A 3-year-old boy presented with abnormal neck posturing and rapidly growing lumps over the neck. The presence of typical malformed great toes and ectopic calcification (figure, A–C) and an affected family member confirmed the diagnosis of fibrodysplasia ossificans progressiva (FOP). MRI of the cervicothoracic region (figure, C, E–G) revealed diffuse hypointense signals extending along the neck, thoracic wall, and paraspinal muscles. The presence of dentate nucleus hyperintensity on brain MRI indicates that FOP may not be purely a musculoskeletal disorder, but a multisystem disorder including CNS involvement (figure, D). There was pseudodystonia of the neck without any other neurologic signs. Because ectopic ossification might not be present in the early stages, diagnosis of FOP based on radiographs alone may be delayed. In the presence of characteristic skeletal malformations, MRI can detect presosseous lesions and avoid additional diagnostic procedures, including biopsy.

REFERENCES