World Academy of Science, Engineering and Technology
International Journal of Psychological and Behavioral Sciences
Vol:9, No:09, 2015

Cretinism Muscular Hypertrophy: An Unorthodox Reflection

Authors : Harim Mohsin, Afshan Channa, Beena Saad

Abstract : The Kocher Debre Semelaigne Syndrome (KDSS) is known as cretinism muscular hypertrophy. It is an unusual presentation in intellectually deficit children, commonly associated with congenital or iatrogenic hypothyroidism. The creatinine phosphokinase (CPK) is usually elevated and it's commonly found in males, consanguineous marriage and ages 18 months to 10 years. It might be misdiagnosed without the classical features of hypothyroidism at first presentation. We present a case of 15 year old intellectually deficit female with epilepsy managed on phenytoin. She had rigidity, myxedema, calf muscle hypertrophy and agitation. The patient was managed as Neuroleptic Malignant Syndrome due to raised CPK of 40,680 IU/L and mixed presentation. Nevertheless, no improvement was noticed and thyroid profile was done to exclude alternative resources. Thyroid stimulating hormone (TSH) was 74.5 IU, Free T3 1.22 ng/dl, and Free T4 0.43 ng/dl. Thyroxine was started along with change in antiepileptic leading to recovery. This case report highlights the inconsistent finding of KDSS. The female gender, non-consanguineous marriage, delayed onset with primarily neuromuscular symptoms, and raised CPK is a rare demonstration in KDSS. Additionally, thyroid profile is not routinely done, which can lead to misdiagnosis and mismanagement.

Keywords : cretinism, hypothyroidism, intellectual deficit, KDSS
Conference Title : ICCPBS 2015 : International Conference on Cognitive, Psychological and Behavioral Sciences
Conference Location : Paris, France
Conference Dates : September 21-22, 2015