Original Article

Congenital Hypothyroidism: An Underestimated Clinical Entity

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ABSTRACT

Objective: The aim of study was to determine the clinical features of Hypothyroid Children in various age groups residing in Rahim Yar Khan.

Study Design: Prospective, descriptive, study, over a span of one year.

Place and Duration of Study: This study was conducted in Pediatrics department at Sheikh Zayed Hospital and Medical College Rahim Yar Khan from 1st March 2010 to March 2011 .

Materials and Methods: The thirty children of both sexes in age group of 1 day – 12 years presenting with clinical features, suspicion of Hypothyroidism were included in study. Children with subtle as well as full fledged sign and symptoms of hypothyroidism were considered for further evaluation. The clinical history, feeding pattern and clinical examination of these cases were recorded. Their blood was examined for T4, T3 and TSH. The X-rays of chest, X-rays of Skelton, particularly X-ray wrist and X-ray knee joint were obtained. Serum cholesterol, glucose, electrolytes (Na^+ & K^+) and perchlorate ($KClO_4$) discharge test were obtained. Thyroid scintigraphy was done for isotope uptake. Ultrasonography for thyroid position, its size and detection of solid and cystic lesions was performed.Cardiac monitoring was gained by E.C.G. The data analysis was done by software SPSS 16.

Results: All children with clinical features and investigations in favor of hypothyroidism were kept in four groups according to the age. The sluggish behavior, bradycardia, typical coarse faces and TSH in values above normal range were consistent findings in all thirty (n=30) cases.

Conclusion: Congenital Hypothyroidism is usually under diagnosed that leads to mental retardation in a child. Therefore any child having suspicion of hypothyroidism should be evaluated by thyroid functions test (TFT), including new born screening.

Key Words: Congenital Hypothyroidism, TFT, Children, New born screening.

INTRODUCTION

Congenital hypothyroidism (CH) has worldwide prevalence of 1:2,000 to 1:4,000 newborns.[1] It is the commonest cause of mental retardation in children.[2] The clinical features may not present in the early infancy due to trans-placental passage of small quantity of thyroxin; therefore the clinical diagnosis is usually missed at even established maternity centers.[3] Congenital hypothyroidism in 85% of the cases is due to thyroid dysgenesis while in the remaining 10-15% the dyshormonogeneses is present.[4] Thyroid dysgenesis may occur as agenesis in 40% of cases and ectopic or rudimentary in the remaining 40% of cases. Congenital hypothyroidism presents as syndromic, nonsyndromic, familial, endemic and sporadic. The familial cases like Pendred's syndrome are autosomal recessive.[5] In Down syndrome the occurrence of hypothyroidism is 1:400[6]

The meticulous clinical examination of the new born and any infant with delayed milestones is an early essential step for the detection of hypothyroidism. The epiphyseal center for the lower end of femur develops under the influence of thyroid hormone. In our setup even at the basic health unit the simplest investigation is X-ray of the knee joint for location of epiphysis center of femur in a new born which can detect the hypothyroidism. Thyroid hormone is also essential for the growth of body tissues, particularly brain, in fetal life.[7] Thereafter the absence of this epiphyseal center in the presence of clinical features like hoarse cry, hypothermia and bradycardia must warrant the sampling for TSH(Thyroid stimulating hormone) .The aim of our study is to emphasize the importance of clinical presentation, X-rays, TFT and new born screening to diagnose CH for early intervention by starting oral thyroxin to reduce mental retardation cases in our community.The new born thyroid screening tests has entirely changed the spectrum of mental retardation in children in the countries where this strategy has been applied for the last many decades.[8]

MATERIALS AND METHODS

The thirty children of both sexes in age group of 1 day – 12 years having clinical features suspicion of hypothyroidism referred to pediatrics unit Sheikh Zayed

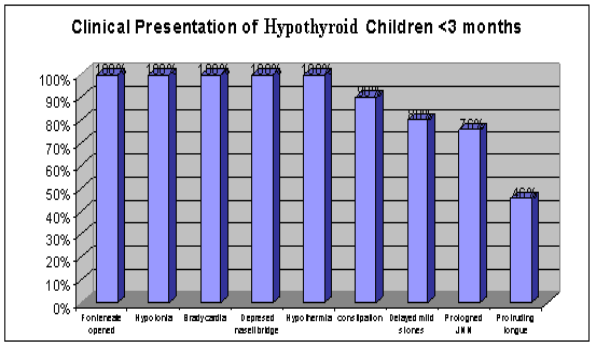
Hospital Rahim Yar Khan were included in the study. The transient hypothyroidism and Down syndrome cases were excluded. The four groups were constructed according to the age. The maternal history, birth history, feeding, developmental milestones, weight and height with clinical sign and symptoms were recorded on a Proforma. The new born babies born in labor room of this hospital presenting with apneic spells, hypothermia, bradycardia and hoarse cry were immediately transported to nursery of the pediatrics unit, managed and evaluated for the study. The other clinical features like prolonged icterus neonatarum, opened fontnellae, feeding and developmental milestones were assessed in the cases in postnatal period. Constipation, pallor, periorbital edema, coarse skin, sleepiness, poor appetite, hearing defects, intelligence quotient (IQ), schooling, social activities, were the features considered in other groups. Goiter size was evaluated according to WHO grading i.e.; grade I and grade II.

Their blood was evaluated for T4, T3 and TSH. The-rays of chest, X-rays of Skelton, particularly X-ray wrist and X-ray knee joint were obtained. Serum cholesterol, glucose, electrolytes (Na^+ & K^+) and per chlorate ($KClO_4$) discharge test were obtained. Thyroid scintigraphy was done for radioisotope uptake. Ultrasonography for thyroid position and description of lesion was performed. Cardiac monitoring was gained by ECG. Hearing was assessed by distraction method. These cases were referred for audiometry. Pure Tone Average (PTA) audiometry, otoacoustic emission (OAE) testing/Auditory brainstem response (ABR) were recommended. The data analysis was done by SPSS16.

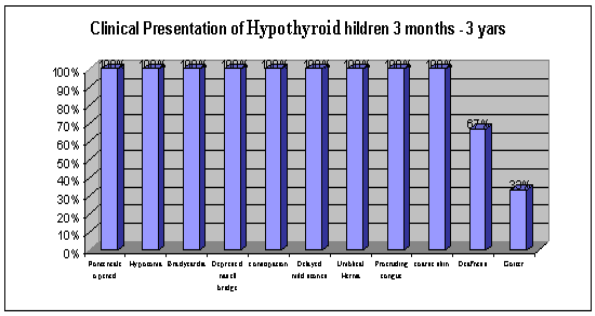
RESULTS

Among the thirty children seventeen (n=17) were male and thirteen (n=13) were female with the male to female ratio of M:F 1:3. They were in age range of 1 day –144 months. The mean weight (group 1, <12 months) was 3.8786 kg. ± 1.54331 std, and the mean height was 56.07 cm ± 6.604 std. The mean weight (group 2, 13-36 months) was 10.500 kg ± 1.3228 std, the mean height was 83.33 cm ± 5.686 std; the mean weight (group 3, 37-60 months) was 12.400 kg ± 9.6177 std, the mean height was 92.00 cm ± 1.414 std; the mean weight (group 4, 61-144 months)was 21.500 kg ± 4.37526 std, the mean height was 106.12 cm ± 7.549 std .All the children were short stature. The figure 1 depicts the clinical features of a ten year hypothyroid, whose carpal bones are showing only two ossification centers on X-ray wrist, therefore his bone age was < 1 year(figure 2). The sign and symptoms of hypothyroid children in different four age groups are shown in graphs 1,2,3,4 respectively. The TSH was above the

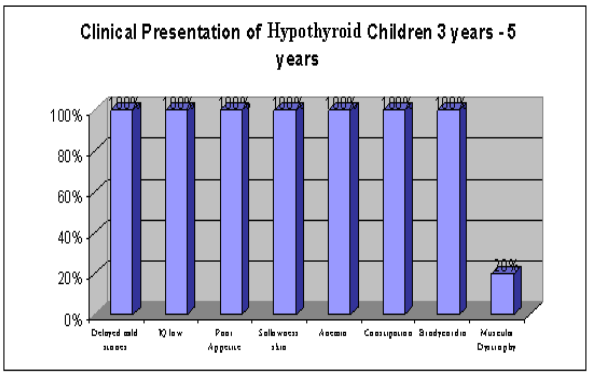
Graph No.1



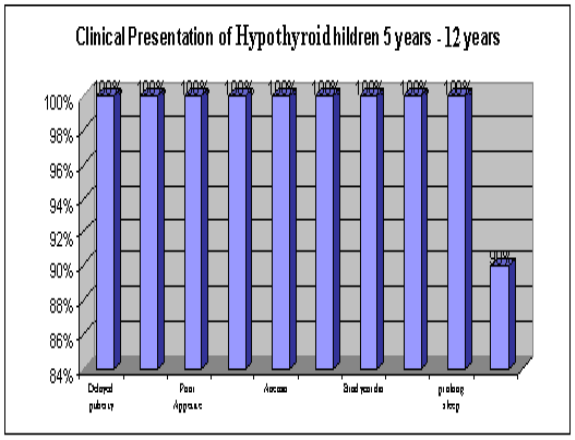
Graph No.2:



Graph No.3:



Graph No.4:



normal reference range of 20 Uml/l and serum cholesterol was also above 200 mg in all cases. The per chlorate discharge was > 40% in three (n=3) cases. The clinical presentation of hypothyroid children different age groups are shown in the graphs.

Figure No. 1: A ten year hypothyroid

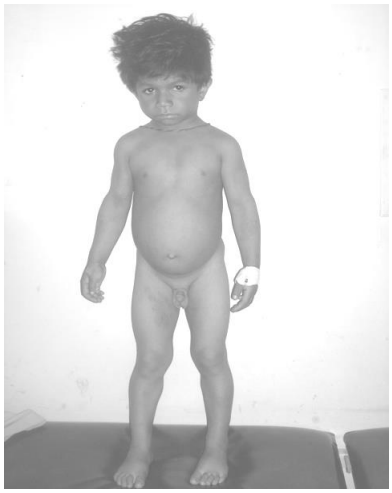
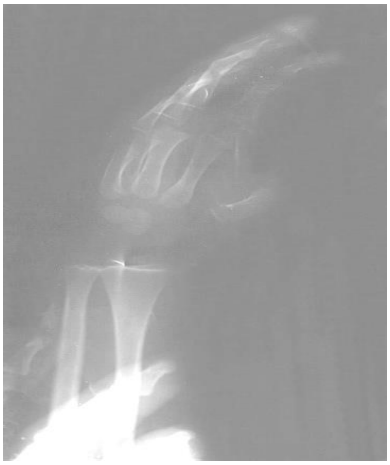


Figure No.2: X-ray wrist for bone age



DISCUSSION

Congenital Hypothyroidism is one of the commonest endocrine disorders in children.[9] The prevalence of disease in Pakistan has been reported 1:1000.[10] The occurrence is even higher 1:140 in Down syndrome. The endemic hypothyroidism has been recorded in the northern areas of Pakistan like Gilgat and Sawat.[11] UNICEF, in 1994, has declared Pakistan[12] as one of the severely affected regions of the world regarding iodine deficiency disorders (IDD) but another study has shown the severity was not much. Rahim Yar Khan is a plane area at a distance from the sea and there is no exact record of iodine deficiency in this area. In the study therefore the endemic hypothyroidism is less commonly seen but the sporadic cases due to maternal thyroid disease, familial like pendred's syndrome have been seen.[13] The clinical features in the study have a pivotal role in the diagnosis of congenital hypothyroidism right from the delivery of the new born to adulthood. Most of the studies on IDD depend upon the evaluation of TFT but this study has highlighted the importance of clinical features and X-rays Skelton.[14] Of course this approach will be suitable in our undeveloped areas where facilities for TFT and other advance investigations are lacking. However later on TFT may be obtained but there will be no delay in commencing the thyroid hormone replacement.

Surprisingly more than thirty percent (n=10)children were diagnosed in outpatient department for treatment of other ailments. A few cases were brought for medical certificates to get the financial help from the Government sector provided to disabled children. It is pity that the children of twelve years or even more then twelve year age were still in the lap of their parents. These children were taken as innocent and generous "God given Bhola". They were brought up as such without looking the advice of clinician. The different studies have focused the clinical assessment as a main diagnostic tool in congenital hypothyroidism. Thyroid hormone replacement is the main stay of treatment. Luckily the treatment is cheaper as compared to treatment of other endocrinal disorders.[15]

Dwarfism is difficult to diagnose and treat in other endocrine disorders in children.[16] The limitation of treatment by growth hormone due to high cost and unavailability to poor patients is one of the drawbacks in the treatment of short stature children. However the replacement of thyroid hormone in the early stages of congenital Hypothyroidism can prevent the stigmata of dwarfism in hypothyroids.

Thyroid hormone replacement in the form of tablet thyroxin can also prevent the neurodeficit like nerve deafness and mental retardation. Maintainence therapy and strict follow-up are prudent. Initially the follow-up should be at 2and 4 weeks after starting of therapy, then every 1-2 months for the first year, and 2 weeks after change of doses. The prognosis, particularly neurological depends upon the age of hypothyroid at the time of initiation of thyroid homone. The earliest start of Thyroxin in early neonatal life can prevent nerve deafness and mental retardation.[17] New born screening for congenital hypothyroidism is an urgent need in our country.[18] The new born screening program had been launched in developed countries since decades.[19] This strategy has reduced the incidence of mental retardation in those countries due to in born errors of metabolism.[20]

CONCLUSION

Congenital Hypothyroidism is usually under diagnosed that leads to mental retardation in a child. Therefore any

child having suspicion of hypothyroidism should be evaluated by thyroid functions test (TFT) including new born screening. Least but not the last the clinical features plus bone age on X-rays be enough to start thyroid hormone replacement till the availability of TFT.

REFERENCES

1. Park SM, Chatter jee VKK. Genetics of Congenital Hypothyroidism: J of Med Genetics 2005; 41(5): 379-389.
2. Haneef SM, Sajid M, Arif MA. Disease of Thyroid Gland: Hypothroidism: In: Text book of pediatrics. 5th ed. Pakistan Pediatric Association; 2007-2008. p.625-627
3. Maynika V, Rastogi, Stephen H, Franchi LA. Congenital hypothyroidism: Orphanet J Rare Dis 2010;5: 17.
4. Stephen H, Franchi LA. Disorders of the Thyroid Gland; Hypothyroidism: In: Behrman RE, Kliegman RM, Jenson H, Stanton, editors. Nelson Textbook of Pediatrics. 18th ed. Philadelphia: W.B. Saunders Company; 2007.p.1872-1873.
5. Meena Desai: Hypothyroidism. In: Nair MKC, Menon PSN, editors. IAP Text Book of Pediatrics 3rd ed. New Delhi: A Parthasarathy; 2006.p.587-588:
6. Gomella, Cunningham, Eyal. Zenk; Thyroid Disorders: Congenital Hypothyroidism. In: Graw Mc, Hill, editors. Lange Neonatolog. 5th ed. 2007.p.586-587.
7. Khan HI. Mandatory newborn screening for congenital hypothyroidism. Pak Pead J 2010; 34(3):121-2.
8. Najam Y, Khan M, Ilahi F, Alam A. Neonatal Thyroid Screening – The Shifa Experience J Pak Med Assoc 2002; 52(2):58-61.
9. Abhyankar S, Michele A, Puryear L, Goodwin R, Copeland S, Eichwald J, et al. Standardizing Newborn Screening Results for Health Information Exchange. AMIA Annu Symp Proc 2010;1–5.
10. Lone SW, Ibrahim MN, Atta I, Leghari T, Khan YN, Raza J. Nine years experience of congenital hypothyroidism; an urgent need for mandatory newborn screening. Pak Pead J 2010; 34(3):123-7.
11. Akhtar T, Zahoor Ullah, Paracha PI, Ghosia L. Impact assessment of salt iodization on the prevalence of goiter in district Swat. Pak J Med Sci 2004; 20(4):303-7.
12. Ghayur S, Siddiqui S, Alam MM, Shaukat A, Khan FA. Spectrum of lodine. Deficiency in School Children of Rawalpindi. Pak Armed Forces Med J 2001;51(1):27-32
13. Khurram IM, Choudhry KS, Muhammad K, Islam N. Clinical presentation of Hypothyroidism: A case control analysis. J Ayub Med Coll Abottabad 2003;15(1):45-9.
14. Golbahar J, Al-Khayyat H, Hassan B, Agab W, Hassan E, Darwish A. Neonatal screening for congenital hypothyroidism: a retrospective hospital based study from Bahrain. J Pediatr Endocrinol Metab 2010;23(1-2):39-44.
15. Jabbari A, Besharat S, Razavianzadeh N, Moetabar M. Common signs and symptoms in hypothyroid-dism in central part of Iran. Pak J Med Sci 2008;24(1):44-7.
16. Luxon LM, Cohen M, Coffey RA, Phelps PD, Britton KE, Jan H, et al.l Neuro-otological findings in Pendred's syndrome. Int J Audiol 2003; 42: 82-8.
17. Malik BA, Butt MA. Is delayed diagnosis of hypothyroidism still a problem in Faisalabad, Pakistan. J Pak Med Assoc 2008;58(10):545-9.
18. Abbas HG, Shan Elahi, Khan MS, Naeem M. Serum Thyroxine (T4) and Thyroid Stimulating Hormone (TSH) levels in cord blood of Newborns in Lahore. Pak J Med Sci 2003; 19(3):211-6.
19. Najam Y, Khan M, Ilahi F, Alam A. Distribution of T4 TSH values in children – the Shifa experience. J Pak Med Assoc 2003; 53(1):26-8.
20. Ahmed B, Hussain T, Memon AR, Solangi GA. Clinical Presentation of Primary Hypothyroidism. J Coll Physicians Surg Pak 2001; 11(11):676-8.

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