The Turkish Journal of Pediatrics 35: 319-322, 1993

LEPRECHAUNISM (DONOHUE'S SYNDROME): A CASE REPORT*

*Ayşegül Tokatlı MD***, Şinasi Özsoylu MD***, Şencan Özme MD*****

SUMMARY: Tokatlı A, Özsoylu Ş, Özme Ş. (Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara, Turkey). Leprechaunism (Donohue's Syndrome): A case report. Turk J Pediatr 1993; 35: 319-322.

Features of leprechaunism, including low-birth weight, a grotesque elfin face with clubbed nose, hirsutism, large low-set ears, enlarge external genitalia, distended abdomen with emaciated extremities, and growth and motor retardation were present in a 3-month-old girl. Because of parental consanguinity in this patient, a recessive mode of inheritence seems likely. *Key words: leprechaunism, female infant.*

In 1948, Donohue WL[1] described an infant with grotesque elfin facies, a flat nasal bridge, flaring nostrils, thick lips, hirsutism, large low-set ears, enlarged external genitalia and psychomotor retardation. Because of an associated endocrine disturbance, he called the syndrome "dysendocrinism". Since Donohue's original description, 37 additional cases have been described[2].

The name of dysendocrinism has been abandoned and the term "leprechaunism" is generally accepted.

We report here an additional patient who has the features of leprechaunism.

Case Report

A.K., a 3-month-old girl, was admitted to Hacettepe Children's Hospital with growth retardation. The patient was born to parents who were first cousins after a normal, full-term pregnancy. The birth weight was 2750 g. The mother noticed failure to thrive and delay in the motor developmental milestones of baby. One sibling had been similar in appearence to this child. The sibling had been admitted to the hospital when she was 4 months old with loose skin, elfin facies, hepatosplenomegaly and retarded bone age. Her liver biopsy revealed non-specific changes.

On admission, the general appearence of the patient was that of an emaciated, underdeveloped infant with an unusual face; her weight was 4100 g (<3rd

* From the Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara.
** Associate Professor of Pediatrics, Hacettepe University Faculty of Medicine.
*** Professor of Pediatrics and Hematologist, Hacettepe University Faculty of Medicine.
**** Professor of Pediatrics and Cardiologist, Hacettepe University Faculty of Medicine.

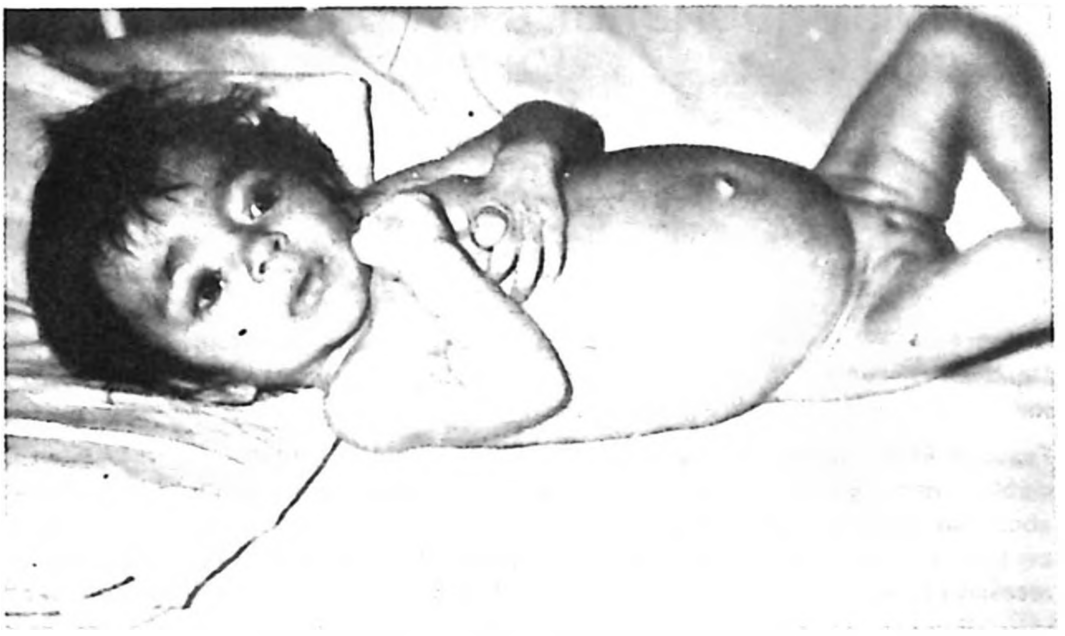

Fig. 1: General appearance of the patient.

percentile), height was 58 cm (25th percentile), head circumference was 39 cm (25th percentile) (Fig. 1). The skin was loose with minimal subcutaneous fat. There was generalized hirsutism, involving mainly the forehead, back and arms. The scalp hair was sparse but long and silky and extended down over the forehead. Examination of the eyes revealed long eyelashes, thick eyebrows and hypertelorism. The ears were large and low-set. The nasal bridge was flat, and clubbed nose was observed. The lips were thick, the palate was narrowly arched and micrognathia was present. The nipples were somewhat prominent and the abdomen was distended (Fig. 2). The liver and spleen edges were palpated 7 cm

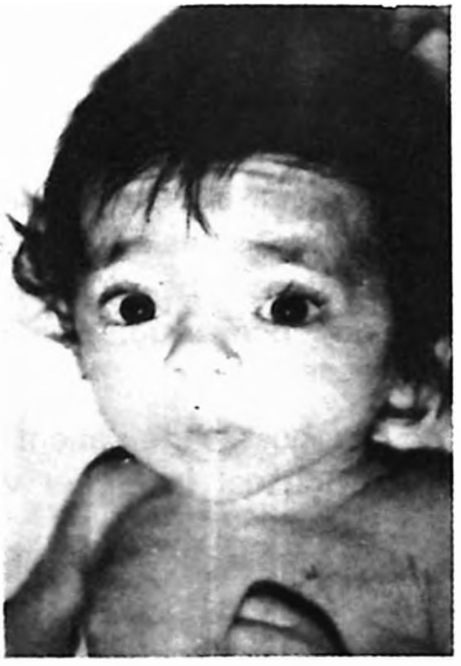

Fig. 2: Facial appearance of the patient.

below the respective costal margins. External female genitalia appeared to be normally formed but were markedly prominent. The extremities were normal but markedly thin. A generalized decrease in muscle mass and muscle tone was noted. The patient could not hold up her head, but she could follow objects with her eyes.

The complete blood count, urine analysis, blood urea nitrogen, levels of chloride, sodium, potassium, calcium, phosphorus, uric acid, creatinine, cholesterol and total lipids were all within normal limits. The blood glucose level was low (28 mg/dl). Serum GOT, GPT, alkaline phosphatase, bilirubin, PT and PTT were elevated, and PT and PTT did not return to normal after vitamin K administration. Serologic markers of viral hepatitis were negative. A mild non-specific generalized aminoaciduria was detected. Urine mucopolysaccharides were negative, and bone age was found to be somewhat retarded (bone age of one month with chronological age of three months). The bone marrow aspiration did not show any storage cells, and liver biopsy could not be obtained because of elevated PT and PTT.

Discussion

It has been emphasized that "the description of any patient as an example of Donohue's syndrome is based upon the clinical appearance, since no characteristic laboratory or histologic features are known"[3].

The diagnosis of our patient was based on clinical findings and comparison with Donohue's original cases and the previously reported cases. Some cases had enlarged liver and spleen[4,5], but none of the previous cases had massive hepatosplenomegaly, which was found in our case.

Recently, an inherited defect in a high-affinity insulin receptor was considered in the pathogenesis of this syndrome[6-8]. The biochemical evidence for this insulin resistance includes elevated plasma insulin levels, impaired glucose homeostasis, absence of anti-insulin-receptor antibodies and altered insulin-receptor binding. Although the plasma insulin level or anti-insulin receptor antibodies could not be studied, hypoglycemia was also documented in our case.

Because of parental consanguinity noted in several of the earlier cases, recessive inheritance is considered to be a possible factor[1,9,10]. In the present report, there was a history of a sibling who was similar in appearance, and the parents were first cousins These factors emphasize that the mode of inheritance is autosomal recessive.

REFERENCES

1. Donohue WL, Uchida I. Leprechaunism. *J Pediatr* 1954; 45: 505-519.
2. Norton KI, Glicklich M, Kupchik G, et al. Leprechaunism: a case report with radiographic features. *Dysmorph Clin Genet* 1990; 4: 57-62.
3. Summitt RL, Favara BE. Leprechaunism (Donohue's syndrome): A case repot. *J Pediatr* 1969; 74: 601-610.
4. Evans PR. Leprechaunism. *Arch Dis Child* 1955; 30: 479-483.
5. Patterson JH, Watkins WL. Leprechaunism in a male infant. *J Pediatr* 1962; 60: 730-739.
6. Bier DM, Schedewie H, Larner J, et al. Glucose kinetics in leprechaunism: accelerated fasting due to insulin resistance. *J Clin Endocrinol Metab* 1980; 51: 988-994.
7. Elsas LJ, Endo F, Strumlauf E, et al. Leprechaunism: An inherited defect in a high-affinity insulin receptor. *Am J Hum Genet* 1985; 37: 73-88.
8. Geffner ME, Kaplan SA, Bersch N, et al. Leprechaunism: in vitro insulin action despite genetic insulin resistance. *Pediatr Res* 1987; 22: 286-291.
9. Kallo A, Lakatos I, Szijarto L. Leprechaunism (Donohue's syndrome). *J Pediatr* 1965; 66: 372-379.
10. Rogers DR. Leprechaunism (Donohue's syndrome). *Am J Clin Pathol* 1966; 45: 614-619.