A boy with blue sclera and recurrent fractures

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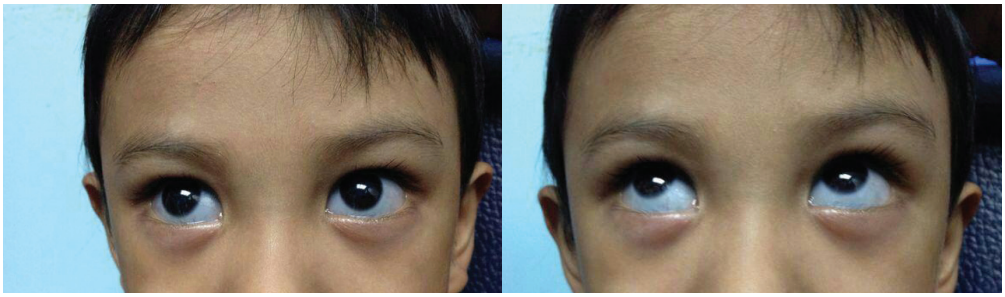
Case summary

A 5-year-old boy was presented with a recurrent history of fractures since the age of 3 years. He had 2–3 admissions per year due to fall and had a history of fractures at his right ankle, left femur and three times at his left elbow. On further questioning, it was found that he was born via spontaneous vaginal delivery. Antenatal and postnatal periods were uneventful and neonatal and development histories were similar to other siblings. No similar illnesses, history of osteoporosis, recurrent fractures or eye problem were found in the family. Examination showed that he was a thin-built boy with a normal gait, vision, speech and hearing. It was noted he had an eye abnormality (Figure 1), but the ear, nose, throat and dentition were normal. The cranial nerve and other neurological tests were also normal. There was no joint hypermobility and the other systems and pure tone audiogram were normal.



Questions

1. What is the likely diagnosis?
2. Which is the most important condition to be ruled out?
3. What is the inheritance in the patient described?
4. What is the treatment?

Answers

1. The presentation is typical of osteogenesis imperfect a (OI). It is a genetic disease with a defect in Type I collagen.[1] This can be caused either by a reduction in the synthesis of Type I collagen or the production of structurally abnormal forms of the collagen or both. The mildest form is Type I.[2] The other types of OI are Type II, III and IV. As for Type V and VI, it is due to the absence of COL1A1/2 allele mutation.[2] Type VII and VIII are newly discovered forms and inherited in recessive manner.[3]
2. One of the most important diagnosis need to be ruled out in a boy with recurrent fall is non-accidental injury (NAI). Presence of multiple fractures in various degrees of healing states should trigger a possibility of NAI.[4]
3. The inheritance pattern of Type I OI is autosomal dominant in which 50% of its defect is due to reduction in the amount of collagen Type I.[5]
4. Management will require a multidisciplinary team consisting of orthopaedic specialists (fracture), otorhinolaryngologists (hearing loss), physicians and dental surgeons.[6] Besides surgery, treatment can involve the use of pharmacological agents, psychological support and preventive medicine. Oral and intravenous biphosphonate are commonly prescribed for all types of OI.[7] Regular surveillance for hearing impairment every 3–5 years is recommended for all types of OI after adolescence as disease progression is invariable.[6]

References

1. Silence DO, Senn A, Danks DM. Genetic heterogeneity in osteogenesis imperfect. *J Med Genet*. 1979;16:101–16.
2. Glorieux FH et al. Type V osteogenesis imperfecta: A new form of brittle bone disease. *J Bone Miner Res.* 2000;15(9):1650–58.
3. Ward LM, Rauch F, Travers R, et al. Osteogenesis imperfect type VII: An autosomal recessive form of brittle bone disease. *Bone.* 2002;31:12–8.
4. Carty H, Pierce A. Non accidental injury: A retrospective analysis of a large cohort. *Eur Radiol.* 2002;12:2919–25.
5. Dalgleish R. The human collagen mutation database 1998. *Nucleic Acids Res.* 1998;26:253–5.
6. Steiner RD, Pepin MG, Byers PH. Osteogenesis imperfect. In: Pagon RA, Bird TD, Dolan DR, Stephens K, eds. *Gene Reviews.* Seattle: University of Washington; 1993.
7. Dwan K, Phillipi CA, Steiner RD, Basel D. Bisphosphonate therapy for osteogenesis imperfecta. *The Cochrane Collaboration.* England: John Wiley & Sons Ltd; 2014:2–74.