Pictorial CME

Muscle Weakness in Prader Willi Syndrome

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A 15-year-old boy presented in PMR OPD with generalized obesity(BMI 29.2) (Fig 1) and muscle weakness since 2 years of age. He has history of hyperphagia since early days. On thorough assessment in indoor it was found that he had significant hypotonia and small hands and feet without any polydactyly or syndactyly (Fig 2). On inspection we noted high arch palate without almond shaped eyes, V shaped mouth, dysplastic ears, high nasal or flat nasal bridge ((Fig 3). He was suffering from nasal speech and clinical signs of hypogonadism ((Fig 4).

When we assessed IQ we found that he was suffering from moderate mental retardation (IQ 30). His TSH, free T4, testosterone levels were 2.9, 1.19 and 10ng/dl respectively. Generalised obesity ruled out the diagnoses of Ahlstrom, Cohen and Carpenter syndrome. Presence of craniofacial abnormality, nasal speech, limb hypotonia and absence of polydactyly excluded the diagnoses of Laurence- Moon - Biedle syndrome. These clinical features clinched the diagnoses of Prader- Willi Syndrome.

We counselled his parents regarding the disease prognoses. Non-pharmacological therapy improved his muscle strength and gait pattern slightly.

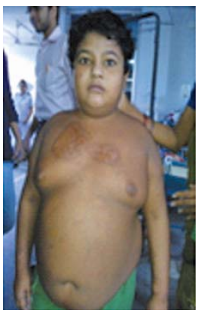

Fig 1

Fig 2

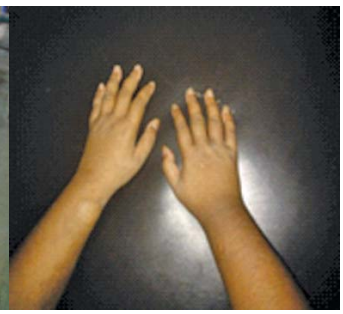

Fig 3

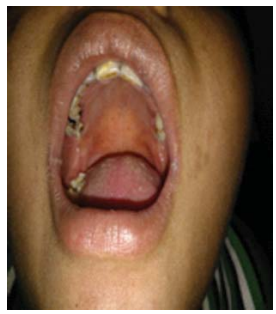

Fig 4

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