Abstract

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Journal of Immunology and Microbiology

2022
Vol 6. No. 1

A patient presented with pigmentation and generalized weakness

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Abstract

X-linked adrenoleukodystrophy (X-ALD) is a devastating genetic disease with three main phenotypes (childhood cerebral forms, adrenomyeloneuropathy(AMN) and Addison disease(AD). Tuberculosis is the commonest cause of AD in underdeveloped countries whereas autoimmune destruction accounts for 75-80% cases in developed countries. This is the first case report of X-AMN/ALD in two brothers in Bangladesh confirmed by raised very long chain fatty acid (VLCFA). Our index patient of 19 years presented with anorexia, weakness, vomiting, weight loss for three years with increased pigmentation in perioral skin, lips, tongue, mucus membrane, palmar creases, knuckles, reduced scalp hair(figure-1a-d), postural hypotension, low BMI, was found to have low basal Cortisol (9.00 am), ACTH >1250 pg/ml (normal<46), was diagnosed with AD. After three months he developed progressive spastic paraparesis along with cognitive declination and behavioral abnormality. His only brother was normotensive, clinically asymptomatic at presentation had only increased pigmentation on tongue, palmar crease, scanty scalp hair(figure-1,2a-d) and extensor planter reflexes. He had high ACTH(>1250 pg/ml) with normal basal cortisol (9.00am). Plasma VLCFA analysis revealed significantly high C26:0 with abnormal ratio of C24:0/C22:0 and C26:0/C22:0 in both brothers, with normal level in mother(table-1). MRI (T2 & FLAIR) showed bilateral hyperintense basal ganglia in index patient(Figure2:3a-c). Electrophysiological studies of nerve (SEPs, VEPs, BAEPs, NCS, EMG) were normal in both. Both were given steroid replacement. After three years, index patient became bed bound with slurred speech, urinary urge incontinence and his asymptomatic brother developed spastic paraparesis with cognitive impairment. None had gonadal impairment. Follow up MRI revealed lesion in brainstem in both along with atrophy of thoracic segment of spinal cord in index patient(Figure2:4a-f) and cerebellum, internal capsule involvement in his brother(Figure3:5a-d). Both were of pure AMN variety but due to presence of cognitive impairment and behavioral abnormality they can be categorized as rare cerebral variety of AMN.

Received: January 07, 2022; **Accepted:** January 17, 2022; **Published:** January 28, 2022

Biography

Syeda Nur-E-Jannat graduated from Dhaka Medical College, the best medical college in Bangladesh. She started her carrier in medicine and completed fellowship in internal medicine from Bangladesh College of Physicians and Surgeons (BCPS), worked as medical officer in district health complex, as Junior consultant (Medicine) at Government Employee Hospital, Dhaka, She completed her specialization in gastroenterology from Bangabandhu Sheikh Mujib Medical University (BSMMU) at Dhaka. Now she is working as assistant professor, gastroenterolology at Dhaka Medical College She is involved in teaching of undergraduate and post graduate students at Dhaka Medical College and is working as co-guide of a number of thesis. She has a number of publications in the field of medicine and gastroenterology in national and international journals.

