



UNIVERSITY of
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MEDICAL CENTER

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DEPARTMENT OF IMAGING SCIENCES

Imaging Sciences Interesting Cases

CASE 209

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CLINICAL PRESENTATION: Patient is a 19-year-old male following a video assisted thoracic surgery (VATS) procedure. Multiple prior exams available for comparison.

IMAGING FINDINGS: Left-sided cardiac pacing device is present, unchanged from multiple prior exams. Interval placement of chest tube in left lung apex. Elevation of the left hemidiaphragm, both relative to the right hemidiaphragm and relative to prior exams. Radiopaque components of electronic devices project over the hemidiaphragms bilaterally.

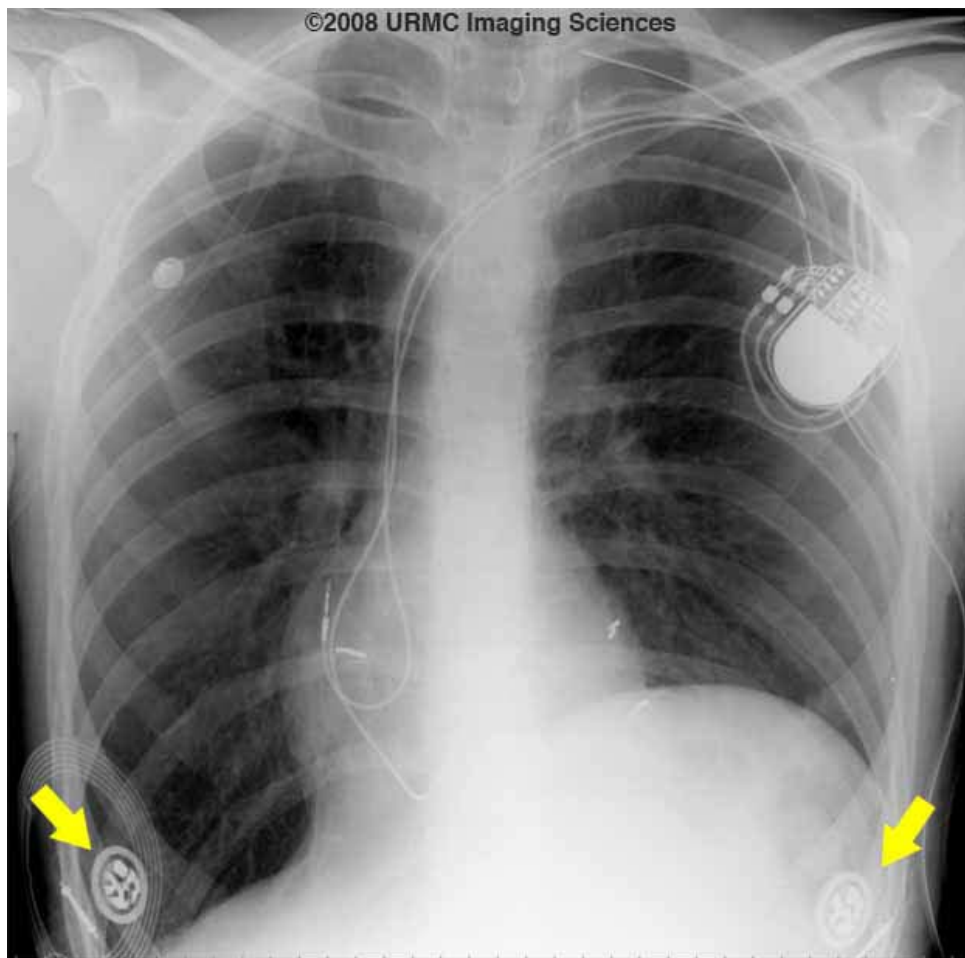


Figure 1: PA radiograph demonstrating elevation of the left hemidiaphragm relative to the right hemidiaphragm. Cardiac pacing device and bilateral electronic devices projecting over the hemidiaphragms are also identified (arrows).

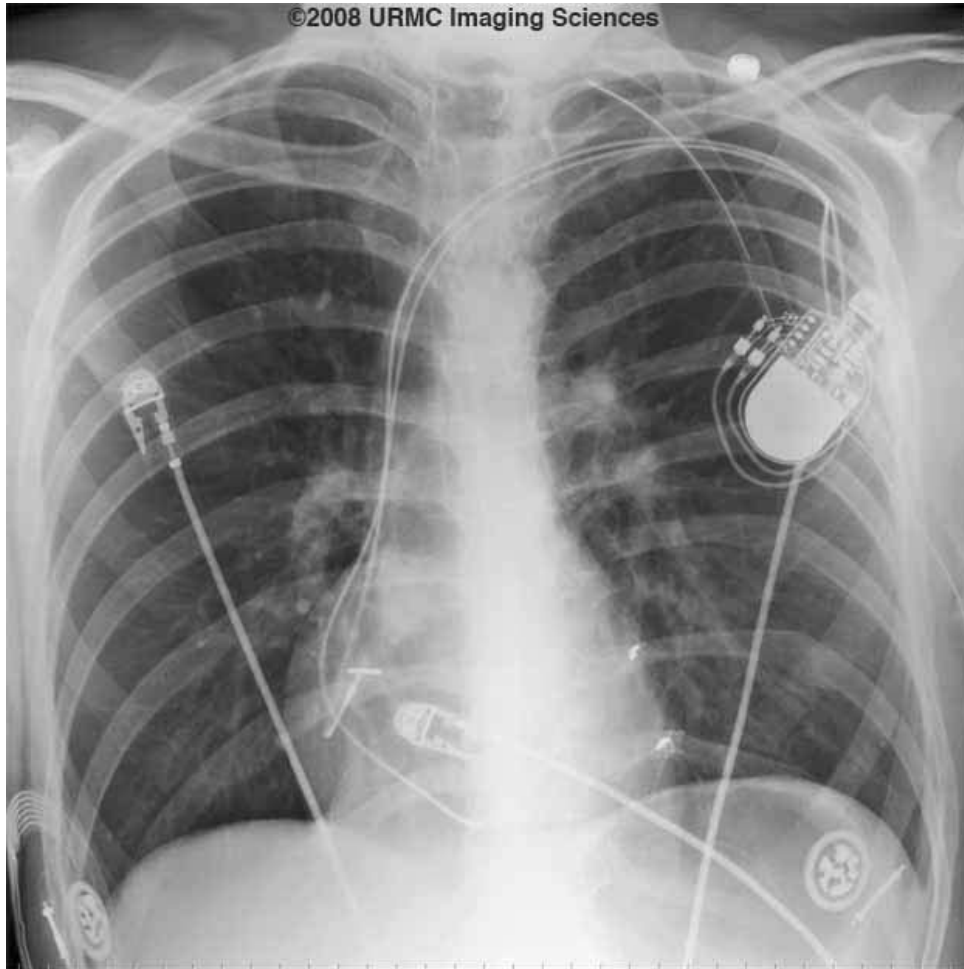


Figure 2: Subsequent radiograph demonstrating normal appearance of the hemidiaphragms following repair of left diaphragmatic pacing device.

DIAGNOSIS: Congenital hypoventilation syndrome with malfunction of left diaphragmatic pacemaker

DISCUSSION: Congenital central hypoventilation syndrome, or CCHS, is a rare congenital disorder of the central nervous system in which autonomic control of breathing is absent or impaired. Response to hypercapnea and/or hypoxia is severely impaired during waking hours and may be completely absent during sleep and/or stress, often requiring 24 hour ventilatory support. CCHS is now associated with a specific gene mutation (PHOX2B) in at least 90% of patients, however, the mutation is highly variable in size and expression, which may account for the range of phenotypic presentations. Other organ systems are almost universally affected, including cardiac disease (approximately 20%), seizures (40%), Hirschsprung's disease (20%), and errors of thermoregulation (40%), among others.

In this patient, the history of CCHS was not mentioned in the requisition. The presence of striking asymmetry of the hemidiaphragms suggested phrenic nerve palsy / dysfunction, which is an uncommon finding in a patient of this age group (as is the presence of a cardiac pacing device). The small devices projecting over the diaphragms bilaterally were a second clue as to the etiology of the elevated left hemidiaphragm, and investigation of the patient's medical record suggested the ultimate diagnosis -- malfunction of the left diaphragmatic pacemaker. The malfunctioning pacing lead was repaired, and subsequent imaging demonstrated restoration of symmetric diaphragmatic excursion.

REFERENCES:

1. Vanderlaan M, Holbrook CR, Wang M, Tuell A, Gozal D. Epidemiologic survey of 196 patients with congenital central hypoventilation syndrome. *Pediatr Pulmonol.* 2004 Mar;37(3):217-29. [PubMed]