Metabolic bone disease: Newer perspectives

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Metabolic bone disease is the third most common endocrine disorders after diabetes and thyroid diseases. The common metabolic bone diseases (MBD) include osteoporosis, rickets/ osteomalacia, flurosis and primary hyperparathyroidism (PHPT), while the rare MBDs include Paget's disease, tumor induced osteomalacia, fibrous dysplasia, osteogenesis imperfecta and so on.

Primary hyperparathyroidism (PHPT) is the common metabolic bone disease and unlike West, majority of the patients in our country presents with florid symptoms of the disease. Bone pains, fractures, renal stone disease, gall stone disease and pancreatitis are the usual manifestations in our scenario, while in the West, the majority of the subjects are asymptomatic.[1] High calcium, low phosphate and elevated iPTH are the hall mark of PHPT. However, with the use of autoanalyzer, the frequency of normocalcemic PHPT has remarkably decreased despite vitamin D deficiency. Rarely, PHPT is associated with low or undetectable Parathormone (PTH) possibly because of mutant PTH secretion or PTHrP secretion by the parathyroid adenoma. In such situation, it is difficult to take a decision to go ahead for surgery.[2] The localization of adenoma in PHPT is of importance in our scenario, where expert parathyroid surgeons are not easily available. Ultrasonography has a reasonably good sensitivity of 70% with positive predictive value of 100% and is a useful and inexpensive tool in establishing the etiology of PHPT.[3] Anemia is quite prevalent in patients with PHPT and etiology is PTH induced myelofibrosis.[4] Histologically, clear cell and chief cell adenoma are the two main variants, whether it does have impact on clinical and biochemical expression of the disease is not known. However, the data recently published by us shows that histology does not influence the clinical and biochemical outcome. Various genes have been implicated in parathyroid tumorigenesis including PTH receptor gene, vitamin D receptor gene and cyclin D1 gene. Our work on cyclin D1 gene was novel and showed that decreased expression of cyclin D1 may contribute to parathyroid adenoma formation.

Sporadic hypoparathyroidism is another common endocrine disorder and is associated with spectrum of neurologic presentations including carpopedal spasm, paresthesias, seizures and memory loss.[5] These patients also have a predisposition for increased carotid- intima media thickness and consequently may result in increased cardiovascular events.[6]

The Paget's disease has been reported from India and most of the case reports are described from South India. We described a study of 21 patients from North India having a Paget's disease with varying presentations including the patients who presented with recurrent paraparesis.[7] McCune Albright syndrome is associated with café-au-lait mascules, fibrous dysplasia and varying endocrinopathies. We reported our experience of 26 patients of fibrous dysplasia, among them 9 had McCune Albright syndrome.[8] Fibrous dysplasia in McCune Albright syndrome is treated with bisphosphonates and our experience of intra-lesional Zoledronic acid is very novel. Hypophosphatemic osteomalacia, particularly tumor induced osteomalacia (TIO) is quite uncommon and we have described our experience in 17 patients of hypophosphatemic osteomalacia, out of these 3 had TIO.[9]

Osteogenesis imperfecta (OI) is not an uncommon disorder at a tertiary care referral center[10] and we have demonstrated expression of HSP 47 is reduced in type-III OI and mutation in exon 9 is associated with severe OI. Recently, we have also described a novel mutation in a young female with increase BMD, syndactyly and progressive vision loss

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due to sclerosteosis and she had a frame shift mutation at c 296-297 in SC (Bone. 2012 Oct 16 Accepted Article).

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