Heterochromia Iridis with Primary Hypoparathyroidism

A 10 year old girl presented with the history of recurrent seizures of three month duration. She was product of non-consanguineous marriage with normal antenatal, perinatal and postnatal history. There was no family H/o similar complaints. There was no H/o delayed dentition, recurrent attacks of infections, exertional dyspnoea or cyanosis. Her mental and social milestones of development were normal and her scholastic performance was average.

On examination, her height was 130 cms (less than 50th centile), weight was 29 kg (less than 50th centile) with normal body proportions; Chvosteck’s and Trousseau’s signs were positive. Albright’s hereditary osteodystrophy (AHO) phenotype and the bony signs of rickets were absent. She had heterochromia iridis involving inferomedial quadrant of right eye (Fig. 1). Her systemic examination was normal.

On investigation, serum creatinine was 0.8 mg/dl, serum calcium 4.3 mg/dl, serum phosphate 8.7 mg/dl, serum albumin 4.2 gm/dl, serum alkaline phosphatase 352 IU/L, and iPTH 14.5 pg/ml (normal value 10-70 pg/ml). EEG revealed generalized seizure disorder. CT scan revealed multiple bilateral symmetrical calcification in basal ganglia (Fig. 2).

On basis of low values of iPTH, serum calcium, high serum phosphate and basal ganglia calcification, the diagnosis of idiopathic variety of primary hypoparathyroidism was made. Heterochromia iridis is usually congenitally present in association with Waardenburg’s syndrome or may be acquired as in Horner’s syndrome. This is the first case report of heterochromia associated with idiopathic hypoparathyroidism.

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Fig. 1 : The iris of right eye shows heterochromia.

Fig. 2 : CT scan showing calcification of the basal ganglia.