Measures to Improve Newborn Screening System in Korea

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Mandatory newborn screening is primarily applied to the diseases that are treatable if caught early enough but can cause severe disability if not detected and treated at a right time. The Ministry of Health and Social Affairs adopted a newborn screening program to detect inborn errors of metabolism for low-income families in 1991 and expanded it to cover all newborns in 1997. Inborn errors of metabolism are diseases that should be detected within the period between 48 hours and one week after birth, which require lifelong treatment and management.

After 10 years since the inception of the newborn screening program, now is the time for evaluation for improvement. For this purpose, a questionnaire-based postal survey was conducted in 2000 on 241 health centers across the country in 2000. Additionally, the authors conducted a supplementary survey on the families of the patients, analyzed a vast range of existing data and research results, gathered expert opinions on the policy measures, reviewed the current status and problems of the newborn screening policy, and presented policy measures to improve the screening program.

Key Word: Newborn screening, Inborn errors of metabolism

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Ⅰ. Introduction

In 1991, the Ministry of Health and Social Affairs adopted a newborn screening program for the low-income families and expanded it in 1997 to cover all newborns. At the beginning of the program 6 diseases were selected for screening. These were Congenital hypothyroidism (CH), Phenylketonuria (PKU), Galactosemia (GAL), Maple syrup urine disease (MSUD), Homocystinuria (HU), Histidinemia (HIS). The number of screening items then was reduced to two (CH and PKU) from the year 1995 because other four diseases had been rarely detected and cost-effectiveness issues were raised. The government program has a ten year history, and now is a good time to review the program in the context of effectiveness, efficiency and equity, and to present policy measures for further improvement of the program.

The purpose of the study is to review the current status of the newborn screening policy, to identify problems and to present policy options to improve the program. In order to gauge the current status of newborn screening in Korea, this paper presents a wide range of information from various sources.

The authors have reviewed the achievement of the government program. We also present the data, the report on the detected patient and quality of the laboratory screening tests analyzed by Korea Family Health and Welfare Association (KFHWA) in order to evaluate the effectiveness of screening program. We have reviewed

the National Fertility and Family Health Survey. It showed the aspects of equity in terms of coverage of the screening test on the all newborns in Korea .

In order to identify characteristics of the detected patients and to review the status of screening system in Korea, a nationwide postal questionnaire survey was conducted at 241 health centers from 20 June to 20 July in 2000.

In addition, a supplementary survey using semi-structured questionnaire was conducted on the patient's family at the meeting of PKU Family Association and the Family Association of Other Inborn Errors of Metabolism to gather information (Han, 2000).

Ⅱ. Status of Newborn Screening in Korea

Table 1 shows the increase in funding that took place in 1997, when the screening program was expanded from low-income newborns to all newborns. Some 40% of the total cost was provided by the central government and 60% by local governments. Follow-up supports, including special milk provision, were given only to low income newborns.

Table 2 describes the breakdown of expenditures on inborn errors of metabolism. An estimated 40% of the total fund was provided by the central government and other 60% by local governments. The total cost for all screening tests in 2002 was 3,705,000 thousand won (US$ 3M) or 89.4% of the total budget. For low-income patients with inherited metabolic diseases, special milk and patient management cost is

provided by the governments. The patient management cost for PKU and CH patients was expanded to include Organic Aciduria since 2001. The patient management cost was 439,616 thousand won (US$ 360,341) in 2002 and occupied 10.6% of the total budget.

Table 1. Achievement of the Government Screening Program

(Unit: thousand won)

Year	Project Budget			Source of Fund
	Total Cost	Screening Test	Follow up	
1991	297,600	240,000	57,600	Social Welfare Fund
1992	320,000	281,400	38,600	Central Government Subsidy, Social Welfare Fund
1993	544,160	491,360	52,800	Central Government Subsidy, Social Welfare Fund
1994	757,600	702,674	54,926	Central Government Subsidy, Local Government Budget, Social Welfare Fund
1995	837,400	766,432	70,968	Central Government Subsidy, Local Government Budget, Social Welfare Fund
1996	641,980	562,144	79,836	Central Government Subsidy, Local Government Budget
1997	5,367,673	5,273,700	93,973	Central Government Subsidy, Local Government Budget
1998	3,910,439	3,768,309	142,070	Central Government Subsidy (40%), Local Government Budget (60%)
1999	3,890,595	3,748,525	142,070	Central Government Subsidy (40%), Local Government Budget (60%)
2000	3,849,903	3,641,442	208,461	Central Government Subsidy (40%), Local Government Budget (60%)
2001	4,045,859	3,705,000	574,860	Central Government Subsidy (40%), Local Government Budget (60%)
2002	4,144,616	3,705,000	439,616	Central Government Subsidy (40%), Local Government Budget (60%)

Sources: 1) MOHW, *Internal Material*, 1991~1999.
2) _____, *Family Health Program Guideline*, 1991~2002.

Table 2. Budget for the Inborn Errors of Metabolism of the Government Program (2002)

(Unit: thousand won)

Classification	Contents	Central Government Budget (40%)	Local Government Budget (60%)
Cost for Test	9,500 won × 390,000 persons = 3,705,000 (US$ 3,036,885)	1,482,000 (US$ 1,214,754)	2,223,000 (US$ 1,822,131)
Cost for Management of Patient	439,616 (US$ 360,341)	175,846 (US$ 144,136)	263,770 (US$ 216,205)
PKU	2,540,000 won × 61 persons = 154,940 (US$ 127,000)	61,976 (US$ 50,800)	92,964 (US$ 76,200)
CH	276,000 won × 301 persons = 83,076 (US$ 68,095)	33,230 (US$ 27,238)	49,846 (US$ 40,857)
Organic Aciduria	6,720,000 won × 30 persons = 201,600 (US$ 165,246)	80,640 (US$ 66,098)	120,960 (US$ 99,148)
Total	4,144,616 (US$ 3,397,226)	1,657,846 (US$ 1,358,890)	2,486,770 (US$ 2,038,336)

Source: MOHW, *Family Health Program Guideline*, 2002, p.41.

The cost of newborn screening was 8,000 won for 6 kinds of test in 1991 and 9,500 won for 2 kinds of test in 2002 (see Table 3).

Table 3. Cost for Newborn Screening of the Government Program (1991~2002)

(Unit: Won)

Year	Cost	Test Items
1991	8,000(US$ 6.2)	6 kinds: PKU, CH, HU, MSUD, GAL, HIS
1992~1994	14,000(US$ 10.8)	6 kinds: PKU, CH, HU, MSUD, GAL, HIS
1995~1996	8,000(US$ 6.2)	2 kinds: PKU, CH
1997	8,640(US$ 6.5)	2 kinds: PKU, CH
1998~2002	9,500(US$ 7.3)	2 kinds: PKU, CH

Note: PKU: Phenylketonuria, CH: Congenital Hypothyroidism, HU: Homocystinuria, MSUD: Maple Syrup Urine Disease, GAL: Galactosemia, HIS: Histidinemia

Source: MOHW, *Family Health Program Guideline*, 2002.

During the period between 1991 and 2001, 2,222,110 newborns were screened under the government program. 488 Congenital Hypothyroidism patients and 49 Phenylketonuria patients were detected with the detection rates of 1 per 4,553 for CH and 1 per 45,349 for PKU (see Table 4).

Table 4. Number of Tested Newborns and Detected Patients (1991～2001)

(Unit: event, person)

Year	No. of Tested Newborns	No. of Detected Patients	
		CH	PKU
1991	28,286	7	1
1992	20,372	7	0
1993	35,094	8	0
1994	51,045	12	1
1995	74,880	8	2
1996	62,542	6	1
1997	345,013	62	6
1998	416,115	132	7
1999	398,444	88	9
2000	407,981	89	9
2001	382,338	69	13
Total	2,222,110	488	49

Sources: MOHW, *Internal Material*, 1991～2001.
KFHWA, *2001 Report on the Statistical Analysis of the Blood Collection Papers of the Screening Test*, 2002.

KFHWA plays a coordinating role in the newborn screening program. Through health centers, it distributes blood test paper to

the blood collection agencies and screening laboratories. Blood test samples are taken at health centers or medical facilities and sent to laboratories either directly or by mail. The test results are being sent back to the client organization. Any positive result is notified immediately to both the client organization and the district health center. Any newborn with positive results on the first screening test should be taken a close examination and the cost is on his own. However, local government supports the cost for the low-income family. The laboratory responsible for the screening test will notify the family, health center and client organization immediately via telephone and let the patient take further treatment and management.

The test laboratories claims health center test fee every month. Health center pays 9,500 won per test and 900 won out of 9,500 won sent to the KFHWA for management cost of the screening test. The important by product of this process is producing statistics on the newborn screening by KFHWA. KFHWA also conduct external inspection on the laboratories and produce statistics on the quality of the screening tests.

In 1994, the Quality Assurance Committee (QAC) on the Inborn Errors of Metabolism was established in KFHWA to conduct inspections on the government designated laboratories. The number of these laboratories increased rapidly since 1997, when the government screening program expanded to cover all newborns. KFHWA sampls 10 percent of all newborn blood test papers and analyzes them. Table 5 shows the distribution of the newborns by the number of tests taken, based on the sampled data from KFHWA. The laboratories also report screening tests not only on government supported PKU and CH but also on HU, MSUD, GAL, HIS that is not covered by

the government program. The government supported two items occupied a majority share. The proportion of the two items only had decreased from 72.7% in 1998 to 66.2% in 1999 and then increased to 73.2% in 2000 and stayed at 70.0% in 2001.

Table 5. Distribution of Newborns by the Number of Screening Tests Taken

(Unit: %, person)

No. of Screening Tests	1998	1999	2000	2001
2 kinds[1)]	72.7	66.2	73.2	70.0
3 kinds	0.3	2.1	1.7	7.1
4 kinds	0.8	1.2	0.7	0.8
5 kinds[2)]	10.9	17.0	14.3	13.5
6 kinds[3)]	15.3	13.5	10.1	8.6
Total	100.0 (28,274)	100.0 (27,000)	100.0 (27,000)	100.0 (27,000)

Notes: 1) PKU, CH, government provided tests
2) PKU, CH, HU, MSUD, GAL
3) PKU, CH, HU, MSUD, GAL, HIS

Source: KFHWA, *2001 Report on the Statistical Analysis of the Blood Collection Papers of the Screening Test*, 2002.

According to the KFHWA data, the test rates for both CH and PKU were 100%, while those for GAL, HU and MSUD included in DRG were about 30 % or less (see Table 6).

According to the survey on the 69 laboratories participated in external quality assurance program in 2001, all of them conducted screening on the government supported diseases, PKU and CH. However, as for other test items, the percentage of laboratories conducting screening test on GAL was 75%, MSUD 57%, HU 38%, and CAH 44%. Guthrie method (BIA) was used for PKU at 10

laboratories and ECA/EIA method at 59 laboratories. However, contracted screening laboratories with many test cases usually used cheap Guthrie method (see Table 7).

Table 6. Test Rate on the Test Item[1)]

(unit: %)

Test Item	1999	2000	2001	Remarks
PKU	100.0	100.0	100.0	Government support
CH	100.0	100.0	100.0	Government support
HU	30.7	24.8	23.5	DRG
MSUD	30.7	24.7	23.3	DRG
GAL	31.6	25.0	22.5	DRG
CAH	16.4	11.9	14.4	DRG

Note: 1) Result on the sample of 27,000 newborns.
Source: KFHWA, *2001 Report on the Statistical Analysis of the Blood Collection Papers of the Screening Test*, 2002.

Table 7. Methods of Newborn Screening[1)](2001)

(Unit: post, %)

Disease	Test Items	Method of Screening Test					No. of Screening Laboratory	%
		BIA	ECA/EIA	RIA	others	total		
Phenylketonuria	Phenylalanine	10	59	-	-	69	69	100.0
Congenital Hypothyroidism	TSH	-	62	7	-	69	69	100.0
	T4	-	18	5	-	23		
Maple Syrup Urine Disease	Leucine	22	-	-	-	22	39	56.5
	Leu, Ile, Val	-	17	-	-	17		
Homocystinuria	Methionine	26	-	-	-	26	26	37.7
Galactosemia	Galactose	6	44	-	-	50	52	75.4
	Galactose-1-PUT	-	-	-	4	4		
Congenital Adrenal Hypertrophy	17-OHP	-	30	-	-	30	30	43.5

Note: 1) Survey Result from 69 Screening Laboratories in 2001.
Source: MOHW · KFHWA, *Education for Clinical Pathologists in 2001*, p.125.

'Quality Assurance Committee on the Inborn Errors of Metabolism' produced ten test samples—including abnormal ones—four times a year (March, June, September, November), sent them to government-designated laboratories and analysed the reports they produced.

In 1997, the committee set a rejection standard regarding laboratory reports based on the external inspection of PKU and CH. According to this standard, more than 1 false negative on the test of PKU and CH or more than 2 false positive on each item is subject to rejection. A laboratory rejected more than three times within a period of one year may be disqualified as a designated laboratory. The pass rate has been increased recently, however there has been fluctuations even within the year by time period. Most of the laboratories failed to pass PKU tests are found to have used Guthrie method. (see Table 8).

In order to enhance the quality of screening tests, test procedure should be taken concurrently with test sample collection on a daily basis. However, only 10.5% of the total laboratories conducted daily tests, while those undertaken weekly tests occupied 41.9% in 2000 (see Table 9).

Among the 86 screening laboratories, 25.6% conducted tests on the two items only under the government support. The percentage of laboratories conducted tests on 5 items took up 32.5% (see Table 10).

Table 11 shows the distribution of the laboratories according to the cost per additional test, which ranges from free of charge to more than 10,000 won per each test item. Proportion of the laboratories by cost were almost evenly distributed, ranging from 'less than 5,000 won (28.1%)', '5,000~less than 10,000 (28.1%)' to more than 10,000 won (26.6%).

Table 8. Result of External Inspection on Newborn Screening

(Unit: post, %)

Year	Quarter	No of Participating Laboratories	No of Passed Laboratories	No of Rejected Laboratories	Pass Rate
1994	1	17	15	2	88.2
	2	17	15	2	88.2
1995	1	22	21	1	95.5
	2	22	17	5	77.3
1996	1	24	24	0	100.0
	2	24	22	2	91.7
	3	23	22	1	95.7
	4	23	23	0	100.0
1997	1	74	67	7	90.5
	2	79	76	3	96.2
	3	78	78	0	100.0
	4	76	67	9	88.2
1998	1	75	45	30	60.0
	2	75	73	2	97.3
	3	76	68	8	89.5
	4	76	69	7	90.8
1999	1	76	74	2	97.4
	2	77	74	3	96.1
	3	78	70	8	89.7
	4	78	75	3	96.2
2000[1)]	1	76	74	2	97.4
	2	75	71	4	94.7
	3	74	70	4	93.3
	4	71	70	1	94.6
2001[2)]	1	71	71	0	100.0
	2	70	68	2	97.1
	3	69	68	1	98.6
	4	68	68	0	100.0

Note: 10 test samples were produced for inspection each time and conducted test on 8 items on each sample. 8 test items are Phenylalanine, TSH, T4, Leucine(Leu, Ile, Val), Methionine, Galactose, Galactose-1-PUT, 17-OHP.

Sources: Survey Result on the Inborn Errors of Metabolism, *Quality Assurance Committee on the Inborn Errors of Metabolism*, 2000.

1) MOHW·KFHWA, *Education for Clinical Pathologists in 2001*, p.115.

2) KFHWA, *2001 Report on the Statistical Analysis of the Blood Collection Papers of the Screening Test*, 2002.

Table 9. Number of Screening Laboratory by Test Interval (2000)

(Unit: post, %)

Test Period	No. of Screening Lab	%
Daily	9	10.5
4~5 times per week	5	5.8
3 times per week	12	13.9
2 times per week	19	22.1
Weekly	36	41.9
Every 10 days	1	1.2
Every other week	2	2.3
Others	2	2.3
Total	86	100.0

Source: KFHWA, *Internal Material*, 2000.

Table 10. Number of Screening Laboratory by Test Items (2000)

(Unit: post, %)

Test Items	No. of screening Lab	%
2	22	25.6
4	6	7.0
5	28	32.5
6	14	16.3
7	15	17.4
8	1	1.2
Total	86	100.0

Source: KFHWA, *Internal Material*, 2000.

Table 11. Number of the Screening Laboratory by Cost Per Additional Test[1)]

(Unit: post, %)

Cost for Test	No. of Screening Lab	%
Free of Charge	10	15.6
Less than 5,000 won	18	28.1
5,000~less than 10,000 won	18	28.1
More than 10,000 won	17	26.6
Others	1	1.6
Total	64	100.0

Note: 1) Analysis based on the 64 laboratories except 22 Labs without additional test.

Source: KFHWA, *Internal Material*, 2000.

Ⅲ. Survey Results

1. National Fertility and Family Health Survey

According to the survey result on the National Fertility and Family Health, the newborn screening rate was increased from 35.7% in 1994 to 88.8% in 2000. The rate has been higher for rural areas that for urban areas since 1997, presumably due to the active program delivery by health workers at health centers in rural area. The rate was high among the women with a university degree and above (see Table 12).

Table 12. Taking Screening Test of the Last Birth to 15~44 Year Old Married Women

(Unit: %, person)

Classification		1994[1)]	1997[2)]	2000[3)]
Region	Urban	35.7	73.4	88.4(1,242)
	Rural	35.8	74.4	93.0(109)
Education	Middle School	31.2	65.7	81.5(53)
	High School	33.5	71.6	86.7(768)
	University and Above	43.2	79.0	92.5(530)
Age of mother at delivery	15~24	36.6	70.2	83.3(201)
	25~29	33.9	72.7	90.9(695)
	above 30	39.3	77.3	87.9(455)
Total		35.7	73.6	88.8(1,351)

Sources: 1) Hong M. S. et al., *1994 National Fertility and Family Health Survey*, KIHASA, 1994.
2) Cho, N. H. et al., *1997 National Fertility and Family Health Survey*, KIHASA, 1997.
3) Kim, S. K. et al., *2000 National Fertility and Family Health Survey*, KIHASA, 2000.

Although the government-supported newborn screening tests have expanded to cover all newborns, most women tend to get their newborns take additional tests at their own expense. Very often, hospitals demand a high cost on an additional test without requesting government to pay for the 2 items due to the complicated reimbursement system. The proportion of self-paid for the newborn screening was 81.0% (see Table 13). Therefore, it seems some hospitals have provided two tests free of charge and offset the cost by offering other charged items.

Table 13. Cost of Screening Test for the Last Birth to 15~44 Year Old Married Women (2000)

(Unit: %, person)

Region	Self-paid	Free of charge	Self-paid+ government	Others	Total
Whole country	81.0	13.5	2.8	2.7	100.0(1,020)
Urban	81.6	12.6	2.9	2.9	100.0(947)
Rural	72.3	25.0	2.7	--	100.0(73)

Note: Government funded only 2 items(Congenital Hypothyroidism and PKU).
Source: Kim SK et al., *2000 National Fertility and Family Health Survey*, KIHASA, 2000.

2. Health Center-based Survey on the Management of Inborn Errors of Metabolism

This study utilized structured questionnaires to conduct a survey covering 241 health centers in June 2000 to identify characteristics of the detected patients and to review status of screening system in Korea. In addition, a supplementary survey was conducted on the patient's family at the patient's family association to gather

information from them (Han, 2000).

In order to identify the number of Inborn Errors of Metabolism, the information on the patients was gathered from the survey and various existing sources and made into an duplication-free data set by utilizing patients' ID numbers. The total number of cases identified through this process was 481, including 378 CHs, 73 PKUs, 11 MSUDs, 7 PAs, 9 UCDs and 3 GALs (see Table 14). Most of the cases were identified after 1997 with the expansion of the government program.

Table 14. Distribution of Inborn Errors of Metabolism Identified

(Unit: person)

Year of birth	CH		PKU		MSUD		Propionic Acidosis		Urea Cycle Disorders		Galactose-mia	
	M	F	M	F	M	F	M	F	M	F	M	F
2000[1)]	24	18	1	-	2	-	1	1	-	1	1	-
1999	44	44	9	1	3	-	3	1	3	3	1	1
1998	49	37	3	3	1	-	1	-	1	1	-	-
1997	34	41	4	3	3	-	-	-	-	-	-	-
1996	10	10	1	2	1	-	-	-	-	-	-	-
1995	5	12	4	4	-	-	-	-	-	-	-	-
1994	8	7	1	3	-	-	-	-	-	-	-	-
1993	9	4	0	0	-	-	-	-	-	-	-	-
1992	4	1	2	2	-	1	-	-	-	-	-	-
1991	1	2	0	5	-	-	-	-	-	-	-	-
~1990	6	8	11	14	-	-	-	-	-	-	-	-
Total	194	184	36	37	10	1	5	2	4	5	2	1

Note: 1) Only the patient born from Jan. to May in 2000 were included

Among the 260 patients whose time of discovery was known, 83.5% was reported to be discovered within 2 months after birth. Especially the discovery rate within 2 month after birth had been increased rapidly from 23.5% in 1994 to 90% in 1997 (see Table 15).

Table 15. Time of Discovery of Inborn Errors of Metabolism

(Unit: person, %)

Year of Birth	Less than 2 Month		2 Months~ Less than One Year		1 Year ~ Less than 3 Years		More than 3 Years		Total	
	N.	%	N.	%	N.	%	N.	%	N.	%
2000	31	96.9	1	3.1	-	-	-	-	32	100.0
1999	59	93.7	4	6.3	-	-	-	-	63	100.0
1998	60	92.3	3	4.6	2	3.1	-	-	65	100.0
1997	45	95.7	2	4.3	-	-	-	-	47	100.0
1996	6	75.0	1	12.5	1	12.5	-	-	8	100.0
1995	8	72.7	1	9.1	2	18.2	-	-	11	100.0
~1994	8	23.5	2	5.9	8	23.5	16	47.1	34	100.0
Total	217	83.5	14	5.4	13	5.0	16	6.1	260	100.0

From the year of 1997, most cases of Inborn Errors of Metabolism were identified by screening test. However, 48.5% of the cases in 1994 were identified due to abnormal symptom and 30.3% was due to growth retardation before 1994 (see Table 16).

Table 16. Moment of Discovering Inborn Errors of Metabolism

(Unit: person, %)

Year of Birth	Screening Test		Abnormal Symptom		Growth Retardation		Total	
	N.	%	N.	%	N.	%	N.	%
2000	32	100.0	-	0	-	0	32	100.0
1999	61	96.8	2	3.2	-	-	63	100.0
1998	60	98.4	1	1.6	-	-	61	100.0
1997	47	100.0	-	-	-	-	47	100.0
1996	7	77.8	1	11.1	1	11.1	9	100.0
1995	8	80.0	1	10.0	1	10.0	10	100.0
~1994	7	21.2	16	48.5	10	30.3	33	100.0
Total	222	87.1	21	8.2	12	4.7	255	100.0

Note: Excluded 5 persons without response.

Family history was asked and 258 cases responded. Although the small size of the sample does not allow any generalization, this information will prove to be useful for further studies on the influence of family upon the occurrence of disease. 17.7% of PKU patients and 4.2% of the Congenital Hypothyroidism cases have family history (see Table 17).

Table 17. Distribution of Patient with Inborn Errors of Metabolism by Family History

(Unit: person, %)

Family History	CH		PKU		MSUD		PA		UCD		GAL	
	No.	%	No.	%	No.	%	No.	%	No.	%	No.	%
Yes	8	4.2	9	17.7	2	33.3	1	16.7	1	25.0	-	-
No	182	95.8	42	82.3	4	66.7	5	83.3	3	75.0	3	100.0
Total	190	100.0	51	100.0	6	100.0	6	100.0	4	100.0	3	100.0

Ⅳ. Identified Problems

The identified problems through the study are as follows.

- Absence of an organization responsible for coordination and control of national newborn screening services.
- Too many screening laboratories. There are 69 laboratories for newborn screening tests in 2002 in Korea.
- Test items are not selected according to the prevalence of diseases except PKU and CH and the number of tests, and costs varies across medical facilities.

- Follow up treatment and services are not adequate.
- Remuneration system is very complicated and requires a lot of administration process.

Ⅴ. Recommendations

- Forming an organization responsible for national newborn screening, and provide services such as quality assurance and accreditation of the screening laboratory, technical assistance and research.
- Forming a newborn screening and patient management model consisting of test sample collection system, screening laboratory work system, follow-up retest system and confirming diagnosis and treatment system.
- Reducing the number of screening laboratories from current 69 posts to 3~4 posts by putting strict requirement on the standards and also induced by cost-competitiveness. Adoption of Tandem Mass Screening as an alternative for quality assurance and cost-effective way of improving the current screening method.
- Strengthening the follow-up management of the identified patient. It will not give much benefit to the improvement of health of the people if the patient are not treated properly even with money for screening tests.
- Simplifying the current complicated remuneration system. There are several alternative method such as providing coupon, commissioning health insurance company payment or payment

through health insurance system.

- In order to improve the patient management, activities of the health center should be strengthened supported by the central government with patient management guidelines and health education materials.
- As a part of the research, dietary guidelines with a standard menu for the Inborn Errors of Metabolism was developed. It was based on the analysis of component of the favorite Korean food selected by the patient family association. This guidelines should be supplemented and revised in the future.

References

Cho, N. H., et al., *1997 National Fertility and Family Health Survey*, KIHASA, 1997.

Han, Y. J., Lee, D.H., Kim, K.S., *Measures to Improve Newborn Screening System*, KIHASA, 2000.

Hong M. S., et al., *1994 National Fertility and Family Health Survey*, KIHASA, 1994.

KFHWA, *Internal Material*, 2000.

_____, *1999 Report on the Statistical Analysis of the Blood Collection Papers of the Screening Test*, 2000.

Kim, S. K., et al., *2000 National Fertility and Family Health Survey*, KIHASA, 2000.

MOHW, *Family Health Program Guideline*, 2000~2002.

_____, Internal Material, 1991~1999.

_____, *Plan of Family Health and Welfare Program*, 1991~2000.

MOHW·KFHWA, *Education for Clinical Pathologists in 2001.*

Survey Result on the Inborn Errors of Metabolism, *Quality Assurance Committee on the Inborn Errors of Metabolism*, 2000.

Summary

先天性代謝異狀 檢查

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국가에서 의무적으로 실시하는 신생아에 대한 스크리닝검사는 검사 및 치료시기를 놓칠 경우 돌이킬 수 없는 심각한 장애를 초래하는 질환 중에서 치료효과가 확실한 질환을 우선적으로 하고 있다. 보건복지부는 1991년 저소득층 신생아에 대한 선천성대사이상 검사사업을 실시하였으며, 1997년 1월부터는 이 사업을 모든 신생아로 확대하였다. 선천성대사이상 검사는 출생후 48시간 이후부터 7일 이내에 검사를 실시해야 하며, 환자가 발견된 후에는 평생 치료 및 관리가 필요한 질환이다.

정부사업으로 동 검사를 실시한 이래 10여 년이 경과하였으므로 사업에 대한 평가가 필요한 시점이다. 이를 위해 전국 241개 보건소 담당자를 대상으로 동 사업 현황과 보고된 환아에 대한 조사를 실시하였으며, 환자 가족모임을 통해서도 자료를 수집하였다. 그리고 선천성대사이상 검사기관의 정도관리 실태와 기존 연구자료를 분석하였으며, 국내외 전문가들의 의견을 수렴하였다. 본 연구에서는 선천성대사이상 검사사업과 환아에 대한 현황과 문제점을 파악하여 추후 정책 수립에 기초자료로 제공하고자 하였으며, 사업 개선을 위한 정책 대안을 제시하였다.