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Case Report

Congenital hypothyroidism: a case report

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Hypothyroidism resulted from deficiency of thyroid hormone production due to a defect in thyroid gland. The disorder may be manifested earlier. When symptoms occur after a period of normal function of thyroid gland, the disorder may be either truly "acquired" or only appear as a variety of congenital defects in which the manifestation of the deficiency is delayed. Normal level of triiodothyronine (T3) in children is 100-260 ng/dl, thyroxin (T4) 7.3 - 15 mg/dl and thyroid-stimulating hormone (TSH) 2-10 mU/mL.[1,2] The age at which symptoms appear depends on the impairment degree of thyroid function. In many cases, the deficiency of thyroid hormone is severe, and symptoms tend to develop in the early weeks of life.[1,3] The prevalence of congenital hypothyroidism has been found to be 1 in 4,000 infants world wide; it is lower in Japan (1 in 5,500 infants) and in African American population (1 in 32,000 infants). Most infants with congenital hypothyroidism are asymptomatic at birth, even when there is a complete agenesis of the thyroid gland.[1,3] But in other cases, during the first few months of life, the symptoms of hypothyroidism such as feeding problems, failure to thrive, constipation, hoarse cry, and somnolence usually can be found.[1,3,4] The purpose of this study is to report a case of congenital hypothyroidism in a 14 months old child.

The case

A 14 month old Balinese girl was admitted to the Child Health Department, Sanglah Hospital, Denpasar on November 22, 1996 with a chief complaint of delayed development. She could neither hold up her head at the age of 6 months nor sit at the age of 14 months. She was not able to walk and talk at that time.

Other complains were a large protruding tongue, protuberant abdomen and also difficulty in defecation. She was born spontaneously, a term gestational age with body weight of 3500 grams. When she was 12 days old, she was icteric and hospitalized in Denpasar Military Hospital for 7 days. The diagnosis at that time was congenital megacolon with icterus neonatorum. At the age of 4 months the defecation was normal. She had a complete basic immunization.

On physical examination, the girl was alert which usually winkled. The respiratory rate was 32x/minute regular, pulse rate was 136x/minute regular, and her body temperature was 36°C. She looked weak and myxedematous. The body weight was 10.1 kg with body heights 71 cm. The head circumference was 49 cm and fontanel was still open. Her hair was scanty, coarse, and brittle. The hairline reached far down on the forehead. The nasoorbital configuration was retained with narrow and swollen palpebrae fissures, with normal con-

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junctivae. The mouth was always open with a protruding, thick and broad tongue. She had flat nose, also short and thick neck. No abnormalities of the heart and breath sound were found.

The abdomen was distended and there was an umbilical hernia. The external genital was edematous. The extremities were broad, with short fingers and myxedematous. The skin of lower and upper extremities was scaly and dry on palpation. The laboratory findings revealed a hemoglobin level of 10.9 gram/dl, the leukocyte count was 5.18x103 / mL, the hematocrit was 38.1 vol %, and the levels of TSH > 375 mU/ml, T3 < 0.35 ng/ml, T4 < 0.40 mg/dl. Liver function tests and urinalysis were normal. Bone age analysis was appropriate for an infant of less than 3 months old.

Based on these findings, the diagnosis was congenital hypothyroidism. This patient was consulted to the Development and Behavior Department as well as to the Medical Rehabilitation Department. The consultant expertise was delayed (abnormal) with Vineland social maturity scale of 54 and she was to be rehabilitated in a positioning treatment, massage, stretching exercise, and range of motion exercise such as sitting and walking.

Since then she had been treated with L thyroxin 5 mg/kg/day. Within three months after the initial treatment, the laboratory examination was reevaluated. T3 and T4 were increased up to 2.59 ng/ml and 15.6 mg/dl, while TSH level decreased to 0.18 mU/ml. After 6 months of therapy she was able to stand up by holding object, sit with her hands holding on her knee, and smile. The lingual protrusion was absent and the umbilical hernia disappeared.

Discussion

Hypothyroidism is caused by deficiency of thyroid hormone production or a defect in thyroid gland. It may be congenital or acquired. The incidence of congenital hypothyroidism is 1:4000 live births, thyroid a/dysgenesis 80%, and dyshormonogenesis 20%. Thyroid gland disorder (aplasia or hypoplasia) is an important cause of congenital hypothyroidism and usually known as thyroidal dysgenesis. If the hypothyroidism developed early during fetal life, the effect might appear in certain organ systems, including the central nervous system and skeleton. However, most infants with congenital hypothyroidism are normal at birth. The diagnosis of hypothyroidism at birth based on clinical evidence is rarely possible.[6,7,8] The diagnosis of congenital hypothyroidism is not always easy to make due to nonspecific symptoms. Usually specific symptoms do not appear in newborns, so most infants are diagnosed by the neonatal screening program. The specific clinical signs are prolonged jaundice, lethargy, feeding problems, constipation, hypothermia, mottling and dry skin, umbilical hernia, macroglossia, anterior and posterior open fontanels, hoarse cry, and slow deep tendon reflexes. The physical development is usually retarded, such as delayed sitting, standing, and talking ages. The degree of physical and mental retardation increases with the age of diagnosis and treatment. The child is stunted in growth.

In our case, her clinical signs were prolonged jaundice, feeding problems, constipation, hypothermia, open fontanel, retained nasoorbital configuration, narrow and swollen palpebrae fissures, flat nose, short and thick neck, umbilical hernia, external genital edema, short extremities and fingers, and myxedema. This patient was able to sit at age of 14 months, but yet she was not able to walk and talk.

According to the literature, the roentgenogram showed a delayed epiphyseal development (bone age). Ossification centers, mainly on the hips, usually showed multiple small centers or a single, stippled, porous or fragmented center (epiphyseal dysgenesis).[1,9]; coxavara and coxaplana might also be found, as well as delayed formation and eruption of teeth.[1,9] In our case, radiological examination showed retarded bone age of less than 3 months, epiphyseal dysgenesis, delayed formation and eruption of teeth, but no femoral epiphysis

Special USG and CT scan are important to detect the cause of hypothyroidism and thyroid dysgenesis. The disorder of thyroxin production can be detected by thyroid function test. In our case, USG and CT scan examinations were not

done, because there were no USG and CT scan facilities for detecting the possibility of thyroid dysgenesis.

Early diagnosis and adequate treatment result in normal linear growth and intelligence compared with that of unaffected siblings. Thyroid hormone is critical for normal early brain development and effective treatment must be initiated promptly to prevent irreversible brain damage.[4,9,11]

The drug of choice for congenital hypothyroidism is thyroxine. The dosage of L-thyroxine for 0-6, 6-12, 12-59, 60-144, and > 144 month old child were 25-50 mg (8 mg/kgbw/day), 50-75 mg (6 mg/kgbw/day), 75-100 mg (5 mg/kgbw/day), 100-150 mg (5 mg/kgbw/day), and 150 mg (3 mg/kgbw/day), respectively. According to Klien RZ *et al*, when the treatment is started at the age of less than 3 months, elementary school school performance is similar to that of normal children. Treatment must be continued and guided by the monitoring of T3 and T4 levels every 3 months of the second year and there after every 6 months.[10]

This patient was treated by L-thyroxin 5 mg/kg/day. One month later, laboratory findings revealed T3 2.5 ng/ml, T4 15 mg/dl, and TSH 0.18 mU/ml. Based on the result, treatment of L-thyroxine was discontinued. Two weeks later, T3 level was 0.92 ng/ml, level T4 was 1.0 mg/dl and level TSH was 161 mU/ml, so that she was treated by 25 mg/day L-thyroxine up to now, and the laboratory test will be done every three months.

After 3 month of therapy, the laboratory results were T3 2.59 ng/ml, T4 15.6 mg/dl, and TSH 0.18 mU/ml. She came to the hospital with good condition, although umbilical hernia, lingual protrusion, and open mouth were still present. Myxedema on upper and lower extremities were reduced. After six months of therapy, her umbilical hernia and lingual protrusion were disappeared. She was able to sit with her hands holding on her knees. She had delayed psychomotor development and mental retardation, while the physical growth and the motoric development were within normal range.

Although congenital hypothyroidism has characteristic signs and symptoms, the disease must be differentiated from Down syndrome, Turner syndrome, and short stature. Clinically, all of them have similar symptoms, such as delayed growth and development, and mental retardation. The differentiation can be determined by laboratory examination. In congenital hypothyroidism, we will find increased TSH level and decreased T3 and T4 levels.

Early diagnosis and adequate treatment in the first weeks of life result in normal linear growth and intelligence. The longer delay of diagnosing congenital hypothyroidism, the higher the risk of mental retardation and various neurological sequelae. In this patient, although the treatment has been started at the age of 14 month, there was a significant improvement on her social interaction and motoric development.

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