

CONCLUSION

Genetic mutations in children with NDM are common. Insulin remains the mainstay of therapy in INS-gene mutation. Genetic testing should be done to facilitate management.

EP_P003

ZOLEDRONIC ACID THERAPY FOR MONO-OSTOTIC LANGERHANS CELL HISTIOCYTOSIS: A CASE REPORT

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INTRODUCTION/BACKGROUND

Langerhans cell histiocytosis (LCH) is a rare disease. It can affect any organ in the body but is primarily characterized by osteolytic bony lesions. Skeletal LCH may range from a unifocal, self-limiting, asymptomatic lesion to severe, painful, destructive lesions that are prone to pathological fractures. Treatment decisions are individualized according to location, size, surgical accessibility, and functional impairment. Hence, there is no standard of care at the moment. Zoledronic acid (ZA) has been used in some neoplastic bone conditions to slow down the progression and reduce the bone pain.

CASE

We report a 6-year-old male with unifocal bony LCH at the left tibia who responded well to ZA. He presented at 4 years old with limping and was subsequently not ambulatory due to severe pain. He had a tender swollen left shin without skin changes.

X-ray showed a poorly defined 4.7 x 1.1 cm permeative lytic lesion in the medullary cavity of the midshaft of the left tibia with endosteal thinning. Subsequent MRI and isotope bone scans (Tc-99m MDP) confirmed a suspicious primary bone malignant lesion. A bone biopsy showed a neoplastic proliferation of histiocytoid cells with strong diffuse positivity for CD 1a, which was in keeping with LCH.

This confirmed a symptomatic mono-ostotic LCH with significant cortical destruction that was at risk of fracture. ZA was initiated after careful evaluation. His pain completely resolved with a return of function six weeks after the first dose of ZA. He received 4 doses of ZA in total and demonstrated radiographic evidence of regression and remained in remission.

CONCLUSION

This case demonstrates the potential role of ZA therapy as the first line treatment for mono-ostotic LCH stabilisation and symptomatic control.

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CASE REPORT: MUCOLIPIDOSIS BONE DISEASE RESEMBLING NEONATAL RICKETS

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INTRODUCTION/BACKGROUND

Mucopolipidosis II (I-cell disease) is a rare genetic metabolic disorder of lysosomal metabolism with a combined frequency of 1:422,000, that is characterized by coarse facial features, disproportionate short stature, hyperplastic gums, organomegaly, and retarded psychomotor development. These physical changes however require time to develop and are not apparent at birth. Opportunistically, neonates with I-cell disease are diagnosed after revealing typical spine changes in skeletal survey. Unlike nutritional rickets, their underlying cause and course of the bone disease are different.

CASE

We report a premature 34-week-old male with a birth weight of 1.6 kg. He was born in an ambulance while his mother was being transferred from a district hospital. He required cardiopulmonary resuscitation at birth and was admitted to the neonatal intensive care unit where he stayed for another 6 months due to respiratory reason. During his 1st to 4th months of life, he had markedly raised ALP level (>1000 IU/L), together with severe hyperparathyroidism that gradually resolved with time. He had radiographic changes resembling rickets which persisted and progressed to "chronic osteitis fibrosa cystica". His serum calcium, phosphate, and 25(OH)D₃ levels had always been normal. He never had a fracture. His diagnosis was later confirmed by marked elevation of plasma b-hexosaminidase, b-mannosidase and a-mannosidase. He was discharged home with tracheostomy and CPAP. He continues to survive at 11 months old by the time of this report.

CONCLUSION

Mucopolipidosis osteodystrophy could resemble nutritional rickets during the neonatal period but it is not related to either deficient vitamin D or minerals. Failure to respond to conventional treatment (vitamin D and calcium or phosphate), should prompt for a search for other causes of increased bone turnover. In I-cell disease,