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Clinical Image: Avascular necrosis of the femoral head driven by the *COL2A1* gene mutation

A 14-year-old boy was brought to the rheumatology clinic with a two-year history of pain in the bilateral hip. The hip pain emerged after walking and subsided upon taking a break. There had been no preceding trauma or fever or history of receiving glucocorticoids. Physical examination showed no limitation in hip range of motion. The patient had no low back pain. Laboratory tests revealed normal levels of erythrocyte sedimentation rate, C-reactive protein, interferon-γ release assay, and endocrine hormones. His HLA-B27 was negative. (A) Pelvic X-ray and (B and C) computed tomography revealed a focal area of collapse in the bilateral femoral head (arrow). (D) Magnetic resonance imaging showed rough articular surface, (E) bone marrow edema and synovitis, and (F) "double-line sign" (arrow). These findings indicated an avascular necrosis of the femoral head (ANFH). His parents and sister were in good health. Nevertheless, the peripheral blood samples were collected from the patient and his family members, and the patient's blood sample was analyzed via the whole-exome sequencing. (G) It identified a de novo *COL2A1*: 3508G>T, p.Gly1170Cys heterozygous mutation in the patient and this gene in the patient's pedigree of the family was tested wild type confirmed by Sanger sequencing. A diagnosis of Legg–Calvé–Perthes disease (LCPD) was made. LCPD is an idiopathic ANFH in preadolescent children. Pain is primarily localized in the hip, occasionally accompanied by leg and knee pain, and most of the patients show limited hip internal rotation.[1] Its incidence ranges from 0.4 of 100,000 to 29.0 of

100,000 in children younger than 15 years old.[1] The cause of LCPD remains unknown. Mutations in the type II collagen, which are encoded by the *COL2A1* gene, are associated with type II collagenopathies, including LCPD and ANFH. A heterozygous mutation (c.1888G>A, p.Gly630Ser) in exon 29 of *COL2A1* leading to LCPD and ANFH had been reported in a Chinese family.[2] Our patient demonstrates a novel heterozygous mutation in the *COL2A1* gene that causes LCPD. The patient received a conservative treatment regimen of diclofenac sodium for pain relief and a brace to reduce pressure on the hip. Consent was obtained directly from the patient and parents. T1WI, T1-weighted imaging; T2WI, T2-weighted imaging.

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