

Congenital Hypothyroidism

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ABSTRACT We report a case of congenital hypothyroidism seen at the Division of Endocrinology, Medical School, University of North Sumatera / Dr. Pirngadi Hospital. A 13-month old Batak girl presented with typical signs and symptoms of congenital hypothyroidism. On physical examination, umbilical and inguinal hernias were present. Treatment with L-Thyroxine using initial dose 45 µg/day for one month was initiated, and after two months the signs and symptoms disappeared. Laboratory finding showed normal T₄, but T₃ was slightly low. Radiological examination was done to evaluate the bone age. The treatment was continued using the follow-up dose of L-Thyroxine 45 µg. Although the treatment has resulted in good outcome, evaluations for the child's growth and development should be continued. [*Paediatr Indones* 1994; 34:170-174]

Introduction

Congenital hypothyroidism is an uncommon clinical and biochemical syndrome entity due to the decrease of thyroid hormones produced by the thyroid gland, while the abnormality of thyroid gland is present at birth.^{1,2} Thyroid gland disorder (aplasia or hypoplasia) is an important cause of congenital hypothyroidism and is usually called as thyroidal dysgenesis.²

The thyroid gland functions under hypophyseal control, where the thyrotrophic hormone is produced and it is this hormone that regulates thyroid gland to produce thyroid hormones, including thyroxine (T₄) and triiodo-tyronine (T₃). The biosynthesis of these thyroid hormones requires iodine in which the process consists of two phases: binding of iodides by thyroid gland, and the organification of iodine. The iodides are bound by the cells of thyroid gland, and under normal condition this process is stimulated by the thyroid stimulating hormone (TSH).³

In some cases, this symptomatic deficiency of thvroid gland may occur in the

first weeks of life; but at a lower degree of deficiency, the symptoms appear and show a slower manifestation within months or years.³ The incidence of congenital hypothyroidism is about 1 per 3980 in Europe; 1 per 4250 in North America; 1 per 5370 in Japan; 1 per 5140 in Australia; and 1 per 4000 in Southern Thailand.³ In Indonesia, in the period of 1970-1982, The Department of Child Health, Medical School, University of Indonesia-Cipto Mangunkusumo Hospital treated 73 cases of congenital hypothyroidism.⁴ Congenital hypothyroidism is a serious problem in childhood and adolescence because serious complications may occur when the diagnosis and treatment are delayed.⁵

Case

A 13-month old Indonesian Batak girl was admitted to the Division of Pediatric Endocrinology, Medical School, University of North Sumatera-Pirngadi Hospital, Medan on March 3, 1992, with the complain that she could only lie supine. Besides, there were signs and symptoms of thick, large and protruding tongue, protruding navel and groin area, especially when she was crying. She slept almost all the time, and her body looked fat with short looking neck. Since she was one month old, her skin was dry and squamous, she had constipation with the frequency of defecation once in 5-6 days.

The history of pregnancy and delivery showed that her mother was apparently healthy during pregnancy, with regular antenatal care visits. The patient was

born spontaneously in hospital, on January 22, 1991, assisted by a midwife, with the body weight of 3700 g and body length of 52 cm without evidence of asphyxia. The patient had been completely immunized against tuberculosis, diphtheria, pertussis, tetanus, poliomyelitis, and measles.

On examination she was alert, not dyspneic, jaundiced, or cyanotic. Her body weight was 9 kg, body length 56 cm (the average normal BW and BL for 13 months age are 10 kg and 75 cm, respectively, according to our standard). Her body temperature was 37°C. Head examination showed thick hair; hypertelorism was noted with positive light reflex and isocoric pupils. The nose was flat. Her relatively small mouth was filled with large and protruding tongue and it was drooling. Her neck was short looking. The chest was symmetrical. The heart and respiratory rates were normal. No abnormality was found on heart and lung examinations. The abdomen was large, non-tender, with protruding navel and groin area. The liver was 3 cm below the costal margin, with smooth surface and sharp edge; the spleen was not palpable. The skin at the lower and upper extremities was squamous and dry on palpation. The genital was normal.

Laboratory investigations disclosed T₃ =15.4 µg/dl (normal=100-260 µg/dl); T₄ =1.2 µg/dl (Normal =7.3-15 µg/dl); TSH =9.8 µU/ml (Normal=0.2-1 µU/ml). The results of blood, urine and stool examinations were normal.

The working diagnosis was congenital hypothyroidism. Treatment was started with L-Thyroxine 45 µg/day. After treatment with L-Thyroxine 3 x 15 µg/day for

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three months, the patient did not take any medication and no follow-up within 1 month. On the 4th month she came with an improved condition showing no lingual protrusion, and the umbilical and inguinal hernias had disappeared. She had been able to sit with her hand held on the knee. One month later, the laboratory examination demonstrated that T3=90 µg/dl and T4= 10.4 µg/dl. She was able to stand up by holding objects. Radiological examination of her hands and legs indicated delayed bone growth, showing the bone age between 6 months to 1 year; whereas her calendar age was 1 year and 8 months.

The treatment was continued for the next 5 months; then, within 4 months she had no control and treatment. She was again lost of observation and no treatment given for 4 months. On examination, April 7, 1993, she was 2 years and 2 months; with body weight of 12 kg and the height was 82 cm (Normal body weight and height were 12.5 kg and 87 cm, respectively).

At present, the patient is only able to stand by holding subjects but unable to walk. She can only speak one or two words. Laboratory findings showed T3= 80 µgr/dl and T4= 6.5 µgr (N=7.3-15 µg/dl). Treatment with L-Thyroxine with total dose of 45 µg/day is continued.

Discussion

The diagnosis of congenital hypothyroidism in this case was established at the age of 13 months, based on history clinical and laboratory findings. The clinical symptoms were partly seen at birth such

as protrusion and enlargement of the tongue, flat nose, hypertelorism, short neck, umbilical and inguinal hernias, associated with dried skin and obstipation since 1 month of her life.

These conditions are similar to the signs and symptoms as stated in the literatures (Table 1).^{1,3,6} The laboratory examination indicated low T3 and T4 levels, while TSH increased.

Table 1. Signs and symptoms in congenital hypothyroidism

Signs	Symptoms
Skin mottling	Prolonged jaundice
Umbilical hernia	Lethargy
Jaundice	Constipation
Macroglosia	Feeding problems
Large fontanelle	Hypothermia
Hoarse cry	
Hypotonia	
Slow deep tendon reflexes	
Dry skin	

The diagnosis of congenital hypothyroidism is not always easy, because the symptoms are not characteristic, and in newborn baby there is often no early symptom.^{2,7} The first laboratory findings showed low levels of T3 and T4, but TSH level was elevated; this indicates that the causative factor is in the thyroid gland itself. Radiological examination for bone age was done only once, and the result showed retarded bone growth.⁸

It is difficult to make early diagnosis so that it may often be misdiagnosed with

mongoloid, besides other differential diagnosis such as Hurler's syndrome and imperfect osteogenesis.⁶

USG and CT scan are important to detect the cause of hypothyroidism whether there is dysgenesis of thyroid gland, while synthetic disorder of thyroxine can be detected by thyroid-function test using 125I sodium or 99mTc sodium pertechnetate.³

Sutan Assin⁹ reported 73 cases of hypothyroidism, and 35 of them were with thyroidal defects consisting of 17 (48.6%) cases with athyroid, 13 (37.1%) ectopy and 5 (14.3%) dysmorphogenesis.⁹ In this patient, diagnosis and treatment were instituted after the age of 13 months. This would certainly affect the growth and development of the child, especially her intelligence.^{2,7,9,10} The treatment was adapted from the Tuchinda's standard (Table 2),² i.e., initial dose of 45 µg/day (5 µgr/kg/day) and maintenance dose 5 µg/kg/day.¹¹

Table 2. Maintenance dose of L-Thyroxine

Age (month)	Dose
0-	25-50 µg (8µg/kg/day)
6-	50-75 µg (6 mg/kg/day)
12-	75-100 µg (5 µg/kg/day)
60-	100-150 µg (5 µg/kg/day)
144-	150 µg (3 µg/kg/day)

The treatment seems to be satisfactory. Umbilical and inguinal hernias disappeared, and there was no more constipation and squamous skin. At present, the patient has been able to stand by holding objects, although this is ra-

ther late than normal.^{2,3,7,10} Treatment will be continued while monitoring T4 and T3 levels will be done every 3 months for the second year and every 6 months thereafter.^{2,5} According to Klien R.Z. *et al* (1990), when the treatment is started at the age of less than 3 months, schooling performance in elementary-school level is similar to that of normal children.⁵

In this patient, the treatment was irregular by taking medication for total ten months and no medications for five months. The patient has already developed motoric and intelligence retardations. Laboratory findings demonstrated that T3 and T4 levels were low, and even lower than that of the second examination. It was shown that psychomotor delayed was present, however her physical growth was within normal range.

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