Prevention and Control of Thalassemia at Saraburi Regional Hospital

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Abstract

Objective : To evaluate the program in prevention and control of thalssemia among pregnant women and their spouses, prevention of new cases by screening tests, confirmatory test, genetic counselling, prenatal diagnosis, and selective abortion.

Subjects : The pregnant women, attending antenatal care unit, Saraburi center hospital, as well as their spouses. 1 January 2000 - 31 December 2001

Method : As part of the antenatal care assessment, pregnant women before 16 weeks gestation were screened, with pre- and post- test counselling, by osmotic fragility (OF) and dichorophenol indophenol precipitate (DCIP) tests, and confirmed by complete blood count (CBC), mean corpuscular volume (MCV), hemoglobin typing and polymerase chain reaction for αthal1 (PCR αthal1) if any of two screening tests was positive. The husbands of those who were carriers of severe thalassemia were encouraged to have thalassemia screening and confirmation. When both the pregnant women and their husbands were carriers of severe thalassemia, the pregnant women would voluntarily perform the prenatal diagnosis. Termination of pregnancy would be offered when the fetus had severe thalassemia.

Results : There were 3,739 from 4,214 women (88.7% of all antenatal women), who participated in the program. OF and/or DCIP were positive in 1,742 of 3,739 subjects (46.5%). Of those, 960 from 1,742 (55.1%), had husbands who were willing to have the testing, and OF and/or DCIP were positive in 443 of 960 cases (46.1%). The confirmatory tests revealed carrier and disease of thalassemia, and hemoglobinopathies in 931 of 1,742 women (53.9%), and 135 of 960 husbands (14.0%). The 20 couples who had the possibility of having severe thalassemic newborns, were strongly advised to have prenatal diagnosis. The 12 risk pregnancies had been performed cordocentesis. Finally 3 of 12 (25.0%) fetuses were documented to have severe thalassemia and all of them decided to have selective abortion.

Conclusions : The screening model for thalassemia carriers by using the combination of OF and DCIP is the easy screening model. It can be done quickly, it is inexpensive, therefore it is suitable

for large numbers of population screening. The systematic screening, confirmatory of thalassemia diagnosis and prenatal diagnosis are the measure of thalassemia prevention and control, and aims to decrease the number of newborns with severe thalassemia.

Key word : Thalassemia, Prevention and Control, Screening and Confirmation, Prenatal Diagnosis, Selective AbortionIntroduction

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Thalassemia and hemoglobin disorders are the most common hereditary diseases and are important health problems in Thailand[1-3]. They result from unbalanced hemoglobin synthesis caused by decreased production of at least one globin polypeptide chain (α, β, γ, δ). In its homozygous state, thalassemia genes cause severe and often lethal disease. Hemoglobin Barts' hydrops fetalis (homozygous α-thalassemia), homozygous β-thalassemia (β-thalassemia major) and β-thalassemia/hemoglobin E, are three severe types. The first is lethal, while the second and third bind most of the patients to the hospital for life. Thalassemic diseases can give rise to abnormality in any organ system. Apart from the common symptoms of anemia, jaundice, retarded growth and puberty, thalassemic face, hepatosplenomegaly, gall stones and cholecytitis, and frequent upper respiratory tract infection. In Thailand, the prevalence of α-thalassemia, β-thalassemia, hemoglobin E, and Hemoglobin Constant Spring, are 20-30, 3-9, 13, and 1.6 per cent, respectively[4]. Their distributions vary from region to region and among different ethnic groups.

Overall, the carriers of thalassemia amount to around 18-20 million of the Thai population, and each year 48,500 couples are estimated to be at risk of thalassemia, and one fourth of which result in thalassemic newborns[5,6]. Recently, some studies, hospital based data, have shown the prevalence of thalassemia and hemoglobinopathy carriers among pregnant women, vary from 42 to 45.3 per cent[7,8]. In 1998, the Department of Health, Ministry of Public Health, initiated a program for thalassemia prevention and control among pregnant women. The comprehensive strategy includes 1) health education and promotion; 2) personnel training; 3) laboratory facilities; 4) counselling service; 5) prenatal diagnosis; and 6) selective abortion[9,10]. Prevention and control are necessary in order to decrease the number of thalasssemic newborn.

Ideally, a screening test for thalassemia should be reliable and specific yet not expensive or time-consuming. A combination of osmotic fragility (OF) and dichorophenol indophenol precipitate (DCIP) tests are promising as appropriate screening models, whereas, the sensitivity, specificity, positive and negative predictive values are 100, 97.1, 94.9 and 100 per cent, respectively[11]. Couples who are confirmed to be severe thalassemia carriers, should be advised to have prenatal diagnosis, amniocentesis or cordocentesis. Counselling on selective abortion will exclusively be confined to cases with an affected fetus with severe thalassemia. The thalassemia prevention and control program, large-scale carrier screening, was established in Saraburi Center Hospital in 1999. The aims of this study were to determine the prevalence/incidence of carriers of thalassemia and hemoglobinopathy in Saraburi province, and to evaluate the entire intervention program in terms of acceptability, operational database, positive and negative lessons, and advantages and disadvantages.

METHOD

The study received approval from the institutional review board (IRB). The step by step intervention was performed, with the local network collaboration of community hospitals, provincial hospitals, and Saraburi Center Hospital (focal point), as well as technical support from the Faculty of Medicine, Chiang Mai University. The subjects, 4,214 cases, were recruited from the antenatal care clinic, from January 1, 2000 to December 31, 2001. Inclusion criteria included voluntary pregnant women, gestational age 16 weeks or less, and non-directive counselling related to every procedure of thalassemia prevention and control program. Gestational age was determined by the last normal menstrual period, and cases of uncertain date were confirmed by ultrasound assessment.

After the study was explained and verbal consent obtained, the pregnant women were asked to complete a questionnaire on antenatal general information. Within six hours of collection of 10 ml blood, and transfer to the central laboratory for complete blood count (CBC), Mean Corpuscular Volume (MCV), and was screened by OF, and DCIP tests.

Osmotic fragility test

Khon Kaen osmotic fragility test, commercial kit, was used. Twenty μl of whole blood collected in ethylenediaminetetraacetic acid (EDTA) was pipetted into the glass test tubes which contained 2 ml of 0.36 per cent saline solution. Tubes were shaken well and allowed to stand at room temperature. After fifteen to thirty-minutes, contents of the tubes should be left unshaken and then held immediately against a piece of dark paper. Theoretically, normal red blood cells can maintain themselves without hemolysis in 0.85 per cent saline solution, but are hemolyzed in a lower concentration. While the red blood cells of thalassemia and hemoglobin E are partially hemolyzed. Clearly visible content through the tube containing buffered saline, the test was considered negative, implying hemolysis had occurred in more than 90 per cent. Whereas, a cloudy content was considered positive, indicating partial hemolysis[11,12].

Dichorophenol indophenol precipitate (DCIP) test

Khon Kaen DCIP test, commercial kit, was applied. Twenty μl of whole blood collected in ethylenediaminetetraacetic acid (EDTA) was pipetted into the glass test tubes which contained 2 ml of DCIP clearing solution. Tubes were shaken well and incubated in a water basin at 37°C. After 15 minutes, the tubes were left at room temperature. Clearly visible content through the tube containing buffered saline, the test was considered negative, and cloudy content was considered positive[13].

From all the subjects who had a positive result of OF and/or DCIP, the aliquot of blood was confirmed hemoglobin typing by high performance liquid chromatography (HPLC). The subjects who had genetic risks of α-thalassemia, β-thalassemia, and Hemoglobin E, their husbands were advised to be tested. In addition, the DNA analysis by polymerase chain reaction (PCR) was conducted for confirmation of α-Thalassemia 1 at the Faculty of Medicine, Chiang Mai University.

Only couples, with the possibility of the severe thalassemia had the prenatal diagnosis carried out by cordocentesis. The fetal blood was also tested by HPLC, Variant™[14-16]. The Variant is a fully automated HPLC system which can be used to separate and determine area percentages for hemoglobin A_2 and F and to provide qualitative determinations of abnormal hemoglobin.

Selective abortion was offered for those who had confirmed severe thalassemia. This prospective description analysis was carried at Saraburi Center Hospital, for means and comparisons of means, using SPSS.

RESULTS

During two years of intervention, pregnant women whose gestational ages were less than 16 weeks, 3,739 of 4,214 (88.7%) were voluntarily enrolled to the study. By two screening tests, OF and/or DCIP were positive in 1,742 of 3,739 subjects (46.5%). Of the positive screening, a confirmatory test by HPLC was performed. Of those, 960 of 1,742 (55.1%), their husbands were willing to have the thalassemia testing, and OF and/or DCIP were positive in 443 of 960 cases (46.1%), according to their personal reasons: inconvenience, ignorance, and low cooperation. The confirmatory test revealed carriers and disease of thalassemia, and hemoglobinopathies in 931 of 1,742 women (53.9%), and 135 of 960 husbands (14.0%). A pregnancy in which both the couples were carriers, was considered at risk. The couples, who had the possibility of having severe thalassemic newborns, were strongly suggested to have prenatal diagnosis. Of the twenty couples, who were at risk of severe thalassemic newborns, twelve had the prenatal diagnosis, cordocentesis performed. The cordocentesis was per-

formed on 12 of 3,739 cases (0.3%). Finally, in 3 from 12 (25%) the fetuses were documented to have hemoglobin Barts' hydrops fetalis. All of the spouses decided to have selective abortion carried out after couple counselling (Table 1).

Of 3,739 pregnant women who were screened, 1,742 cases (45.6%) of positive OF and/or DCIP were detected. The incidence of positive screening was 1 in 2.2 pregnant women. Of those, 931 cases (24.9%) were thalassemic carriers: heterozygous HbE 715 (76.8%), homozygous HbE 107 (11.5%), heterozygous β-thalassemia 53 (5.7%), heterozygous α-thalassemia 22 (2.4%), β-thalassemia/HbE 16 (1.7%), Hb H disease 15 (1.6%), HbD 2 (0.2%), and Constant Spring 1 (0.1%) (Table 2).

Of 960 husbands who were screened, 444 cases (46.2%) of positive OF and/or DCIP were detected. The incidence of positive screening was 1 in 2.1 husbands. Of those, 135 cases (14.0%) were thalassemic carriers: heterozygous HbE 108 (80.0%), homozygous HbE 8 (5.9%), Heterozygous β-thalassemia 7 (5.1%), heterozygous α-thalassemia 6 (4.4%), Hb H disease 3 (2.2%), and HbH/homozygous HbE 3 (2.2%) (Table 3).

DISCUSSION

Thalassemic diseases pose a challenge to the public health system at provincial level, in applying new appropriate technology for the prevention and control of severe thalassemia. Thalassemia control programs by public health education, mass carrier detection, genetic counselling, prenatal diagnosis, and termination of pregnancies with affected fetues have been applied successfully in many Western societies (17). Thalassemia presents an individual, social and economic burden: a key question is whether the medical and economical point of view converge or not. The ideal option of prevention and control of thalassemia is community screening. The optimized approach thereto is antenatal screening(18).

Cost-effectiveness of the one tube Osmotic Fragility (OF) test combined with DCIP test was 74.07 bahts per diagnosis, the automated analyzer was 94.34 bahts per diagnosis and the hemoglobin electrophoresis was 188.68 bahts per diagnosis(19). There are several strategies for prevention and control of thalassemia in the newborn, antenatal approach. Firstly, hemoglobin typing among pregnant women is carried out, and if thalassemia and hemoglobinopathy are detected, then their spouses will be included. Secondly, OF and DCIP are conducted among pregnant women, and confirmed by hemoglobin typing in the case of positive screening tests. Only if the screening and confirmatory tests are concordant, being thalassemia and hemoglobinopathy, then their spouses will be enrolled. Lastly MCV may be one of screening tests, with or without OF and DCIP.

Self-preparation for OF and DCIP is cheaper, while internal quality control has to be concerned. Buffered saline solutions with concentrations of 0.36 per cent were prepared by diluting a 10 per cent stock solution of sodium chloride (90 g), disodium hydrogen phosphate (13.65 g) and sodium dihydrogen phosphate (2.43 g) in 1000 ml of distilled water (pH 7.4). Twenty μl of whole blood collected in ethylenediaminetetraacetic acid (EDTA) was pipetted into the

Table 1. The characteristics of the couples.

	Number	%
Pregnant women from the Antenatal Clinic	4,214	100
Pregnant women: screening tests (n = 3,739)	3,739/4,214	88.7
OF and/or DCIP positive	1,742/3,739	45.6
Pregnant women: confirmatory test (n = 1,742)		
Thalassemia trait, hemoglobinopathies	931/1,742	53.9
Husbands: required thalassemia testing (n = 1,742)		
Acceptability to test	960/1,742	55.1
OF and/or DCIP positive	443/960	46.1
Thalassemia trait, hemoglobinopathies	135/960	14.0
Couples at risk for severe thalassemia	20/1742	1.1
Prenatal diagnosis; cordocentesis	12/1,742	0.7
Affected fetuses; severe thalassemia	3/1,742	0.4
Selective abortion (n = 3)		
Carry through selective abortion	3/3	100

Table 2. The results of screening and confirmatory tests among the pregnant women.

	Number	%
Pregnant women (n = 3,739)		
Screening tests, OF and DCIP		
OF and DCIP negative	1,997	53.4
OF positive only	596	15.9
DCIP positive only	361	9.6
OF and DCIP positive	785	20.9
Confirmatory test, Hb typing		
Heterozygous HbE	715	19.1
Homozygous HbE	107	2.8
Heterozygous β-thalassemia	53	1.4
Heterozygous α-thalassemia	22	0.58
β-thalassemia/HbE	16	0.42
HbH disease	15	0.40
HbD	2	0.05
Constant Spring	1	0.02

Table 3. The results of screening and confirmatory tests among the husbands.

	Number	%
Husbands (n = 960)		
Screening tests, OF and DCIP		
OF and DCIP negative	517	53.8
OF positive only	191	19.8
DCIP positive only	74	7.7
OF and DCIP positive	178	18.5
Confirmatory test, Hb typing		
Heterozygous HbE	108	11.2
Homozygous HbE	8	0.83
Heterozygous β-thalassemia	7	0.72
Heterozygous α-thalassemia	6	0.62
HbH disease	3	0.31
HbH/homozygous HbE	3	0.31

glass test tubes (100 mm x 10 mm) which contained 5 ml of 0.36 per cent saline solution. The tubes were shaken well and allowed to stand at room temperature. After thirty-minutes, the contents of the tubes were again shaken and the tubes were held immediately against a piece of paper. Words were clearly visible and could be easily read, through the tube containing distilled water (control). If the words were similarly visible through the tube containing buffered saline, the test was considered negative, whereas, if the words were not clearly visible, the test was considered positive(11,12).

The working agent of DCIP contained the Trisma base (4.36 g), EDTA $Na_2.2H_2O$ (2.68 g), DCIP (0.02776 g), and Saponin (0.05 g), adjusted pH to 4.5 at room temperature, and volume to 500 ml with sterile water. After centrifuging EDTA whole blood for 10 minutes at 1,000 RPM, twenty µl of red cells, was pipetted into the glass test tubes. The tubes were left at room temperature for 10 minutes, and then incubated at 37.0°C for one hour, without being shaken. The results were reported as: level 0 = clear solution, blue green color, and no precipitance; level 1^+ = cloudy solution, green color, and no precipitance; level 2^+ = cloudy solution, green color, and precipitance in the solution; level 3^+ = cloudy solution, green color, precipitance in the solution plus sediment at the bottom of the tube; and level 4^+ = clear solution, green color, a lot of sediment at the bottom of the tube(13).

Advantageously, OF and DCIP are simple screening tests and can be utilized in primary health services. Nevertheless, a false positive can occur in a number of proportions. Ideologically, the best screening is hemoglobin typing, however, in a poor resource setting the OF and DCIP are practical to cover the mandatory screening program.

There is no magic bullet in the clinical preventive strategies, because thalassemia prevention and a control program among the newborn in the continuum process; 1) screening and confirmatory tests of pregnant women before 16 weeks gestation; 2) confirmation test of their spouses if the pregnant women are thalassemic carriers or have the disease, and hemoglobinopathy; 3) prenatal diagnosis in couples who have the possibility of severe thalassemia in the newborn; and 4) selective abortion. Roughly, around two-thirds of the pregnant women attend the antenatal care clinic before 16 weeks gestation. Thus, promotion of early antenatal care visits is essential. The husband compliance to be tested is 55.1 per cent, and it reflects to the awareness and concern of the public toward thalassemia prevention and control program. However, that can be achieved by various kinds of education; mass, group, and individual education. Prenatal diagnosis needs individual counselling, as well as selective abortion, creating the step by step to health consciousness; awareness, concern, knowledge, attitudes, willing to co-operate, stable behavior, and specific consciousness. The incidence of thalassemia carriers in this study is 24.9 per cent as the other other studies(20). The heterozygous HbE is 76.7 per cent, it is quite high because Saraburi is a industrial town

where the people from the north-east are immigrated to. We used cordocentesis to get the fetal blood for hemoglobin typing test, with high performance liquid chromatography technic to diagnose severe thalassemia in the fetus of the couples at risk. This procedure can be setting in many provincial hospitals. Though, amiocentesis, chorionic villi sampling for DNA analysis are the other ways in prenatal diagnosis of severe thalassemia but, it is rather expensive and technology is hardly setting. Average time used for cordocentesis operation 30 minutes, it is in learning curve in operation after cordocentesis training from the Perinatology Unit, Faculty of Medicine, Chiang Mai University. There was fetal loss of 1-3 per cent in a large series from Chiang Mai University(21). Fortunately there was no fetal loss in the present study which may be because of the the small number of cases and time used in operation.

Now, many hospitals in Thailand provide a full-scale prenatal diagnosis service, either by cordocentesis followed by HPLC, or by amniocentesis followed by DNA analysis. This can be a focal point of thalassemia prevention and control program, and networking collaboration with primary and secondary health care, or vertical cooperation with research institutes. However, more systematic prevention and control programs, are needed, appropriately conducted in a long-term process, with collaboration between the public and private sectors. The standardization of personnel, facilities and support, medical services, and economic evaluation, are suggested.

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โครงการป้องกันและควบคุมการเกิดโรคเลือดจางธาลัสซีเมีย : โรงพยาบาลศูนย์สระบุรี

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วัตถุประสงค์ : เพื่อประเมินการดำเนินงานโครงการควบคุมและป้องกันการเกิดโรคเลือดจางธาลัสซีเมียในเด็กแรกเกิดรายใหม่ การใช้การตรวจคัดกรอง การตรวจยืนยัน การให้คำปรึกษาทางพันธุศาสตร์ การวินิจฉัยทารกในครรภ์ และการยุติการตั้งครรภ์

กลุ่มศึกษา : หญิงตั้งครรภ์และสามีในหน่วยฝากครรภ์ โรงพยาบาลสระบุรี ระหว่างวันที่ 1 มกราคม 2543 ถึง 31 ธันวาคม 2544

วิธีการ : หญิงตั้งครรภ์ที่อายุครรภ์น้อยกว่า 16 สัปดาห์ จะได้รับการให้คำปรึกษาก่อนและหลังการตรวจคัดกรอง osmotic fragility (OF), dichlorophenol indophenol precipitate (DCIP) และตรวจยืนยันด้วย Complete blood count (CBC), mean corpuscular volume (MCV), Hemoglobin typing, polymerase chain reaction (PCR) ในรายที่ผลการตรวจคัดกรองผิดปกติ สามีจะได้รับคำแนะนำเพื่อตรวจเช่นเดียวกัน ส่วนคู่สามีภรรยาที่พบว่ามีโอกาสให้กำเนิดบุตรโรคเลือดจางธาลัสซีเมียชนิดรุนแรง จะได้รับคำปรึกษาการวินิจฉัยทารกในครรภ์ และการยุติการตั้งครรภ์หากพบว่าทารกในครรภ์เกิดโรคเลือดจางธาลัสซีเมียชนิดรุนแรง

ผลการศึกษา : หญิงตั้งครรภ์ 3,739 รายจาก 4,214 ราย (คิดเป็น 88.7% ของหญิงตั้งครรภ์ทั้งหมด) พบ OF และ/หรือ DCIP ผิดปกติ 1,742 รายจาก 3,739 ราย (46.5%) คู่สามีของหญิงตั้งครรภ์ที่ยินดีรับการตรวจ 960 รายจากคู่สมรส 1,742 คู่ (55.1%) พบ OF และ/หรือ DCIP ผิดปกติ 443 รายจาก 960 ราย (46.1%) การตรวจยืนยันพบความผิดปกติของฮีโมโกลบิน 931 ราย จาก 1,742 ราย (53.9%) และ 135 รายจาก 960 ราย (14.0%) ในหญิงตั้งครรภ์และสามีตามลำดับ พบคู่สมรส 20 คู่มีโอกาสให้กำเนิดทารกธาลัสซีเมียชนิดรุนแรง คู่สมรสที่ยินดีรับการตรวจทารกในครรภ์ 12 คู่ พบทารกฮีโมโกลบิน บาร์ท 3 รายและตัดสินใจยุติการตั้งครรภ์

สรุป : การตรวจคัดกรองพาหะโรคเลือดจางธาลัสซีเมีย โดยการใช้ OF และ DCIP เป็นวิธีการที่สะดวกรวดเร็วและราคาไม่แพง ซึ่งเหมาะที่จะใช้ในประชากรกลุ่มใหญ่ ดังนั้นการตรวจยืนยันการเป็นพาหะของโรคและการวินิจฉัยก่อนคลอดจึงเป็นแนวทางในการป้องกันการเกิดทารกโรคเลือดจางธาลัสซีเมียชนิดรุนแรง

คำสำคัญ : โรคเลือดจางธาลัสซีเมีย, การควบคุมและป้องกัน, การตรวจคัดกรองและยืนยัน, การวินิจฉัยก่อนคลอด, การยุติการตั้งครรภ์

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