

Research Article

Reflections on the First 100 Plus Babies with Congenital Hypothyroidism (CH) Seen after the Start of the National Screening Programme (NSP) in 1998 in the United Arab Emirates (UAE)

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Submitted: 20 January 2020

Accepted: 31 January 2020

Published: 02 February 2020

ISSN: 2373-9312

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Keywords

- Congenital hypothyroidism
- National screening programme
- Babies

Abstract

Background: A comparison of outcomes for CH before and after the start of the NSP in the UAE.

Methods: Review of the findings of all the babies with CH seen in the period 1996 to 2004.

Results: Before the UAE NSP babies were usually diagnosed very late often with a bad outcome. The NSP resulted in much earlier diagnosis and better outcome. The incidence of CH was high compared to most other studies and eutopic hypothyroidism was nearly as common as ectopic hypothyroidism. Some of the former could come off treatment later and some of the latter were very mild.

Conclusion: The most important conclusion is that the NSP is very worthwhile with generally good outcomes.

INTRODUCTION

I worked in Tawam hospital, Al Ain, UAE from 1996-2004. I was one of the doctors involved in setting up of the National Screening Programme for CH (and Phenylketonuria, Congenital Adrenal Hyperplasia and Sickle Cell disease) in 1978. During my time there I was involved in the diagnosis and care of over 100 babies with CH plus a few that came my way from earlier.

Each time I saw a baby with CH I sent a letter to Dr. Reem M Zayed at the preventive medicine department in Al Ain-so many that the weight of them is over 3kg, about the weight of a newborn baby.

What follows is some conclusions and lessons learned from reviewing these letters and recollections from my time there.

METHODS

Before the National Screening Programme

Tawam Hospital was measuring cord FT4 and another

Hospital, the Oasis Hospital, was measuring cord TSH both for about a decade before the start of the NSP [1].

The seven children I have seen and have records on before the screening programme make grim reading.

The mother of 1 girl diagnosed at 2yrs old made the comment that before treatment "she sleeps all the time". Subsequently she did poorly at school.

One boy diagnosed at 2yr 5 months was initially suspected to have rickets because of bone abnormalities. At the correct diagnosis the FT4 was 7.1 and TSH 182!

He has an older sister who had a goitre seen at 2 months old, but was not investigated until 4 years old when she had a larger goitre. Fortunately both are developmentally normal. The girl was top of her class. The parents are first degree cousins.

Another child with a cord blood of FT4 of 7.6 was not treated until 3 weeks old when the FT4 was 3.1 and TSH >100. He was developmentally delayed and attended a school for retarded children.

Another boy treated from 3 weeks was delayed especially in speech.

One boy is hypothyroid and growth hormone deficient.

Of the 7 children who came my way diagnosed before the NSP 3 are delayed. Of course the outcome depends both on the delay before treatment and the severity of the hypothyroidism [2].

After the NSP started a number of children diagnosed with CH have older siblings with developmental delay giving further weight to the well known fact that early diagnosis and treatment is important [3].

The National Screening Programme

Overall it has been a great success, but there were a number of teething problems and some ongoing problems. In the Emirates there was a high take up except from Dubai. Even in Al Ain some pediatricians did not participate resulting in late diagnosis. One newborn baby had a 3 week hospital stay- poor feeding, then jaundice, then constipation leading to a suspicion of Hirschsprungs disease before the correct diagnosis was made. However, the coverage (percentage of neonates screened) in the Al Ain district went from 58% in 1998 to 98% in 2002. (Personal Communication from Dr Reem Zayed, head of Maternal and Child Health, Al Ain)

Non compliance occurred in some families. It was not uncommon in treated children for them to have a normal FT4 and high TSH from not taking the tablets until shortly before the clinic visit, not giving time to for the TSH to come down. Failure of understanding sometimes occurred with parents not realizing that the treatment had to continue. Mistakes in dosage and preparations sometimes occurred, for instance instead of crushing the tablets on the spoon and swallowing from the spoon, putting the whole dose in a glass of water. Going on holiday with insufficient tablets or leaving them behind occurred on 2 or 3 occasions.

Sometimes delayed starting treatment occurred because of a hold up somewhere along the chain of events leading to informing the treating doctor. Despite some shortcomings the programme overall has been very successful.

THE AETIOLOGY OF CH

The figures I have are:

Eutopic thyroids 55, males 28, females 27 (a few with maternal antibodies, but most presumably metabolic).

Ectopic 52, males 24, females 28. Athyreosis 13, males 7, females 6. Unknown 11, males 7, females 4 (most of the unknown are from the protocol not been followed and treatment started before a Tc scan).

Transient raised TSH with normal FT4 7, 4 males, 3 females. (Of course how one classifies this group depends on definition. We took over 25 as raised, but a Scottish study took over 40 as raised on the initial test) [4].

What is clear is that the ratio of Eutopic to Ectopic hypothyroid infants is much higher here than in Western studies and the

incidence of CH is also much higher than in most other studies (approximately 1 in 1500 here and 1 in 3500 elsewhere).

Transient hypothyroidism

With children with eutopic thyroids quite a number can come off treatment later even when the initial results show the hypothyroidism was severe. For instance baby KB had an umbilical cord FT4 of 6.4 and on the NSP a TSH of >300 and was able to stop treatment at the age of 3.5 years. It is known that even with a known metabolic defect some children can come off treatment [5]. The 3 with thyroid antibodies from the mother were able to come off treatment.

ECTOPIC THYROIDS

All, but one, of the children with ectopic thyroids were put on thyroxin presumably forever. However, some appear to be very mild. For instance baby OA with a small sublingual thyroid at 2 weeks before treatment started had an FT4 15.4 and TSH 47.5 and on just 25micrograms of thyroxin at 5 months had an FT4 of 17.1 and TSH 2.8 making me doubt whether I was right to treat him. Also baby girl BA with a sublingual thyroid has never had a low FT4 (subclinical hypothyroidism) and is only on 25micrograms at the age of 2. The one baby not put on thyroxin had an ectopic thyroid lower than sublingual, but in the hypoglossal region. Anecdotally I know of one man who only was found to be hypothyroid as an adult with an ectopic thyroid. I would not be surprised if there are some adults in good health not knowing they have an ectopic thyroid.

The children with Athyreosis not surprisingly had initial very low FT4s, one with an FT4 of 1 and one of 0. Serum thyroglobulin was very useful in confirming the diagnosis with levels less than 1. On occasions babies were initially diagnosed as having no thyroid but a measurable thyroglobulin showed there must be some thyroid activity somewhere [6].

Clinical findings before treatment

Most parents had no complaints about the baby and thought all was well, but at least two said the baby sleeps all the time. One mother said the only way that she knew the baby was hungry was when she moved her head. The words lazy babies quite often are used. Jaundice of course was common but phototherapy was only required in two cases. Baby girl MM with a screening TSH 116 with a Eutopic thyroid required phototherapy but at 2yrs was able to come off treatment. At the other end of the scale baby boy SM with athyreosis, FT4 0, was the other one who needed phototherapy. Only a few were diagnosed clinically before the Screening result, but there were more that should have been.

GOITRES

With children with eutopic thyroids goitres were quite common. With 2 children the goitres were so large that there were breathing problems. The goitres shrank once treatment was started.

Family history

There was a family history of thyroid problems in many cases. Baby NN's parents were second cousins and the baby ectopic hypothyroid. The father's brother was mentally retarded from

hypothyroidism presumably diagnosed late. Cousin marriage was quite common, at least 11 that I know of in this study. Baby AI was originally thought to be Athyreotic as no thyroid was seen on the Tc scan. However, he had a thyroglobulin of 516, so has a thyroid somewhere. He has a 5 year old brother who was diagnosed at 9 months and required special schooling. 3 mothers have a past history of thyrotoxicosis and 2 mothers were hypothyroid. The mother of baby FD was thyrotoxic 15yrs previously and had a thyroidectomy. The baby girl had a eutopic thyroid and needs thyroxine permanently.

The mother of baby boy AAD is on thyroxin. Her thyroglobulin antibodies were > 5000 (cut off point 225) and thyroid peroxidase antibodies 3577 (cut off point 35) His hypothyroidism was transient. His mother's history is interesting. 13 years prior to the baby's birth her hair was falling out and she was depressed and said she wanted to die. She was seen by a psychiatrist and given some medicine, but was no better. After the diagnosis of hypothyroidism was made and she was treated her depression went and she was fine since. It shows that in adults as well as children it can be very important to make the diagnosis early.

Nationality

Most were local but of the 131 total there were 11 Indian, 5 Pakistani, 1 Bangladeshi, 1 Omani, 1 Irani and 2 Sudanese families (21 in all).

ASSOCIATED CONGENITAL ANOMALIES

3 Downs syndrome (2 transient hypothyroidism and 1 with an AV canal cardiac defect and Hydronephrosis); 1 Growth hormone deficiency; 2 Ventriculo Septal defects; 1 Choanal atresia; 1 Bilateral glaucoma; 1 Accessary auricle; 1 Glandular hypospadias; 1 Cleft lip and palate and frontal meningoencephalocele; 1 Beta thalassaemia intermedia (This baby also had many acquired problems related to prematurity); 1 Diaphragmatic hernia. 12 in all out of 131, in some the hypothyroidism being the least of the problems

CONCLUSIONS

The National Screening Programme is certainly worthwhile

resulting in earlier diagnosis and treatment and better outcomes than before even early treated babies with athyreosis do well [7]. I know of only one infant with developmental delay probably attributable to hypothyroidism since the start of the NSP}

The incidence Of CH found here is about 1 in 1500 as opposed to about 1 in 3500 in Western studies. The ratio of Eutopic to Ectopic or Athyreotic is also much higher. Some of the Eutopic infants are able to come off treatment later even if there is a metabolic defect [5].

In the eutopic infants there is often a family history of thyroid problems. Some of the ectopic infants have very mild hypothyroidism. Thyroglobulin levels are useful in confirming Athyreosis.

There is a high incidence of associated congenital anomalies. Of course I agree with Robert Guthrie who stated "No child should die or suffer disabilities if a simple blood spot can prevent it"

REFERENCES

1. Hardy JD, Zayed R, Doss I, Dhatt GS. Neonatal Screening for Congenital Hypothyroidism: A Retrospective Hospital Based Study from Bahrain. *J Pediatr Endocrinol Metab.* 2010; 21: 245-249.
2. Bongers-Schokking, Koot HM, Wiersma D, Verkerk PH, de Muinck Keizer-Schrama SM. Influence of timing and dose of thyroid hormone replacement on development in infants with congenital hypothyroidism. *J Pediatr.* 2000; 136: 293-297.
3. Fuggle PW, Grant DB, Smith I, Murphy E. Intelligence, motor skills and behavior at 5yrs in early treated congenital hypothyroidism. *Eur J Paediatr.* 1991; 150: 570-574.
4. Ray M, Muir TM, Murray G, Kennedy R, Girdwood R, Donaldson M. Audit of screening programme for congenital hypothyroidism in Scotland, 1979-93. *Arch Dis Child.* 1997; 76: 411-415.
5. El- Desouki ML. Thyroglobulin measurements in Congenital Hypothyroidism. *Emirates Med J.* 1998; 16: 162-164.
6. Maynika V Rastogi, Stephen H LaFranchi. Even transient congenital hypothyroidism may be due to a genetic defect of thyroid hormone synthesis. *New Eng J Med.* 2002; 347: 95-102.
7. Wolter R, Noel P, de Cock P. Neuropsychological study in treated thyroid dysgenesis. *Acta Paediatr Scand Suppl.* 1979; 277: 41-46.

Cite this article

Hardy JD (2020) Reflections on the First 100 Plus Babies with Congenital Hypothyroidism (CH) Seen after the Start of the National Screening Programme (NSP) in 1998 in the United Arab Emirates (UAE). *Ann Pediatr Child Health* 8(1): 1167.