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Sanjad-Sakati Syndrome: A Case Report

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Case report

A 8-month-old boy from a consanguineous marriage, and having as ATCD two deceased sisters: one who died at the age of 4 years in an unexplored table of poor weight gain and the 2nd died at the age of 4 months in a picture of convulsion. The boy was admitted to the service at birth with a picture of generalized neonatal convulsions. The biological assessment showed a low calcemia, a high phosphoremia and very low parathyroid hormone levels PTH. Transfontanelle, cardiac, cervical and abdominal ultrasound are normal. The EEG showed signs of pain. In front of this table, a congenital hypoparathyroidism was suspected and Sequencing analysis of the Tubulin Folding Cofactor E (TBCE) gene revealed a homozygous 12-bp deletion .the diagnosis Sanjad-Sakati syndrome was retained. The evolution is favorable under vitaminocalcic supplementation.

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