ORIGINAL ARTICLE

Mental Retardation Due To Congenital Hypothyroidism and Its Prevention

by

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Abstract.

Thyroid hormone deficiency early in life causes severe and irreversible brain damage.

Fetal and neonatal hypothyroidism usually occurs in association with :

a) Environmental Iodine deficiency producing endemic cretins

b) Sporadic congenital hypothyroidism.

In Indonesia 12 million people are suffering from endemic goiter and there are roughly 100,000 endemic cretins. Adequate Iodine supply in affected areas, which is now a National Health Programme, will diminish the birth of endemic cretins in the next decades.

The sporadic congenital hypothyroidism (CHT) is due to anatomical abnormality or malfunction of the tyroid gland. If treated before the age of 3 months, fourfifths of the children will attain an IQ above 92.

The only alternative is screening of all newborns for CHT, which is now a standard procedure in most of the developed countries. The incidence around the world is 1 : 3000–4000 live births.

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In our clinic we have encountered 73 CHT children in 13 years time (1970–1982). Most of them were misdiagnosed as Down's syndrome.
The majority were recognized and treated after one year of age. Almost all of them had neuropsychological defects, besides physical growth retardation.

Replacement therapy with tyroid hormone gave good results in physical growth restoration, but the mental development remained retarded.
Prevention of this kind of mental retardation is only possible by a neonatal thyroid screening programme. This should start at all modern hospitals and maternity clinics and in the future nationwide.

Introduction.

Thyroid hormone deficiency early in life causes severe and irreversible brain damage (Morreale de Escobar, G and Escobar del Rey, F., 1980). Mental retardation and CNS damage an association with inadequate or altered thyroid function during fetal or neonatal period usually occurs in association with :

a) Environmental Iodine deficiency producing endemic cretins.

b) A congenital anatomical or/and functional defect of the thyroid gland (sporadic congenital hypothyroidism).

Endemic cretinism is mainly found in developing countries, while the sporadic type is encountered everywhere in the world.
Two forms of endemic cretinism is known, the neurologic cretinism and the hypothyroid cretinism, which may have a different pathogenesis.

Early detection and treatment of endemic cretins, particularly in its neurological form, doubtfully prevent their mental retardation (Morreale de Escobar, G. and Escobar del Rey, F., 1980). Only adequate iodine prophylaxis before the first trimester of pregnancy can prevent the birth of endemic cretins.

The sporadic Congenital Hypothyroidism (CHT) appears at random and is due to anatomical abnormality or malfunction of the thyroid gland
Few disorders have such devastating effects on growth and development as untreated CHT. If, however, CHT is diagnosed and treated before three months of age, four fifths of the children will attain an IQ above 92 (Klein, 1980). With the introduction of mass neonatal thyroid screening technic many children have been saved from permanent brain damage.

Most of the developed countries have now a neonatal screening program not only for the sake of the child but for financial reasons as well.
The incidence of CHT throughout the world is 1 in 3000 – 4000 live births, while the extra costs of care and special education for 10 children with mental retardation caused by CHT is five times higher than to detect 10 infants through the screening program (Burrow, 1980).

Hypothyroidism in Indonesia.

Endemic goiter still belongs to the four major nutritional problems in Indonesia. According to estimation approximately 12 million suffers from this disease and ± 1% among them or around 100.000 are cretins (LIPI, 1983). The government has started an eradication program ("Panca Karsa Husada and Panca Karya Husada") to reduce this disease in to a minor problem in the year 2000.

The non-endemic Congenital Hypothyroid (CHT), however, is still an unfamiliar problem in the Indonesian medical society. Few reports have been published on CHT in Indonesia. The incidence throughout the world is around 1 . 3000 – 4000, the incidence in Indonesia is not known. If this figure is the same for Indonesia and the brithrate of 35% will be the same in the coming years (5 million births each year) then every year roughly 1500 babies with CHT will be born.

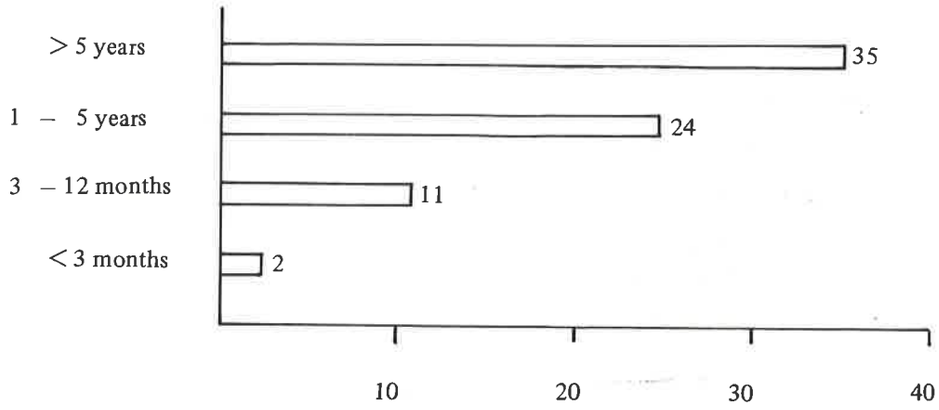
In our clinic 73 cases of CHT have been encountered in 13 years time (1970 – 1982; table 1).

Table 1. *Number of CHT cases 1970 – 1982*

	F	M	Total
1970 – 1974	8	5	13
1975 – 1978	20	11	31
1979 – 1982	18	11	29
	46	27	73

Due to the unfamiliar clinical pattern to our community and the practitioners as well, most of the patients were misdiagnosed as Down's syndrome and they came to our clinic at a very advanced age (Fig.1).

Fig. 1. *Number of cases in age groups at time of diagnosis.*



In general only one child in a family was affected, but there were four families with more than one sibling with CHT (table 2). It seems to be sex linked.

Table 2. *Four families with more than one sibling affected.*

FAMILY	Male	Female	Total
A	2	–	2
B	3	–	3
C	–	2	2
D	–	2	2

The type of thyroid defect in 35 patients is shown in table 3.

Table 3. *Type of thyroid defect*

	Female	Male	Total
Athyroid	13	4	17
Ectopic	12	1	13
Dyshormonogenesis	1	4	5
	26	9	35

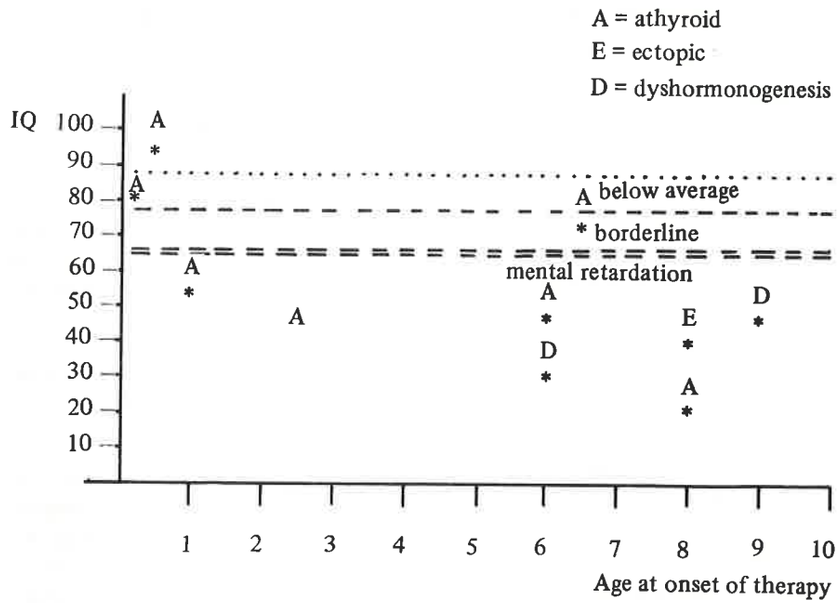
Fortyfour children with CHT (60.3%) were delivered by trained medical personnel in hospitals, maternity clinics or at home (table 4).

Table 4. *Number of CHT patients delivered by trained medical personnel and tradisional midwives.*

Hospitals	21
Maternity clinics	17
Midwives	6
Traditional midwives (dukun)	5
Not clear	24
Total	73

As soon as the diagnosis was established, replacement therapy with dissicated thyroid hormone or L–thyroxine was immediately initiated. Almost all of the patients were mentally retarded and even with adequate thyroid hormone therapy, the results were very disappointing (Fig. 2). Although the numbers of psychological evaluations were very small, but it shows, that only one of the patients reached a normal IQ.

Fig.2. *Correlation between age at onset of therapy, achieved IQ and type of thyroid defect in 11 CHT children.*



Discussion.

Endemic goiter, still a major problem in Indonesia these days (Djokomoeljanto, 1974) will certainly diminish in the next one or two decades due to our national health programme. The target for the year 2000 is to reduce this into a minor problem. With it the endemic cretins will disappear.

The problem of CHT is different. We see more and more cases every year. Our experience of the last 13 years can be summarized as follows :

1. Out of 73 cases only 2 came to our clinic at the age less than 3 months.
2. Almost all of the patients were not immediately recognized as CHTs. Most of them were mistaken as Down's syndrome, the reason why the initia-

tion or therapy almost always started a very advanced stage.

3. The majority of our patients were born in the hands of doctors and experienced midwives either in big hospitals, clinics or at home.
4. Although physical growth could be achieved satisfactory with adequate thyroid hormone treatment, the mental retardation remained.
5. The costs of education for CHT children are very high and not always available.

In Metropolitan Jakarta 227550 babies are born every year (Birthrate 35%) and if the incidence of CHT is the same as everywhere in the world, then there will be around 50 cases of CHT every year. That means, that between 1970 and 1982 roughly 700 CHT children were born. In the same period we have seen less than 100 cases, which means that there are still many unrecognized cases around.

Most of our CHT patients were born in modern hospitals and clinics in the hands of well trained medical personnel, even specialist. At birth the signs and symptoms of CHT are not specific, mostly absent. The clinical picture becomes clear after six to eight weeks after birth. If these babies could be treated before this age, the neuropsychological sequelae will be minimal and will mainly be : short attention span, fine motor coordination difficulties and impaired spatial orientation, the so called "minimal brain dysfunction syndrome" (Vandeschueren–Lodeweyckx at al, 1980).

Our patients, however, were treated much later that this age, which explains their below normal IQ and neuropsychological defects.

Replacement therapy with thyroid hormone almost always gave astonishing results in the first two to three years in particular the physical growth and the social contact (Sutan Assin and Pramuljo, 1983).
After that we often noticed a leveling off in the improvements.
Speech, learning disabilities and behaviour disorders were the main problems. Agressiveness and restlesness developed in many of our patients after several years of replacement therapy.

Our experience is in agreement with most of the reports from other countries. Availability of thyroid hormone is not enough.
Early diagnosis and treatment are vital for therapeutic success (Klein, 1980). Children with CHT treated after three months of age will unlikely attain normal levels of intellegence.

The only solution lies in screening all newborns for CHT.

This is already a routine procedure in most of the developed countries. Here in Indonesia only the Neonatal Ward of the Dr. Cipto Mangunkusumo Hospital in Jakarta is doing this as a pilot study in coordination with the Medical Faculty of the University of Indonesia.

In the future a national thyroid screening program should be developed.
In the mean time all modern hospitals and maternity clinics should realise the need of screening their babies because these are their responsibility.

This is the only way to minimize the occurence of this kind mental retardation.

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