

# Screening for congenital hypothyroidism in North-West region of Saudi Arabia

Mohammad S. Al-Maghamsi, DCH, CABP, Zakaria M. Al-Hawsawi, MBBS, CABP,  
Ghulam N. Ghulam, MBBS, MD, Ahmad M. Okasha, PhD(UK).

---

## ABSTRACT

**Objectives:** The aim of this study is to demonstrate the prevalence of congenital hypothyroidism (CH) in Madina Al-Munawara region, Kingdom of Saudi Arabia (KSA) and to compare the results with other reported studies.

**Methods:** All deliveries conducted at the Ministry of Health Hospitals, Health Centers and Private Hospitals in Madina Al-Munawara region, KSA during the period from 1989 to 1999 were included in this study. Cord blood samples for thyroid stimulating hormone (TSH) measurement was used for screening.

**Results:** Out of 200,040 deliveries 193,613 infants were screened for CH during the study period. Forty-six infants had hypothyroidism with an overall prevalence of 1:4208

live births. Thirty-three (72%) of the diagnosed infants were Saudi and 13 (28%) were not Saudi. The male to female ratio was 1:3. The mean age at the start of treatment was 9 days.

**Conclusion:** The screening program based on the initial measurement of TSH in cord blood is detected by nearly 97% of the total infants born in Madina region over 10 years. The overall prevalence of CH was 1:4208 lower than other local studies. Mental retardation can be prevented by neonatal screening program; however, Neonatologists and Pediatricians should remain vigilant in searching for signs and symptoms of CH.

Saudi Med J 2002; Vol. 23 (12): 1518-1521

---

Congenital hypothyroidism (CH) is a relatively common endocrine disease, its prevalence showing marked geographical and racial variations, incidence varies from 1:3500-1:5000 live birth.<sup>1,2</sup> Al-Nuaim et al<sup>3</sup> reported an incidence of 1:2666 live birth from Kingdom of Saudi Arabia (KSA). Recent study at Riyadh Al-Kharj Hospital Programme, KSA showed an incidence of 1:2759 live birth,<sup>4</sup> most of the studies suggested that the incidences of CH in KSA is higher than other countries. Congenital hypothyroidism fulfills most of the criteria for disease screening as set out by Wilson and Jungner.<sup>5</sup> Congenital hypothyroidism leads to mental and physical retardation; however, early diagnosis and treatment can prevent neurological complications.<sup>6</sup> Thyroid hormone plays an important role in the

development and growth of the brain. Early treatment with thyroxine is essential to prevent neurological, mental and physical deficiencies.<sup>7</sup> The aim of this study is to demonstrate the prevalence of CH at Madina Al-Munawara region, KSA and to compare it with other similar local studies.

**Methods.** The CH screening program was started in Madina region, KSA on September 1989. All deliveries conducted at the Ministry of Health Hospitals, Health Centers and Private Hospitals during the period from 1989 to 1999 were included in this study and were screened for CH. Madina region is located in the North Western Province of KSA, with an estimated population of 1.5 million.

---

From the Department of Pediatrics (Al-Maghamsi, Al-Hawsawi), Department of Pediatrics & Neonatology (Ghulam), Medina Maternity & Children's Hospital, and the Department of Laboratory (Okasha), Madina Al-Munawara, Madina, Kingdom of Saudi Arabia.

Received 10th July 2002. Accepted for publication in final form 10th September 2002.

Address correspondence and reprint request to: Dr. Mohammad S. Al-Maghamsi, Consultant Pediatrician, Department of Pediatrics, Medina Maternity & Children's Hospital, PO Box 5019, Madina Al-Munawara, Kingdom of Saudi Arabia. Tel. +966 (4) 8361000 Ext. 2616. Fax. +966 (4) 8383385. E-mail: msfaleh@yahoo.com

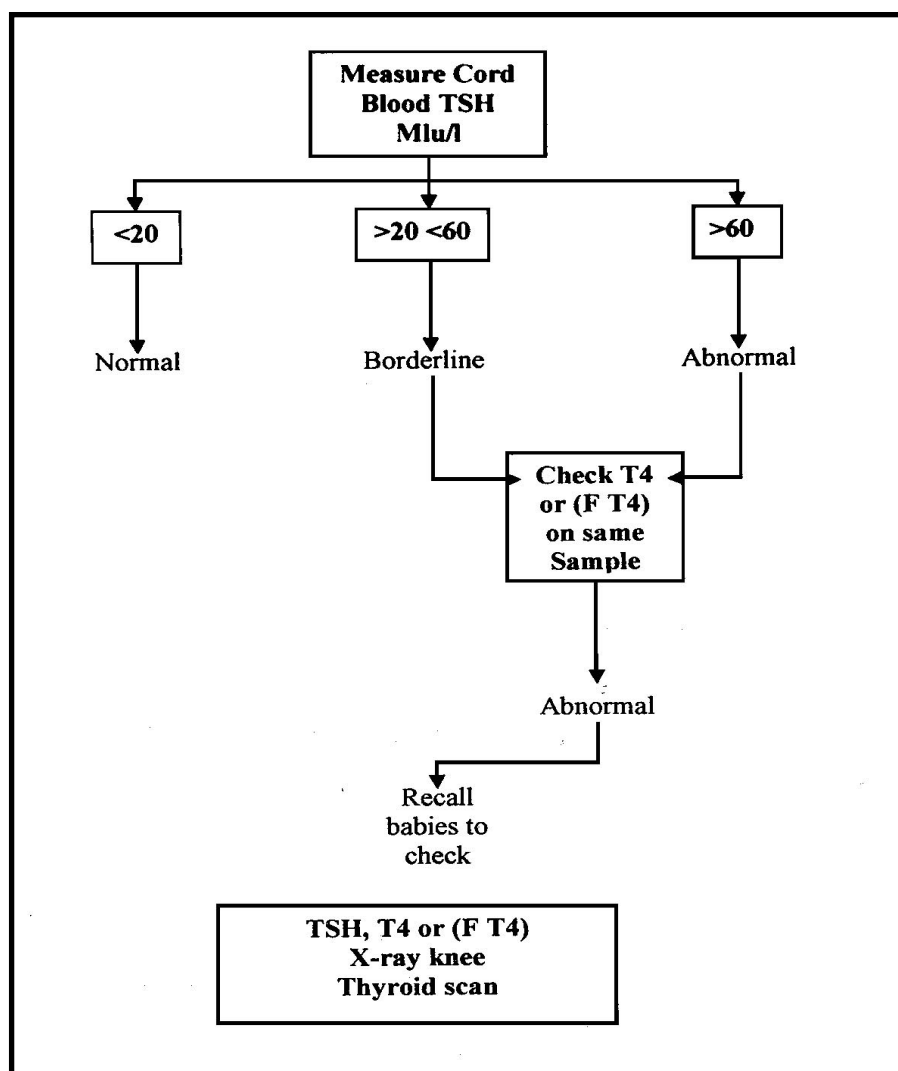


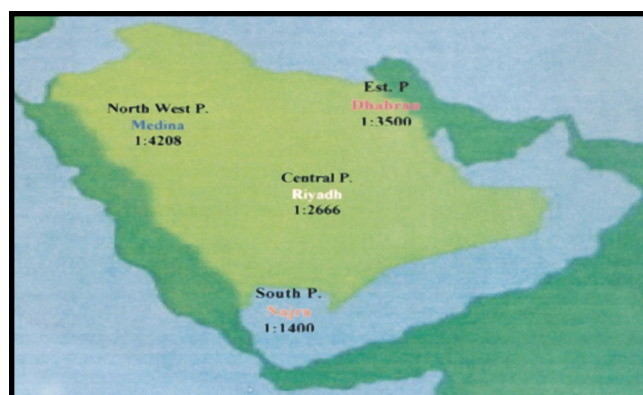
Figure 1 - Algorithm for TSH interpretation. TSH - thyroid stimulating hormone, T4 - thyroxine, FT4 - free thyroxine.

Cord blood samples was used for screening. Three ml of cord blood was collected in sterile tube. Thyroid stimulating hormone (TSH) measurement was used for screening, Delphia immunofluorescent kits (Pharmacia Diagnostic Wallacoy, Finland) were used, and the cut-off level was 60 mlu/L. **Figure 1** shows the algorithm of TSH interpretation. Thyroid scan was performed only in 20 patients due to non availability of Isotope material during the study. Technetium <sup>99m</sup> pertechnetate was used in imaging of thyroid gland. The infants with confirmed hypothyroidism were started on thyroxin treatment and they were followed up regularly at the Endocrinology Clinic.

**Results.** The number of infants born between September 1989 to September 1999 in Madina region, KSA were 200,040. A total of 193,613

infants were screened for CH representing 96.8% of the total live births, and 46 were diagnosed with CH. The overall prevalence was 1:4208 live birth, 33 (72%) infants were Saudi and 13 (28%) were non Saudi, the male to female ratio was 1:3. In 20 patients, thyroid scan was performed; 11 patients had athyreosis, 6 patients had thyroid ectopia and 3 patients had dysmorphogenesis. The mean age at the start of treatment was 9 days. **Figure 2** demonstrates the prevalence in different region of KSA.

**Discussion.** This is the first report on large-scale regional screening program to detect infants with CH in Madina Al-Munawara region. A similar study was reported earlier from Riyadh region of KSA by Al-Jurrayan et al.<sup>8</sup> The prevalence of CH in



**Figure 2** - Prevalence of congenital hypothyroidism in neonates in different regions of the Kingdom of Saudi Arabia. P - province.

our study was 1:4208, which was consistent with world wide incidence of 1:4000 live birth.<sup>9</sup> However, it was lower compared to the reports from other part of KSA: Najran 1:1400<sup>10</sup> Riyadh 1:2666<sup>11</sup> and Dhahran 1:3500<sup>12</sup> It is also lower than they reported from the West Bank part of Palestine 1:2070.<sup>13</sup> The possible reason for low prevalence of CH in Madina is that Madina is a Holy place and Moslems had migrated over the years from all over the world to this place. The population is heterogeneous including Afro-Asian races. Racial factor is known to influence the prevalence.<sup>14</sup> All affected infants were full term; however, high incidence has been reported in preterm infants.<sup>15</sup> The male:female ratio was 1:3 which is consistent with most international studies.<sup>4,16</sup> The screening program in Madina region was based on the measurement of cord blood TSH which is considered the most sensitive test for the diagnosis of CH,<sup>17</sup> all the cases with borderline TSH level (20-60mlu/g proved to be normal on further evaluation, and the cut-off level of 60 mlu/L is more sensitive. However, cut-off of 50 mlu/L which was used in several screening programs would be more appropriate.<sup>18,19</sup> The thyroid scan is an important test in determining the etiology of CH. Among the investigated cases, athyreosis was accounted for 55%, followed by thyroid ectopia with 30% and dyshormonogenesis with 15%. These findings in line with those of Henry et al<sup>4</sup> and in contrast sharply with those of Al-Jurrayan et al,<sup>8</sup> who found 24% athyreosis, 50% thyroid ectopia and 26% defective organification, none of our affected infants had clinical features of CH in the neonatal period, the characteristic signs and symptoms of CH are rarely seen in the neonatal period. Clinical feature of CH can be present approximately in 10% of infants in the first month and in 35% of infants within 3 months of life,<sup>11</sup> the mean age of starting the treatment was 9 days (range 5-30). This is the real measure of the success of the screening program, as the

conventional wisdom supported by various studies<sup>20,21</sup> is that initiation of treatment before the age of 3 months generally conduces to satisfactory physical and mental development in affected infants. Follow up of affected infants revealed a normal physical and mental development. It has been observed that the progress for psychomotor development of infants with CH is quite good provided that the treatment is started in the first 6 months of life.<sup>22</sup> Although neonatal screening program for CH is considered as one of the advances in diagnosis and treatment of endocrine disease in the newborn period; however, it is not 100% perfect and approximately 8-10% of cases can be missed in the screening program, for such reason Pediatricians must remain alert to the possibility of missed CH presenting in infancy.<sup>23,24</sup>

In conclusion, the screening program based on initial measurement of TSH in cord blood, detected nearly 97% of the total infants born in Madina region between 1989 and 1999. The overall prevalence of CH was 1:4208 live births lower than other screening programs in KSA. We also conclude that mental retardation can be prevented by a neonatal screening program; however, Neonatologists and Pediatricians should remain vigilant in searching for signs and symptoms of CH.

**Acknowledgment.** The authors would like to express their thanks to the staff of the Pediatric and Laboratory Departments for their help and cooperation. Thanks also to Dr. Ahmad Habib Mahmoud (former Director of the Hospital), for allowing us to undertake this study, and to Miss. Darna Sarail Alie for Secretarial Assistance.

## References

1. Barnes ND. Screening for congenital hypothyroidism the first decade. *Arch Dis Child* 1985; 60: 587-592.
2. Fisher DA, Dussault JF, Foley TP, Klein AH, Lafranchi S, Larsen PR et al. Screening for congenital hypothyroidism: results of screening one million North American infants. *J Pediatr* 1979; 94: 700-705.
3. Al-Nuaim A, El-Desouki M, Al Jurrayan N, Eissa M. Neonatal screening for congenital hypothyroidism: incidence, imaging feasibility and difficulties for nationwide programme. *Annals of Saudi Medicine* 1992; 12: 129-134.
4. Henry G, Sobki SH, Othman JM. Screening for congenital hypothyroidism. *Saudi Med J* 2002; 23: 529-535.
5. Wilson JMG, Jungner G. Principles of screening for disease. Public health papers 34. Geneva: World Health Organization; 1968. p. 26-39.
6. Tan SH, Wong HB. Congenital hypothyroidism in Singapore: pre-screening era. *Aust Paediatr J* 1988; 24: 148-152.
7. Hsiao PH, Chiu YN, Tsai WY, SU SC, Lee JS, Soong WT. Intellectual outcomes of patients with congenital hypothyroidism not detected by neonatal screening. *J Formos Med Assoc* 1999; 98: 512-515.
8. Al-Jurrayan NA, Al Nuaim AA, Redha MA, Al Dessouki MI, Al Herbish AS, Abo-Baker AM et al. Neonatal screening for congenital hypothyroidism in Riyadh: analysis of six years experience. *Annals of Saudi Medicine* 1996; 16: 20-23.

9. Dreimane D, Varma SK. Common childhood thyroid disorders. *Indian J Pediatr* 1997; 64: 3-10.
10. Al Jurrayan NA, Shaheen FL, Al Nuaim AA, El Desouki MI, Faiz A, Al Herbish AS et al. Congenital hypothyroidism: Increased incidence in Najran Province, Saudi Arabia. *J Trop Pediatr* 1996; 42: 348-351.
11. Al Jurrayan NA, Al Nuaim A, El-Desouki M. Congenital hypothyroidism: Clinical and laboratory characteristics in infants detected by neonatal screening. *Annals of Saudi Medicine* 1992; 12: 135-139.
12. Abuosba YK, Mallouh A, Salamah M, Hann R, Thalji A, Hamdan J et al. Comprehensive newborn screening program: Aramco experience, the national need and recommendations. *Annals of Saudi Medicine* 1992; 12: 235-240.
13. Sack J, Feldman I, Kaiserman I. Congenital hypothyroidism screening in the West Bank, a test case for screening in developing regions. *Horm Res* 1998; 50: 151-154.
14. Brown AI, Fernhoff PM, Milner J, Mcewen C, Eisas L. Racial differences in the incidence of congenital hypothyroidism. *J Pediatr* 1981; 99: 934-936.
15. Delange F, Dalhem A, Bourdoux P, Lagasse R, Glinoeir D, Fisher DA et al. Increased risk of primary hypothyroidism in preterm infants. *J Pediatr* 1984; 105: 462-469.
16. Mikelsaar RV, Zordania R, Viikmaa M, Kudrjartser G. Neonatal screening for congenital hypothyroidism in Estonia. *J Med Screen* 1998; 5: 20-21.
17. Christenson RA. Thyroid function test and their interpretation. *Indian J Pediatr* 1997; 64: 21-26.
18. Verkerk PH, Buitendijk SE, Verlove Vanhorick SP. Congenital hypothyroidism screening and the cut-off for thyrotropin measurement; recommendation from the Netherlands. *Am J Public Health* 1993; 83: 868-871.
19. Price DA, Erlich RM, Wolfish PG. Congenital hypothyroidism. Clinical and laboratory characteristics in infants detected by neonatal screening. *Arch Dis Child* 1981; 56: 845-851.
20. Jacobsen BB, Brandt NJ. Congenital hypothyroidism in Denmark: incidence, type of thyroid disorders and ago at onset of therapy in children 1970-1975. *Arch Dis Child* 1981; 56: 134-136.
21. Klein R. History of congenital hypothyroidism. In: Burrow GN, Dussault JH, editors. Neonatal thyroid screening. New York (NY): Raven Press; 1980: p. 51-59.
22. Kmianou F, Makaronis G, Lambaridis J, Sarafidou E, Varchni F, Mangreli C et al. Psychomotor development in congenital hypothyroidism, the Greek Programme. *Eur J Pediatr* 1988; 147: 275-278.
23. Willi SM, Moshang T. Diagnosis dilemas results of screening tests for congenital hypothyroidism. *Pediatr Clin North Am* 1991; 38: 555-566.
24. Fisher DA. Effectiveness of newborn screening for congenital hypothyroidism, prevalence of missed cases. *Pediatr Clin North Am* 1987; 34: 881-890.

## Related Abstract

### Source: Saudi MedBase



Saudi MedBase CD-ROM contains all medical literature published in all medical journals in the Kingdom of Saudi Arabia. This is an electronic format with a massive database file containing useful medical facts that can be used for reference. Saudi Medbase is a prime selection of Abstracts that are useful in clinical practice and in writing papers for publication.

#### Search Word: congenital hypothyroidism

##### Authors:

Nasir A. M. Al-Jurrayan, Abdul-Rahman A. Al-Nuaim, Merfat A. Redha, Mahmoud I. El-Desouki, Abdullah S. Al-Herbish, Abdullah M. Abo-Bakr, Abdulrahman A. Al-Swailem, Yaqoub Y. Al-Mazrou, Abdullah Al-Deress

##### Institute:

King Khalid University Ministry of Health, Riyadh, Saudi Arabia

##### Title:

Neonatal screening for congenital hypothyroidism in Riyadh: Analysis of six years experience

##### Source:

Annals of Saudi Medicine 1996 January, 1 : 20-23

#### Abstract

The regional screening program for congenital hypothyroidism (CH) in Riyadh province was started as a pilot study in December 1988. By september 1990, the program covered all deliveries at the Ministry of Health (MOH) and King Saud University Hospitals. The program utilizes cord serum thyroid-stimulating hormone (TSH) with a low recall rate of 0.1%. The average cost per specimen was SR 12 (US\$3.20). Among 283,647 infants screened, 83 infants were confirmed to have CH (incidence 1:3417). In 17 infants, however, the diagnosis was not confirmed due to difficulties in recall. Eleven infants with cord serum TSH of more than 100 mu/l proved to be clinically and biochemically euthyroid at recall. Three of these were secondary to maternal propylthiouracil (PTU) therapy. The female to male ratio was 1.5: 1. The majority of infants lacked clinical symptoms and signs of hypothyroidism. Thyroid scans showed the most common etiology to be thyroid ectopy (50%), followed by dyshormonogenesis (26%) and athyrosis (24%). Although there was no significant difference in the mean cord T4 values among the different groups, the mean T4 value at recall in the athyrotic group was significantly ( $p < 0.001$ ) lower than the cord results (14.8 versus 62.7) and that is of recall for the ectopic and dyshormonogenesis groups (14.8 versus 47 and 51.3 respectively). There was no significant difference in the mean TSH among the different groups in the cord and recall samples. Skeletal maturation was more delayed in the athyrotic group. The mean age at the time of recall was 16.4 days (range 4 to 64), and the mean age at the start of therapy was 17.8 days (range 5-64).