the chance of significant air or fat embolism with this procedure. I would not approach this as a rapid sequence induction. Achondroplasia is associated with a difficult tracheal intubation due to midface hypoplasia and micrognathia. Minimal manipulation of the cervical spine is advisable because of the potential of atlanto-axial joint instability. Rigid videolaryngoscopy or flexible fiber-optic laryngoscopy may be necessary to secure the airway with minimal cervical spine manipulation. Careful evaluation of the airway during the preoperative visit is crucial. In the operating room, airway access could be assessed after providing sedation in a spontaneously breathing patient.

2. An electively prepared patient should not ordinarily need a rapid sequence induction, and I would not use succinylcholine. Succinylcholine causes fasciculations that may produce atlanto-axial instability and spinal cord compression. Achondroplasia is also associated with hypotonia, and the use of succinylcholine may produce clinically significant hyperkalemia. Even if the patient demonstrated good range of motion of the neck, it would not necessarily be reassuring with regard to routine laryngoscopy and intubation.

   Intubation of trachea in a conscious child is performed after effective intravenous sedation, with incremental doses of a short-acting opioid such as fentanyl 0.5 mcg/kg IV repeated at 5 min intervals to effect supplemental IV non-opioid sedation with midazolam in 25 mcg/kg increments, and should achieve a state of anxiolysis, antegrade amnesia, and sedation while maintaining the patient's ability to respond to verbal commands. The airway mucosa of the nares, oropharynx, and supraglottic area can be anesthetized with topical local anesthetic and/or glossopharyngeal and superior laryngeal nerve blocks to markedly reduce the discomfort associated with instrumentation and reduce the requirement of intravenous sedation. Lidocaine 1 % and cocaine 4 % spray are effective topical anesthetics for the mucosa. The advantage of cocaine over lidocaine is that it produces vasoconstriction and minimizes bleeding with turbinate instrumentation. The oropharynx is anesthetized with an intraoral glossopharyngeal nerve block with submucosal infiltration into each palatoglossal fold. The supraglottic mucosa, including the epiglottis, is anesthetized with bilateral superior laryngeal nerve blocks. This is accomplished by percutaneous injection of 2–3 mL of lidocaine 1 % at the junction of greater cornu of the hyoid bone and thyroid cartilage. Alternatively, the supraglottic mucosal anesthesia is achieved by placing lidocaine-soaked gauze pad into the pyriform fossae.

3. After the first 3 h of the case, six transfemoral Ilizarov struts have been placed; the pulse oximeter reads 94 % on 50 % nitrous and oxygen. What do you make of this? Why/why not?

   There are at least three more hours of distraction to go. What will you do? Is PEEP effective for fat embolism syndrome? How would you choose the most effective PEEP level? What are the problems with PEEP? Any particular problems with this patient? What are they? Is this different than any other patient?

4. After the first 4 h of the case, you note that the current nasopharyngeal temperature is 39.7°; the heart rate, which started at 100/bpm, is now 140/bpm. What do you think is going on? What else could it be? How will you evaluate? Is there any way to test? Would you place an arterial line at this point? Give dantrolene prophylactically? What if the end-tidal CO_2 was 43? 57? What if you increased the minute ventilation by 100 %, and the end-tidal CO_2 decreased from 53 to 50? What could this be? Why?

A light wand can be used for blind oral intubation while the neck is stabilized. An anterior commissure scope may not be useful because of restricted neck mobility during stabilization. Often, excessive neck extension is required to visualize the glottis with an anterior commissure scope. The advantage of the light wand is it allows blind oral intubation without the need for excessive neck extension and flexion movement. The disadvantage is that advancing of the endotracheal tube may require flexion of the neck that can be a substantial risk in the presence of atlanto-axial instability. The advantage of an anterior commissure scope is that it facilitates visualization of an anteriorly located larynx. The disadvantage is that it requires hyperextension of the neck.

3. The potential for fat embolism should be considered. Optimize ventilation. Increase the inspired oxygen to maintain oxygen saturation above 95 %. Notify the surgeon. Place an arterial cannula to monitor arterial blood gasses. PEEP is an effective strategy for fat embolism syndrome. Optimal application of PEEP involves a stepwise increase of PEEP to optimal arterial saturation without compromising hemodynamics. It may compromise right ventricular function by increasing intrapulmonary pressure and left ventricular function by interventricular septal shifting. The hemodynamic effect of PEEP could be exaggerated in this patient due to scoliosis and restricted intrathoracic capacity.

4. With these findings, malignant hyperthermia must be included in the differential diagnosis list. Passive hyperthermia from active patient heating and the use of insulating drapes are also likely. Other less likely causes are mismatched blood transfusion reaction, drug-induced pyrexia, a cerebral bleed or thermogenesis from fat embolism, and an allergic reaction. Monitoring closely for a progressive rise of end-tidal carbon dioxide ($ETCO_2$) concentration is crucial, as is obtaining a venous or arterial blood gas. Active cooling of the patient should be started. Depending on the clinical context and monitoring results, I might place an arterial line at this point, particularly if the tachycardia and elevated $ETCO_2$ were out of proportion to efforts to deepen the anesthetic and increase minute ventilation or were accompanied by muscle contracture. I would administer dantrolene if the arterial blood gas analysis reflects uncompensated metabolic and respiratory acidosis and a wide A-a oxygen gradient despite increased minute ventilation. The values presented are not suggestive of malignant hyperthermia. These low values could be due to increased dead space ventilation. However, an inability to reduce $ETCO_2$ despite doubling the minute volume is suggestive of an inability to compensate for excessive production of carbon dioxide. This may suggest MHS. If the hypercarbia is due to increased dead space, it would decrease with hyperventilation.

Postoperative Course

Questions

1. Should this patient be extubated (assume odontoid hypoplasia and decreased neck range of motion)? Why/why not? Is it safer if he pulls his tube out in the middle of the night in the ICU? What criteria will you use for extubation? Why? Do you think this patient would meet spirometric or inspiratory force criteria for extubation? Why/why not? Is there any reason for him to have weak muscles? Should he be extubated in the operating room?

2. Is this patient at any higher risk for PCA methods of pain management? Would a preoperative blood gas have been likely to reveal this problem? How would you write the orders for his PCA?

Additional Questions

Questions

1. A 1.5-year-old with osteogenesis imperfecta is on the add-on list for open reduction internal fixation of a femur fracture. His preoperative temperature is 39.4°, and he has no signs of a URI. How will you evaluate this? Could this be a urinary tract infection? Is this temperature normal? Why? What else will you evaluate preoperatively? What if his platelet count is normal, but he has a history of easy bruising, as well as easy breaking? How can platelet aggregation abnormalities be evaluated? How will it affect your anesthetic management? Hematologic support? Does he need platelets? What if he is 6 kg; how will you manage his fluid therapy? Would you use succinylcholine to gain rapid control of the airway?

Postoperative Course

Answers

1. Determining whether the patient with a difficult airway should get extubated will depend on the intraoperative course as well as the potential for postoperative complications. If the intraoperative course was uneventful (i.e., if the above events did not occur), then careful extubation after meeting standard criteria would be reasonable. For concerns about the ease of reintubation, the endotracheal tube could be removed over an airway exchange catheter for a period of time, facilitating replacement if necessary. I will use the following criteria for extubation and ensure that the patient is alert and responds appropriately; there is a leak around the endotracheal tube, and patient does not have residual effect of neuromuscular blockade. This patient may not fulfill standard spirometric or inspiratory force criteria due to restrictive chest wall disease because of scoliosis and hypotonia. Many patients with achondroplasia are cognitively impaired and will not cooperate with spirometric assessment. For those patients with a normal mental status, it often suffices to ask them if they are getting enough air to breathe as an aid to extubation.

2. Yes. Chronic respiratory insufficiency with carbon dioxide retention may accompany patients with severe scoliosis and/or kyphosis as well as hypotonia. A preoperative arterial blood gas may reveal hypercarbia with a relatively normal pH as a result of a compensatory metabolic alkalosis, although the primary process with be the respiratory acidosis. A more conservative PCA dosing regimen may be called for, as well as nurse-controlled analgesia if the patient is cognitively impaired.

Additional Questions

Answers

1. The preoperative evaluation should rule out pulmonary and other infections. This temperature can, however, be normal for children with osteogenesis imperfecta. Hyperthermia occurs in OI patients due to a baseline hypermetabolic state from the high turnover of bone degeneration and reformation. The tendency toward bruising is due to defect in platelet adhesion; the platelet count is expected to be normal. Bleeding time is a good clinical indicator of this platelet aggregation defect, which can cause excessive bleeding during surgery. Transfusion of platelets will improve coagulation function and minimize surgical bleeding. Platelet transfusion is indicated if there is unexpected excessive intraoperative

What are the drawbacks? One of your partners suggests that this patient needs an arterial line instead of a blood pressure cuff. Do you agree? Why or why not?

2. Describe the findings associated with the following Radial dysplasia and associated syndromes:

   Radial aplasia and Hallermann-Streiff syndrome
   Radial aplasia and Cornelia de Lange syndrome
   Radial aplasia and Holt-Oram syndrome
   Radial aplasia and Fanconi anemia
   TAR syndrome
   VATER syndrome

3. A 6-month-old comes in for a hernia repair; he has rickets. What are the anesthetic implications? Any lab tests needed for this particular patient? Which ones? Is there any particular anesthetic technique that would be safer than any other?

bleeding. Fluid replacement requires careful management because some patients with OI may experience hyperhidrosis and may need addition fluid replacement of insensible losses. I would not use succinylcholine because succinylcholine-induced contractures increase the risk of fracture of the brittle bones. An arterial line is a safe alternative to a blood pressure cuff if the OI is severe. Alternatively, the use of an ultrasonic Doppler or aneroid strain gauge blood pressure device allows the use of the lowest inflation pressures. Repeated inflation of the blood pressure cuff to high pressures such as with the use of oscillometric device may cause fracture of the fragile bones.

2. Radial aplasia and Hallermann-Streiff syndrome: Narrow upper airways, obstructed nares, micrognathia, tracheomalacia, obstructive sleep apnea and cor pulmonale, and structural heart disease. Birdlike facies and ocular defects such as microphthalmia, cataracts, coloboma, glaucoma, and retinal degeneration.

   Radial aplasia and Cornelia de Lange syndrome: Short neck (66 %), anticipate difficult airways, and difficult access. Severe developmental delay. High-arched palate.

   Radial aplasia and Holt-Oram syndrome: It is an autosomal dominant heart disorder (ASDs, VSDs, and conduction system defects) associated with skeletal malformations including hypoplastic thumb and short forearm. Patients may have radioulnar synostosis, accessory carpal bones, or carpal coalition or tarsal coalition. Hyperphalangism and preaxial polydactyly are additional anomalies seen in some patients.

   Radial aplasia and Fanconi anemia: skeletal defects in association with bone marrow failure. Microcephaly with ptosis, strabismus, and microphthalmia may occur, along with hydrocephalus. Patients have short stature, with small or aplastic thumbs and radial aplasia, clinodactyly, syndactyly, or other radial abnormalities. There can be additional axial defects in the ribs or vertebral bodies as well as abnormalities of the kidneys and genitals.

   Thrombocytopenia-absent radii syndrome (TAR syndrome) similar to Fanconi anemia may be associated with micrognathia and congenital heart disease (tetralogy of Fallot, coarctation, ASD). Bilateral radial aplasia ulnae. The thumb is always present. Thrombocytopenia with diminished or absent megakaryocytes in the bone marrow.

   VATER syndrome: V (vertebral anomalies), A (anal atresia), T (tracheoesophageal fistula), E (esophageal atresia), and R (radial and renal dysplasia) in any combination. The VACTERL association is an acronym for V (vertebral anomalies), A (anal atresia), C (cardiac/malformations), T (tracheoesophageal fistula), E (esophageal atresia), R (renal anomalies), and L (limb anomalies).

3. Patients with rickets usually have hypocalcemia that may cause arrhythmias and potentiate non-depolarizing muscle relaxants. Patients with untreated rickets may have soft bones, which predispose to fracture with mild pressure. Poor

Why/why not? Risk of postoperative mechanical ventilation? Why is the serum calcium level typically normal? Then how is the metabolic component of the diagnosis made?

4. With what degree of curvature would you counsel a family for postoperative mechanical ventilation? Why? How will this influence your choice of monitors? At what degree of curvature would you expect to have to use a pulmonary artery catheter? Why?

5. A 16-year-old is scheduled for a sagittal split osteotomy and mandibular advancement; she has severe juvenile rheumatoid arthritis. What are your airway considerations? Why do these patients have airway problems? Are these mainly at the level of the mandible, the temporomandibular joint, or the larynx? Is there any cervical spine problem?

muscle tone is associated with kyphosis. Lab tests should include serum ionized calcium, phosphorus and alkaline phosphate concentrations, and parathyroid hormone. Avoidance of muscle relaxants may reduce the potential of enhancing the muscle weakness. Hypocalcemia and hypophosphatemia potentiate non-depolarizing muscle relaxant effects. The need for postoperative ventilatory support is high due to baseline poor muscle tone, restrictive chest wall disease (pectus, scoliosis), residual inhalation anesthetic effect, and postoperative opioid-induced suppression of central respiratory drive. The serum calcium is normal because of the compensatory increase of parathormone secretion in response to the initial low serum calcium. Increased parathormone secretion mobilizes calcium and phosphorus from the bone (producing osteomalacia), normalizes serum calcium, raises the serum alkaline phosphatase, and lowers serum phosphorus due to inhibition of reabsorption of phosphorus from the renal tubules. The metabolic component of rickets is completed by analysis of plasma calcium, phosphorus, alkaline phosphatase, and vitamin D hepatic and renal components (vitamins D3, D2, 25(OH)D3, 1,25(OH)2D3, 24,25(OH)2D3).

4. A curve of 65° or greater increases the chances for postoperative mechanical ventilation because of a significant decline in ventilation-perfusion matching in addition to an increased dead space to tidal volume ratio (>0.6). If the scoliosis curvature progresses beyond 65°, adequacy of ventilation should be monitored with serial arterial blood gas analysis. A pulmonary artery catheter or transesophageal echo monitoring may be required in the presence of a curve of 90° or greater, clinical evidence of right ventricular compromise, or the presence of preoperative hypoxemia and carbon dioxide retention. The use of TEE in the postoperative period can be very useful to monitor right ventricular function during positive pressure ventilation and to guide fluid and inotropic therapy.

5. In advanced or severe rheumatoid arthritis, the airway can be compromised. The earliest manifestation is micrognathia due to ankylosis of the temporomandibular joints. Other manifestations are ankylosis of the cricoarytenoid joint and a small glottic aperture, cervical spine ankylosis, flexion deformity, and subluxation of the atlanto-axial joint. A small mouth opening, limited neck range of motion, severely restricted neck extension, and small glottis due to restricted movement of cricoarytenoid joint may be present in varying combinations and severity. Airway difficulties should be expected at all levels of the upper airway passages. Flexion deformity of the cervical spine and subluxation of the vertebral bodies and atlanto-axial joint should be anticipated.