

How frequent is osteogenesis imperfecta in patients with idiopathic osteoporosis?: Case reports: Erratum

In the article, “How frequent is osteogenesis imperfecta in patients with idiopathic osteoporosis?: Case reports”,^[1] which appeared in Volume 96, Issue 35 of *Medicine*, the authors would like to provide an updated Table 1 with information essential to the article. The new version is included below.

Table 1

Describes the clinical and radiographic phenotype and the genotype of the index cases-and siblings.

Index Case	Age	Clinical presentation	Clinical phenotype	Radiologic phenotype	Siblings-clinical phenotype	Family history –affected subjects-	Genotype of the index case
Index case: 1	10-years-old-boy	Frequent elbow dislocation	Short stature (-1SD), frontal bossing, opalescent teeth and ligamentous hyperlaxity, and easy bruisability	Wormian bones, and skeletal survey revealed osteoporosis	Two sisters with normal phenotype and with no history of fractures, apart from ligamentous hyperlaxity as the dominating features	Four affected subjects: Father, two sisters and grandfather	Mutation COL1A2 p.G1078D
Index case 2	13-years-old-boy	Persistent backpain	Normal height, blue sclera and moderate ligamentous hyperlaxity and myopia	Compressive vertebral fractures of T5-10 and skeletal survey showed osteoporosis (figure 1)	One younger brother with opalescent teeth	Three affected subjects: Mother and grandmother	Mutation in COL1A1, c.3233 G>A
Index case: 3	10-years-old girl	Irritable hip pain	Blue sclera, and opalescent teeth	Wormian bones of the skull (figure 2A). 3D reformatted CT scan of the cranium showed massive sclerosis with trace of wormian bones after administration of pamidronate therapy for two years (figure 2B)	Two sisters with ligamentous hyperlaxity and blue sclerae were the dominating clinical feature-no history of fractures	Three affected family subjects: Grandmother, mother and a male cousin	Mutation in COL1A1 (c.1541G>C, Pg514A)
Index case:4	13-years-old girl	Genu varum (bowing of the legs)	Short stature, frontal and temporal bossing, blue sclera	Progressive deformity and bowing of the demineralized long bones with no fractures	One elder brother with a history of asymptomatic mitral valve prolapse	Grandfather, mother, and a maternal sister	Mutation in COL1A2, pGly634Asp
Index case: 5	9-years-old-boy	At birth she manifested unilateral hip dislocation and was treated with Pavlik harness – recently she developed scoliosis	Normal height, but opalescent teeth-myopia (-1diopter)	Wormian bones of the skull and generalized osteoporosis	One elder brother with a history of ligamentous hyperlaxity and asymptomatic mitral valve prolapse.	Three affected subjects:, a female cousin, grandmother, and grandmother sister Mutation in COL1A1, C. 1541G>=C, p. g514A	Mutation in COL1A1, C. 1541G>=C, p. g514A
Index case: 6	17-years-old girl	Frequent elbow dislocation	Obesity and ligamentous hyperlaxity	Bilateral mild bowing of the radius and ulna associated with osteoporosis	Two younger male siblings -14-years old with a history of two fractures of radius -9-years-old with ligamentous hyperlaxity and blue sclera	Four affected subjects: Mother, two sisters and a grand father	Mutation COL1A1 (c.589–2A>C
Index case: 7	14-years-old – boy	Irritable hip pain started at age of 9-years	Frontal bossing, opalescent teeth, blue sclera	Bilateral fragmentation of the capital femoral epiphyses, coxa vara and osteoporosis	One younger sister with bowing of the long bones (no history of fracture)-Echo-Cardio-doppler showed asymptomatic mitral prolapse	Four affected subjects: Grandmother Grandmother sister Two index case uncles Father showed mutation in COL1A2(p.G322S)	Homozygous Mutation in COL1A2(p.G322S)
Index case: 8	13-years-old-boy	Frequent elbow dislocations and two times fracture fingers	Opalescent teeth and generalized ligamentous hyperlaxity, and easy bruisability	Thin and demineralized long bones	Two male cousins with a history of fractures	Three affected family subjects: father, brother and grand father	Mutation in COL1A2 c.2827G>A

(continued)

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Table 1**(continued).**

Index Case	Age	Clinical presentation	Clinical phenotype	Radiologic phenotype	Siblings-clinical phenotype	Family history –affected subjects-	Genotype of the index case
Index case: 9	15-years-old-girl	Pre-adolescent scoliosis	Short stature and opalescent teeth and easy bruisability	Defective ossification of the skull associated with spinal osteoporosis	One older sister recently developed headache, and sleep apnea	Four affected family subjects: Father, two paternal siblings and grand mother Father Genotype: Carrier of the heterozygote mutation of <i>in COL1A2</i> , <i>NM_000089.3c.1801 > A,p. (Gly601 Ser)</i> <i>Male Sibling:</i> Carrier of the heterozygote mutation of <i>in COL1A2</i> , <i>NM_000089.3c.1801 > A,p. (Gly601 Ser)</i>	<i>Heterozygote mutation in COL1A2</i> , <i>NM_000089.3c.1801 > A,p. (Gly601 Ser)</i>
Index case: 10	16-years-old -boy	Scoliosis	Short stature, opalescent tooth	Defective ossification of the skull, and demineralized spine	Two younger sisters with ligamentous hyperlaxity	Three affected family subjects: father, uncle and grand father	<i>Mutation in COL1A1 (17q21.31-q22) OI type I</i>
Index case: 11	14-years-old-girl	History of frequent elbow fractures	Opalescent teeth, and generalized ligamentous hyperlaxity	Mild platyspondyly and osteoporosis	One younger brother was born with bilateral hip dislocation	Two affected family subjects: Father and grandmother	<i>Mutation in COL1A1 (c.4103A > G, p.Y1368C)</i>

Reference

- [1] Al Kaissi A, Windpassinger C, Chehida FB. How frequent is osteogenesis imperfecta in patients with idiopathic osteoporosis?: Case reports. *Medicine*. 96;35: e7863.