Case Presentation

Juan Pablo Ghisi, MD
Argus Diagnóstico Médico
Chief of MRI Department, Hospital General de Agudos "Dr. Juan A. Fernández" (Buenos Aires, Argentina)

Adrián Trache, MD
MRI Staff Member, Hospital General de Agudos "Dr. Juan A. Fernández" (Buenos Aires, Argentina)
Argus Diagnóstico Médico

See case resolution on page 434.

On this paper, we discuss the case and the radiological findings of a 26-year-old woman whose chief complaint was thickening of both Achilles tendons. The patient reported that the pain and swelling in both ankles suddenly started when she was about 14 years old. At the time, she was treated with anti-inflammatories, since a sprain was suspected. Over the years, the swelling of both ankles not only did progress, but became more obvious (Figure 1). The patient also presented with new, or enlarged, nodules in the hands and elbows (Figure 2).

The level of serum cholestanol measured by gas chromatography-mass spectrometry was 20.30 µmol/L (reference value: <5.0 µmol/L), and the analysis of bile acids in urine by electrospray ionization determined an increase in 27-Nor-cholestane-5β-pentol, cholestanetriol and 27-Norcholestanehexol. The biochemistry panel and the imaging findings suggested an ongoing diagnosis, which was then confirmed by an Achilles tendon biopsy. Chenodeoxycholic acid was prescribed. This replacement therapy improved the tendonitis, which recurred once the patient went off the medication temporarily. The patient underwent surgery on both Achilles tendons to reduce tendon volume.

Neurological symptoms have been scarce over the years, and the patient has only had headaches and occasional dizziness. She never reported any visual disturbances or changes in bowel movements. Genetic tests have not yet been performed on the patient.

FINDINGS AND INTERPRETATION OF IMAGING STUDIES

The patient was evaluated by an Achilles tendon ultrasound, an ankle X-ray (not shown here) and an ankle, brain, spine, hand and elbow MRI.

On the ankle MRI, both Achilles tendons had fusiform diffused thickening, variable signal intensity and a reticulate pattern within, as well as iso- to hyperintensity on T1WI and fat-suppressed PD-weighted sequences. Normal tendon fibers with low signal intensity were also observed. When scanning the ankles, a similar MRI pattern was incidentally found on both peroneus longus tendons (Figures 3 and 4).

MRIs of the other areas showed bilateral fusiform lesions on the extensor tendon of the middle finger (Figures 5 and 6) and on the triceps muscle (Figures 7 and 8).

The brain MRI revealed minor anomalies in the posterior fossa (Figure 9), with no evident alterations in the supratentorial region (thus the scarce neurological symptoms of this patient). The spine MRI was normal.

JUAN PABLO GHISI, MD • jpghisi@gmail.com

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Figure 1. Photograph of the left ankle showing gross swelling of the Achilles tendon. The skin covering the area looks healthy with no color changes.

Figure 2. Photograph of the right hand, showing an oval lesion along the extensor tendon of the third finger, at the metacarpophalangeal joint level.

Figure 3. Axial view of a fat-suppressed PD-weighted MRI (3990/36) of the left Achilles tendon. In addition to tendon thickening, this view shows the telltale reticular pattern of the disorder in question. A similar finding is observed in the peroneus longus tendon (arrowheads).

Figure 4. Sagittal view of a T1-weighted MRI (660/25) of the left Achilles tendon, showing prominent fusiform thickening. While there is variable signal intensity, a generalized hyperintensity is mixed with linear trabeculations that could suggest the presence of pathologic tissue (probably with related inflammatory changes) and normal residual collagen fascicles, respectively.

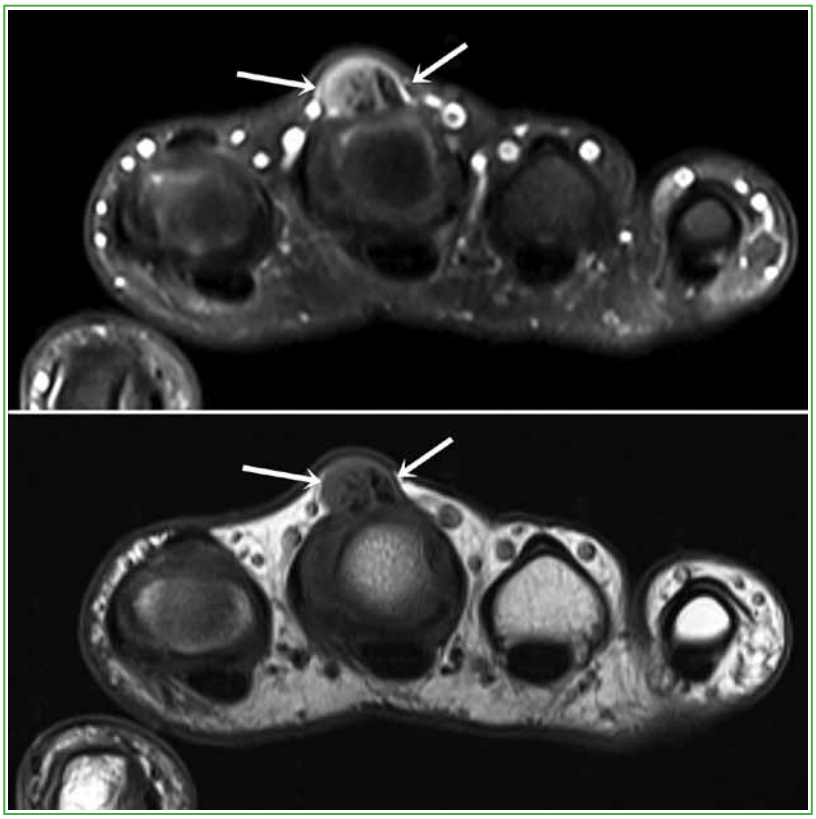

Figure 5. Axial view of a fat-suppressed PD-weighted MRI (2960/29) and a T1-weighted MRI (636/13) of the right hand. There is a nodule (white arrows) over the extensor tendon of the third finger protruding over the integumentary plane. Normal tendon fibers are mixed with tumor tissue.

Figure 6. Sagittal view of a fat-suppressed PD-weighted MRI (2100/25) and a T1-weighted MRI (587/13) of the right hand. In the sagittal view, the nodule (yellow arrows) is fusiform.

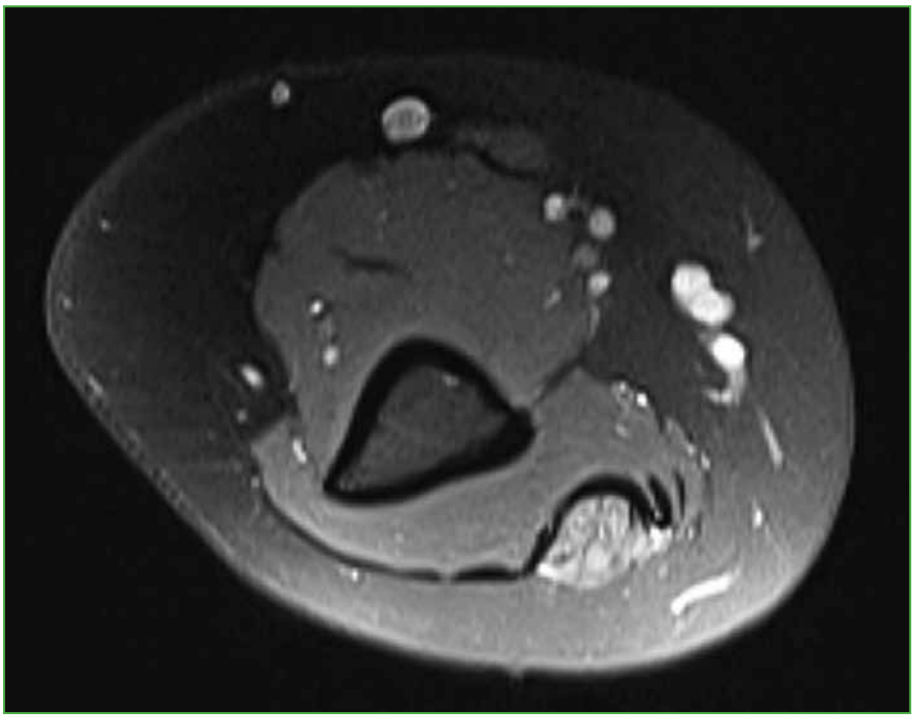

Figure 7. Axial view of a fat-suppressed PD-weighted MRI (2770/39) of the left elbow showing a tumor along the triceps tendon, which had been discovered through palpation.

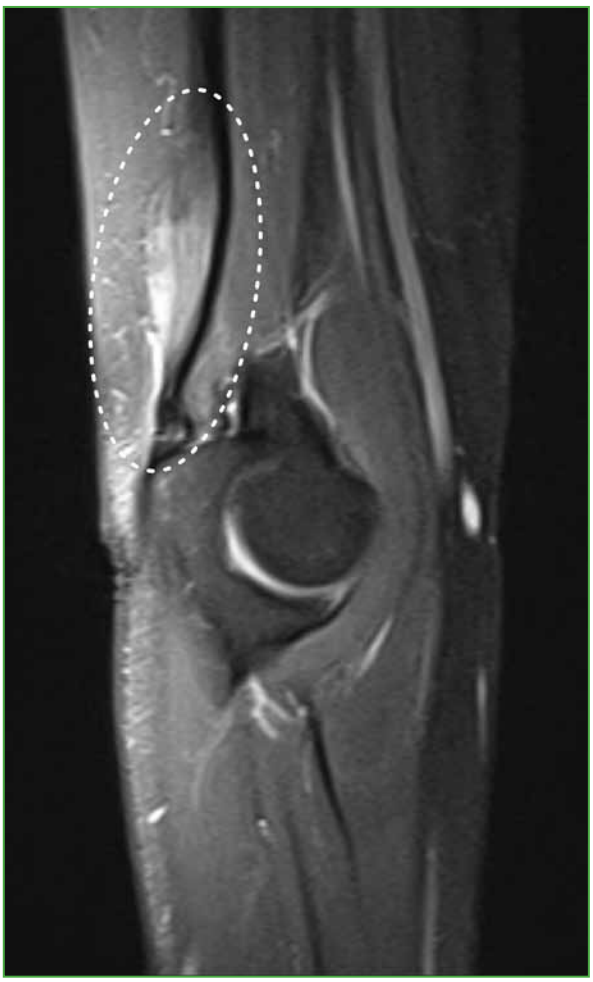

Figure 8. Sagittal view of a fat-suppressed PD-weighted MRI (3340/37) of the left elbow showing the telltale reticulated pattern of this type of lesions.

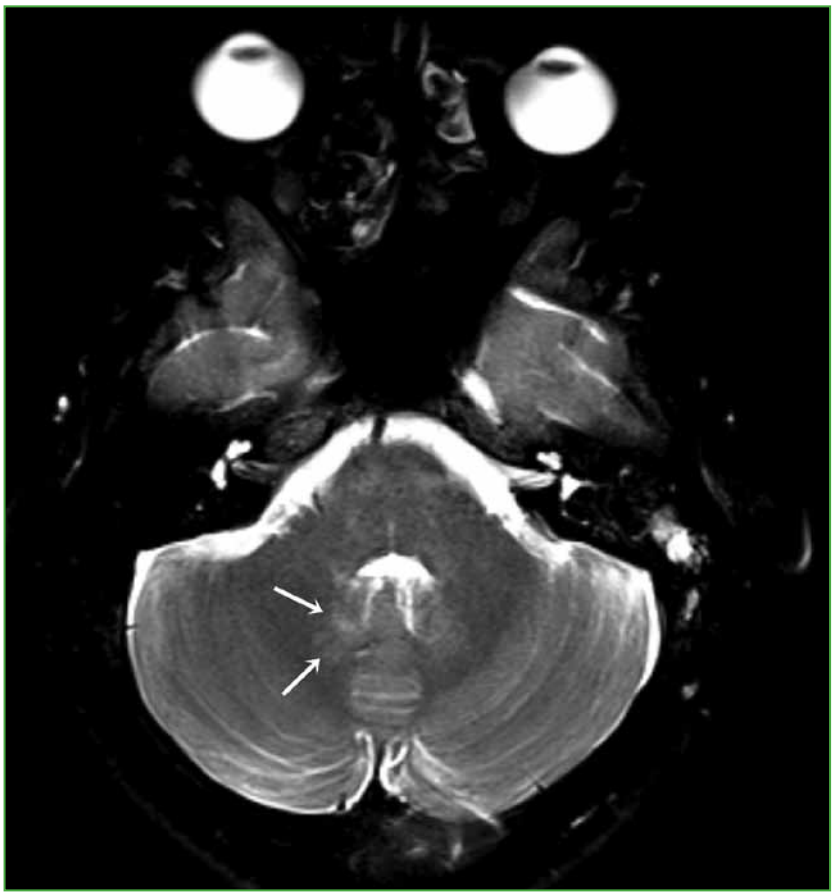

Figure 9. Axial view of a fat suppressed T2-weighted MRI (5470/95) of the brain showing a subtle signal alteration at the right dentate nucleus (arrows).

Conflict of interest: Authors claim they do not have any conflict of interest.

A. Trache ORCID ID: http://orcid.org/0000-0002-5111-3828