

## A patient presented with pigmentation and generalized weakness

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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 (Figure 2:3a-c). Electrophysiological studies of nerve (SEPs, VEPs, BAEPs, NCS, EMG) were normal in both. Both were given steroid replacement.

### Recent Publications

[Global Summit on Clinical & Medical Case Reports; May 22-23, 2020; Paris, France](#)

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### Biography (150 word limit)

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, gastroenterology 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.