A 5-month-old, otherwise healthy, former 35-week male infant, presented to our pediatric emergency department with the chief complaint of cough and “sucking in” at the chest. The patient was in his usual state of health until 1 day prior to presentation when he developed nasal congestion. Before coming into the hospital, the family noted decreased oral intake and only 3 wet diapers in the past 24 hours. No other past medical history was notable except intubation at time of delivery due to respiratory distress requiring surfactant administration. Past surgical history was significant for bilateral inguinal hernia repair and circumcision. He is on no medications currently and immunizations are up to date. He lives at home with his nonsmoking parents.

In the emergency department, the patient’s vitals were as follows: heart rate, 137; blood pressure, 94/51; temperature, 36.8; respiratory rate, 56; and pulse oximetry, 97% on room air. Physical examination revealed an infant in mild respiratory distress with tachypnea, subcostal retractions, clear nasal discharge, and coarse breath sounds throughout. The patient was treated with nasal suctioning, and the decision was made to send for chest radiograph.

Question What is the current American Academy of Pediatrics’ recommendation regarding the use of imaging when diagnosing bronchiolitis?

Discussion

The American Academy of Pediatrics Clinical Practice Guidelines for the diagnosis, management, and prevention of bronchiolitis from 2014 recommend that the diagnosis of bronchiolitis be made clinically and that the use of imaging is not warranted in most cases. Current evidence does not support routine chest radiography in children with bronchiolitis as data are insufficient to demonstrate that chest radiography correlates well with disease severity. One randomized trial revealed that children with suspected lower respiratory tract infection who had radiography performed were more likely to receive antibiotics without any difference in clinical outcomes. Initial radiography should be reserved for cases in which the patient clinically worsens unexpectedly, respiratory effort is severe enough to warrant ICU admission, or where signs of airway complication (ie, pneumothorax) are present.
CASE CONTINUATION

Our patient’s chest radiograph revealed normal lungs and a healing posterior fracture of the right seventh rib (Fig 1). This finding prompted concern for inflicted injury and physical abuse. Social work, the Department of Children and Families, and our Child Protection Team were consulted upon discovery of the rib fracture. A full Child Protection medical workup was initiated, and the patient was admitted to the hospital. Current suggested guidelines for the workup of suspected child abuse/neglect were recently published in May 2015 in the Journal of Pediatrics. The protocol at our institution is based on these recommendations and includes standard age specific tests/interventions (Table 1). Based on this protocol, a skeletal survey was completed. These radiographs revealed bilateral irregularities to the distal radial and ulnar metaphyses consistent with healing metaphyseal fractures, as well as healing metaphyseal corner fractures of the right and left distal femurs (Fig 2). The survey also revealed more than 5 wormian bones of the skull, and an unusual pelvis configuration. The patient underwent a computed tomography (CT) scan of the head that revealed no acute intracranial abnormalities, except the aforementioned wormian bones. Due to the lack of any history to explain these injuries, the case was highly concerning for child physical abuse.

The patient was removed from parental custody by the Department of Children and Families.

Question What is the sensitivity and specificity of radiologic findings, such as posterior rib fractures and metaphyseal fractures, for the diagnosis of physical abuse in pediatric patients?

Discussion

Some authors suggest that rib fractures in children younger than 3 years of age have a positive predictive value of 95% as an indicator of abuse with posterior medial rib fractures having the highest specificity. The positive predictive value of rib fractures as an indicator of child abuse was reviewed in a meta-analysis and the probability of abuse with rib fracture present was 71% (study excluded children with significant noninflicted trauma and postoperative injury). Data also suggest that inflicted rib fractures are associated with a high risk of mortality, likely due to the degree of force required to produce them.

Classic metaphyseal lesions (CMLs), or “bucket handle/corner fractures,” are considered the injury with the highest specificity for diagnosing child physical abuse. Data suggest that they are discovered in ~39% to 50% of abused children younger than 18 months of age and they are seen almost exclusively in children younger than 2 years of age. CMLs are the most common long bone fracture found in infants who die of nonaccidental injury. CMLs are linear fractures through the primary spongiosa of long bone metaphysis. The injury is inflicted when torsional and/or tractional shearing strains are applied across the metaphysis, such as with vigorous pulling or twisting of an infant’s extremity. CMLs most often occur in the distal femur, proximal tibia, distal tibia, and proximal humerus.

CASE CONTINUATION

After further examination, the patient was noted to have mild hypotonia and what seemed like abnormal hair; thus a multidisciplinary discussion ensued and genetic and dermatologic consults were

![Figure 1](Chest radiograph upon initial presentation. Notable finding of focal expansion of right posterior seventh rib (arrow).)

### TABLE 1

<table>
<thead>
<tr>
<th>Age</th>
<th>Intervention (Age Specific)</th>
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| 0–6 mo | - Skeletal survey  
- Head CT or brain MRI  
- Ophthalmology consult, if neurologic abnormalities or intracranial bleeding  
- Serum AST/ALT  
- Urine comprehensive toxicology and drugs of abuse |
| 6–24 mo | - Low threshold for head CT or brain MRI  
- Serum AST/ALT  
- Urine comprehensive toxicology and drugs of abuse |
| >24 mo | - If >5 y: consider history from child  
- If >24 mo and history of developmental delay, proceed with skeletal survey  
- Consider serum AST/ALT  
- Consider urine comprehensive toxicology and drugs of abuse |

ALT, alanine aminotransferase; Alk Phos, alkaline phosphatase; AST, aspartate aminotransferase; Ca, calcium; iPTH, intact parathyroid hormone; Mg, magnesium; Phos, phosphorus; PT/PTT, prothrombin time/partial thromboplastin time; vWF, Von Willebrand. |
Menkes disease is a rare X-linked recessive disorder of infantile-onset. It was first described in 1962 and later demonstrated to result from a defect in the ATP7A gene. The prevalence is 1 in 100,000-250,000 live births, with a male predominance. Menkes disease is a disorder of copper transport leading to low serum copper levels. The typical physical manifestations of Menkes disease include the following: coarse/steely hair; skin pallor; neurologic sequela (ie, seizures), tortuous vasculature, developmental delay, wormian bones, pectus excavatum, sacral dimple, bilateral inguinal hernia, blue sclera, hypotonia, failure to thrive, hypothermia, metaphyseal widening, and abnormal pelvic structure. Age of presentation is typically 2 to 4 months of age in boys with death around 3 years of age if left untreated. These patients often present to the emergency department due to seizure activity, or to their primary care physician with failure to thrive. Definitive treatment requires daily subcutaneous or intravascular copper supplementation.

**CASE RESOLUTION**

Once Menkes disease was added to our differential diagnosis, further workup was initiated. Light microscopy examination of the patient’s hair revealed a “pili torti” appearance. Kidney and bladder ultrasonography revealed a possible bladder diverticulum and the presence of mild bilateral hydronephrosis. In addition to the previously mentioned findings, the patient had a history of neonatal hypothermia and bilateral inguinal hernia repair. The serum copper level was found to be 18 μg/dL (normal: 24–152 μg/dL) with a serum ceruloplasmin level of 8 mg/dL (normal: 15–48 mg/dL). Therefore, based on all of these findings, a clinical diagnosis of Menkes disease was made, and after further discussion, the patient was reunited with his family. The patient was started on intravenous copper chloride 250 μg twice daily and was discharged to continue daily subcutaneous injections at home. The patient’s respiratory distress followed a course consistent with viral bronchiolitis and gradually improved over the course of the admission.

**DISCUSSION AND CONCLUSIONS**

Previous authors have suggested including Menkes disease on the differential for child abuse. In previous case reports, most patients with misdiagnosed Menkes disease suffered from intracranial hemorrhage and seizures, thus mimicking abusive head trauma. This case demonstrates the importance of remaining open to considering additional explanations for clinical findings, even when a patient presents with common symptoms suggesting a common diagnosis.

**LEARNING POINTS**

- In the majority of cases, radiographic studies are not indicated for the diagnosis of bronchiolitis in infants.
- Anterior and posteroi medial rib fractures in children are highly specific for child abuse and are associated with a high risk for mortality.
- CMLs are a highly specific radiologic finding for inflicted injury in children younger than 2 years of age.
- Standardized protocols exist for the suggested radiographic and laboratory workup of suspected child abuse in children (Table 1).
- Menkes disease is a relatively rare X-linked genetic disorder that is due to abnormal function of cellular copper absorption or transport from the gastrointestinal tract. The disease has a well-defined list of clinical criteria and the definitive diagnosis is made by the finding of low serum copper levels.
- Even though a metabolic cause may be identified to explain injuries consistent with child abuse, it is important to consider child abuse in the differential until the full multidisciplinary workup has been completed.

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**FIGURE 2** Radiograph of distal femur from Skeletal Survey consistent with healing metaphyseal corner fracture (arrow).


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