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May 2023 Critical Care Case of the Month: Not a Humerus Case

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History of Present Illness:
A 57-year-old woman with history of bone disease presented with a 3-day history of cough with thick yellow phlegm and progressive shortness of breath. No fever, chest pain or abdominal pain was noted. In the emergency department, she had SpO2 of 55% on room air, and then 90% on 15L NRB.

Past Medical History/Social History/Family History

- Bone disease since birth
- Asthma
- Severe scoliosis
- Gastrointestinal reflux disease
- Cholecystectomy
- Spinal growth rods
- Lives in adult care home, supportive family
- No smoking or alcohol use
- No illicit drug use
- There is no family history of any bone disease

Home Medications:

- Albuterol MDI PRN
- Alendronate 10mg daily
- Budesonide nebulizer BID
- Calcium carbonate BID
- MVI daily
- Lisinopril 10mg daily
- Loratadine 10mg daily
- Metformin 500mg BID
- Metoprolol 12.5mg BID
- Montelukast 10mg daily
- Naprosyn PRN
- Omeprazole 20mg daily
- Simvastatin 10mg daily
- Tizanidine PRN
- Vitamin D 2000 IU daily

Allergies:

- Cefazolin, PCN, Sulfa - all cause anaphylaxis

Physical Examination :

- Vital signs: BP 135/95, HR 108, RR 36, Temp 37.0 C Noted to desaturate to SpO2 in 70-80s off of Bipap even when on Vapotherm HFNC
- General: Alert, slightly anxious woman, tachypneic, able to answer questions
- Skin: No rashes, warm and dry
- HEENT: No scleral icterus, dry oral mucosa, normal conjunctiva

- Neck: No elevated JVP or LAD, short length
- Pulmonary: Diminished breath sounds at bases, no wheezes or crackles
- Cardiovascular: Tachycardic, regular rhythm without murmur
- Abdomen: Soft nontender, nondistended, active bowel sounds
- Extremities: Congenital short upper and lower limb deformities
- Neurologic: Oriented, fully able to make health care decisions with family at bedside

Laboratory Evaluation:

- Na^+ 142, K^+ 4.3, Cl^- 100, CO2 29, BUN 15, Cr 0.38, Glu 222
- WBC 21.9, Hgb 13.6, Hct 42.9, Plt 313 with 83% N, 8% L, 1% E
- Normal LFTs
- Lactic acid 2.2
- Venous Blood Gases (peripheral) on Bipap 10/5, FiO2 90%: pH 7.36, pCO2 58, pO2 55
- COVID-19 positive

Radiologic Evaluation:

A thoracic CT scan was performed (Figure 1).

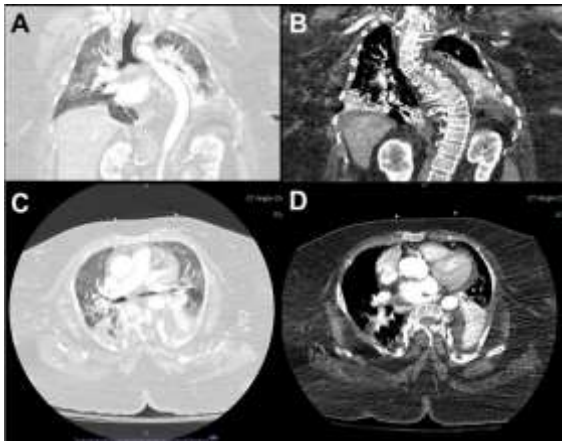

Figure 1. Representative images from thoracic CT scan in lung windows (A,C) and soft tissue windows (B,D). Click here to open Figure 1 in a separate enlarged window.

The CT images show all the following ***except***:

1. Severe scoliosis
2. Diffuse ground glass opacities
3. Right lower lobe consolidation
4. Pneumothorax
5. Atelectasis in bilateral lower lobes

Correct!
4. Pneumothorax

No pneumothorax is detected. The CT images do demonstrate severe scoliosis leading to chest cavity restriction with bilateral atelectasis. In addition, right lower lobe consolidation and ground glass opacities are seen and are concerning for infectious process. No pulmonary embolus was identified.

What is the ***most likely diagnosis***?

1. Achondroplasia
2. Gaucher's Disease
3. Marfan's Syndrome
4. Neurofibromatosis
5. Osteogenesis imperfecta

Correct!
5. Osteogenesis Imperfecta

Achondroplasia is a genetic disorder with an autosomal dominant pattern of inheritance whose primary feature is dwarfism. Gaucher's disease is a rare, inherited lipodystrophy in which deficiency of the enzyme glucocerebrosidase results in accumulation of glucocerebroside, throughout the body especially within the bone marrow, spleen and liver. The symptoms and physical findings associated with Gaucher disease vary greatly from patient to patient. Some individuals develop few or no symptoms (asymptomatic); others may have serious complications. Marfan syndrome is an inherited disorder that affects connective tissue, most commonly the heart, eyes, blood vessels and skeleton. People with Marfan syndrome are usually tall and thin with unusually long arms, legs, fingers and toes.

Regarding osteogenesis imperfecta, which of the following statements is ***false***?

1. Genetic mutations affect osteoblast and osteoclasts leading to high bone mineral density and decreased fracture risk
2. Clinical manifestations can include hypermobile joints, hearing loss, tooth discoloration, easy bruising and blue sclera
3. Aortic root dilatation, aortic dissection, heart valve defects and atrial fibrillation are associated with osteogenesis imperfecta
4. The cause of osteogenesis imperfecta is gene mutations, most commonly COL1A1 and COL1A2, affecting the amount of type 1 collagen and/or its structure leading to disorganized collagen matrices affecting osteoid production
5. Osteogenesis imperfecta is present in 1 in 10,000 to 20,000 births.

Correct!
1. Genetic mutations affect osteoblast and osteoclasts leading to high bone mineral density and decreased fracture risk

Osteogenesis imperfecta (OI) does have genetic mutations affecting osteoblasts and osteoclasts, but it is associated with low bone density leading to a higher incidence of fractures, scoliosis, short stature, long bone length discrepancies and bowing of long bones. OI is associated with chest wall deformities such as scoliosis, pectus carinatum, pectus excavatum, barrel chest along with rib and vertebral fractures. These chest wall deformities can lead to restrictive lung disease.

The other statements regarding osteogenesis imperfecta are true. Osteogenesis imperfecta has a broad range of severity ranging from fractures in utero to normal adult stature with low fracture incidence as well as variable extra skeletal manifestations.

Our patient is in danger of requiring intubation and mechanical ventilation. What ***potential risks are associated with intubation*** in patients who have osteogenesis imperfecta?

1. Fractures of the cervical spine, mandible, teeth
2. Occipital-atlas dislocation
3. Hyperpyrexia
4. Vertebral artery compression
5. All the above

Correct!
5. All the above.

Risk of fractures, occipital-atlas dislocation, hyperthermia, and vertebral artery compression as well as short neck lengths make intubations high risk in patients with these osteogenesis imperfecta manifestations. Respiratory infections, restrictive lung disease related to scoliosis, and rib as well as vertebral fractures are the major cause of morbidity and mortality in osteogenesis imperfecta. The decreased functional residual capacity and cardiac valvular disease can complicate a difficult airway during intubation and peri-operatively or in the intensive care unit.

Hypermetabolism and hyperpyrexia, possibly malignant hyperthermia has been reported in the anesthesia management of osteogenesis imperfecta patients. Malignant hyperthermia is reported as a potential complication, but some studies propose that the hyperthermia may be related to increased metabolic rate.

What are ***techniques for airway management*** in patients with osteogenesis imperfecta?

1. Ketamine is preferred agent (along with propofol, etomidate, thiopental)
2. Non-depolarizing muscle relaxants are preferable since fasciculations associated with succinylcholine can cause fractures and muscle damage
3. Fiberoptic intubation
4. Avoid overextension of the cervical spine
5. All the above

Correct!

5. All the above

Hyperthermia is a reported risk, and it is unclear whether this is malignant hyperthermia or hyperthermia related to increased metabolism. Given the uncertainty, avoidance of anticholinergics as well as medications with increased risk of inducing malignant hyperthermia such as succinylcholine and halothane should be avoided. Awake intubations using topical anesthesia or video laryngoscopy with ketamine, propofol, remifentanil and/or rocuronium can be considered. Use of an intubating laryngeal mask (LMA) is an additional consideration. The head and neck must be maintained in a neutral position given the heightened fracture risks.

What are ***management strategies for adults*** with osteogenesis imperfecta?

1. Early and ongoing optimization of bone health with bisphosphonates, advanced osteoporosis therapies such as denosumab and teriparatide, and maintenance of mobility and muscle strength working with a multidisciplinary team
2. Routine dental exams to prevent tooth loss
3. Routine hearing evaluations to detect early hearing loss
4. Cardiovascular assessments for valvular and aortic root measurements
5. All the above

Correct!

5. All the above.

Early diagnosis is important and an evaluation for osteogenesis imperfecta be considered for people presenting with short stature, minimal trauma fractures and short, long bones. Genomic testing confirms the genetic diagnosis but a normal genetic test does not exclude a clinical diagnosis of osteogenesis imperfecta.

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