Case Report

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Failure of treatment of Coxa Vara in Gauchers Disease: A Case Report

Orthopaedics Section

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ABSTRACT

Gaucher's disease (GD) is an autosomal recessive storage disorder which occurs due to the deficient functioning of the lysosomal hydrolase enzyme [1, 2]. In this case report, coxa vara occurred after the union of a stress fracture in the basicervical and the intertrochanteric region of the left femur of a male child who had Gaucher's disease. Femoral Valgus osteotomy and fixation with cannulated screws was done. The disease showed a progressive course even after the surgery, resulting in refracture, increase in the coxa vara and shortening of the limb. An incidental association of Perthe's disease in the contralateral hip was seen.

Key Words: Gaucher's disease, Coxa vara, Osteotomy, Treatment failure, Perthe's disease

CASE REPORT

This is a case of a male child who developed coxa vara of the left hip after a stress fracture in the neck of the femur due to Gaucher's disease. The disease was diagnosed after a thorough investigatory exercise in 2006, when at the age of 2 years, the child presented with failure to thrive, recurrent abdominal distension and bone pain. Splenectomy was done a year after the diagnosis. In December 2009, the patient presented with the complaints of pain in the left hip and limp for a few days. He had a trivial trauma around one year back. Clinically, there was an irregular bony prominence at the level of the left greater trochanter. The hip region showed no signs of sepsis, fracture or dislocation. The patient walked with a short limp gait, with a true shortening of 2.5 cm in the left leg. The range on the motion was restricted only in the abduction and internal rotation, which were $25^0$and 20^0 respectively. The Trendelenberg's test was positive. The radiological scans of both the hips showed a callus in the trochanteric region, with coxa vara on the left side, with a normal right side (the neck shaft angle was 108° on the left side and 125° on the right side). The Hilgenreiner's epiphyseal angle (the angle which is subtended by the horizontal line through the triradiate cartilages and an oblique line through the proximal femoral capital physes) was 72° on the left side. The physes was relatively vertical on the left side. Ultrasonography of the hip joint was unremarkable, with no avascular necrosis in either femoral head.

As the Hilgenreiner's epiphyseal angle was more at 60°, the neck shaft angle was < 120° and as he had vertical epiphysis (risk of Slipped Capital femoral epiphysis) with gait abnormality, the patient was managed by femoral valgus osteotomy and was fixed with screws and Hip spica. At 6 weeks post-operatively, the radiological scans of the patient's left hip showed fracture of the femoral bone at the osteotomy site. The right hip was normal. Hip Spica was continued for another 6 weeks. Subsequent X-rays films showed that the fracture was united, but the degree of the coxa vara had increased. The patient was allowed sequential weight bearing and was followed up every 3 months with clinical and radiological examinations.

After about 1 year of the surgery, the patient presented with pain in the opposite hip region and increasing deformity on the operative site. Avascular necrosis of the femoral head on the right side was noticed and increased coxa vara (neck shaft angle at 88°) with a Hilgenreiner angle of 80° was observed. The patient was prohibited from putting weight on the right leg and his last X-rays showed sclerosis of the head of the femur on the right side. The patient is being regularly followed up to check the progress of the disease condition and its complications, if any. The patient's parents were made fully aware of the condition, its prognosis and the associated rates of morbidity and mortality.

DISCUSSION

Various bony sequelae and their effect on the treatment were observed in our case of Gaucher's disease. Jachson and Simon, on the contrary, stipulated that the bone involvement in Gaucher's disease was very unusual in the first decade [3]. Mohindroo et al. has documented a case report of bilateral osteonecrosis of the femur in type 3 Gaucher's disease, leading to a varying amount of morbidity [4]. Wounlund and Lohmann reported a case of septic necrosis of the capitate in a young patient with Gaucher's disease and with no fracture of the carpus [5].

The investigatory work up in the diagnosis of Gaucher's disease includes the measurement of the glucocerebrosidase activity in leukocytes and fibroblasts and/or the mutation analysis of the cerebrosidase gene [6, 7]. High levels of the angiotensin converting enzyme is also an associated finding [8]. Typical Gaucher's cells may be seen in the bone marrow or in the live biopsy [9,2]. In our case, the disease was confirmed with the histopathological examination of both the soft tissues and the bony tissue from the pathological fracture site.

Enzyme replacement therapy has established itself as the treatment of choice in Gaucher's disease. However, the improvement in the extensive bone marrow infiltration and the loss of bone mass takes a longer time due to fewer uptakes of imiglucerase and slow bone turn over. Enzyme therapy cannot reverse establish the

osseous deformities including fractures and joint collapse [10,11]. In our case, enzyme treatment was advised to the patient, but it could not be given due to very its high costs and the lack of immediate availability of the same.

The surgical options target at the fixation of the fractures and various osteotomies to correct the deformities. In our case, we performed sub-trochanteric osteotomy of the femur for the correction of coxa vara, which is a well recognized method for the treatment of coxa vara (Heimke et al, in 2009) [12]. Gaucher's disease, in our case, was associated with a pathological fracture at the neck of the femur on one side and avascular necrosis of the contra lateral femur head. There was an increased rate of infections in the joints, which was probably related to splenectomy, which was done for the disease.

Various surgeons found poor surgical results in the patients of Gaucher's disease [13-15]. Catonne et al, in 1992, found aggravation of the coxa vara, pseudarthrosis or osteo-arthritis after femoral valgus sub-trochanteric osteotomies [13]. Surgeons have recommended the initiation of enzyme replacement therapy for at least one to two years prior to total hip replacement, to facilitate the bone remodelling in these young patients [14, 15].

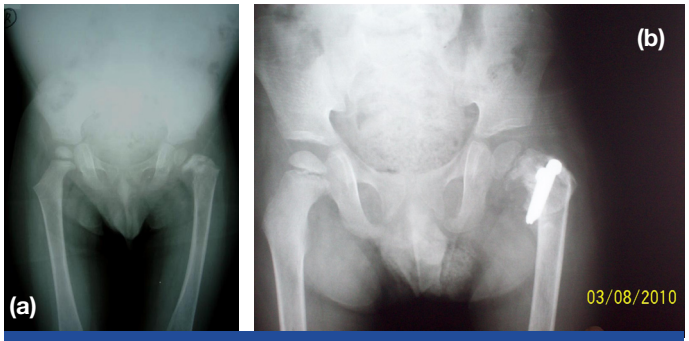
A) Congenital		B) Acquired	
1	Cleidocranial Dysplasia	1	AVN: (DDH or Perthes
2	Chondrodysplasia Punctata	2	Fibrous Dysplasia
3	Metaphyseal Chondrodysplasia	3	SCFE
4	Gauchers Disease	4	Rickets
5	Multiple Epiphyseal Dysplasia	5	Paget's Disease
6	Proximal Focal Femoral Deficiency	6	Osteomyelitis of Hip /septic arthritis
7	Spondyloepiphyseal Dysplasia Congenita	7	Traumatic

[Table/Fig-1]: Differential diagnosis of Coxa vara

Types of Gauchers disease		
Type 1	**Type 2**	**Type 3**
Most frequent type.Onset at any age. Presents with Massive splenomegaly, thrombocytopenia with/without anemia, hepatomegaly, bone lesions vary from growth retardation in children, delayed puberty in adolescents to AVN of hip in adults. Erlenmeyer flask shaped deformity of femur or lytic lesion in bones. There are no neurological abnormalities or intelligence defects in this type. The life span can be expected to be normal with ERT. Rarely pulmonary hypertension, infiltrative lung disease, portal hypertension or renal involvement is seen [4].	Type 2 or the infantile type presents with visceromegaly, strabismus, and severe neurological abnormalities with spasticity. Bony crisis are uncommon. Onset is in infancy with less than 2 years of life span.	Type3, the adolescent or Norrbotten form, have early onset of organomegaly and slow progressive neurological dysfunction. Onset is in childhood and expected life span varies from 2-60 years.

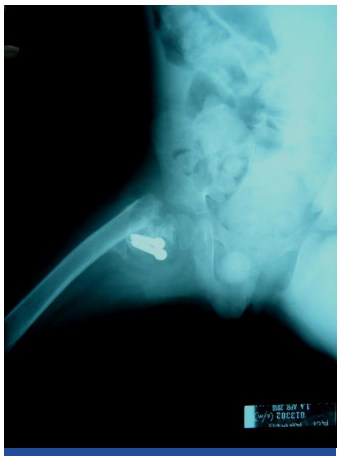
[Table/Fig-2]: Types of Gauchers disease

We found an incidental involvement of contralateral hip with Perthe's disease. To our best of knowledge and after a thorough research of the literature, we found such a combination for the first time.

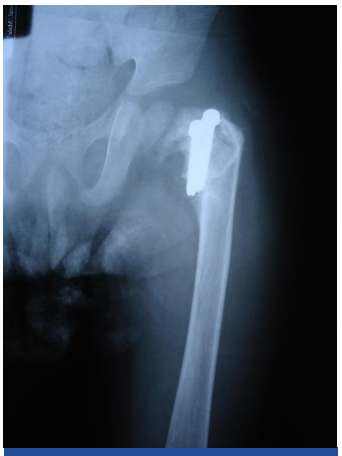


[Table/Fig-3]: (a) Pre op Xray showing coxa vara left hip with normal right hip. The coxa vara occurred due to fracture of neck of left femur which united in varus position. The neck shaft angle was 108^0 as compared to 125^0 on right side. The He angle was 72^0

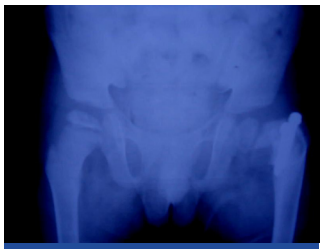
(b) Post op Xray showing Femoral valgus Osteotomy with screw fixation



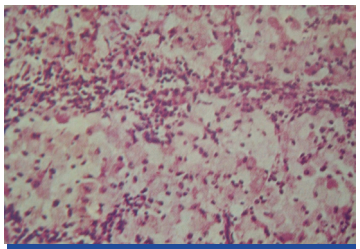
[Table/Fig-4]: Failure at osteotomy site was evident by 6 weeks.



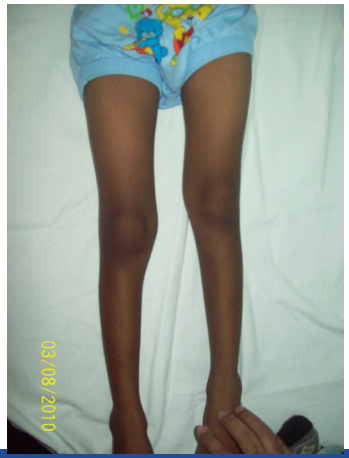
[Table/Fig-5]: Subsequent displacement of fracture at osteotomy site with union in increased varus position was visible



[Table/Fig-6]: Increased coxa vara on left side with neck shaft angle 88^0 and H-E angle of 80^0 and avascular necrosis of right femoral head. This was evident by 1 year after the corrective osteotomy surgery



[Table/Fig-7]: Gauchers cells which are seen as large foamy cells with their cytoplasm filled with lipid. These cells are a part of reticuloendothelial system and form an important part in diagnosis of Gauchers disease.



[Table/Fig-8]: The picture shows true shortening of left lower limb

CONCLUSION

Gaucher's disease had a detrimental effect on the results of our treatment method (which is an otherwise recommended form of treatment in such cases) and it resulted not only in the failure of the osteotomy, but it also ended up increasing the deformity and the limb length discrepancy. The rate of healing of the fracture was also found to be delayed with respect to the apediatric population. Our aim is to emphasize that untreated Gaucher's disease may be an important cause of the failure of surgical treatment . Regular follow up of the disease with early treatment is the most important factor in minimizing the failure of the treatment for the bony sequelae and the deformities in Gaucher's disease.

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