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Familial early onset paget's disease: A case report

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Introduction: Familial Early - Onset of Paget's Diseases is an Autosomal dominant disorder associated with Mutation in gene TNFRSF11A which encode the Receptor Activator of Nuclear Factor Kappa (RANK).It is rare in Asians and usually presents in teens or twenties. Its features are similar to those of the classic form of the disease, although it is more likely to affect the skull, spine, and ribs (the axial skeleton) and the small bones of the hands. The serum alkaline phosphatase urinary hydroxyproline and serum C- telopeptide are elevated.

Case: We report a case of 21 years old boy who presented with complain of generalized bone pain for 12years, multiple bone fractures on slight trauma leading to progressive bone deformities for 5years and gradual hearing Loss for 4 years. His parents are cousins and one of their cousin had a similar problem but she died undiagnosed at 40.Patient's younger sister (20years) has similar problem since the age of 10 yrs. On investigation his CBC, UCE, LFTs , Uric acid, Ca, PO4, PTH, vit D3 levels were all normal, but his Alkaline phosphatase was markedly elevated at 2526 U/L (100 – 290).His sister also had similar laboratory results. His bone biopsy was consistent with Familial early – onset paget 's disease.

Conclusion: Familial Early onset Paget's disease is a rare disease in Asians and usually presents in young age in which alkaline phosphatase is markedly raised, confirmation of diagnosis is by bone biopsy and gene studies, in our case we were unable to do genetic studies because of lack of funding, and Treatment of choice is bisphosphonates. Long-term follow-up is mandatory in these patients in order to identify and treat any subsequent complications

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