Images in Clinical Practice

Berardinelli-Seip Syndrome

A 12-year-girl, product of consanguineous marriage, presented with abnormal facies and dark discoloration of flexural creases of skin. She had masculine features over face and body, with generalized loss of subcutaneous fat and prominent muscularity. Acanthosis nigricans was present over neck, axilla, elbow, waist and knee, involving the flexural creases. Abdomen was protuberant with hepatosplenomegaly. There was mild clitoral hypertrophy.

Berardinelli-Seip syndrome (or congenital generalized lipodystrophy) is a rare autosomal recessive disorder characterized by generalized loss of subcutaneous fat and a well defined musculature. Anabolic features are observed and visceral organs are enlarged. Females have a masculine habitus, fatty steatosis of liver with hepatosplenomegaly may be present. Acromegalic gigantism with advanced dentition is an early and constant feature. All patients have *Acanthosis nigricans*, to some degree. *Diabetes mellitus* usually begins in the teenage years. Hyperlipidemia and cardiomegaly is seen in few cases.

Patients tend to survive till young adulthood or early middle age. Common causes of death are renal failure and hepatic failure. The gene for Berardinelli-Seip syndrome has been mapped to 9q34 and 11 q13.

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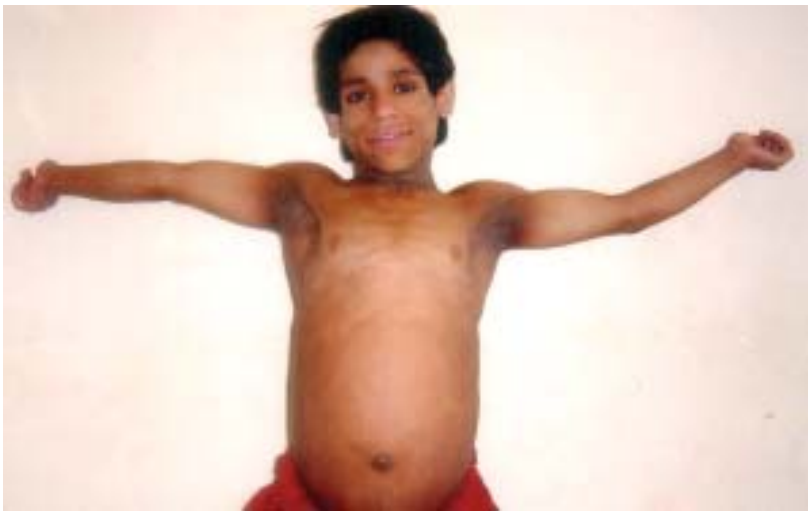

Fig. 1. Masculine facies with muscular prominence, acanthosis nigricans of neck and axilla, protuberant abdomen in a patient with Berardinelli-Seip syndrome.