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Medicine

OSTEOGENESIS IMPERFECTA TYPE I PRESENTING AS SHORT STATURE: A CASE REPORT

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ABSTRACT Osteogenesis imperfecta (OI), a secondary cause of osteoporosis and bone fragility principally manifests as recurrent fracture in childhood. Other major clinical features may include skeletal deformity (short stature), blue sclerae, hearing loss and fragile, opalescent teeth. Here we report the case of 28 year old female patient presenting with short stature, bone deformity, blue sclerae and abdominal lump.

KEYWORDS : Osteogenesis imperfecta, bone fragility, short stature, blue sclera and abdominal lump.

INTRODUCTION

Osteogenesis imperfecta (OI) is a genetic syndrome that is characterized by recurrent long bone fractures in childhood, low bone mass, blue sclera and dentinogenesis imperfecta (DI) (1). It is a rare disorder with an overall incidence of ~1 in 10,000-20,000 births. Their most common pattern of inherited autosomal dominant but recessive inheritance is also seen. Molecular defects seen in type I collagen genes, COL1A1 and COL1A2, were seen in 90% of cases.2 10-15% of all OI are linked to defects other than these two regions like CRTAP, LEPRE1, FKBP10, and SERPINF1.3 Mutations in the a1 (I) or a2 (I) chain, especially glycine (Gly) to cysteine or tryptophan substitutions in the Triple helix is usually seen. (2) Different forms of the disease may have platelet dysfunction, cardiac and respiratory difficulties due to rib cage deformities.(3) A prevalence rate of hearing loss of 46% has been reported.(4)

Case report;

A 28 year old short stature female patient presented with h/o abdominal pain, swelling and loss of appetite since 2 month. The swelling was progressively increasing since 2 month.

The parents were non-consanguineous and healthy and there was no family history of bone disease. She was borne by full term normal vaginal delivery and there was no asphyxia at birth. She attained menarche at the age of 15 year and her menstrual cycles were normal. Patient had history of left femur fracture at the age of 1.5 year after minor impact which required almost 3 months for healing.

She has Short stature (99cm), triangular facies, frontal bossing present, bluish grey colour of sclera, Malocclusion of teeth (Dentinogenesisimperfecta) and lump in abdomen arising from right pelvis region extending up to umbilicus.

Her laboratory investigation including complete blood count, KFT, LFT, Sr .calcium (11mg/dl), alkaline phosphates and CA-125 were unremarkable. Her USG Abdo-pelvis showed solid hypo echoic mass of size 9.8 × 7.7 cm in right renal fossa causing displacement of right kidney inferiorly (claw sign) ,which was confirmed on CECT Abdo-pelvis showing 9.2 ×11.2 × 9.1 cm sized minimally enhancing mass in right lumbar region which can´t be separated from right ovary (Ovaries R- 4.1 × 2.6 cm .L-2.8 ×1.8 cm). Right kidney - 3.7 ×1.7 cm,Left kidney -10.7 ×4.7 cm).

She does not have any hearing loss and her intellectual development also normal. Cardiac examination (including ECG and 2d echo) and neurological examination did not reveal any abnormality. An X-ray of the limb and spine showed diffuse low bone density, the absence of Wormian bones. The treatment given was calcium (1,000 mg/day) + vitamin D (800 U/day).

Figure 1-Short stature (99cm)

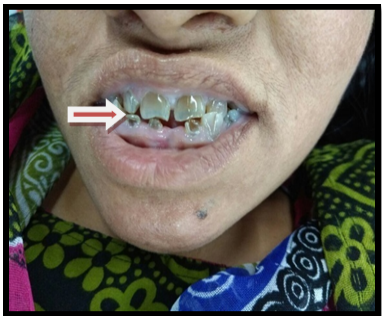

Figure 2-Malocclusion of teeth (Dentino-genesisimperfecta)

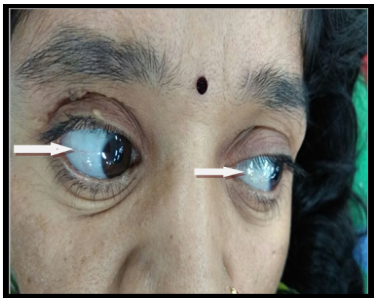

Figure 3-Bluish grey discolouration of sclera (arrowheads),

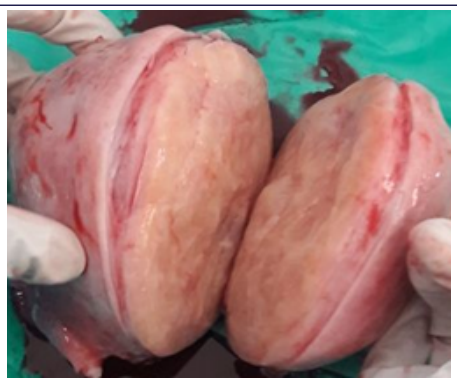

Figure 4: **ovarian mass**

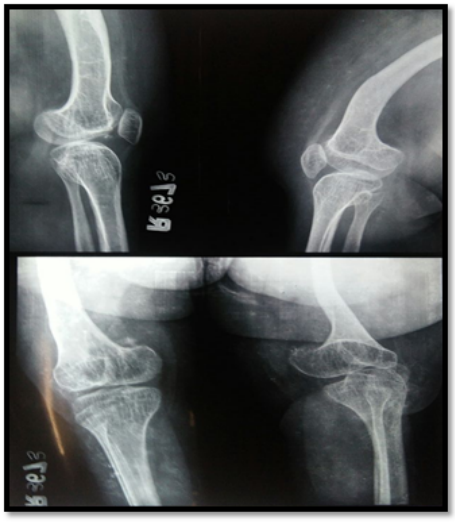

Figure 5: showing severe osteoporosis

DISCUSSION

In our case patient belong to type I OI, presented with abdominal lump. On examination she has short stature, blue sclera, abnormal dentition (Dentino-genesis imperfecta) and history of fracture. In our case patient don't have hearing loss and she presented as abdominal lump (ovarian mass) for that she was referred to gynecologist for further treatment. Patient underwent operative procedure and mass was removed, which was fibro-Thecoma confirmed histopathologically. We didn't find any literature of ovarian mass associated with OI; our finding may be incidental association. Particular pattern of inheritance is not shown by the subject. Probably it is a case of sporadic mutation.

Osteogenesis imperfect also called as brittle bone disease, results from mutations in genes encoding for type I collagen.Collagen is the major structural protein in bone, ligaments, tendons, skin, sclera, and dentin.3 Mutant expression produces non-functional collagen (severe OI) or insufficient quantities of collagen (mild OI).(5-8)

Table No. 1 .Sillence classification (9-12)

Type of OI	Clinical feature
I (mild, most common)	Short stature, prepubertal recurrent fracture, blue sclera, dentinogenesis-imperfecta.
II (very severe)	Usually fatal,with death secondary to intracranial hemorrhage or respiratory insufficiency caused by incompetency of the rib cage; the infant is stillborn or lives only a short time.
III (sever)	Short stature, kyphoscoliosis, recurrent fracture, spinal deformity, normal sclera
IV(moderate)	Short stature, skeletal deformity .normal sclera
V	moderate deforming with normal teeth and sclera
VI	moderate disease with fish scale pattern of bone lamellation, normal sclera and teeth
VII	clinically similar to type II, with the exception that the patients have a smaller head and normal sclera
VIII	defects in growth and mineralization present

Borland and Gaffey (2012) described that fractures in OI patients heal at a normal rate; but have a poor quality callus. Repeated fractures, typically lead to progressive deformity (both shortening and angular). This is due to the poor quality callus being easily deformed by weight bearing forces (13). Our patient also has history of childhood fracture and bony deformities in upper and lower limbs.

The molecular genetic test used for clinic diagnosis confirmation and mutations in COL1 A1 and COL1 A2 genes responsible of synthesis of type I procollagen and confirmed the disease osteogenesis imperfect, but in our case it was not possible due affordability of patient.

As there is no proven treatment which has clear cut benefit. Treatment should be individualized and optimization of OI treatment in adults remains a challenge, because available treatment does not target the underlying collagen defect. Treatment of OI consists of supportive medical management to reduce the occurrence of fractures like bisphonate treatment specifically in childhood, calcium supplementation, vit. D3, physical and occupational therapy to promote gross motor development and maximize functional independence, and surgery to stabilize bones and correct deformities. As our patient is adulthood we have started calcium and vit. D 3. As age OI patient progresses risk of fracture decreases.(2)

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