A Young Boy with Microcephaly, Ichthyosis and Cerebral Dysgenesis: A Rare Case of CEDNIK Syndrome

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ABSTRACT

Cerebral dysgenesis, neuropathy, ichthyosis and keratoderma (CEDNIK syndrome) is an extremely rare condition with only 19 cases diagnosed worldwide. It is an autosomal recessive syndrome occurs as a result of homozygous deletion in 22q11.2 as well as a mutation in Synaptosomal-associated protein 29 (SNAP29) which is a protein that regulate vesicle fusion in the cells. We report a rare case of CEDNIK syndrome which is the first case reported in Bahrain and the Arabian gulf. The patient presented with skin changes, developmental delay and his brain MRI showed significant cerebral dysgenesis.

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