RADY 401 Case Presentation

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23 year-old female w/ history of osteopetrosis

• Recent discharge on 7/12, concern for preseptal cellulitis and chronic osteomyelitis, discharged on Augmentin
• Presented 7/14 after being found down after unwitnessed fall, concern for AMS

• History of developmental delay, blindness, hearing impairment
• Nontoxic appearing, afebrile with stable vital signs
Imaging performed

- CT C-spine screening (w/o contrast)
- CT Head w/o contrast
- X-ray Trauma Hip Bilateral
- MRI C-spine without contrast
- X-rays of hands, hips with Judet views, chest, pelvis, C-spine (AP and lateral)
- US of abdomen – fluid search
CT C-spine screening, transverse, w/o contrast

Nondisplaced fractures of C1 anterior arch and lateral masses—further evaluation with MRI recommended to look for ligamentous injury.
CT head w/o contrast, sagittal

- Note presence of thickened skull and dense bone marrow → difficulty with typical windows to view CT
- Ventriculomegaly of lateral and third ventricles (unchanged compared to 2009)
- No intracranial hemorrhage seen on other cuts
Comparison to “normal bone” window
Oblique x-ray of hand

- Diffuse osseous sclerosis consistent with history of osteopetrosis
- Unchanged partially imaged plate and screw fixation of ulna and radius
X-ray Trauma bilateral, positive arrow signs for lucencies
Previously seen lucencies thought to be non-displaced fractures are not seen here
Assessment and Plan

• For nondisplaced pelvis fractures—no operative intervention indicated
  • Patient is household ambulator and wheelchair dependent outside of home
  • She will likely self-limit weightbearing and gradually return to baseline
  • Follow-up as outpatient in 2 weeks for repeat radiographs

• For stable C-spine fractures—continue C-spine precautions and wearing C-collar until follow-up in 6 weeks with Neurosurgery for repeat radiographs
Imaging Discussion

• Osteopetrosis → defective osteoclastic resorption of immature bone → predisposition to fractures

• Unwitnessed fall
  • Need to evaluate sources for fall as well as consequences
  • CT C-spine, CT head – evaluating for stroke and fracture → AMS
    • MRI C-spine to further assess ligamentous injury

• X-rays to evaluate for fractures – trauma hip, hands, chest, pelvis, c-spine
  • Addition of Judet views to further assess for non-displaced fractures

• Ultrasound of abdomen – trauma evaluation
Classic imaging findings

- Osteoclast dysfunction leads to dense bone and obliterated medullary canals
- Predisposition to fractures
- Increase in osteomyelitis which may be identified on imaging – due to lack of marrow vascularity and impaired WBC function
Classic imaging findings (spine)

- Lumbar spine demonstrates a “sandwich vertebrae” appearance
- May also observe “bone within bone” appearance secondary to sclerosis along inner margins of the endplates
Imaging Discussion

- Radiographs are sensitive for detecting osteopetrosis, but non-specific as many disorders can increase bone density and lead to similar appearances (i.e. Pyknodysostosis)
- Increased density from decreased osteoclast activity or increased osteoblast activity
- There are also many subtypes of osteopetrosis
Estimated costs

- CT C-spine screening (w/o contrast)
- CT Head w/o contrast
- X-ray Trauma Hip Bilateral
- MRI C-spine without contrast
- X-rays of hands, hips with Judet views, chest, pelvis, C-spine (AP and lateral)
- US of abdomen – fluid search

- $1,436 to $3,170
- $1,059 to $2,619
- $309 to $649
- $1,736 to $3,953
- $199 to $429, $309 to $649, $18 to $512, $323 to $726
- $533 to $1,334

***From FairHealthConsumer.org—Zip code 27516
References

• FAIR Health Consumer. https://www.fairhealthconsumer.org/medical