

Ocular Sonographic and Magnetic Resonance Imaging Features of Osteoporosis Pseudoglioma Syndrome

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ABSTRACT

Osteoporosis pseudoglioma syndrome is a rare autosomal recessive disorder, characterised by early-onset osteoporosis and congenital- or juvenile-onset blindness. The combination of multiple osteoporotic fractures and blindness suggests a clinical diagnosis of osteoporosis pseudoglioma syndrome. The main ocular manifestations include microphthalmia, anterior segment anomalies, cataract, vitreoretinal dysplasia, and retrolental mass. This report describes the ocular sonographic and magnetic resonance imaging features of a 23-year-old Chinese woman who was confirmed to have osteoporosis pseudoglioma syndrome by genetic testing. This case report illustrates how to differentiate this rare clinical entity from retinoblastoma. This report also shows how to differentiate osteoporosis pseudoglioma syndrome from Norrie disease, which shares many of the ocular features. As in this patient, the early onset of osteoporosis and subsequent fractures pose a diagnostic challenge for osteoporosis pseudoglioma syndrome. Studies have shown that many such patients have been erroneously diagnosed with osteogenesis imperfecta.

Key Words: Eye manifestations; Magnetic resonance imaging; Norrie disease (pseudoglioma) protein, human; Osteoporosis; Ultrasound