

Executive Summary

Research Project: Assessing the Pilot Study for the Prevention of Down Syndrome

Down syndrome is a hereditary condition that occurs due to a chromosomal abnormality, specifically chromosome 21. Children with Down syndrome typically have various health issues and shorter life expectancies. Symptoms that are clearly seen in people with Down syndrome include intelligence in the mildly-to-moderately low range, congenital heart disease, and issues with blood circulation, hearing, sight, digestion, skin, respiration, and the endocrine system. In Thailand, it is estimated that approximately 800 – 1,000 children per year are born with Down syndrome. One method for detecting Down syndrome is to conduct a prenatal screening test in pregnant women to determine whether the fetus has developed the condition.

A study in Thailand that assessed the cost-effectiveness of prenatal screening and diagnosis of Down syndrome in the country found that screening and diagnosing pregnant women via blood tests, followed by an amniocentesis if the risk of Down syndrome was high, was cost-effective. Thus, the Sub-committee of Health Prevention and Promotion under National Health Security Office (NHSO) approved prenatal screening and diagnosis of Down syndrome as a new benefit for the 2016 fiscal year. Moreover, the Maternal and Child Health Advisory Board under the Department of Health, Ministry of Public Health (MOPH) passed a resolution for the Department of Health to consider providing prenatal screening and diagnosis in pregnant women. Therefore, a pilot study for the prevention of Down syndrome was initiated in the five following provinces: Chiang Mai, Lamphun, Khon Kaen, Nakhon Sawan, and Songkhla. The aim of the study was to learn more about the program's management and services provision, particularly the guidelines for establishing a services provision framework and determining an appropriate method for expanding the project.

Objectives

To assess the pilot study for the prevention of Down syndrome.

Specific Objectives

- 1) To assess the provision of services for the prevention of Down syndrome in the pilot provinces and compare it to the Royal Thai College of Obstetricians and Gynaecologists' guidelines.
- 2) To assess the results of the provision of services for the prevention of Down syndrome in the pilot provinces.
- 3) To provide guidelines in the provision of services for the prevention of Down syndrome for expansion to other provinces.

Methodology

This study aimed to assess the provision of prevention services for Down syndrome. These include genetic counseling about Down syndrome, screening, diagnosis, and abortion if the fetus was diagnosed with Down syndrome. In addition, hospital laboratories in the pilot provinces were also evaluated to determine whether they followed the guidelines created by the Royal Thai College of Obstetricians and Gynaecologists. Finally, a four-pillar framework was used to determine the factors that contributed to the success of the pilot study, namely: 1) output and outcome; 2) services provided; 3) peripheral factors; and 4) stewardship at both national and regional levels. This study was approved by the Research Ethics Committee at the Institute for the Development of Human Research Protections in March 2016.

Both quantitative and qualitative approaches were conducted in this study. Information obtained from the Department of Health's pilot study output reports were reviewed quantitatively. Meanwhile, the qualitative portion consisted of document reviews and in-depth interviews with the project's managers at both the national and regional levels. Document reviews and interviews at the NHSO and the Department of Health were conducted at the national level. Regionally, documents were reviewed and interviews were held in all pilot provinces with each of the following: health promotion centers, provincial public health offices, Maternal and Child Health Advisory Boards at the provincial level, the pilot study advisory board and working group, and representatives of service providers in hospitals in their respective provinces, i.e. one representative among university hospitals, health promotion hospitals, hospital centers or referral hospitals that conduct amniocentesis or abortions; one community hospital with an obstetrician and one community hospital without (two in total); three district health promotion hospitals under the supervision of that province's community hospital; and a personnel representative from every laboratory.

Results

Many organizations played a role in the prevention and treatment services for Down syndrome. On the national level, the Department of Health was responsible for many facets of the project – providing policy, various documents such as operational guidelines for the project, leaflets and flip charts for genetic counseling, data forms, and monitoring performance. Meanwhile, health centers and public health offices in each pilot province were the arms that carried out the Department of Health's policies and liaised with other provincial agencies involved in the project, including monitoring the implementation based on the provided guidelines. Regional health promotion

hospitals, university hospitals, hospital centers/general hospitals, community hospitals, and provincial health promotion hospitals provided the prevention services for Down syndrome, and laboratories in regional health promotion hospitals, university hospitals, hospital centers, and the Faculty of Medical Technology, the Prince of Songkhla University, analyzed the genetics and chromosomes of patients. In addition, each province also formed an advisory board/working group to increase the effectiveness of the pilot study.

The pilot studies in the provinces did not begin at the same time. Some of them were able to begin the pilot on 1 October, 2015, the designated start date set by the Department of Health. However, other provinces had to start the pilot later due to resource and fiscal constraints, particularly Nakhon Sawan and Chiang Mai. Both lacked the experts required to build capacity in personnel and therefore required training courses to be held. In addition, fiscal limitations in Chiang Mai limited its ability to provide screening even though these costs would later be reimbursed by the NHSO since the chemicals used for genetic testing were expensive. Eventually, once the training courses were held and the necessary budget was provided by the NHSO, the remaining provinces began their trials.

During the trial period from 1 October, 2015 until 31 August, 2016, a total of 12,547 pregnant women received prevention and treatment services. The province most receptive to prenatal screening was Lamphun (99%), although the rate of false positives came out to between 4-10%¹. Meanwhile, acceptance rates for amniocentesis were highest in Khon Kaen and Lamphun at 98% and 97%, respectively. The province of Songkhla had the lowest acceptance rate for amniocentesis with 39% of pregnant women who were screened as high-risk being receptive to the procedure. Finally, a total of 25 women went through abortions once it was determined that their fetus had Down syndrome.

Monitoring and assessing the results of the study utilized both supervision and performance reports. Supervision was divided into two types: 1) supervising performance on-site via the advisory board/pilot study working group, where an officer was responsible for coordinating tasks between the health center and the provincial public health office was designated as the Secretary; and 2) supervising performance via meetings in provinces where an advisory board/pilot study working group was not formed. On-site supervision enabled hospitals to provide services and resolve cases if the screening results were deemed high-risk or abnormally low. For supervision via performance reports, a template was designed to provide an alternative in monitoring pilot study records. However, it was found that the template was not sufficient and resulted in misunderstandings about definitions and data discrepancies. Moreover, utilizing Microsoft Excel to record information onto the template was not easy or intuitive and resulted in many provinces sending their reports after the given deadline.

Screening and diagnostic services for Down syndrome begin with hospital staff providing genetic counseling about Down syndrome to pregnant women. Since these staff are also responsible for providing advice about antenatal screening, they have the potential to provide genetic counseling as well but must undergo further training about Down syndrome. This pilot study provided training about Down syndrome from both theoretical and practical perspectives as well as supporting counseling media such as flip charts and leaflets about Down syndrome and screening methods. However, once the staff began providing counseling, the advice provided was shortened even further as the amount of time given only ranged between 10-15 minutes to account for the number of pregnant women or services provided. Consequently, important information was left out such as the severity of Down syndrome. Moreover, only some pages of the flip chart provided were used during counseling while some hospitals decided to use brochures instead of flip charts due to various factors: convenience, concise information, being suitable for the duration of service provision, and the ability to bring the brochure home to study later.

Moreover, medical results from screening for Down syndrome should have been recorded as “high-risk” or “low-risk”. However, this study found that some hospital staff who provided counseling used various terms such as “normal”, “abnormal”, “at risk” or “no risk” in documents such as the Mother and Child Health Handbook – a notebook used to record the development of the mother and child. This may result in the patient misunderstanding the screening results.

When storing and handling the serums used for blood testing, the temperature should be set at an appropriate level to minimize false positive readings. Staff from each laboratory received training on how to store and handle the serums. However, during actual practice, it was found that some laboratory staff were not aware of the correct procedures. This was because these staff were not the ones who attended the training, or they attended but were ultimately indifferent to its importance. As mentioned earlier, transferring the serum to laboratories required temperature levels to be set at an appropriate level. Some provinces monitored serum quality by placing a temperature monitoring device in the storage box. If any problems occurred, the hospital that sent the serum would be notified so they could diagnose and rectify the situation. In addition, patients’ medical information should be included with the serum in order to perform the proper tests. The assessment team found that the information provided by the laboratories were sometimes incorrect, difficult to decipher or incomplete. These factors played a role in the additional time required to analyze the sample and may have contributed to screening errors in both high-risk and low-risk situations.

Many hospitals delivered their Down syndrome screening results late by more than 14 days from the deadline stated in the guidelines. As a result, subsequent procedures were unable to be carried out in a continuous manner, potentially resulting in failure to terminate the pregnancy prior to the 24th week. Only one laboratory was able to deliver their results within 3-4 days due to proficient resource management. In addition, an appropriate amount of serum should have been used for each round of genetic testing to lower costs as much as possible. However, some provinces only had a few number of pregnant women take advantage of the services provided. Consequently, the pilot was run at a loss in these places as the initial costs for genetic testing were higher than what the NHSO was able to reimburse (1,200 baht per person). Finally, the procurement costs for the biochemical reagent might have been higher than the amount that the Head of government sections are authorized to approve based on the Regulations of the Office of the Prime Minister on Procurement B.E. 2535 (1992). With laboratories being under direct supervision of hospitals under the MOPH, these procurement constraints posed compliance risks with the aforementioned regulations.

Diagnosing Down syndrome via an amniocentesis and chromosome testing was conducted by hospitals with obstetricians or university hospitals and hospital centers/general hospitals. These venues provide the abovementioned services for referred pregnant women in addition to diagnosing other genetic diseases such as thalassemia. As such, if diagnosing Down syndrome were to be included together with other diagnostic processes, a system for recording patient data and referring pregnant women should be thoroughly considered. For example, if a pregnant woman comes to undergo an amniocentesis, the first attending medical officer should ask whether she wants to also test for other potential risks which require amniotic fluid. This would reduce the chances of abortion due to undergoing amniocentesis more than once. **Abortions for fetuses diagnosed with Down syndrome** are typically conducted by obstetricians. However, the assessment showed that some obstetricians had negative opinions about abortions, particularly in their beliefs or the laws governing the issue. This may have played a role in the adequacy of abortion services if the fetuses were found to have abnormalities. Regardless, results from all five provinces showed that in cases where the obstetricians were against the idea or refused to go through with the abortions, other physicians were still able to provide this service.

Factors which affected the participation rate of pregnant women in the pilot project can be divided into three main areas: 1) receiving services for the prevention and treatment of Down syndrome without incurring any costs; 2) discrepancies about the understanding and knowledge or not enough information about Down syndrome in pregnant women, e.g. some pregnant women

refused to undergo screening as they believed that their fetus was not at risk of Down syndrome since both herself and her husband were in good health, did not engage in risky behavior, and were young; and 3) individual beliefs or the beliefs of the people around them, e.g. religious beliefs or that having an amniocentesis will result in an abortion.

Conclusion

Overall, from the performance of provincial hospitals in the pilot study, it was found that health care providers were able to sufficiently screen and diagnose Down syndrome in addition to their usual antenatal care services. A reasonable number of pregnant women utilized the screening and diagnostic services for Down syndrome even though this project had just begun. In general, health care providers that were involved and pregnant women both agreed that screening and diagnostic services for Down syndrome should be provided as it is beneficial for fetal care planning. However, some health care providers expressed concern regarding the budget used to provide these services. Therefore, if this project were to be expanded to other provinces, it should be done on a gradual basis. Meanwhile, the provision of services should also be improved upon to ensure that patients are receiving high-quality services that also address policymakers' objectives.

Policy Recommendations

- 1) The NHSO should review the rate of reimbursement for services to synchronize with actual costs, especially the number of pregnant women that comes to receive services from a hospital does not meet the break-even point. This includes costs related to ultrasound, which is used to determine the age of the fetus, and costs related to any Down syndrome tests which reveal other abnormalities in the fetus.
- 2) The Department of Health, together with other involved organizations, should define a standard for genetic counseling that is consistent with actual practice. They should also develop supporting materials with appropriate information in the context of the service provider, e.g. choosing content that is important, short, and concise to maximize results given time constraints; these revisions should also include the Mother and Child Health Handbook.
- 3) The Department of Health should integrate the guidelines for screening and diagnosing Down syndrome and thalassemia in pregnant women, and guidelines for using ultrasound to determine the age of the fetus and detect any abnormalities during antenatal care.

4) The Department of Health and NHSO should jointly revise the method for collecting information about services provided from the current system using Microsoft Excel to a more suitable system oriented towards the users.

5) The Department of Medical Science, together with the Faculty of Medical Technology and other involved organizations, should define requirements and qualifications for laboratories to create a standardized examination for the pilot study. This would enable specimens to be analyzed in a standardized and complete manner.

6) The on-site working group or the maternal and child health advisory boards at the provincial or district levels should consider holding training programs according to the needs or issues in that respective area, supervise performance, provide advice in resolving problems, and assess the quality of performance. This would assist each province in learning and increasing their potential in providing services.

Policy Recommendations for Expanding the Initiative to Other Provinces

1) The MOPH, together with the NHSO and other organizations involved, should share knowledge with the community and private clinics regarding the screening and diagnosis of Down syndrome so that access to services may be improved for pregnant women via referrals to community hospitals.

2) The MOPH, together with the Department of Medical Sciences and laboratories, should jointly formulate a standardized plan to distribute serums, especially for distribution across provinces.

3) The Department of Health, together with the NHSO, should reduce the target time frame for reporting genetic testing results from two weeks to one week or three days so the time saved may be spent on other procedures.

4) The Department of Medical Sciences should develop a regional medical science center under its supervision to provide screening services for Down syndrome via genetic testing. This would lower initial screening costs and enable the project to expand to other provinces.

5) The MOPH, with the Department of Health collaborating with the NHSO, should form a national board for the prevention and treatment of Down syndrome. It would be responsible for formulating both the short-term and long-term strategy and ensure consistency with the National Reproductive Health Policy and Strategy (Rev. 2)

For more information: <http://www.hitap.net/documents/170601>