

CASE REPORT

MALIGNANT INFANTILE OSTEOPETROSIS

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Abstract: *Malignant infantile osteopetrosis is a rare congenital disorder characterized by increased bone density due to defective resorption of bone by osteoclasts. Infantile malignant osteopetrosis usually presents in infancy with bone marrow failure leading to hematological abnormalities like pancytopenia and hepatosplenomegaly due to extramedullary hematopoiesis. Though it commonly presents in early infancy, it should be suspected even in neonates presenting with characteristic features.*

Keywords: *Osteopetrosis, Bone marrow, Osteoclast.*

References

1. Wilson CJ, Vellodi A. Autosomal recessive osteopetrosis: diagnosis, management, and outcome. Arch Dis Child 2000; 83(5):449-452.
2. Stark Z, Savarirayan R. Osteopetrosis. Orphanet J Rare Dis 2009; 4(1):1-2.
3. Helfrich MH, Aronson DC, Everts V, Mieremet RHP, Gerritsen EJA, Eckhardt PG, et al, Morphologic features of bone in human osteopetrosis. Bone 1991;12(6):411-419.
4. Diniz G, Olukman O, Calkavur S, Buyukinan M, Altay C. A histologically diagnosed case with infantile osteopetrosis complicated by hypopituitarism. Case reports in pathology. 2015; Article ID 786836, 5 pages, 2015. <https://doi.org/10.1155/2015/786836>.
5. Srinivasan M, Abinun M, Cant AJ, Tan K, Oakhill A, Steward CG. Malignant infantile osteopetrosis presenting with neonatal hypocalcaemia. Arch Dis Child-Fetal and Neonatal Ed 2000; 83(1):F21-23.
6. Mazzolari E, Forino C, Razza A, Porta F, Villa A, Notarangelo LD. A singlecenter experience in 20 patients with infantile malignant osteopetrosis. Am J Hematol 2009; 84(8):473-479.
7. Key Jr LL, Ries WL, Rodriguiz RM, Hatcher HC. Recombinant human interferon gamma therapy for osteopetrosis. J Pediatr 1992;121(1):119-124.
8. Askmyr MK, Fasth A, Richter J. Towards better understanding and new therapeutics of osteopetrosis. Brit J Hematol 2008;140(6):597-609.
9. Key L, Carnes D, Cole S, Holtrop M, Bar-Shavit Z, Shapiro F, et al. Treatment of congenital osteopetrosis with high-dose calcitriol. N Engl J Med 1984;310(7):409-415.

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