PARTIAL AGENESIS OF CORPUS CALLOSUM IN SANJAD-SAKATI SYNDROME

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Introduction: Sanjad-Sakati syndrome (SSS) was first described in the Middle- East in children of consanguineous parents . It is a rare autosomal recessive disorder known also as Middle-East syndrome or Richadson-Kirk syndrome or hypoparathyroidism-retardationdysmorphism (HRD) syndrome. Children afflicted with this condition are born with intrauterine growth retardation, and present with hypocalcemic tetany or seizures due to hypoparatyroidism at an early stage in their lives . In this paper we report, for the first time, the presence of partial agenesis of corpus callosum in a patient with SSS.
Methods: It is conducted with the utilization of MRI , EEG and review of articles are done .
Results: HRD syndrome was caused by a deletion (c.155-166del12) within the tubulin cofactor E (TBCE) gene that encodes a molecular chaperone that is required for heterodimerization of α-tubulin with β-tubulin and other mutations also reported .The brain MRI in our patient showed a partial agenesis of the corpus callosum characterized by absence of the splenium and part of the isthmus , a finding which has never been reported in this syndrome.
Conclusion: We report, for the first time partial agenesis of corpus callosum in Sanjad-Sakati syndrome (SSS). Based on our data and those reported in the literature, we hypothesize that the partial agenesis of corpus callosum contributes to the pathogenesis of motor and developmental abnormalities in SSS.
(Figures Included in the Poster).