

## Update on Joint Commission on Health Care Study of Newborn Screening HJR 164 in 2004 (Delegate Plum)

In 2004, the Joint Commission was asked in HJR 164 to collect information regarding the screening of newborns for metabolic disorders.

- At the same time we were completing our study, the American College of Medical Genetics (ACMG) released – *Newborn Screening: Toward a Uniform Screening Panel and System* – which advised that the U.S. Department of Health and Human Resources should recommend screening for approximately 30 disorders. (At that time, the federal recommendation was to screen for only 3 disorders.)
- JCHC voted to introduce HB 2511 and SB 1184 (2005).
  - The bills provisions would expand Virginia’s newborn screening panel from 11 to approximately 30 disorders to be consistent with anticipated new recommendations from the U.S. Department of Health and Human Resources.
- Legislation was enacted (HB 2511 was incorporated into HB 1824 and HB 1824 and SB 1184 were enacted) amending Title 32.1 of the *Code of Virginia* to expand Virginia’s newborn screening panel as of March of 2006.
- The Department of General Services increased the cost of each filter paper kit (used to collect the blood for screening newborns) from \$32 to \$53.
  - Hospitals which purchase the overwhelming majority of filter kits have absorbed this cost increase.
- Infants identified as possibly having a metabolic disorder through the newborn screening process receive follow-up services:
  - Virginia Department of Health newborn screening unit coordinates the care until the infant receives a diagnosis (positive or negative for a disorder) or is 6 months of age.
  - Metabolic treatment centers provide “long-term medical management of all panel diagnosed individuals.”
  - Care Connection for Children “provides long-term care coordination for all diagnosed children/families. Pool of funds access for those at or below 300% of the federal poverty level.”

**Source:** *Newborn Screening Services Expanded Panel Implementation: Changes and Challenges March 2006*, Virginia Department of Health and Division of Consolidated Laboratory Services.

137,359 newborns were screened from March 1, 2006 – May 31, 2007  
32 infants were found to have five of the newly added disorders:

**Cystic Fibrosis (CF)**

**23 cases**

Incidence greater than 1 in 5,000

“Cystic fibrosis is one of the most common inherited disorders in the U.S....result[ing] in lung and digestive problems, and death at an average age of 30-35 years. Studies suggest that early diagnosis and treatment improves the growth of babies and children” with cystic fibrosis.

**Very Long Chain Acy-CoA Dehydrogenase Deficiency (VLCAD)**

**4 cases**

Incidence greater than 1 in 75,000

“Symptoms can first appear at any age from the newborn period through adulthood....Without treatment, affected infants often develop heart and liver failure and die during the first year of life. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, avoidance of fasting and prolonged exercise.”

**3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)**

**2 cases**

Incidence greater than 1 in 75,000

Untreated this disorder “can lead to brain damage, seizures, liver failure and death in infancy or no symptoms at all into adulthood....Treatment with a low-protein diet and in some cases, nutritional supplements may be helpful.”

**Propionic Acidemia (Prop)**

**2 cases**

Incidence greater than 1 in 75,000

“Without treatment, brain damage, coma and death can result. Even with treatment, including a low-protein diet and nutritional supplements, some affected children suffer from developmental delays, seizures, abnormal muscle tone, frequent infections and heart problems.”

**Citrullinemia (CIT)**

**1 case**

Incidence less than 1 in 100,000

“Without treatment, seizures, coma, brain damage and death can result. With early diagnosis and treatment, normal development is possible. Treatment includes a low-protein diet, medications to rid the body of amino groups to prevent ammonia build-up, and nutritional supplements.”

**Source:** Department of General Services’ Consolidated Laboratory Services Data; March of Dimes Quick References and Fact Sheets; and American College of Medical Genetics, *Newborn Screening: Toward a Uniform Screening Panel and System*, Final Report, March 8, 2005.

## **Disorders Included in Virginia's Newborn Screening Panel**

1. Argininosuccinic acidemia (ASA);
2. Beta-Ketothiolase deficiency ( $\beta$ KT);
3. Biotinidase deficiency (BIOT);
4. Carnitine uptake defect (CUD);
5. Citrullinemia (CIT);
6. Congenital adrenal hyperplasia (CAH);
7. Congenital hypothyroidism (CH);
8. Cystic fibrosis (CF);
9. Galactosemia (GALT);
10. Glutaric acidemia type I (GA I);
11. Hemoglobin Sickle/Beta-thalassemia (Hb S/ $\beta$ Th);
12. Hemoglobin Sickle/C disease (Hb S/C);
13. Homocystinuria (HCY);
14. Isovaleric acidemia (IVA);
15. Long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);
16. Maple syrup urine disease (MSUD);
17. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
18. Methylmalonic acidemia (mutase deficiency) (MUT);
19. Methylmalonic acidemia (Cbl A,B);
20. Multiple carboxylase deficiency (MCD);
21. Phenylketonuria (PKU);
22. Propionic acidemia (PROP);
23. Sickle cell anemia (Hb SS disease) (Hb SS);
24. Tyrosinemia type I (TYR I);
25. Trifunctional protein deficiency (TFP);
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD);
27. 3-hydroxy 3-methyl glutaric aciduria (HMG), and
28. 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

(Plus screening for hearing loss.)