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A family with homozygous and heterozygous p.Gly337Ser mutations in *COL1A2*

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ABSTRACT

Osteogenesis imperfecta (OI) is commonly caused by monoallelic mutations in *COL1A1* or *COL1A2*. Biallelic mutations are extremely rare. Only five previous reports have identified seven OI patients with homozygous mutations in *COL1A2*. OI is a genetically and phenotypically heterogeneous disorder which challenges an establishment of genotype-phenotype correlation. Notably, more than thirty patients with OI possess the heterozygous mutation, p.Gly337Ser, in *COL1A2*. Their clinical severity ranges from mild OI type I to severe types III and IV. Here, we report a 17-year-old Thai female with recurrent bone fractures, short stature, blue sclerae, triangular face, missing teeth, dentinogenesis imperfecta (DI), skeletal deformities, and scoliosis. She was diagnosed with OI type III. Her parents were second-cousin-once-removed. The father was a professional Thai boxer. Both had normal bone mineral density, no history of bone fractures, and only teeth problems. They were diagnosed with DI without OI. Whole exome sequencing identified that the proband harbored the homozygous mutation, c.1009G > A (p.Gly337Ser), in exon 19 of *COL1A2* while her parents were heterozygous for this mutation. This study reports the eighth child with OI and the homozygous mutation in *COL1A2*; and the first two individuals with the heterozygous p.Gly337Ser mutation in *COL1A2* causing an isolated DI without OI.

1. Introduction

Osteogenesis imperfecta (OI) is a rare heritable disease characterized by bone fragility and deformity. The incidence of OI is 1/10,000–20,000. Common clinical manifestations are short stature, dentinogenesis imperfecta (DI), blue sclerae, hearing loss, and ligamentous laxity (Astrom et al., 2010; Marini et al., 2017). OI has been associated with mutations in at least 17 genes, inherited in an autosomal dominant, autosomal recessive, or X-linked manner (Lindert et al., 2016; Marini et al., 2017). Predominantly, it is caused by monoallelic mutations in *COL1A1* and *COL1A2,* which encode type I collagen. Of those, the heterozygous glycine substitution in the collagen triple helix is the most common type of mutation and leads to misfolding and over-modification of type I procollagen (Marini et al., 2017). Biallelic mutations in *COL1A1* and *COL1A2* are rare. Only 5 families affected with OI have been reported to be homozygous for mutations in *COL1A2* (OMIM *120160) (https://www.le.ac.uk/genetics/collagen/)(Costantini et al., 2018; De Paepe et al., 1997; Nicholls et al., 1984, 2001; Pihlajaniemi et al., 1984). Among them, only 2 families possessed glycine substitutions in *COL1A2* (Costantini et al., 2018; De Paepe et al., 1997).

To date, many studies have demonstrated genotype-phenotype correlation in OI patients (Ben Amor et al., 2011; Forlino et al., 2011; Lindahl et al., 2015; Maioli et al., 2019). However, a solid relationship is difficult to establish due to complexity of causative variants and clinical manifestations of OI. To the best of our knowledge, DI without OI has never been linked with collagen type I mutations.

Here, we report a Thai trio. A Thai woman was homozygous for the mutation c.1009G > A (p.Gly337Ser) in *COL1A2*. She was affected with OI type III (OMIM #259420) and DI whereas her parents who

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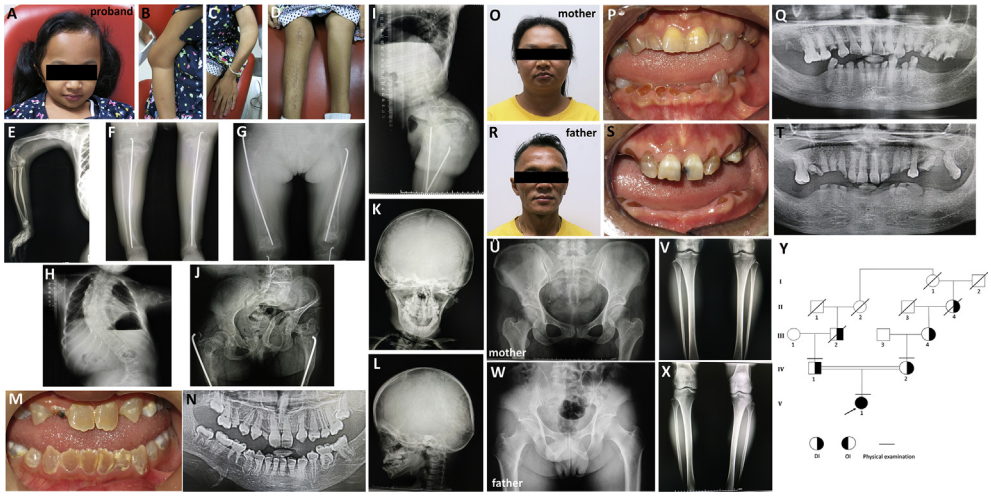

Fig. 1. Clinical and radiographic features of the proband and her parents. A-D. Photographs showed triangular face, blue sclerae, and limb deformities. E-G. Radiographs showed curvature of long bones. Her lower extremities were repaired with rods. H-J. Scoliosis and lumbar lordosis were present. K-L. Skull radiographs showed wormian bones and class III malocclusion. M-N. Her teeth exhibited dentinogenesis imperfecta. Permanent upper lateral incisors and right second premolars were absent. O-X. Clinical and radiographic features of the proband's parents. Both mother and father had dentinogenesis imperfecta. Their teeth were severely deteriorated and pulp cavities were obliterated. Their long bones and pelves were unremarkable. Y. Pedigree of the family.

were heterozygous for the mutation had only DI without OI.

2. Clinical report

2.1. Patients' characteristics

The proband, a 17-year-old Thai woman, was diagnosed with OI type III and DI. She was born at term. Her birth weight was 2800 g. At birth, she had deformities and fractures of upper and lower extremities. Intravenous pamidronate therapy was initiated at the age of 1 year and continued every 3 months. During the first 9 years of her life, the patient had experienced twenty two times of bone fractures, particularly the humeri and femora, and had been wheelchair-bound due to bone deformity. Physical examination at 13 years of age showed short stature, occipital-frontal circumference of 53 cm, triangular face, blue sclerae, DI, missing upper lateral incisors and right second premolars, barrel chest, and severe bone deformities (Fig. 1A–D). Full-body radiographs revealed diffuse osteopenia, wormian bones, class III malocclusion, severe bending of right humerus, curvatures of ulnae and radii, rods in femora and tibiae, scoliosis, and lumbar lordosis (Fig. 1E-L). Panoramic radiograph showed bulbous crown. Pulp cavities of erupting teeth were large while those of erupted teeth became obliterated (Fig. 1M, N). Bone mineral density of lumbar spine was 0.404 g/cm^2. The height-for-age Z-score (HAZ) was applied to predict bone Z-score (Nakavachara et al., 2014; Zemel et al., 2010). The $BMD_{haz}Z$ was −0.08. Her vision and hearing were normal. At 17 years of age, her weight was 18.3 kg and height was 94 cm.

The proband's parents were second-cousin-once-removed. They never had bone fractures. Interestingly, the father was a previous professional Thai boxer. Oral examination revealed that they had DI. There teeth were severely deteriorated and showed obliterated pulp cavities (Fig. 1O–Y). The height of the father was 157 cm and the mother was 150 cm. Lumbar spine BMD of the father at 43 years of age was 0.724 g/cm^2 (z-score −1.7) and of the mother at 37 years of age was 0.972 g/cm^2 (z-score −0.2).

2.2. Genetic analyses

The study was approved by the institutional review board, Faculty of Medicine, Chulalongkorn University (IRB# 346/61). The written informed consents were obtained from all participants. Genomic DNA extracted from peripheral blood leukocytes was subjected for mutation analysis using whole exome sequencing (WES) (Intarak et al., 2019). Briefly, genomic DNA was captured using a SureSelect Human All Exon version 4 kit (Agilent Technologies, Santa Clara (CA), USA) and sequenced using Hiseq2000 (Macrogen, Seoul, Korea). The sequences were aligned to the human genome reference sequence (UCSC Genome Browser, hg19) using Burrows-Wheeler Aligner (http://bio-bwa.sourceforge.net/). Downstream processing was carried out with SAMtools (samtools.sourceforge.net/) and annotated against dbSNP and 1000 Genomes. After quality filtering, the variants were screened using a list of OI genes (Table S1). All calls with coverage < 10x; minor allele frequency ≥1% in 1000 Genomes Project, Exome Aggregation Consortium database (exac.broadinstitute.org), and in-house database of 1876 unrelated Thai exomes; non-coding variants; and synonymous exonic variants were filtered out. The identified variants were confirmed by Sanger sequencing (Table S2).

Exome and Sanger sequencing identified that the proband possessed the homozygous missense mutation, c.1009G > A (p.Gly337Ser), in exon 19 of *COL1A2* (NM_000089.3, NP_000080.2) (Table S1). The variant has been submitted to the ClinVar database (http://www.ncbi.nlm.nih.gov/clinvar/) (SCV001134943.1). Sanger sequencing revealed that the father and mother were heterozygous for the mutation (Fig. S1). More than 30 patients with OI type I, III, and IV have been reported to harbor the heterozygous c.1009G > A mutation in *COL1A2* (https://oi.gene.le.ac.uk). Pathogenic variant in the dentin sialophosphoprotein gene (*DSPP*, OMIM *125485) was not detected in the proband.

3. Discussion

A female patient harboring the homozygous mutation, c.1009G > A (p.Gly337Ser), in *COL1A2* was identified. She had several bone fractures, short stature, low BMD, blue sclerae, and DI, suggesting OI type III. Her parents having the heterozygous mutation, c.1009G > A, in *COL1A2* had only DI. The features of this family are unique.

Majority of OI cases are caused by monoallelic mutations in *COL1A1* or *COL1A2* which are *de novo* or inherited in an autosomal dominant manner (Marini et al., 2017). Biallelic mutations in *COL1A2* leading to OI are rare. Five families affected with OI have been reported to be homozygous for mutations in *COL1A2* (Table 1) (Costantini et al., 2018; De Paepe et al., 1997; Nicholls et al., 1984, 2001; Pihlajaniemi et al., 1984). Of these, only two families possessed homozygous glycine substitutions in *COL1A2* (Costantini et al., 2018; De Paepe et al., 1997). We noticed that the index patient who was homozygous for p.Gly202Ser did not have DI (Costantini et al., 2018) whereas the patient who was homozygous for p.Gly337Ser (this study) and the one who was homozygous for p.Gly841Ser (De Paepe et al., 1997) had DI. In addition, the phenotypes of their heterozygous parents were also different: the heterozygous p.Gly202Ser parents did not have either OI or DI; those with heterozygous p.Gly337Ser had DI without OI; and those with

Table 1
Summary of phenotype and genotype of the patients with homozygous mutations in *COL1A2* and their heterozygous carriers identified in this study and previously reported associated with OI.

Homozygous patients	Homozygous mutations in *COL1A2*	Exon	Age (y)	Sex	Diagnosis reported	Short stature	No. of peripheral fractures	No. of vertebral fractures	Bowing of extremities	BMD lumbar spine	Blue sclerae	DI	Other clinical features
This study	c.1009G > A, p.Gly337Ser	19	17	F	OI type III	+	22	0	+	z-score −5.6	+	+	missing upper lateral incisors and right second premolars
Costantini et al. (2018) (II-4)	c.604G > A, p.Gly202Ser	14	31	F	OI type IV	+	> 10	3	+	z-score −2.2	+	–	–
(II-3)	c.604G > A, p.Gly202Ser	14	38	F	OI type IV	+	> 4	NA	NA	z-score −1.3	+	+	–
De Paepe et al. (1997) (IV-5)	c.2521G > A, p.Gly841Ser	40	6	F	OI type III	+	1	0	+	NA	+	+	asymmetric head, osteopenia, undermineralized calvarium and wormian bones, enlarged fontanel, hyperlaxity of the small joints
(IV-6)	c.2521G > A, p.Gly841Ser	40	4m	F	OI type III	+	0	0	+	NA	NA	NA	large head
Nicholls et al. (2001) (III-6)	c.3105+2T > C	Intron46	9	F	OI/EDS	–	3	0	–	NA	+	–	EDS, joint laxity, pes planus and valgus heels leading to a secondary shortening of the achilles tendon
Pihlajaniemi et al., 1984 Nicholls et al., 1984 (VI-1)	c.4001_4004del, p.Asn1334Serfs*34	52	5	M	OI type III	10th centile	severe	+	+	NA	+	–	osteoporosis, right humeral pseudoarthrosis, popcorn expansion of knee joint, vertebral collapse, anteroposterior compression and wormian bones of lateral skull, hypermobile fingers, severely twist of lower limbs
https://oi.gene.le.ac.uk **(Sheffield, UK)**	c.2175_2187 + 14dup	36	NA	NA	OI type IV	NA	NA	NA	NA	NA	NA	NA	–

Heterozygous carriers	Heterozygous mutations in *COL1A2*	Age (y)	Sex	Condition reported	Height (cm)	No. of peripheral fractures	No. of vertebral fractures	Bowing of extremities	BMD lumbar spine	Blue sclerae	DI	OI features
This study	c.1009G > A, p.Gly337Ser											
Proband's father		43	M	unremarkable	157	0	0	–	z-score −1.7	–	+	–
Proband's mother		37	F	unremarkable	150	0	0	–	z-score −0.2	–	+	–
Costantini et al. (2018)	c.604G > A, p.Gly202Ser											
Father (I-1)		68	M	normal BMD	172	0	0	–	T-score −0.1	–	–	–
Mother (I-2)		59	F	osteopenia	158	0	0	–	T-score −2.1	–	–	–
Sibling 1 (II-1)		42	F	unremarkable	157	0	0	–	z-score −0.2	–	–	–
De Paepe et al. (1997)	c.2521G > A, p.Gly841Ser											
Father (III-5)		36	M	mild OI	150	2 after trauma	0	+	−1.4 SD	–	NA	osteopenia, triangular face, varus deformity and reduced mobility of the hips and mild bowing of the lower legs, severe coxa vara, platyspondyly of the lumbar vertebrae
Mother (III-6)		38	F	mild OI	147	0	0	–	−2.5 SD	–	NA	Osteopenia, diffuse articular pain.
Sibling (IV-1)		17	F	mild OI	< 3rd centile	0	0	–	84% normal value	mild	NA	osteopenia, triangular face, hyperlaxity of the finger joints
Sibling (IV-3)		10	M	mild OI	< 3rd centile	0	0	–	NA	mild	NA	triangular face, hyperlaxity of the finger joints

(*continued on next page*)

Table 1 (*continued*)

Homozygous patients	Homozygous mutations in *COL1A2*	Exon	Age (y)	Sex	Diagnosis reported	Short stature	No. of peripheral fractures	No. of vertebral fractures	Bowing of extremities	BMD lumbar spine	Blue sclerae	DI	Other clinical features
Nicholls et al. (2001)	c.3105+2T > C												
Father (II-6)			NA	M	NA	NA	NA	NA	NA	NA	NA	NA	NA
Mother (II-7)			NA	F	joint laxity	NA	NA	NA	NA	NA	NA	NA	–
Pihlajaniemi et al., 1984 Nicholls et al., 1984	c.4001_4004del, p.Asn1334Serfs*34												
Father (V-1)			NA	M	unremarkable	NA	NA	NA	NA	NA	NA	NA	NA
Mother (V-2)			NA	F	unremarkable	NA	NA	NA	NA	NA	NA	NA	NA
https://oi.gene.le.ac.uk (Sheffield, UK)	c.2175_2187 + 14dup												
Father			NA	M	mild OI type I	NA	NA	NA	NA	NA	NA	NA	NA
Mother			NA	F	mild OI type I	NA	NA	NA	NA	NA	NA	NA	NA

OI, osteogenesis imperfecta; DI, dentinogenesis imperfecta; NA, not available; m, month; M, male; F, female; No., number; +, present; -, absent.

heterozygous p.Gly841Ser had OI without DI. These suggest that the positions of mutations along the collagen chain affect clinical manifestation. Consistent with the p.Gly202Ser carrier, the parents reported here did not have bone fragility and other OI features. It has been shown that mutations in the triple helical N-terminus of collagen chains have milder effect on triple helix stability and milder clinical severity than those in the C-terminus (Marini et al., 2017). Both Gly202 and Gly337 locate close to the N-terminus. These may contribute to the absence of OI features in the parents in this study.

In this study, all three members were affected with DI. This is consistent with a previous observation that substitutions beyond the first 120 amino acid residues of collagen type I triple helix may cause DI (Ben Amor et al., 2011). However, DI without OI has never been associated, as far as we know, to collagen type I mutations. DI with normal bone features is caused by mutation in the *DSPP* gene (Porntaveetus et al., 2018). Surprisingly, the proband's parents who were heterozygous for p.Gly337Ser had only DI. The father was a professional Thai boxer. Both had normal BMD and never had bone fractures indicate that they did not have bone fragility or pathognomonic feature of OI. The p.Gly337Ser in *COL1A2* is considered to be the mutation hotspot for human OI (Zhang et al., 2016). It has been identified in a number of patients with OI type I, III, and IV and with different ethnic backgrounds (Caparros-Martin et al., 2017; Ho Duy et al., 2016; Marini et al., 2007; Zhang et al., 2016). The pathomechanisms to explain why the parents do not have OI need further investigation.

This study demonstrates unique phenotypes of a family associated with homozygous and heterozygous mutations in *COL1A2*. The proband is the fifth individual affected with OI who has been found with homozygous glycine substitution in *COL1A2*, while her parents are the first two individuals having heterozygous p.Gly337Ser mutation in *COL1A2* and isolated DI.

CRediT authorship contribution statement

Wandee Udomchaiprasertkul: Investigation, Writing - original draft. **Chulaluck Kuptanon:** Methodology, Writing - review & editing. **Thantrira Porntaveetus:** Conceptualization, Writing - original draft, Visualization. **Vorasuk Shotelersuk:** Conceptualization, Supervision.

Declaration of competing interest

None of the authors have any conflicts to declare.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.ejmg.2020.103896.

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