Congenital hypothyroidism in Denmark

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SUMMARY Among 436 959 live infants born between 1970 and 1975 in Denmark were 72 (49 girls and 23 boys) who developed primary hypothyroidism, giving an incidence of 1 in 6064 live births. In the oldest group (6 to 8 years of age) the incidence was 1 in 5100. The age at diagnosis ranged from 1 week to 5 years 4 months; 10% were diagnosed within the first month, 40% within the first 3 months, and 70% within the first year of life. Thyroid scintigrams were available for 43 patients, 72% had aplastic or hypoplastic thyroid glands, 12% had ectopic thyroid glands, 16% had goitres. Patients with aplastic or hypoplastic thyroids tended to be diagnosed earlier than those with ectopic thyroid glands. The calculated incidence in the present study is lower than that suggested by the Danish neonatal screening programme.

This study was undertaken to find out the incidence and age at diagnosis of children with congenital hypothyroidism, born during a 6-year period in Denmark. The frequency of the various types of primary hypothyroidism was evaluated, too.

Material and methods

During the summer of 1979 questionnaires were sent to all departments of paediatrics in Denmark (a total number of 20) concerning patients with congenital hypothyroidism born in a 6-year period, 1970 to 1975. Replies were received from all the paediatric wards. Furthermore, information was obtained from 10 departments of internal medicine in areas of Denmark without a paediatric service.

Results

A total number of 72 patients (49 girls and 23 boys) with primary hypothyroidism was reported, and clinical and biochemical data were available for 71 of them. During the 6-year period there were 436 959 live births in Denmark,¹ giving an incidence for congenital hypothyroidism of 1 in 6064 live births

(Table 1). The incidence was 1 in 5150 live births for children born 1970-2 and 1 in 7400 live births for children born 1973-5.

The age at diagnosis is shown in Table 2. 10% of the patients were detected within the first month, 38% (27-50%) within the first 3 months, 68% (55-78%) within the first year, and 85% (74-92%) within the first 2 years of life. The confidence limit for the binomial distribution is given according to Diem.²

Thyroid scintigrams were available for 43 patients (Table 2). Aplastic or hypoplastic thyroid glands were present in 72%, ectopic thyroids in 12%, and goitres in 16% of these patients. There was a tendency towards a lower age at the time of diagnosis in patients with aplastic or hypoplastic thyroids compared with patients with ectopic thyroid glands, but the difference was not statistically significant ($P > 0.1$). Four out of 7 goitrous hypothyroid patients were diagnosed within the first 2 weeks of life. In 5 patients the diagnosis was established after age 3 years.

A psychological examination was performed in only a few patients, but 27 out of 59 patients were recorded as mentally retarded.

Table 1 *Total number of patients with congenital hypothyroidism among children born in Denmark 1970-5*

<i>Year of birth</i>	<i>Age of children (years)</i>	<i>No of patients</i>	<i>No of live births</i>	<i>Incidence</i>
1970-2	6-8	43	221 666	1 in 5150
1973-5	3-5	29	215 293	1 in 7400
1970-5	3-8	72	436 959	1 in 6065

Table 2 Age at diagnosis and type of thyroid disorder in children with congenital hypothyroidism born during a 6-year period (1970-5) in Denmark

Age at diagnosis	Cumulative percentage (95% confidence limit)	Type of thyroid disorder (Scintigrams were available for 43 patients)			
		Aplasia	Hypoplasia	Ectopia	Goitre
1 week (n = 2)	3 (0.3-10)				2
2 weeks (n = 3)	7 (2-18)	1			2
4 weeks (n = 2)	10 (4-19)	1			
6 weeks (n = 9)	23 (13-34)	4	1		
2 months (n = 5)	30 (19-42)	2	1	1	
3 months (n = 6)	38 (27-50)	1	2		
4 months (n = 5)	45 (33-57)	3			
5 months (n = 2)	48 (36-60)				
6 months (n = 7)	58 (45-69)	3	2		
9 months (n = 3)	62 (50-73)			1	
12 months (n = 4)	68 (55-78)	2			
2 years (n = 12)	85 (74-92)	3	2	1	2
3 years (n = 6)	93 (84-98)	2	1	1	
4 years (n = 2)	96 (88-99)				
5 years (n = 2)	99 (92-100)			1	
6 years (n = 1)	100 (95-100)				1
Total number	71	22	9	5	7
Percentage		51	21	12	16
(95% confidence limit)		(35-62)	(10-36)	(4-25)	(7-31)

Discussion

The term 'congenital hypothyroidism' is generally applied if the disorder is believed to have been present *at* or *before* birth.³ The clinical signs of the hypothyroidism gradually develop during infancy and childhood. The signs and symptoms that lead eventually to a diagnosis of the disease are complex, so it was not surprising that 4 out of 5 patients diagnosed within the first 2 weeks of life had goitres. About 40% of the hypothyroid patients were detected within the first 3 months and 70% within the first year of life, which is a similar rate to that reported from the Netherlands and Sweden.⁴⁻⁵

Generally, the manifestations of the hypothyroidism appeared earlier in patients with aplasia than in those with ectopic thyroid glands, as has also been reported by others.⁶⁻⁹

In our study 70% of patients for whom thyroid scintigrams were available had aplastic or hypoplastic thyroid glands. These data were not given in earlier epidemiological studies,⁴⁻⁵ and are similar to those found by neonatal screening programmes in North America.¹⁰

Some patients with a congenital defect of the thyroid gland were not diagnosed until between ages 3 and 5 years. It is possible that other patients exist but have not been reported because they are considered to suffer from an acquired hypothyroidism.

The incidence of congenital hypothyroidism in the total population was found to be 1 in 6000 newborn infants, which is in accordance with the result of a similar study from the Netherlands.⁴ A recent study

from Sweden gave an incidence of 1 in 6900 live births,⁵ but that study included only cases diagnosed before age 2 years.

The incidence of congenital hypothyroidism also depends on the length of the observation period (Table 1). We found an incidence of 1 in 5100 live births in 6- to 8-year-old children, compared with 1 in 7400 in 3- to 5-year-old children.

The incidence of patients with a congenital defect of the thyroid gland is therefore probably higher than that found in the present (or previous) epidemiological study. In fact, the Danish neonatal screening programme for primary hypothyroidism gave an incidence of 1 in 4000 live births,¹¹ which is similar to the incidence reported from screening centres in Europe and North America.^{10 12-13}

The discrepancy between the results of the incidence calculations based on screening programmes and epidemiological investigations is probably explained by the observation that some patients are subclinically hypothyroid during early childhood and that the screening programmes also detect transiently hypothyroid patients. However, the incidence given by screening programmes and that found in epidemiological studies with a longer observation period is quite similar (1:4000 compared with 1:5000 newborns) and shows that neonatal screening programmes probably detect all children who will develop primary hypothyroidism in early childhood.

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