

Bilateral Neck Femur Fractures, Rickets And Severe Osteoporosis In Oculocerebrorenal Syndrome Of Lowe

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Citation

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Abstract

The Lowe syndrome is constellation of eye, central nervous system and kidney involvement. We report about a child with bilateral neck femur, rickets and severe osteoporosis in Lowe syndrome.

INTRODUCTION

Oculocerebrorenal syndrome was first described by Charles Lowe in 1952 [1]. It is rare X-linked recessive disorder involving primarily kidneys, eyes and the nervous system. The non specific radiological features-the white matter abnormalities in MRI have been reported in Lowe syndrome [2]. We describe a bilateral neck femur fracture, severe osteoporosis and active rickets in Lowe syndrome.

CASE REPORT

A five and half year old boy presented with failure to thrive and multiple deformities of his limbs. He was the second child of non-consanguineous parents born of an uncomplicated pregnancy at term. He was operated for congenital bilateral dense cataract at 3 years. The child had history of delayed developmental milestones and seizures. On examination, he had short stature, frontal bossing, prominent parietal prominence, hypotonia, corneal scarring, delayed eruption of all primary teeth, delayed deep tendon reflexes, severe mental retardation, pectus carinatum, rachitic rosary, Harrison sulcus, protuberant abdomen, wrist widening, as well as bowing of the both femur and tibia.

Skeletal survey revealed bilateral neck femur fractures (figure 1), generalized osteoporosis, active rickets, delayed bone age (figure 2) and dorsolumbar kyphosis (figure 3). There was no cervical spine instability was noted. Investigation showed normal calcium, hypophosphatemia, raised alkaline phosphate, low sodium and potassium, dilute urine, hypercalciuria, phosphaturia, proteinuria, aminoaciduria. Blood gas analysis showed metabolic acidosis with respiratory compensation. Ultrasound abdomen

was normal. The Oculocerebrorenal syndrome was diagnosed based on clinical and laboratory data.

Figure 1

Figures 1 Pelvic radiograph shows active rickets with bilateral pathological neck femur fractures with poor healing and bone demineralization



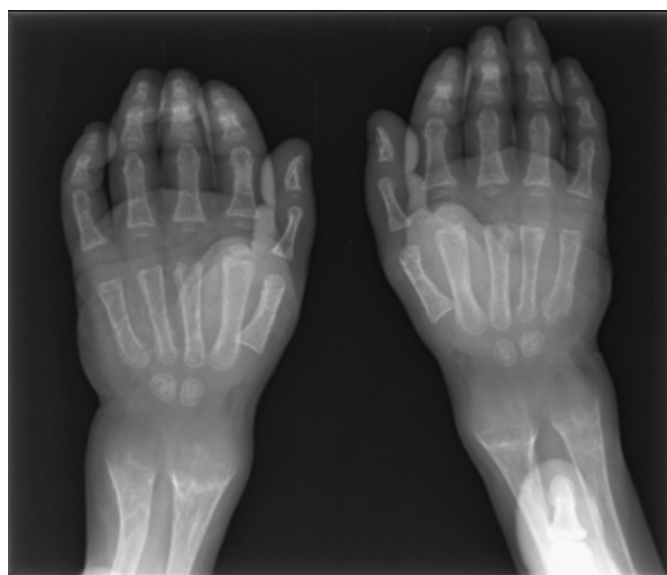
Figure 2

Figure 2 Thoracolumbar spine radiograph shows generalized osteoporosis, mild platyspondyly with dorsolumbar kyphosis



Figure 3

Figure 3 Wrist radiograph shows widening of the growth plate, fraying, splaying and cupping of metaphysis, poor mineralization with two carpal ossifications.



DISCUSSION

Fractures of the neck of the femur in children are extremely rare (1%) [3]. The triad of bilateral neck femur fractures, active rickets and osteoporosis are extremely rare. Hypophosphatasia, renal osteodystrophy, osteogenesis imperfecta, non-accidental trauma, Fanconi syndrome, Cole-Carpenter syndrome, Hadju-Cheney syndrome, Idiopathic juvenile osteoporosis must be considered as differential diagnosis of such a rare entity. Clinical and laboratory investigation are helpful to differentiate between all above mentioned pathology.

Lowe syndrome is constellation of kidneys, eyes and the

nervous system involvement. Cataract is present at birth in all patients and glaucoma is detected within first few years. A renal disease is characterized by proximal renal tubular dysfunction. Delayed motor development, mental retardation and seizure are usual characteristics of Lowe syndrome [4]. The musculoskeletal abnormalities are hypotonia, areflexia, joint hypermobility, osteomalacia, recurrent fractures, rickets, tenosynovitis and joint effusions and contractures [5, 6]. Radiological features described in Lowe syndrome are rickets, multiple fractures, scoliosis, kyphosis, platyspondyly, cervical spine abnormalities and subluxated/dislocated hip [6].

We recommend inclusion of the bilateral femur neck fractures in pathological fractures in inadequately or untreated Lowe syndrome. Osteopenia and pathological fractures should be prevented by correct treatment of rickets.

References

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