

Polyostotic Fibrous Dysplasia of McCune Albright Syndrome responding to intravenous Zoledronate therapy

Introduction:

McCune Albright Syndrome consists of at least 2 of the following 3 features: (1) polyostotic fibrous dysplasia (PFD), (2) Café au lait macules and (3) autonomous endocrine hyperfunction (eg, gonadotropin-independent precocious puberty). Other endocrine syndromes include hyperthyroidism, acromegaly, phosphaturia and Cushing syndrome.

Case

2 year old girl presented with severe hip pain, inability to walk and progressive deformity of right lower limb. Basic workup suggested a hip lesion suspicious for fibrous dysplasia. She was referred to us for further evaluation. On examination she had extensive Café au lait macules over the back. Further investigations showed thyrotoxicosis (T4: 10.92 mcg/dl and TSH 0.01 mIU/ml). Technitium scan of the thyroid showed large toxic nodule in the right lobe. CT brain showed craniofacial fibrous dysplasia. Hence the diagnosis of “McCune Albright Syndrome” (Polyostotic fibrous dysplasia, Café au lait macules and thyrotoxicosis) was made.

She had severe pain in the right lower limb. Hence it was decided to give bisphosphonates. She was started on I.V. Zoledronate 0.0125 mg/kg dosage. She was also started on Carbimazole 5 mg/day. Her pains reduced significantly in the follow up visit (>3 months) and she was able to walk. She was given I.V. Zoledronate 0.025 mg/kg once in every 3 months. Carbimazole was continued at 5 mg/day. Her phosphate excretion (TmP/GFR) was normal.

After 6 months, she had an episode of vaginal bleeding (FSH: 1.27 IU/l, LH: 0.13IU/l). She was given Letrozole 2.5 mg/day, after which she didn't have any vaginal bleed. Her T4 levels increased and TSH suppressed on follow up. We had to uptitrate the Carbimazole dosage for that.

With three monthly Zoledronate injection she is pain free now and able to walk. Corrective hip surgery for the deformity is being planned

Conclusion

I V Zoledronate is a good option in the management of painful fibrous dysplasias. Letrozole is very effective in controlling peripheral precocious puberty in McCune Albright syndrome.

