GENETICS AND GENODERMATOSES

DONOHUE SYNDROME - A DERMATOLOGIST'S PERSPECTIVE

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INTRODUCTION: Donohue syndrome is an extremely rare and severe genetic disorder caused by mutations in the insulin receptor gene. The characteristic features include growth deficiency, elfin facies and fluctuating blood sugar levels. The typical cutaneous manifestations should raise the suspicion of this syndrome with further supportive systematic clinical evidences and laboratory parameters.

CASE REPORT: We report a 5 day old male baby with increased wrinkling of skin, reduced subcutaneous fat, loose skin, abdominal distension and large hands. Baby also had features of elfin facies with large and low set ears, broad nasal tip, flared nares, thick lips. Laboratory parameters showed refractory hyperglycemia and paradoxical hypoglycemia. Mothers antenatal records showed intrauterine growth retardation during last trimester. Baby was low birth weight (1.75 kg). No significant antenatal and family history was present. Before further investigation and genetic analysis, baby died on day 6 of its life secondary to hypoglycemia.

DISCUSSION: Donohue syndrome,also formerly called leprachaunism is a genetic disorder caused by mutations in the insulin receptor gene. It was first identified by William L.Donohue(1906). It is an autosomal recessive disorder. The gene responsible for this disorder is located at chromosome 19p13.2 . It occurs approximately 1 in every 4 million live births. Diagnosis is based on clinical features with blood test showing hypoglycemia with severe insulin resistance. Death is seen in late infancy due to malnutrition. Genetic counseling of parents is useful.

CONCLUSION: This case has been reported for its rarity in India and southern part of Asia. A high index of suspicion of this syndrome must be made by the dermatologist in a baby with classical cutaneous manifestations. An early and systematic multidisciplinary approach along with other specialities must be carried out for the confirmation of this rare and a highly fatal condition.