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A report of two memebers an of Iranian family affected by autosomal recessive osteopetrosis

Abstract

Introduction: In the rare hereditary bone disorder of osteopetrosis, reduced bone resorption function leads to both the development of densely sclerotic fragile bones and progressive obliteration of the marrow spaces and cranial foramina. Marrow obliteration, typically associated with extramedullary hemopoiesis and hepatosplenomegaly, results in anemia and thrombocytopenia; and nerve entrapment accounts for progressive blindness and hearing loss. Severe infantile or malignant osteopetrosis is the worst type of the disease which has poor prognosis. In this study we report two cases of severe infantile or malignant type of the disease in an Iranian family.

Methods: Our two patients were children of a family that wife was the grandchild of husband's aunt. Two weeks after birth, the first patient had episodes of seizure and spastic in extremities. Gradually, the patient found upper and lower respiratory problems and horizontal nystagmus. X-Ray of the hand and foot showed widening and increased bone density. Physical examination showed hepatosplenomegaly and petechiae in extremities. The patient expired due to cardiopulmonary arrest. The some episodes seizure happened for the 2nd patient 2weeks after birth. Gradually, Asymmetry between eyes and eventoal blindness confirmed by ophthalmologist. Finally the patient expired because of severe pneumonia.

Conclusion: Autosomal recessive osteopetrosis has been reported in most ethnic groups although as the disease is very rare it is more frequently seen in ethnic groups where consanguinity is common. For the first time we report two cases of severe infantile or malignant type of the disease in an Iranian family.

Keywords: Osteopetrosis, Autosomal recessive, Consanguinity