CASE REPORT

Not just another case of low back pain

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SUMMARY

A 55-year-old woman presented with persistent, progressive low back and neck pain for 5 years that had failed conservative therapy with analgesics and epidural injections. Physical examination revealed range of motion of lumbar and cervical spine were significantly limited in all directions and she had a bluish discolouration of the pinnae bilaterally. She had elevated urinary excretion of homogentisic acid, diagnostic of alkaptonuria. Further investigation with imaging of the spine revealed diffuse advanced degenerative disease of the cervical and lumbar spine consistent with ochronotic arthropathy.

INVESTIGATIONS

No abnormalities were noted on comprehensive metabolic panel and complete blood count tests.

BACKGROUND

The second most common reason for all physician visits is back pain. This common complaint can be attributed to many aetiologies, most commonly degenerative disease and mechanical low back pain. Physicians must be aware of less common causes of low back pain. Ochronotic arthropathy presents at a young age of onset with advanced degenerative arthritis and has unique imaging and physical examination findings.

CASE PRESENTATION

A 55-year-old Caucasian woman presented with persistent, progressively worsening low back and neck pain and stiffness for 5 years. She had 2–3 h of morning stiffness that improved with activity and heat. She was able to achieve moderate relief with non-steroidal antiinflammatory and narcotic pain medications and cervical epidural injections. She noted bluish discolouration on her ears and her dorsal hands.

She denied a history of trauma, malignancy, intravenous drug abuse or neuropathic symptoms. She had a 20 lb unintentional weight loss over the course of a year, which she attributed to her depression due to limited mobility and pain. Age-appropriate screening was not up-to-date. Her medical history included hypertension, emphysema and cervical stenosis. Her mother had the same pattern of arthritis presenting at a similar age.

Physical examination was notable for cervical spine held slightly flexed and inability to extend beyond neutral. Her Schober examination was abnormal with a 1 cm increase on full forward flexion. She was unable to extend her lumbar spine beyond neutral. Levoscoliosis was present. Pain was elicited on full extension of knees bilaterally with crepitus. There was bluish discolouration of the pinnae bilaterally, and 2 mm blue nodules were seen between the joints on the thumbs bilaterally.

Figure 1 Lateral/anteroposterior views of lumbar spine. Images demonstrate characteristic findings of advanced ochronotic arthropathy. Severe uniform obliteration and calcification of the intervertebral disc spaces throughout the spine are demonstrated.

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Erythrocyte sedimentation rate, ferritin and transferrin were also normal. Urinary organic acids were elevated with an excretion of homogentisic acid of 2563 mmol/mol creatinine (reference value <11 mmol/mol creatinine).

X-ray series of lumbar spine (figure 1) and cervical spine revealed severe uniform obliteration and calcification of the intervertebral disc spaces throughout the spine and marginal intervertebral osteophytes.

OUTCOME AND FOLLOW-UP

A diagnosis of ochronosis was made based on the elevated urinary homogentisic acid and characteristic imaging changes. Patient was advised management is symptomatically and ultimately, joint replacement of the joints that have accrued long term damage. Within 1 year of consultation, the patient underwent total knee arthroplasty.

DISCUSSION

Alkaptonuria is a rare and an autosomal recessive inherited metabolic disease, estimated to occur in 1 in 250 000 to 1 million live-births. An absence of homogentisate 1,2-dioxygenase enzyme results in elevated blood levels of homogentisic acid.¹ Ochronosis is the accumulation of homogentisic acid in tissues leading to a dark bluish discolouration of connective tissues including the joints, eye, skin, cardiovascular and genitourinary systems. Auricular cartilage is commonly involved. The most common manifestation of alkaptonuria is ochronotic arthropathy, which affects large weight-bearing joints and typically manifests in the third or fourth decade. The lumbar spine is affected initially and will often present with low back pain and stiffness with limited range of motion. Radiographic imaging demonstrates characteristic findings, including loss of lumbar lordosis, widespread calcification of intervertebral discs, vacuum disc phenomenon and progressive narrowing of intervertebral spaces.² With advanced disease, obliteration of intervertebral spaces and marginal intervertebral osteophytes may appear (figure 1).² The knee is the most common peripheral joint involved, demonstrating changes similar to osteoarthritis with joint space narrowing and subchondral sclerosis. Characteristic findings on imaging and

elevated urinary levels of homogentisic acid are diagnostic. No effective therapy exists. Management is symptom-directed with analgesics and joint replacement if necessary.

Learning points

- ▶ Back pain is the second most common reason for all visits to physicians.
- ▶ Alkaptonuria is a rare inherited metabolic disease resulting in the accumulation of homogentisic acid in tissues, most commonly involving the joints leading to ochronotic arthropathy.
- ▶ In comparison to more common causes of low back pain, ochronotic arthropathy presents at a young age of onset with advanced degenerative arthritis and has unique imaging and physical examination findings.
- ▶ Characteristic findings on radiographs of the spine include loss of lumbar lordosis, widespread calcification of intervertebral discs, vacuum disc phenomenon and progressive narrowing of intervertebral spaces.² With advanced disease, obliteration of intervertebral spaces and marginal intervertebral osteophytes may appear.²

Contributors All authors had access to the data and a role in writing the manuscript.

Competing interests None.

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