Original Article

Prevalence of neonatal hypothyroidism and phenylketonuria in Southern Thailand: A 10-year report

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ABSTRACT

Background: Congenital hypothyroidism and phenylketonuria, the two major problems of several metabolic errors are presently the focus of attention, in Thailand. These two conditions are assigned as diseases to be controlled under the National Public Health Policies of Thailand. **Materials and Methods:** Here, the authors summarize and report the 10-year study on the prevalence of neonatal hypothyroidism and phenylketonuria in Southern Thailand. **Results:** This report is good representative data from Thailand, a country in Southeast Asia. Another interesting point in this study is the concern of the recalling process. **Conclusion:** It can be seen that there are a considerable number of infants who did not receive the confirmation test due to loss of follow-up after calling for a recheck.

Key words: Congenital, hypothyroidism, phenylketonuria, Thailand

Introduction

The inborn metabolic–endocrine error is an important condition in Pediatrics. There are several problems in this group. There is no doubt that a child with this error cannot develop complete normal growth or developmental process. Nevertheless, some of these inborn errors can be diagnosed early and treated effectively by biochemical supplementations.

Of these several errors, the two important problems that are presently focused upon in Thailand are: congenital hypothyroidism and phenylketonuria. These two conditions are assigned as the diseases to be controlled under the National Public Health Policies of Thailand.[1,2] The screening for these two diseases is promoted. Here, the authors summarize and report on the 10-year report of the prevalence of neonatal hypothyroidism and phenylketonuria in Southern Thailand. This report is good representative data from Thailand, a country in Southeast Asia.

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Materials and Methods

Overview

This study is designed as a retrospective study. The data on the screening program for neonatal hypothyroidism and phenylketonuria in Southern Thailand during 2000 – 2009 were reviewed. The covered screening area in this study was 14 provinces in the Southern Region of Thailand. The screening was performed at the Referencing Neonatal Screening Center of the Southern Region of Thailand, which was located in the Songkhla province. The screening was performed based on the samples collected on filter papers from newborns in every hospital, in every province in the region. The screening was performed based on the standard enzyme-linked immunosorbent assay (ELISA) technique, and was under the control of the Department

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of Medical Science, Ministry of Public Health, Thailand. The intra- and interassay coefficients of variation (CV) were less than 5%. The assay was indigenously developed by the National Referencing Laboratory of the Department of Medical Science and the same assay method was used in all the provinces, for all the 10 years. The summary of the annual prevalence and overall prevalence of hypothyroidism and phenylketonuria has been prepared and is presented in this study.

Process of screening

For congenital hypothyroidism screening, a dry skin prick blood sample was collected for spot thyroid stimulating hormone (TSH) at age 48 hours or older. The cutoff TSH level for a recall test was >25 mU/L. The cutoff level was based on the arbitrary guideline given by the Department of Medical Science (the Department of Medical Science used this level according to the unpublished National Official Reference Finding Study). For abnormal cases, recalling was done and another examination for serum thyroxine (T4), Free T4, and TSH was performed as a confirmatory test.

For congenital phenylketonuria screening, the same blood sample on filter paper was used. Screening was based on the Guthrie method, and then followed by the fluorometric method. Recalling was done in any infant with a phenylalanine level ≥ 4 mg/dl. The cutoff level was based on the arbitrary guideline given by the Department of Medical Science (the Department of Medical Science used this level according to the unpublished National Official Reference Finding Study) and retest using the fluorometric method.

Results

Overall there were 1,118,676 people screened in the 10-year period. For congenital hypothyroidism, the first screening detected 3286 abnormal cases. After recalling, 2932 attended the follow-up. Of the 2932 infants who got confirmatory results, there were 662 confirmed cases with congenital hypothyroidism. For congenital phenylketonuria, the first screening detected 120 abnormal cases. After recalling, 80 attended the follow-up. Of 80 infants who got confirmatory results, there were five confirmed cases with congenital phenylketonuria.

Discussion

Screening for congenital metabolic disease is an important form of prevention in Pediatrics. This activity is very useful in the error detection of many inborn errors. It should be noted that many kinds of congenital organs can be successfully treated if early detection is obtained. Many metabolic disorders can be stopped from further progression to permanent damage in patients, if the specific biochemical supplementation is done in the early phase.

Generally, nearly 100% of all deliveries in Thailand are inside the institution. Hence, the screening program for screening of neonatals is done inside the institution of delivery. Of the several problems, the two disorders that are focused upon in Thailand, at present are: congenital hypothyroidism and phenylketonuria. Congenital hypothyroidism was the first disease to be screened and then the screening for phenylketonuria was added.[1,2] These two screenings became the main neonatal screening practices. They were run under the National Public Health Policies.

Focusing on congenital hypothyroidism, the problem is confirmed for its clinical and epidemiological importance in Thailand.[3] The screening program for this condition is a new, and has been set for less than 15 years. With the implementation of this new program, many cases of congenital hypothyroidism can be detected and controlled. There are some previous reports on the results from the screening. The first report was from the local university hospital in Thailand, which cannot be a representative of the large Thai population.[4] Panamonta *et al*. found that the incidence was 1:3186.[4] Of late, Charoensiriwatana *et al*. has used the geographical system to analyze the four-year result of screening and noted that "all provinces in Thailand suffer from mild to moderate iodine deficiency, in a mild-to-moderate manner".[5] Hence, there is no doubt that screening is important. Here, the author has analyzed the greatest number of population and the longest period of data collection, a decade. The data can be a good representative for the Thai population. Here, the prevalence of the disease is in the ratio of 1:1690. Of interest, this is significantly different from the previous report on the previous period in Thailand (1:2476).[3] The difference in prevalence of congenital hypothyroidism by the current screening as compared to the previous screening is believed to be because the report is the data from only one region of the five regions of Thailand, and this may reflect a lower prevalence in this region compared to the overall prevalence in the country. Furthermore, compared to the data from China, where universal screening has been performed for years, the prevalence in this report is higher (1:2033).[6] This might also show the ethnical difference in the prevalence of the disorder.

Focusing on congenital phenylketonuria, the screening is parallelly done with congenital hypothyroidism screening. According to a nationwide study in Thailand, the incidence was one in 212,535.[7,8] Here, the author found the prevalence equal to 1:223,735, which confirmed the previous reports.

Of interest, the prevalence in Thailand was significantly lower compared to the other settings implementing the universal screening, such as China (1:11,628).[6] Based on the very low prevalence, it seemed that this screening might not be cost-effective in practice, compared to the screening for congenital hypothyroidism. Furthermore, a note of the alarming false positives in phenylketonuria screening (> 95%) raised a query on its authenticity as a screening test.[9]

Another interesting point in this study is a concern on the recalling process. At present, the practice is mail notification of the suspected results to the parents. The information in the mail includes a note of the suspected results and requests the parents to bring the children for further investigation and treatment. However, it can be seen that there are a considerable number of infants who do not receive the confirmation test, due to the loss of follow-up after calling for a recheck. This is the main issue of concern. Also, this implies the necessity to find a better method to resolve this problem. The use of point-of-care testing for screening might be the solution to this problem.

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