January 18, 2013

The Honorable Edward J. Kasemeyer, Chair
Senate Budget and Taxation Committee
3 West Miller Senate Building
Annapolis, Maryland 21401-1991

Re: 2012 Joint Chairmen’s Report, Page 57, M00F03 – Report on the Feasibility of Implementing
Severe Combined Immunodeficiency Disease Screening of Newborns in Maryland

Dear Chair Kasemeyer and Chair Conway:

Pursuant to page 57 of the Joint Chairmen’s Report of 2012, the Department of Health and Mental Hygiene (DHMH) in conjunction with the State Advisory Council on Hereditary and Congenital Disorders respectfully submits this report on the Feasibility of Implementing Severe Combined Immunodeficiency Disease Screening (SCID) of Newborns in Maryland. Specifically, the report provides a cost analysis for start-up and screening, an assessment of insurance reimbursement and the financial impact on Medicaid should a newborn fail to be diagnosed with SCID. The report also advises the Budget Committees on the capability of the Department to fund SCID screening and provides the Department’s prior and future efforts to obtain federal funding to incorporate this panel.

Introduction

Severe Combined Immunodeficiency Disease (SCID) is a primary immune deficiency and is characterized by a severe deficit of both the T and B lymphocyte systems. SCID is more commonly known as “bubble boy disease” because of David Vetter, a young boy who lived in a germ-free bubble for 12 years. The prevalence of SCID is estimated at 1:66,000 live births. Those affected babies who are undiagnosed and untreated rarely survive past their first birthday, as most succumb to complicating infections. These infections are often secondary to the recommended course of pediatric vaccination that include live rotavirus vaccine in the first year of life and live Varicella, measles, mumps and rubella vaccines at 12 months of age. Importantly, hematopoietic stem cell transplantation (HSCT) can cure SCID but only if performed early in life before the baby has had multiple infections, hence the critical need to identify the babies in the newborn period before they are critically ill. Other T-cell disorders are a secondary target of SCID screening and while the impact on health may not be as dramatic, earlier detection and treatment of these disorders, such as DiGeorge sequence, may reduce morbidity in this population as well. If these secondary targets are included, it is possible that 1 in 35,000 to 50,000 infants will be identified as having a disorder.

The Maryland Newborn Screening Laboratory has been at the forefront of SCID research and development from the birth of the idea. Maryland was one of the first newborn screening laboratories to aid Dr. Jennifer Puck with provision of anonymized blood spot specimens so that she could develop the landmark SCID assay. Since that initial collaboration the Maryland newborn screening laboratory has been looking forward to incorporation of SCID into the current screening panel. The Laboratory is set in a prime location to handle the follow-up care of SCID patients, being located around the established children’s centers of Johns Hopkins Hospital, University of Maryland Medical Center, Children’s National Medical Center, and the National Institutes of Health. All of these centers are able to provide expert clinical care and expertise in pediatric immunology and skilled bone marrow transplants for affected patients.
In 2010, the Secretary’s Advisory Committee on Heritable Disorders of Newborns and Children recommended that SCID be added to the routine newborn-screening panel; this was quickly endorsed by the Secretary of the U.S. Department of Health and Human Services. Maryland’s Advisory Council on Hereditary and Congenital Disorders unanimously recommended adding SCID to the Maryland NBS panel in June 2011, however, this screening program has not yet been implemented in the State. Currently at least 11 states are actively screening for SCID. Pennsylvania is planning to begin screening in 2013.

The validation and implementation of SCID testing is contingent upon receiving funds for the project. The DHMH Laboratories Administration has well established Newborn Screening and Molecular Biology divisions which are essential to successful validation and implementation of SCID testing for babies born in the State of Maryland.

Method

The specific assay for population-based SCID screening in newborns is quantification of small DNA fragments called TREC (T Cell Receptor Excision Circle). Specific TREC sequences are detectable by real-time polymerase chain reaction (PCR). Low or absent quantities of TREC sequences indicate a possible underlying genetic defect in the newborn’s ability to generate T lymphocytes.

SCID screening assay performance parameters (as observed by Thomson and Glass):

Sensitivity. The sensitivity to correctly identify babies with SCID is estimated at 93.3%. There have been no known false negatives (no known missed cases of SCID).

Specificity. The specificity to correctly identify babies, who do not have SCID or a related immune deficiency disorder, is estimated at 99.983%.

Estimated Costs of Implementing SCID Screening of Newborns in Maryland

The chart below provides information on the estimated startup costs for SCID screening of newborns in Maryland as well as the costs in subsequent years. The costs are significant:

<table>
<thead>
<tr>
<th></th>
<th>Start up cost</th>
<th>Subsequent Years</th>
</tr>
</thead>
<tbody>
<tr>
<td>Supplies</td>
<td>$250,000.00</td>
<td>$210,000.00</td>
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<tr>
<td>Instruments</td>
<td>$318,609.00</td>
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<tr>
<td>Service Contracts</td>
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<td>$16,000.00</td>
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<tr>
<td>Public Health Lab Scientist (2)</td>
<td>$177,111.30</td>
<td>$182,308.00</td>
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<tr>
<td>Yearly Totals</td>
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<tr>
<td>Instruments (5 years)</td>
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<td></td>
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<tr>
<td>Supplies</td>
<td>$210,000.00</td>
<td></td>
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<tr>
<td>Service Contract</td>
<td>$16,000.00</td>
<td></td>
</tr>
<tr>
<td>Personnel</td>
<td>$182,308.00</td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>$472,029.80</td>
<td></td>
</tr>
</tbody>
</table>

| Cost/Baby (Based on 75,000 babies/year) | $6.29 |
Fiscal/Human Impact

Early identification of babies with SCID is crucial to their health. The mortality is greatly reduced with early treatment and medical costs are dramatically lower compared to babies treated after becoming symptomatic\textsuperscript{11}.

With early diagnosis, an affected baby can receive a bone marrow transplant which drastically improves their long term chances for survival and quality of life. Financially, a family with an undiagnosed SCID child will spend over $100,000 annually in healthcare costs as compared to $23,000 spent annually by families of an early diagnosed patient\textsuperscript{12}. Emotionally, families with undiagnosed SCID children have to bear the weight of seeing their child gravely ill and hospitalized. Sadly, most of these families who do not receive the benefit of early SCID diagnosis have to ultimately deal with the death of their child.

According to the Wisconsin State Laboratory of Hygiene (70,000 annual births), of 5 SCID babies diagnosed late, the average hospital bill was $2,200,000 per child. On the other hand, one early diagnosed SCID baby's treatment was $250,000\textsuperscript{13}.

In Florida there are at least two examples of Medicaid SCID babies who were not screened. The first baby died at the age of 5 months and 10 days in the hospital at an approximate cost of $500,000. The second baby became ill and was hospitalized. In six months the hospital bill for Florida Medicaid was $2.2 million\textsuperscript{14}.

Mortality of cases identified early (transplant < 3.5 months of age) is significantly lower than the cases identified late (transplant ≥ 3.5 months of age) 8.6% vs. 37.5\%.\textsuperscript{11}

The annual treatment costs saved by screening is approximately $465,000 per baby (No Screening = $685,000 vs. Screening= $220,000) as shown by Thomson and Glass\textsuperscript{11}.

Implementation

The Newborn Screening Laboratory applied for but was not successful in obtaining a Federal Grant to Support New Implementation of State or Territorial Public Health Laboratory Capacity for Newborn Bloodspot Screening of SCID (RFA-EH-11-001) in 2011. The Newborn Screening Laboratory intends to continue pursuing such opportunities in the future.

In general, the Maryland State Newborn Screening Laboratory will be able to add SCID testing to the current screening panel within 6-8 months after funds become available. However, the anticipated relocation of all Maryland Public Health Laboratories to newly constructed facilities, projected to occur during the summer of 2014, could extend the time necessary to begin SCID screening once funds are secured. As funding of this screening is not feasible in Fiscal Year 2014 due to the relocation, the Department will consider including funding for this screening in the Fiscal Year 2015 budget process.
I hope this information is useful. For your perusal, I have also attached a list of references relative to the text citations noted above. If you have any questions regarding this report, please contact Ms. Marie Grant, Director of the Office of Governmental Affairs, at (410) 767-6481.

Sincerely,

Joshua M. Sharfstein, M.D.
Secretary

Enclosure

cc: Laura Herrera
    Marie Grant
    Robert Myers
    Patrick Dooley
    John Newman
    Erin McMullen
References


6. Newborn Screening for Severe Combined Immunodeficiency Disorder, Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, May 2011


