Summary and Conclusion

The screening of newborn children for metabolic and endocrinal disorders is one of the marvelous successes of the health service. This study aimed to evaluate the indicators of congenital hypothyroidism (CH) screening program in Fayoum in terms of coverage, effectiveness of detecting and managing the cases. A record review was conducted at the Fayoum health insurance hospital and data from Fayoum Health Directorate during the period from October 2002 to December 2012 to evaluate indicators of the program.

The program for neonatal screening was started at Fayoum governorate at October 2002, where the coverage rate was 63%. In subsequent years the increase in coverage rate progressed relatively fast to 86% in 2003, then in next years (2004-2012) it approaches mean percentage of coverage 96%.

During the study period among the 825737 babies who underwent the screening test, **1849** neonates of them were recalled for confirmatory tests. Thus, the recall rate in our study was estimated as 0.2%. The mean age of the recalled neonates was 12.6 ± 10 days.

400 (21.6%) recalled neonates were diagnosed with CH on the basis of the confirmatory serum T4 and TSH levels. The total positive predictive value of 21.6%.

The incidence of CH was calculated as 1: 2064 screened newborns during this period (2002-2012).

The newborn screening test (1st dry sample) was performed on the 3rd -7th day of life in 358 (89.5%) of the babies diagnosed with CH, 31 babies (7.7%) early in first 2 days, and 11 babies (2.8%) had their tests after the 7th day due to intercurrences at birth.

The mean TSH levels obtained from heel-prick blood samples (1st dry sample) was 85 ±74 mIU/L. The mean serum TSH and T4 levels of neonates diagnosed with hypothyroidism were 86.1 ± 26.6 μIU/mL and 7.3 ± 5.7 ng/dL respectively.

Of the 400 babies diagnosed with CH, 189 (47.2 %) were females, 211 (52.8%) were males, representing a Female/Male ratio of 1:1.1.

Mean age at confirmation (venous sample) was 14 days ±10 (range from 5 to 106). Once the diagnosis was confirmed, therapy was started between day 6 and 113 after birth.

In 259 primarily diagnosed CH patients through the neonatal screening Program from October 2002 to December 2009, 55 patients proved to have transient hypothyroidism (21.7%) and 204 patients with permanent hypothyroidism (78.3%).

CONCLUSION

Based on the statistical data of the high incidence of congenital hypothyroidism in Fayoum Governorate and the success achieved in early detection, treatment and follow up we stress the importance of program continuity and necessity of strengthening program capacity. Indicators of program quality are improving progressively despite of obstacles. Every effort should be done to enhance the performance of the program in all aspects.

The background information gained through studies such as this will be critical for the planning of appropriate genetic and immunologic studies in our Governorate.

RECOMMINDATIONS

In the light of our study present findings, the following are recommended:

1- It should remain one of the program goals to achieve 100% coverage.

2- Efforts to build enhanced direct communication systems, linking curative newborn screening programs to community-based primary care centers, should continue. Such systems would help to enhance timely follow-up for screen-positive infants and facilitate information sharing among those involved in caring for children.

3- Pre-screening education should be emphasized, and all pregnant ladies should receive proper information about this screening program from the Maternity and Child Health units. One of the easiest ways of providing basic education to parents is through a simple programme brochure or programme information sheet. Pamphlets which explain the newborn screening programme, its requirements and its importance, should be developed. Such pamphlets are best distributed to expectant mothers through gynecologists, midwives, in public health maternity clinics and through pre-natal training classes, if available. They should also be available at hospitals as part of any general information on newborns given to the mother.

4- Every effort should be made to carry out newborn screening programs in the ideal proper time in order to ensure early diagnosis and to start treatment as early as possible. To reach this goal, it is important to set time patterns for each step of the screening process, because delays in one or more steps will lead to unacceptable delays in the treatment of the affected neonate.

5- Campaigns directed to healthcare professionals should be conducted in Fayoum Governorate, so that they would instruct parents regarding the importance of early testing (up to the 7th day of life), and alert the basic healthcare unit professionals about the importance of collecting blood samples properly, and quickly sending the patients to the reference laboratory

6- Sample for screening should be taken daily and be sent to the laboratories 3 times a week.

7- Every effort should be done to decentralize services with a view to expand the delivery of health care. This could be accomplished through making central laboratory in Fayoum Governorate for confirmatory samples; this will save much time consumed by the patients and will lead to rapid start of treatment.

8- However it is costly but future plan to use combined TSH-T4 screening technique should be considered in order not to miss the central CH cases.

9- Every effort should be made to ensure that every newborn diagnosed with CH should undergo imaging (thyroid scan ± ultrasound) for distinction between potentially transient and clearly permanent congenital hypothyroidism.

10- However there is no consensus for conducting screening tests for congenital anomalies, careful physical examination is important, and the child should be referred for evaluation if any abnormality is detected. The early detection of other malformations in patients with CH may modify the prognosis of these patients.

11- Start treatment even in low dose for those newborn with elevated confirmatory TSH and normal T4.

12- Updated guidelines to specify the desired time for treatment cessation and follow up of cases after discontinuation of oral hormone substitution are needed.

13- Transient CH has a significantly higher rate than other similar studies. It is reasonable to recommend following these patients for a longer period to rule out probable permanent hypothyroidism that may be detected a few months after drug withdrawal.

14- Care should be taken at the time of delivery not to use iodine or iodine containing antiseptic solutions if alternatives are available, stress on NICUS not to use iodine-containing topical antiseptics.

15- Efforts should be made to improve communication , parent education, and timely follow-up reporting to ease the stress and anxiety related to false-positive screening results.

16- It is important to document program successes and to cultivate parent advocates for convincing policy makers of the importance of the screening program so fund demonstration projects to evaluate technology, quality assurance, and health outcomes of this program are important.

17- Improving monitoring and supervision of the program through: There is need for developing and evaluating (or auditing) quality indicators to improve the overall quality of the newborn screening program. When unacceptable quality or performance is discovered, a plan must be in place to correct this. Thus, it is beneficial to have written protocols for evaluating QA indicators and resolving issues that might arise from the audit process.

• Calculation of the coverage rate at each health zone, and monitoring the diagnosis, treatment and follow-up of positive cases at each health care facility.

• Regular collection and checking of this information to be presented as an annual report about different program components.

18- International cooperation should be enlisted for the exchange and training of personnel, the exchange of information and materials, and the comparison of methods and results.

19 - Further research is required regarding:

• Similar studies describing the program and evaluating its performance should be repeated at suitable intervals in each governorate.

• Large nation-wide study should be undertaken to evaluate the program and its impact on child morbidity.

• A surveillance system should be arranged to detect false negative cases of CH to calculate the sensitivity and specificity of the screening test.

• Studies to measure iodination status in our governorate.

- Evaluation of the relation between newborn and maternal (prenatal) thyroid-function tests.
- Further studies are needed to understand the causes behind higher rates of transient and permanent CH.
- Further studies are needed to know the etiology of CH in Fayoum Governorate through scanning and ultrasonography.