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CASE REPORT

Congenital Hypothyroidism

Report of Two Cases With Emphasis On Clinical Diagnosis And Brief Review of Relevant Literature

by

INDA D. ARIF AND A.S. ONGKIE

(From the Department of Child Health, Faculty of Medicine, University of Sam Ratulangi/Gunung Wenang Hospital Manado)

Abstract

Two cases of congenital hypothyroidism diagnosed clinically have been presented. Clinical improvement during substitution therapy with Thyranon was evident. The importance of clinical diagnosis has been emphasized. Awareness of the condition has been stressed as being of urgent necessity to diagnose accurately.

Relevant literature was briefly reviewed.

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Introduction

Congenital hypothyroidism occurs often enough to warrant attention. Its diagnosis at the earliest possible date is of the utmost importance because upon early diagnosis hangs the success of treatment (Means, 1937 cited by Lowrey et al 1958).
Delayed detection still occurs to a variable extent and, therefore, many cases still escape recognition for periods of several months or more (Burton and Nadler, 1979). Screening programs for the detection of congenital hypothyroidism are highly effective (Novogroder, 1980). However, in areas where modern technical facilities are out of reach, early diagnosis has to rely upon the doctor's clinical diagnostic acumen.

The purpose of this paper is to indicate that a clinical diagnosis of congenital hypothyroidism could be enough to justify treatment and to take stock of earlier published clinical criteria which might serve as a diagnostic tool.

Report of cases.

Case 1.

A 6-month-old female infant of Chinese parentage with mental retardation and snoring was referred by a GP. The mother's pregnancy and delivery were uneventful. She was hospital born at term with a birth weight of 3100 gm. and a baby length of 50 cm. There was no history of neonatal jaundice but there were poor sucking and infrequent bowel movements noted from the age of one month. She slept a lot and only very seldom cried. A hoarse cry was audible right after birth. Developmental milestones were delayed. She could not roll over, lift her head or smile. On examination, she looked pale and had a husky cry and was rather inactive with general hypotonia. There were macroglossia, cold extremities and dry coarse skin with mottling. Laboratory data showed a haemoglobin of 7 gm/100 ml and a serum cholesterol of 107 mg/100 ml. Other findings were within normal limits. The diagnosis of congenital hypothyroidism was then entertained and she was put on Thyranon in an ascending dosage starting with 5 mg twice a day. During the course of treatment, there was evident improvement in the patient's clinical condition.

Case 2.

An 8½-year-old Minahasan girl presented with retarded growth and mental subnormality. History obtained from the mother was that head-lifting was at age 1 year, sitting unsupportedly was at age over 1 year but she could not walk by herself up till now. Since age 5 years she could only say "papa" and "mama". Bowel movement was described as only once in two weeks. Previously, she had been treated several times for similar symptoms without improvement. History of the mother's pregnancy and delivery was uneventful. She was born in a village assisted by a traditional midwife. The mother could remember the development of jaundice in the neonatal period. During infancy she slept most of the day. There was a history of low-voiced cry and poor feeding since infancy. Dentition started at over one year of age. On examination, she was found to be mentally retarded. The body weight was 11.7 kg and the height was 78 cm. She had puffy eyes, sparse hair, a dull expression and the teeth were carious. Her cry was low-pitched. The skin felt coarse and dry. There was a distended abdomen with umbilical hernia. Developmental milestones were found to be very much delayed. She could only sit.

Laboratory data showed a haemoglobin of 8.6 gm/100ml., a haematocrit of 28% and an MCHC of 32%. The WBC count was 172.000 per c.mm. The serum cholesterol was 246 mg/100 ml. An ECG showed low voltage deflections in the standard leads, the unipolar extremity leads and the V_5 and V_6 leads. An X-ray of the left wrist showed a bone age of 0 – 3 months. The diagnosis of congenital hypothyroidism was made and the patient was put on Thyranon in an ascending dosage starting with 5 mg. twice a day. Obvious clinical improvement was then seen.

Discussion.

Case 1 was born in a hospital and the diagnosis of congenital hypothyroidism could be actually suspected in her neonatal period. It is suggested by the history of poor sucking, infrequent bowel movements, sleeping most of the day and having an infrequent and hoarse cry. We must admit that delayed diagnosis could well be due to the parents. This is exemplified by case 2 being born in the village to an unobservent mother.

In medicine, unless we begin to suspect, we often miss the diagnosis. The question arises as to how do we start having a suspicion ? What features should ring a bell ? Smith et al (1975) included in their publication on congenital hypothyroidism in the newborn period a list of features obtained by history prior to 3 months of age in 31 patients diagnosed as having congenital hypothyroidism (Table 1).

Table 1. *Initial signs and symptoms in congenital hypothyroidism in 31 patients who had signs and symptoms present before 3 months.* *

Signs and symptoms	No.	Percentage
Cold to touch	6	19
Mottling	18	58
Decreased activity	12	39
Feeding problem	12	39
Enlarged tongue	14	45
Hoarse cry	12	39
Constipation	16	52
Jaundice during newborn period	5	16
Dry skin	14	45
Umbilical hernia	18	58

* Unpublished data from Drs. Frederic M. Kenney and Alan H. Klein University of Pittsburg Children's Hospital.

They concluded that most athyrotic infants do manifest signs and symptoms in the newborn period. They reported on a case of congenital hypothyroidism, who was born in the Southmead Hospital, and was diagnosed clinically as early as 5 hours of age. They further stated that until an effective screening test is generally utilized, the early clinical signs and symptoms will be the major basis for the detection of congenital hypothyroidism in the newborn nursery. All the features included in table 1 could well bring the baby to the attention of the medical profession. It is but awareness that is needed for an early detection.

Early diagnosis of congenital hypothyroidism is of paramount importance. Klein et al (1972) found that patients treated before 3 months of age had a significantly higher average IQ in later life than those treated after 3 months.

In infancy, it may be expected that with the advancement of age, signs and symptoms would be more numerous so as to herald this pathological state. In case 1, on the basis of the typical history forwarded by the mother and the finding of pallor, lethargy, hypotonia, macroglossia, cold extremities and dry coarse skin with mottling and an audible husky cry, the correctness was proven by the drastic clinical response following treatment. The history of case 2 was illustrative for the presence of delayed developmental milestones, lethargy and mental retradation. A lowpitched cry, infrequent bowel movements and prolonged neonatal jaundice were additionally described. She had most of the typical signs and symptoms of congenital hypothyroidism evident from infancy to deserve recognition.

Because of the lack of facilities for sophisticated studies in most parts in our country, and seen from the practical point of view, it is important that the condition in question can be diagnosed clinically. Statements of mothers such as, "She was unusually quiet and never moved much", "She almost never cried" and "She was the best baby I ever had" (Lowrey et al 1958) should make one consider congenital hypothyroidism. La Franchi (1979), listing signs and symptoms separately presented the following tables pertaining to infancy (Tables 2 and 3).

Table 2. *Symptoms of congenital hypothyroidism in 25 infants detected by the Northwest Regional Screening Program.*

Symptoms	Per cent
Constipation	40
Lethary	32
Prolonged jaundice	28
Poor feeding	24
Hypothermia	8

Table 3. *Signs of congenital hypothyroidism in 25 infants detected by the Northwest Regional Screening Program.*

Symptoms (sic)	Per cent
Hypotonia	36
Umbilical hernia	28
Skin mottling	24
Large anterior fontanel	20
Macroglossia	20
Hoarse cry	20
Distended abdomen	20
Dry skin	20
Jaundice	20
Pallor	16
Slow deep tendon reflex	16
Large posterior fontanel	12
Hypothermia	8

Signs additional to those put forward by Smith et al (1975) are hypotonia, large anterior and posterior fontanel, distended abdomen, pallor and slow deep tendon reflex of which the detection is evidently the concern of the medical profession.

In childhood, a greater number of features of congenital hypothyroidism may be expected to present. It is illustrated by case 2 who had most of the typical signs and symptoms pertinent to hypothyroidism. La Franchi (1979), in his review article on hypothyroidism, presented another list of of signs and symptoms in childhood (Table 4).

Table 4. *Clinical features of hypothyroidism in childhood.*

Symptoms
Slow growth rate
Puffiness
Swollen thyroid gland
Lethargy and decreasing school performance
Cold intolerance
Galactorrhea
Menometrorrhagia in pubertal girls

Signs
Bradycardia, low pulse pressure
Short stature and decreasing growth velocity
Mild overweight condition
Immature (increased) upper to lower body segment ratio
Myxedema
Delayed dentition
Goiter
Precocious sexual development
Pale, thick, carotenemic, or cool skin
Flabby muscles, or rarely, pseudohypertrophy
Delayed deep tendon reflex return
Dull, placid expression

Hence, it is evident that the diagnosis of congenital hypothyroidism in childhood should be more readily made rather than overlooked. The cause of failure to diagnose is unawareness.

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"Learn ye well all the conspicious peculiarities but also the capricious and hidden hints of thyroid failings, and much misery will be spared your patients and much satisfaction added unto you".

Lisser, 1951.