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"Osteogenesis Imperfecta, Type Xvii" - A Rare Genetic Entity

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ABSTRACT

Introduction: Osteogenesis imperfecta (OI) is a genetic disorder with the incidence of 1 in 15,000 to 20,000 births. Around 85%–90% are dominant mutations in collagen type I α1 (COL1A1) or COL1A2 genes, while a plethora of other genes have been associated with non-collagen OI in recent years. Osteogenesis imperfecta type XVII is described as an autosomal recessive form which is rare, occurring due to mutation in SPARC gene on chromosome 5q33, characterized by bone deformity, significant reduction of bone density and short stature.

Case Report: A 7 year male child product of non-consanguinous marriage presented with complaints of multiple bony deformities in the form of forward bowing of bilateral lower limbs, outward bowing of bilateral upper limbs, suggestive of multiple fracture lines and severe osteopenia along with thinned out cortex on skeletal survey. Although metabolic profile was normal. Child also had short stature and cross bite teeth.

Child was diagnosed to have OI based on characteristic clinico-radiological features and thereafter whole genome sequencing was done to know the subtype. Whole genome sequencing was suggestive of Osteogenesis imperfect type XVII (SPARC mutation) on Exon 9.

Conclusion: This case is being reported because of the rarity of OI type 17 especially in Indian population. Besides, the aim was also to review the literature to highlight the challenges and recent advances in the management. There is a great need for further research to decrease the morbidity associated with this disease and also to find a cure for the same.

1. INTRODUCTION

Osteogenesis imperfecta (OI) is an inherited connective tissue disorder marked by reduced bone density and mass, leading to increased bone fragility and frequent fractures over time, often resulting in bone deformities and joint instability. The condition can manifest in different ways, including dentinogenesis imperfecta, blue sclerae, short stature, progressive hearing loss, and cardiac abnormalities. [1,2] Osteogenesis imperfecta (OI) or brittle bone disease is a genetic disorder occurring in 1 in 15,000 to

20,000 births. [3] Around 85%–90% are dominant mutations in collagen type I α1 (COL1A1) or COL1A2 genes, while a plethora of other genes have been associated with non-collagen OI in recent years.[4] Osteogenesis imperfecta OI type XVII is described as an autosomal recessive form which is rare, characterized by bone deformity, significant reduction of bone density, short stature, and, in some patients, blue sclera. We hereby present a case of osteogenesis Imperfecta Type XVII who was diagnosed on the basis of characteristic clinico-radiological features and confirm by whole genome Sequencing to know the subtype.

2. CASE REPORT

A 7 year male child product of non-consanguinous marriage presented with complaint of multiple bony deformities. Child was apparently well till one and half years of age, bony deformities appeared which increased progressively over a period of time. Motor milestone were delayed however other are normal.

His height was < 3rd Percentile for his age.

The general physical examination reveals forward bowing of both lower limbs, outward bowing of both upper limbs, a depressed lambdoid suture with frontal bossing, conjunctival xerosis at stage X1A, and dental malocclusion.

Skeletal survey showed generalized osteoporosis, thinned out cortex, thin multiple fracture lines, over tubulation of metacarpals, metatarsals and phalanges along with vertebra plana.

Child was diagnosed to have osteogenesis Imperfecta Type XVII clinico-radiologically, and confirmed by Whole Genome Sequencing.

3. DISCUSSION

In the present case, the patient had bony deformities, short stature with dental malocclusion - meeting the diagnostic criteria of Osteogenesis imperfecta. The radiological images were suggestive of brittle bones.

Differential diagnosis are Juvenile Idiopathic Osteoporosis , Cole-Carpenter Syndrome, Hypophosphatasia, Achondroplasia, and Ehlers–Danlos syndrome.Physiotherapy, rehabilitation and orthopedic surgery are the mainstays of management.Bisphosphonates can also be helpful .Surgical intervention is done when medical therapy fails.Bone marrow and stem cell transplantation, in addition to gene-based therapy, provide potential cures for OI. However, these approaches are currently not ready for clinical trials.

4. CONCLUSION

Patient with Osteogenesis imperfecta mainly have brittle bones due to mutation in collagen synthesis leading to bony deformities with or without grey/ blue sclera. Follow up and multidisciplinary approach. Prenatal diagnosis can be done by ultrasound scans. Gene mutation analysis is of paramount importance for accurate classification. Genetic counselling should be offered to all such patients.

Images:

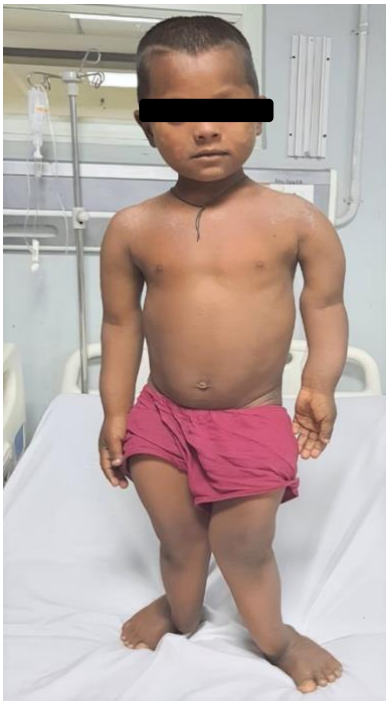
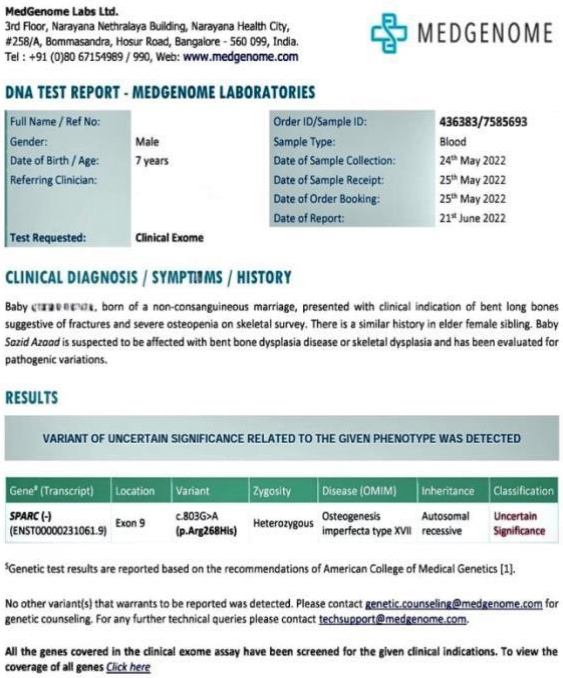


Fig. 1: Clinical examination of child showing bowing of bilateral upper and lower limbs.



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MEDGENOME

DNA TEST REPORT - MEDGENOME LABORATORIES

Full Name / Ref No:		Order ID/Sample ID:	**436383/7585693**
Gender:	Male	Sample Type:	Blood
Date of Birth / Age:	7 years	Date of Sample Collection:	24th May 2022
Referring Clinician:		Date of Sample Receipt:	25th May 2022
		Date of Order Booking:	25th May 2022
		Date of Report:	21st June 2022
Test Requested:	**Clinical Exome**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Baby [redacted], born of a non-consanguineous marriage, presented with clinical indication of bent long bones suggestive of fractures and severe osteopenia on skeletal survey. There is a similar history in elder female sibling. Baby *Sazid Azaad* is suspected to be affected with bent bone dysplasia disease or skeletal dysplasia and has been evaluated for pathogenic variations.

RESULTS

VARIANT OF UNCERTAIN SIGNIFICANCE RELATED TO THE GIVEN PHENOTYPE WAS DETECTED

Gene[1] (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
***SPARC* (-)** (ENST00000231061.9)	Exon 9	c.803G>A **(p.Arg268His)**	Heterozygous	Osteogenesis imperfecta type XVII	Autosomal recessive	Uncertain Significance

[1]Genetic test results are reported based on the recommendations of American College of Medical Genetics [1].

No other variant(s) that warrants to be reported was detected. Please contact genetic.counseling@medgenome.com for genetic counseling. For any further technical queries please contact techsupport@medgenome.com.

All the genes covered in the clinical exome assay have been screened for the given clinical indications. To view the coverage of all genes *Click here*

Fig.2: Genetic study reveal Osteogenesis imperfect type XVII.

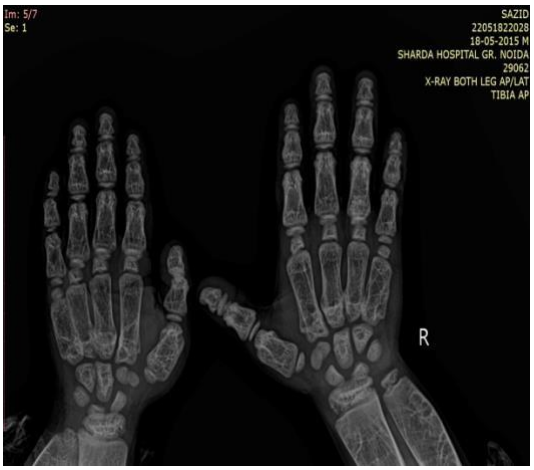

Fig 3: Image showing Metacarpals, metatarsals and phalanges are overtubulated.

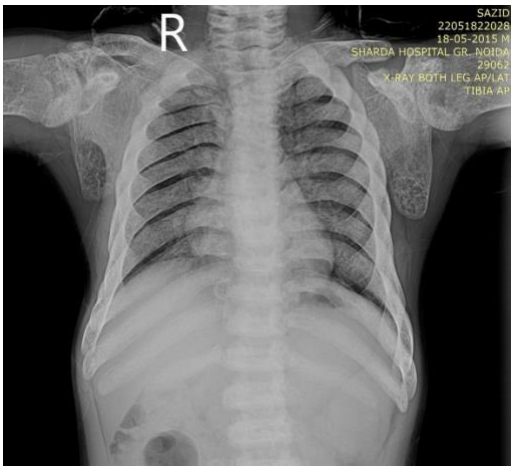

Fig 4: Image showsing vertebra plana.

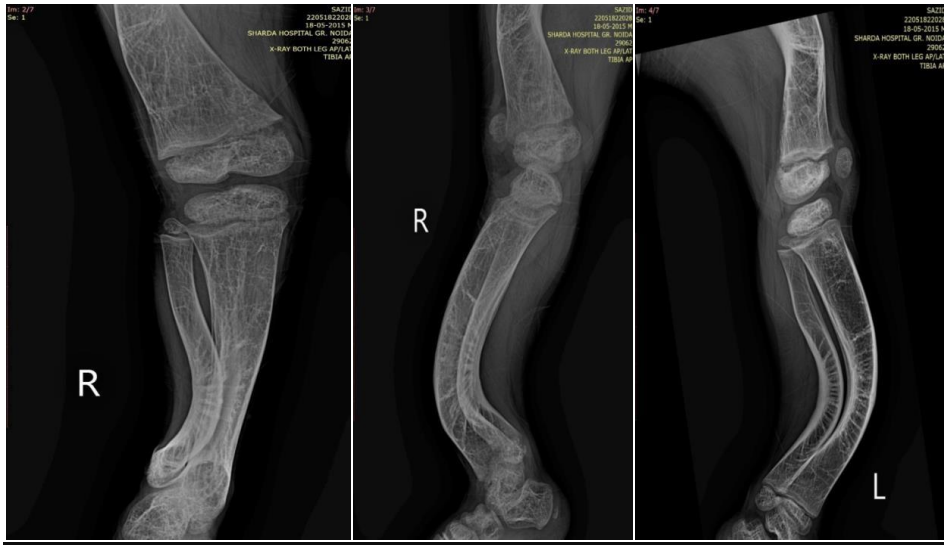

Fig.5: Long bones are showing generalized osteoporosis, thinned out cortex, severe deformity, multiple thin fractures lines are seen.

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