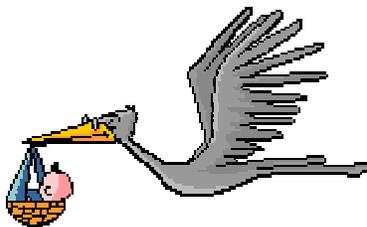


# Infant Screening Task Force Recommendations and Report

**Implementation of House Bill 817**

*Screening, Identifying, Diagnosing, and Managing Newborns At  
Risk for Metabolic, Endocrine, and Genetic Disorders and  
Hemoglobinopathies*



Allen W. Root, M.D., Chairman

Infant Screening Task Force

**September 1, 2002**



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September 1, 2002

Jeb Bush, Governor  
John M. McKay, President of the Florida Senate  
Tom Feeney, Speaker of the House  
John O. Agwunobi, M.D., M.B.A., Secretary of the Department of Health

Dear Governor Bush, President McKay, Speaker Feeney, and Secretary Agwunobi:

Attached please find the report of the Infant Screening Task Force implemented in response to House Bill 817. The members of the Task Force ardently support recommendations for improvement of the current infant screening program and its expansion to include a number of metabolic disorders. These diseases result in death or severe neurologic impairment unless detected and treated in the period immediately after birth. Appropriate therapy effectively ameliorates the medical, neurological, and socioeconomic consequences of these dreadful illnesses.

The Task Force recommends this report to you and requests your consideration and support for its implementation. If you require additional information, please contact me.

Sincerely,

Allen W. Root, M.D.  
Chairman, Infant Screening Task Force  
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## Executive Summary

*In response to House Bill 817 concerning improvement and expansion of Florida's neonatal screening program, this report presents the recommendations made by the Florida Infant Screening Task Force. They result from the research and deliberations of the Task Force at meetings on May 13, June 10, July 11, July 26, August 7, and August 8, 2002. The Florida Infant Screening Task Force consisted of all members of the Genetics and Infant Screening Advisory Council of Children's Medical Services and a member from each of the following: Florida Hospital Association, Florida Statutory Teaching Hospital Council, and Florida Chapter of the March of Dimes.*

*The Florida Infant Screening Task Force members reviewed the findings and recommendations regarding the expansion of the newborn screening program previously put forth by the Genetics and Infant Screening Advisory Council. On June 10, 2002, the Task Force met with George Cunningham, M.D., California Department of Health, and Joseph Muenzer, M.D., Ph.D., University of North Carolina, who provided information regarding their states' experiences with expanded newborn screening using tandem mass spectrometry technology. Using all the information available to them, the Task Force made the following recommendations delineated in the sections of the report:*

- *The Florida Infant Screening Program should correct deficiencies identified in the current infrastructure in order to reduce the amount of time from birth to diagnosis and treatment, thus decreasing the risk of morbidity and mortality.*
- *The Florida Infant Screening Program should expand the neonatal screening program from five (5) disorders to twenty-eight (28) disorders in order to identify potentially catastrophic, but treatable, illnesses that, when detected early, can preserve useful lives and reduce future costs of healthcare and special services.*
- *The improvements in, and expansion of, the Florida Infant Screening Program should be funded with state General Revenue because neonatal screening is a public health function.*

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## Introduction

Neonatal screening is the process by which all newborns are tested shortly after birth for selected disorders with potentially adverse consequences that can be identified and treated before the illness becomes apparent. Florida's Infant Screening Program presently screens all newborns to identify, diagnose, and treat those newborns with one of five endocrine-metabolic or genetic disorders, or significant hemoglobinopathies such as sickle cell anemia. This is done in coordination with hospitals, birthing facilities, and endocrine-metabolic, genetic and hematology referral centers.

It is the goal of the Florida Infant Screening Program to screen every newborn in Florida for a selected group of disorders so that she or he may be diagnosed and treated before the consequences of the disease become apparent in order that she/he may have the greatest opportunity to live a normal, productive life. The program is only successful if the affected newborns are identified and treated **before** the damaging effects of the disorder occur. The long-term benefits are a better quality of life for the child and his/her family, and considerable cost savings for the insuring payers and the taxpayers of the state of Florida.

In 1965, Florida began screening newborns for phenylketonuria (PKU). Today, the Florida Infant Screening Program also tests for: galactosemia, congenital hypothyroidism, congenital adrenal hyperplasia, and hemoglobinopathies, including sickle cell disease. Currently, this program is funded by assessment of birthing facilities. According to s. 383.14, F.S., a hospital assessment fee of \$20 is charged for each live birth, as recorded by the Office of Vital Statistics, occurring in a hospital licensed under Part I of Chapter 395, F.S., (up to 3000 births per year), or a birth center licensed under s. 383.305, F.S., (over 60 births per year).

For each live birth in Florida, a blood specimen is obtained (by heel stick) by the hospital or birthing facility and placed on special filter paper. It is mailed to the Department of Health, Infant Screening Program Laboratory, in Jacksonville for testing. The laboratory performs the infant screening tests and reports the results to the Children's Medical Services (CMS) Program, birthing facility, and the physician of record.

The CMS Infant Screening Program staff is immediately notified by the State Laboratory of all presumptively abnormal test results. In turn, the CMS staff immediately notifies the appropriate referral center designated to provide diagnostic, treatment, and follow-up services. The CMS staff tracks each newborn with an abnormal test result to be certain that the at-risk newborn receives prompt and effective care. Since initial screening tests give only preliminary information, more precise testing must follow it. Thus, a presumptively abnormal screening test result indicates that further testing is necessary in order to confirm or eliminate the diagnosis suggested by the screened disorder. Infants with presumptively abnormal test results are referred to one of three CMS endocrine-metabolic or genetic referral centers, or one of nine CMS hematology-oncology centers that provide medical expertise in endocrinologic, metabolic, genetic, and hematologic disorders of newborns. All referred neonates undergo confirmatory testing and receive treatment if indicated. Confirmatory testing is the precise testing that

provides the definitive diagnosis of a suspected disorder identified through the screening process.

Between 1980 and 2000, more than 2.6 million newborns in Florida were screened for phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell disease, and congenital adrenal hyperplasia. Of that total, 2,799 newborns were found to have one of these disorders; that is, more than one in every 1000 newborns had a significant illness that was not apparent at birth, and, without the screening program, would likely not have been identified until he/she was severely ill or had died. Many of the 2,799 newborns identified by the Florida Infant Screening Program during these two decades would have been severely developmentally disabled if early diagnosis had not been available. It must be noted that the average cost of residential care for an individual who is developmentally disabled is greater than \$101,000 per year. This is an estimated cost for a typical state operated facility in Florida, but daily care is dependent on the level of services needed by the individual and could exceed \$400 per day. Hospitalizations, diagnostic services, and specialty treatments are not included in the cited amount. The emotional cost to a family, who must rear a child with a permanent disability, cannot be calculated.

The contribution of the neonatal screening program must be considered not only in abstract numbers and dollar figures, but also in relationship to the human element. The program has afforded the affected newborns the opportunity to grow, develop, and mature into educated and productive adults. The families of these infants have been spared the disruptive effects and extraordinary emotional and financial costs of serious illness, specialized schooling, and long-term residential care. With early diagnosis and treatment, most of these children have progressed through the educational system, often as honor roll students; enrolled in college; pursued careers in the trades or business; and, have become contributing citizens to the tax base of the state and country. It has been reported that newborn screening programs for congenital hypothyroidism save or return to the state \$7.00 for every \$1.00 expended. In this context, improvement and expansion of the present newborn screening program is needed to facilitate still further its mission. Newly developed technologies such as tandem mass spectrometry provide the opportunity to expand the screening program – specifically, to identify newborns with metabolic diseases (i.e., disorders of the metabolism of certain amino, organic, and fatty acids) that result in sudden infant death and/or severe neurologic disabilities.

During the 2002 Legislative Session, Senator Wasserman-Schultz and Representative Sobel introduced legislation to create a task force to study the Florida Infant Screening Program. The task force was charged with the following: 1) to identify deficiencies in the current neonatal screening program that when corrected will result in improved service to Florida's newborn population; 2) to consider expansion of the neonatal screening program to include additional disorders; 3) to consider the costs of improving the current testing program and of expansion; 4) to suggest sources for reimbursing the costs of testing; and, 5) to determine the appropriate location for such testing. This report provides the recommendations of the Infant Screening Task Force.

## **Part I: Improvements to Existing Program**

After careful examination of the current neonatal screening program, the Task Force identified areas and procedures in the program that could be improved. The Task Force further determined that these deficiencies must be corrected prior to program expansion. Thus, the Task Force recommends the following steps to correct identified deficiencies:

- Improve specimen collection rate including the percentage of properly collected specimens.
- Reinstigate overnight transportation of specimens to the Department Of Health State Laboratory for more rapid testing.
- Ensure processing and testing of the specimen by the State Laboratory in a timely and accurate manner.
- Streamline laboratory testing and follow-up procedures to ensure immediate reporting of presumptively abnormal test results to CMS Infant Screening staff, the collection facility or provider, and referral centers that will provide confirmatory testing and follow-up care of the newborn.
- Increase staff coverage to seven days each week at the State Laboratory, CMS, and referral centers.
- Expedite notification of specialists and consultants at the referral centers of presumptively abnormal screening test results in order to hasten diagnosis and initiation of treatment.
- Ensure long-term follow-up that includes continuity of care in a medical home with assessment of outcomes.
- Expand education of primary care physicians on the disorders being screened.
- Enhance quality assurance and evaluation to ensure the program's efficacy.

These improvements will reduce the amount of time from birth and testing to diagnosis and treatment, thus minimizing the risk of morbidity and mortality. There is also a recommendation to change the name of the Florida Infant Screening Program to Florida Newborn Screening Program to allow Florida to conform to nationally accepted terminology when referring to newborn screening. The above areas must be addressed and improvements completed before new disorders are added to the screening program.

**Recommendations and costs to improve the existing program:**

Recommendations to Improve Existing Program	Estimated Costs
<p><b>Utilize a courier service:</b> In order to reduce the time that elapses between specimen collection and its receipt by the State Laboratory, contract with a courier service to ensure overnight delivery from all birthing facilities seven days a week.</p> <p><i>*Cost calculation: \$10/package x 150 facilities x 365 days/yr.</i></p>	<b>547,500</b>
<p><b>Implement a “Weekend and Holidays” on-call system:</b> In addition to the routine weekday schedule, CMS Infant Screening Staff will rotate “on call” assignments to receive screening test results from the State Laboratory on weekends and holidays. Designated “on call” CMS, genetic, endocrine-metabolic and hematology referral center staff will be then contacted and requested to arrange rapid evaluation of the at-risk neonate. In order to accomplish this task, CMS will expand contractual responsibilities and funding of genetic, endocrine-metabolic and hematology centers for staff to accept referrals of newborns with presumptively abnormal test results on a daily basis. The referral center will then contact parents to arrange for timely follow-up of the at-risk newborn. Contract funding and service provisions will include professional time and cell phone costs for weekend and holiday follow-up for referral center and CMS staff.</p> <p><i>**Cost calculation: State holidays, Saturday and Sunday = 113 days/ yr. Based on an hourly rate of \$25 x ¼ state hourly rate = \$50.00/ day (8 hours). 113 days X \$50 = \$5,650.00/ person. 17 persons (1 person/center and CMS office)= \$96,050. Estimated annual cell phone cost = \$1,430 annually/person x 17 persons= \$24,310.</i></p>	<b>120,360</b>
<p><b>Initiate an intensive educational campaign:</b> Provide an intensive educational program to birthing facilities regarding the importance of submitting timely and correctly collected blood specimens to the State Laboratory. Contract for services of an educator for targeted on-site educational programs for hospital and medical staff, to prepare Infant Screening Program (ISP) printed materials and monthly newsletter, and to develop and maintain a current educational website. Materials shall include: (a) information regarding appropriate times to obtain screening samples in order to reduce the number of inappropriate Day Of Birth screening specimens; (b) documents describing responsibilities of hospitals, birthing centers or health care providers to ensure an initially collected satisfactory blood sample; and, (c) detailed review of each neonate with a presumptively abnormal test result to demonstrate timely confirmation of the illness within 1-2 days after receipt of the report.</p>	<b>100,000</b>
<p><b>Revise the ISP brochure:</b> Revision will emphasize the importance of neonatal screening and need for prompt response by parents when repeat screening or follow-up medical evaluation is necessary, e.g., “Help Us Keep Your Baby Healthy.”</p>	<b>30,000</b>
<p><b>Reduce laboratory-testing timeline:</b> Revise newborn screening rules (Florida Administrative Code, F.A.C., 64C-7) to reduce laboratory-testing timeline from 10 days to 5 days after receipt of a specimen and revise name of program to reflect national language. Rule promulgation will be required.</p>	<b>2,000</b>
<b>Total</b>	<b>\$708,360</b>

**Infant Screening Task Force recommendations to improve the on-going screening program that are currently being implemented within existing resources include:**

- Review laboratory testing methodology to determine if a reduction in time to test completion can be achieved.
- Expand the Infant Screening Laboratory hours for operation on weekends and holidays.
- Update the Infant Screening Laboratory's computer system.
- Fax laboratory results to appropriate parties in order to avoid mailing time.
- Revise reporting criteria for all test results to decrease the time from screening to follow-up.
- Revise referral center reporting requirements to reflect more frequent updates.
- Implement a "pending list" to facilitate required updates of confirmatory findings from the referral centers.
- Develop a notification and tracking system for congenital hypothyroidism and congenital adrenal hyperplasia.

**The appropriate location for specimen collection:**

The Task Force members agreed that the hospital or birthing center is the appropriate location for the initial specimen collection. Birthing facilities submitting unsatisfactory specimens should be required to collect the repeat specimen and ensure that it is done correctly. It was determined that repeat specimens, required for other reasons, could be addressed by the Infant Screening Advisory Council.

## **Part II: Expansion of Current Program**

It is a prerequisite that the prior recommendations designed to improve the current program be funded and implemented before addition of new disorders to the newborn screening program.

With these improvements in place, the Task Force then unanimously recommends that infants born in Florida be offered expanded newborn screening for disorders of amino, organic, and fatty acid metabolism. Expansion of newborn screening to include these disorders will result in the prevention of catastrophic illnesses leading to death or neurological impairment of infants with serious metabolic diseases. Furthermore, families at risk for giving birth to infants with serious metabolic diseases will be identified so that genetic counseling and early treatment of the affected infants, and his/her future siblings, can be provided.

The Task Force emphasizes that all elements of an expanded newborn screening program must be in place before its implementation, in order to maximize benefits and minimize risks. These elements include:

- Equipment, procedures, and personnel for tandem mass spectrometric analysis of amino, organic, and fatty acids in filter paper dried blood specimens.
- Personnel with expertise in the discipline of Biochemical Genetics to provide consultative services to the State Laboratory, including technical advice and aid in interpretation of screening results, and supervisory support in regard to definitive diagnostic studies and treatment at the peripheral referral centers.
- Personnel at the metabolic referral centers with expertise in the provision of confirmatory diagnostic and treatment services for those neonates with a presumptively abnormal metabolic screening test; these services encompass those provided by physicians, nutritionists, and genetic counselors.
- Procedures to ensure quality assurance and evaluation of the newborn screening program.

Elimination of any of these elements will place the effectiveness of the entire program in jeopardy. The Task Force does not support a pilot project to implement expansion and does not recommend the implementation of a partial expansion plan.

In CY 2000, there were 204,030 live births and 291,285 newborn screening tests.

**Table 1: Disorders currently screened in the Florida Infant Screening Program:**

<i>Disorders currently screened in Florida</i>	<i>Number of abnormal screening results</i>	<i>Number of newborns with disorder</i>
➤ Sickle cell disease	309	150
➤ Galactosemia	16	7
➤ Congenital hypothyroidism	808	63
➤ Congenital adrenal hyperplasia	872	11
➤ Phenylketonuria	113	22
	<b>2118</b>	<b>253</b>

**The benefits of expanded newborn screening include:**

1. Saving lives and preventing catastrophic illnesses that are treatable when detected early.

*Nicholas was born four years ago with a fatty acid oxidation acylcarnitine disorder. The disorder was not diagnosed until he sustained a stroke that caused significant brain damage. He appeared to be a normal, happy, healthy boy until a bout of the flu reduced his food intake, leading to hypoglycemia (low blood sugar) that led to the stroke. This was a direct consequence of his undetected metabolic disorder. After the stroke, he could no longer walk, talk, or eat. He has endured multiple hospitalizations and today has weakness on his right side, uncontrolled seizures, behavior disorders, and global developmentally delayed. He receives speech, physical, and occupational therapies. He is scheduled to have brain surgery that hopefully will decrease his seizures. His mother estimates that, between insurance and the family, almost one million dollars has been spent on Nicholas' care and treatment. Another little boy with the same disorder was tested and diagnosed at birth because his sibling died from this disorder. Placed immediately after birth on the correct diet (low fat with supplemental carnitine), he has developed normally, does not receive any therapy, and has had only two brief hospitalizations.*

2. Reducing healthcare costs associated with expensive hospital diagnostic studies and treatment of extremely ill neonates with undiagnosed catastrophic illnesses.

*Jakob, aka "Cowboy Jake", was born on January 27, 2000, in Key West, Florida. Jake was rushed to the emergency room at nine days of age because of decreased reflexes, lethargy, and irritability that led to seizures and swelling of the brain. His deteriorated condition necessitated helicopter transport by LifeFlight to a tertiary hospital where he spent thirty-one days in the neonatal intensive care unit. Numerous painful and costly diagnostic tests, including CT scan and MRI, were performed before the diagnosis of*

*Maple Syrup Urine Disease was made. As a result of the late diagnosis, Jake needed physical, occupational, and speech therapy until he was seventeen months old. Had Jake's newborn screening tests included Maple Syrup Urine Disease, he could have begun a special diet with supplemental formula, thereby avoiding the crisis. Jake is a very lucky boy in that he is now progressing well and is developmentally normal.*

3. Decreasing the number of children who develop neurological damage, and reducing the need for special education services and life long residential care.
4. Providing genetic counseling to families at risk for delivering children with treatable metabolic disorders and preemptive management of the affected newborn.

**Disorders recommended for expansion:**

The 23 disorders to be included in an expanded neonatal screening program as recommended by the Task Force are listed in Table 2. To better understand the effect that these disorders have on newborns, a brief description is included, as well as the outcome, if no treatment is provided. It should be noted that most of these disorders are treated with appropriate diet restrictions or medication.

*Fatty acid oxidation:*

Newborns with fatty acid oxidation disorders may appear to be very healthy for months or years after birth; but, during their first significant cold or flu-like illness, they may develop very low blood sugars (hypoglycemia) and lapse into a coma with resulting death or brain damage. Untreated, about half will die of the disease; about 30% of children with the disease suffer significant brain damage (stroke) during the period before the diagnosis is made. If parents and physicians are aware that the newborn has the disease, medications, diet, and appropriate measures, instituted during times of illness, can prevent hypoglycemia and other consequences.

*Amino acid and organic acid:*

Newborns with amino acid and organic acid disorders are often normal at birth. After a few days or weeks, however, they generally become listless, begin to vomit, and feed poorly. Because their symptoms are so nonspecific, and because few specialists are trained to recognize these disorders, diagnosis is often delayed. The outcome may be severe coma leading to permanent neurological impairment or even death. The best hope for a normal life is through screening and treatment with special diets and medications before damage occurs. Newborns with homocystinuria, who are untreated, develop mental retardation, eye problems, and stroke. With a specialized diet and medications, these complications may be prevented.

*Biotinidase deficiency:*

Newborns with biotinidase deficiency have developmental delays and severe seizures, hearing problems, and skin problems if not promptly diagnosed and treated. Providing high doses of biotin daily to the affected newborn effectively "cures" this disorder.

Table 2 lists the 23 additional disorders, recommended by the Infant Screening Task Force, to be added to the newborn screening program. The third column in Table 2 presents the expected number of presumptively abnormal screening test results requiring referral center diagnosis and confirmation. The fourth column in Table 1 2 indicates the expected number of newborns actually diagnosed with the disorder based on Florida's 200,000 births per year. Each referral center evaluation was estimated to cost \$1,455 and includes: follow-up of presumptively abnormal test results by the Infant Screening Program staff at the CMS Program office; initial evaluations at genetic, endocrine-metabolic and hematology referral centers; first response clinic appointments; and, confirmatory testing costs. For disorders with no incidence data available, it was projected that there would be one positive result per eight presumptively abnormal screening tests.

Please note that new methodology will permit screening for phenylketonuria by tandem mass spectrometry. In its place, the Task Force recommends that screening for biotinidase deficiency be substituted. Testing for this disorder can be accomplished with presently available instruments, but will require purchase of test-specific reagents.

**Table 2: Disorders recommended for expansion:**

<i>Disorders recommended for expansion</i>	<i>Annual incidence</i>	<i>Number of abnormal screening results</i>	<i>Number of newborns with disorder</i>	<i>Projected costs</i>
<i>Acylcarnitine Disorders, Fatty acid oxidation:</i>				
➤ Carnitine Palmitoyl Transferase Deficiency-Type I (CPT-1)	~1/200,000*	8	1	11,640
➤ Carnitine Palmitoyl Transferase Deficiency-Type II (CPT-2)	~1/200,000*	8	1	11,640
➤ Carnitine/Acylcarnitine Translocase Deficiency (CAT)	~1/200,000*	8	1	11,640
➤ Long Chain hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHAD)	1/50,000	32	4	46,560
➤ Multiple Acyl-CoA Dehydrogenase Deficiency (Glutaric Acidemia-Type II)	~1/200,000*	8	1	11,640
➤ Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	~1/200,000*	8	1	11,640
➤ Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	1/10,000	160	20	232,800
➤ Very Long-Chain Acyl-CoA dehydrogenase deficiency (VLCAD)	~1/200,000*	8	1	11,640

**Table 2: (continued)**

<i>Disorders recommended for expansion</i>	<i>Annual incidence</i>	<i>Number of abnormal screening results</i>	<i>Number of newborns with disorder</i>	<i>Projected costs</i>
➤ Long-Chain Acyl-CoA Dehydrogenase deficiency (LCAD)	1/200,000	8	1	11,640
<i>Acylcarnitine disorders, Organic acids:</i>				
➤ Glutaric Acidemia-Type 1 (GA-1)	1/30,000	56	7	81,480
➤ 3-Hydroxy 3 Methylglutaryl-CoA Lyase Deficiency (HMG)	1/300,000	8	1	11,640
➤ Isovaleric Acidemia (IVA)	1/50,000	32	4	46,560
➤ 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	1/75,000	24	3	34,920
➤ Methylmalonic Acidemia (MMA)	1/50,000	32	4	46,560
➤ Mitochondrial Acetoacetyl-CoA thiolase deficiency (3-ketothiolase)	~1/200,000*	8	1	11,640
➤ Propionic Acidemia (PA)	1/50,000	32	4	46,560
<i>Amino acid disorders (phenylalanine, tyrosine, valine, methionine, and citrilline):</i>				
➤ Argininosuccinate lyase deficiency (ASA)	1/60,000	32	4	46,560
➤ Citrullinemia	1/60,000	32	4	46,560
➤ Tyrosinemia type I	1/163,000	16	2	23,280
➤ Tyrosinemia type II	~1/200,000*	8	1	11,640
➤ Maple Syrup urine disease	1/250,000	8	1	11,640
➤ Homocystinuria	1/50,000	32	4	46,560
➤ Biotinidase deficiency (not an amino acid disorder)	1/72,000	24	3	34,920
Total costs for 23 additional disorders:		<b>592</b>	<b>74</b>	<b>\$861,360</b>

*\*Note: It was presumed that there would be one confirmed positive result per eight presumptively abnormal screening results for the disorders in which the incidence rate is unknown. Because the disease is presumed to occur infrequently, the incidence rate was estimated to be ~1/200,000. The incidence rates shown above were obtained from multiple sources.*

**Estimated costs of Florida Infant Screening Program expansion:**

The following represents estimated year one start up costs and recurring annual costs to expand the newborn screening program from five to 28 disorders.

<i>Estimated costs of Florida Infant Screening Program expansion</i>	<i>Year one start up costs</i>	<i>Recurring annual costs</i>
Infant Screening Program Laboratory funding for four tandem mass spectrometry units at \$350,000 each	1,400,000	0
Infant Screening Program Laboratory expenses for mass spectrometry reagents, supplies, maintenance and software.	500,000	500,000
Infant Screening Program Laboratory expenses to add three instrument operators (Chemist II) at \$42,000 each and one senior clinical chemist (Chemical Administrator) at \$65,247. (Centers for Disease Control and Prevention guidelines recommend an operator for each instrument plus a back-up/supervisor. One operator will come from existing laboratory staff.)	191,247	196,984
Cost of reagent for biotinidase testing (assumes transition of PKU testing to tandem mass spectrometry)	94,250	29,000
Newborn Screening Biochemical Geneticist Consultant(s) (physician) to guide the expansion and assure the integrity of the expanded program. This person(s) will assist in the development of laboratory testing protocols, interpretation, and quality control; and, provide medical consultation to the Infant Screening Program, CMS Metabolic Centers, and Florida medical providers.	160,000	160,000
Enhancement of CMS Infant Screening Program (ISP) database to track newborns through the confirmation process with screening results, reporting and quality evaluations. These costs include implementation of a web-based CMS ISP database. Year one includes development and implementation. Year two reflects maintenance costs.	350,000	35,000
CMS direct patient cost for confirmation based on \$75 to track each newborn with a presumptively abnormal screening test result for 592 newborns per year (see Table 2).	44,400	46,800
CMS Metabolic or Genetic Program costs associated with confirmation based on \$1380 for 592 newborns per year (see Table 2).	816,960	816,960
CMS Metabolic or Genetic Center Program costs associated with geneticist and genetic counselor activities related to treatment and follow-up and nutritionist consultation for dietary management.	150,000	253,125
Registered Nurse Consultant in CMS to provide education to the health care providers, physicians, hospital laboratory and nursery staff, obstetrical services staff at birthing facilities, and parents. This person will also be responsible for development of educational materials regarding the additional disorders such as brochures, posters, and web-based information.	75,000	78,000
<b>Total:</b>	<b>\$3,781,857</b>	<b>\$2,115,869</b>

**Part III: Funding Required for Improvements and Expansion:  
Options and Recommendations**

According to section 383.14, Florida Statutes (F.S.), a hospital assessment fee of \$20 is charged for each live birth, as recorded by the Office of Vital Statistics, occurring in a hospital licensed under Part I of Chapter 395, F.S., or a birth center licensed under s. 383.305, F.S., up to a cap of 3,000 live births per licensed hospital per year or more than 60 births per birth center per year. (A total of 18 hospitals delivered more than 3,000 newborns in 2000.)

The billing summary for FY 2000-2001 indicates that all but \$14,500 of the \$3.2 million collected in program fees were paid by hospitals. In addition to paying the assessment, hospitals are required to collect a blood specimen from each baby born in the facility and to send the specimen to the State Laboratory for processing. The Florida Hospital Association (FHA) has estimated the collection cost at \$10 - \$30 per specimen tested. The cost estimates, multiplied by the total number of specimens tested (291,285 first and repeat specimens in CY 2000), could result in additional services contributed by hospitals to the screening program. This does not include costs incurred for repeat specimens necessitated by collection of an initially unsatisfactory specimen, most recently reported by the Infant Screening Laboratory at a rate of 3 percent. According to survey responses received by the Florida Hospital Association, hospitals that bill for specimen collection receive no reimbursement from Medicaid and limited or no reimbursement under managed care contracts. The Task Force recognizes that the contribution of Florida hospitals to the Florida Infant Screening Program is substantially greater than the \$3.2 million paid in fees.

<b>Improvements and Expansion</b>	<b>Year One Start Up Costs</b>	<b>Recurring Annual Costs</b>
Cost of current program:	3,649,556	3,649,556
Cost to improve existing program:	708,360	708,360
Cost to expand current program to 28 disorders:	<b>\$3,781,857</b>	<b>\$2,115,869</b>
<b>Total costs:</b>	<b>\$8,139,773</b>	<b>\$6,473,785</b>
Hospital fees collected in FY 2000-2001	3,195,869	3,195,869
<b>Projected deficit with existing fee structure:</b>	<b>\$4,943,904</b>	<b>\$3,277,916</b>

**Options for funding the Florida Infant Screening Program improvements and expansion:**

The Task Force evaluated a series of options to fund the program improvements and proposed expansion:

- Repealing the fee requirements and funding the Infant Screening Program with General Revenue.
- Retaining the current fee requirements and funding the improvements and expansion with General Revenue.
- Eliminating the current “cap” or “exemptions”. The Task Force determined that based on 200,000 live births, this option would result in additional funding of \$800,000 generated from fee collection from birthing facilities.
- Requiring payment of screening services by third party payers (medical insurance companies, Medicaid, etc.).
- Identifying a new funding source, such as an increase in health care provider licensing fees or other tax.
- Increasing the current fee from \$20 to \$40 per live birth and eliminating the current “cap” or “exemption” resulting in an additional collection of \$4 million, with individual birthing facilities experiencing fee increases up to 372%.

**Recommendations for funding improvements and expansion:**

The Infant Screening Program is a core public health function. Therefore, it is the opinion of the Task Force that the state of Florida should provide adequate funding so that these services are available to Florida’s tiniest citizens.

Therefore, the Task Force recommends that:

- The current fee be repealed and the entire program be funded with General Revenue;
- OR
- The current fee be retained and improvements and expansion be funded with General Revenue.

The Task Force suggests that the state of Florida explore and seek federal funding, such as block grants or a Medicaid waiver, as a means of offsetting the state’s obligation to fund the program with General Revenue.

**Florida Infant Screening Task Force final recommendation:**

Infant screening is a public health function. Florida should fund core public health functions. The Task Force members agreed that either the entire program be funded with General Revenue, or with a combination of General Revenue and federal funding.