

QUIZ

Mohammed Mohy El-Din Selim, MD.

Consultant Dermatologist

Khalid Mansoor

Dermatology & Venereology Department

Hamad Medical Corporation

P.O. Box: 3050 - Doha - Qatar.

An Infant when born was 910 grams weight. His features showed broad nose, low set ears, large globular eyes, hypertelorism, hypertrichosis of forehead and cheeks.

The skin of the hands, feet, legs and thighs was corrugated. The skin at the flexures was loosely folded and acanthotic. There was hypertrophy of the genitalia.

What is your diagnosis?

Answer:

Leprechaunism (Donohue's syndrome)

Leprechaunism is a rare syndrome characterized by elfin-like face, acanthosis nigricans, absence of subcutaneous fat and intrauterine and neonatal growth retardation⁽¹⁾ other clinical features^(2,3,4) include low birth weight which can be as low as 910 grams⁽⁵⁾, broad nose, low set ears, large globular eyes, hypertelorism, micrognathia, hypertrichosis of forehead and cheeks, The skin appears too large for the body and is loosely folded at the flexures and may be corrugated on hands and feet, pachyderma and prominent rugal skin around the body orifices (Fig. 1-6). The breast, penis or clitoris may be hypertrophic.

The hands and feet may be disproportionately large. The bone age is retarded and there may be metaphyseal and epiphyseal dystrophy. Progressive muscle wasting with poor nutritional status and high susceptibility for infection is usually present.

The biochemical hallmark of the syndrome is disordered carbohydrate metabolism manifested by postprandial hyperglycemia, fasting hypoglycemia, insulin resistance and notable hyperinsulinemia^(3,6). It is sug-

gested that the skin changes in leprechaunism may be causally related to insulin resistance⁽³⁾. Post receptor defects in insulin action have been reported in fibroblasts isolated from two patients with leprechaunism⁽⁷⁾. Receptors for insulin like growth factor 1 are defective in fibroblasts cultures from a patient with leprechaunism and may contribute to in utero growth retardation⁽⁸⁾.

The skin changes in 25 leprechaunism cases were in the following order of frequency⁽³⁾, decreased subcutaneous fat (23/25), hypertrichosis of body and face (17/25), wrinkled loose skin (15/25), thick lips (11/25), hyperkeratosis (8/25) gingival hypertrophy (8/25), acanthosis nigricans (7/25), short thin concave dysplastic nails.

There is excessive proliferation of epidermis and epithelia of bronchi, collecting tubules of kidneys, bile duct, pancreatic duct and almost complete atrophy of lymphoid tissue⁽⁹⁾.

Leprechaunism is a rare autosomal recessive inherited disorder^(10,11,12,13).

Acanthosis nigricans seen in leprechaunism is possibly due to an interaction between excessive amounts of circulating insulin with insulin like growth factor receptors on keratinocytes and dermal fibroblasts⁽¹⁴⁾, some ovarian tissue and in the heart⁽¹⁵⁾. Acanthosis nigricans is a feature of HAIR-AN syndrome (hyperandrogenemia insulin resistance and acanthosis nigricans).

The relation of insulin resistance and hyperandrogenemia may explain the hyperandrogenemia in leprechaunism (enlarged penis or clitoris)⁽¹⁶⁾.

The diagnosis of leprechaunism is essentially clinical as there are no specific laboratory tests⁽¹⁷⁾.

Leprechaunism has to be differentiated from Patterson syndrome⁽¹⁸⁾, which includes bronzed hyperpigmentation, cutis laxa of hands and feet, bodily disproportion, severe mental retardation and major bony deformities. Its etiopathogenesis is unknown although quantitative and qualitative abnormalities of mucopolysaccharides excretion were found in one case⁽¹⁸⁾.



Fig-1 : Corrugated skin of hand



Fig-2 : Corrugated skin of legs and feet

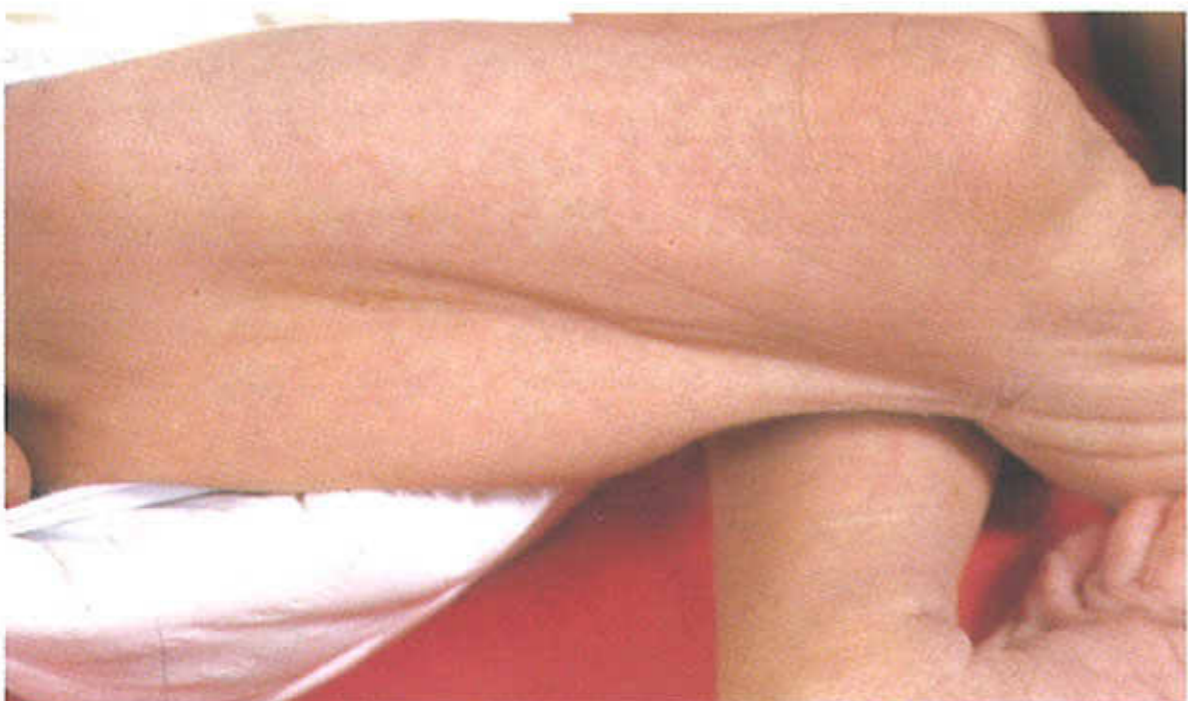


Fig-3 : Corrugated skin of thighs



Fig-4 : Broad nose, hypertelorism, low set ears



Fig-5 : Corrugated hand



Fig-6 : Corrugated skin of legs and feet

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