



Osteopetrosis in Children: A Case Report

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Case report

The study is about a 6 years old boy who was hospitalized during the neonatal period for 10 days in the neonatal intensive care unit for a status epileptical with a normal check-up (CT, ETF, EEG and biological check-up).

At the age of 1 year, the parents noticed a delay in psychomotor acquisition, a delay in dental growth and blindness.

At the age of 3 years, the child was hospitalized for a hemorrhagic syndrome with epistaxis of medium abundance and gingivorrhagia. He had developmental delay, a psychomotor

delay, a blindness, a retrognathism and no teething. His biological check-up showed a normocytic normochromic anemia with thrombocytopenia. The myelogram was normal, and the thoracic and skeletal radiography showed a condensed aspect of the bone frame with symmetrical and bilateral periosteal appositions at the upper extremity of the two humeri.

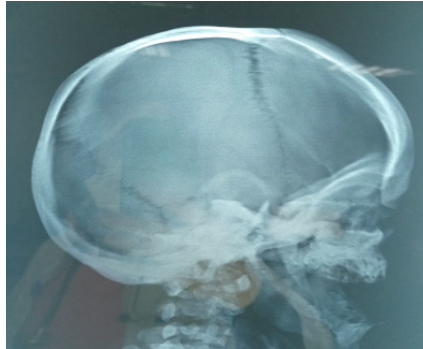
This clinical aspect, combined with the radiological and biological work-up, concluded to osteopetrosis confirmed by genetic study, which has identified a double heterozygosity.



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Condensed aspect of the bone frame



Condensed aspect of skull base



Osteopetrosis face.