Frequently Asked Questions (FAQS)

Secondary Conditions of the Recommended Uniform Screening Panel

What is the Recommended Uniform Screening Panel (RUSP)?

The Primary (Core) Recommended Uniform Screening Panel consists of 31 conditions. This panel is recommended for state newborn screening programs and is endorsed by the U.S. Secretary of Health and Human Services. The DSHS Laboratory currently screens for 29 of these conditions through blood spot testing. Healthcare providers screen for two conditions, hearing loss and critical congenital heart disease, at the point of care. The RUSP identifies an additional twenty-six (26) conditions as secondary conditions.

What is a secondary condition?

Secondary conditions are believed to be clinically significant, but some may have an unclear natural history or lack appropriate medical therapy that affects long-term outcome. They are detected during screening for core conditions. Some secondary conditions may be as severe and life threatening as core conditions.

What is changing in Texas?

Texas law requires screening for core and secondary conditions to the extent funding is available. The DSHS Newborn Screening Laboratory currently screens for the 29 core conditions tested via dried blood spots. In 2015, the DSHS Laboratory will begin reporting results for 24 of the 26 secondary conditions listed in the RUSP.

The DSHS Newborn Screening Laboratory already could detect 18 of these secondary conditions. This expansion will allow DSHS to optimize detection, enhance follow-up, and expand services for these conditions. The laboratory also will be able to identify 6 previously undetectable conditions.

Which two secondary conditions will not be added?

Galactose epimerase deficiency and galactokinase deficiency have been excluded due to an exemption in the Texas law.

Will there be a fee increase?

The initial laboratory and clinical care coordination costs of the expansion fit within the current DSHS budget. No additional fees will be assessed by the DSHS Laboratory at this time. However, the cost of the newborn screen will be evaluated as part of a routine laboratory-wide assessment of fees to be conducted in 2015 in accordance with Senate Bill 80 (82R). At that time, the Newborn Screening fee may be adjusted.

Will more blood be required to be collected?

No additional blood will need to be collected.

What are the conditions that will be added?

Metabolic Disorders - Amino acid disorders

(1) Argininemia (ARG)
(2) Citrullinemia, type II (CIT II)
(3) Hypermethioninemia (MET)
(4) Benign hyperphenylalaninemia (H-PHE)
(5) Biopterin defect in cofactor biosynthesis (BIOPT (BS))
(6) Bioterin defect in cofactor regeneration (BIOPT REG)
(7) Tyrosinemia, type II (TYR II)
(8) Tyrosinemia, type III (TYR III)

Metabolic Disorders - Fatty acid disorders

(9) Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
(10) Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency (M/SCHAD)
(11) Glutaric acidemia type II (GA2)
(12) Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
(13) 2,4 Dienoyl-CoA reductase deficiency (DE RED)
(14) Carnitine palmitoyltransferase type I deficiency (CPT IA)
(15) Carnitine palmitoyltransferase type II deficiency (CPT II)
(16) Carnitine acylcarnitine translocase deficiency (CACT)

Metabolic Disorders - Organic acid disorders

(17) Methylmalonic acidemia with homocystinuria (Cbl C, D)
(18) Malonic acidemia (MAL)
(19) Isobutyrylglycinuria (IBG)
(20) 2-Methylbutyrylglycinuria (2MBG)
(21) 3-Methylglutaconic aciduria (3MGA)
(22) 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

Hemoglobin Disorder

(23) Various other hemoglobinopathies (Var Hb)

Other Disorder

(24) T-cell related lymphocyte deficiencies

How common are these conditions?

Prevalence rates in the United States for some of the conditions are unknown. However, these conditions are generally estimated to be very rare. As an example, Tyrosinemia type II is estimated to occur in ~ 1 in 250,000 births. Other conditions have had only a few known cases worldwide.

Are there medical treatments for these conditions?

Most of the conditions are treatable. The medical community continues to work on improving treatments to make them more effective.

Which specialists will be involved in the follow-up of secondary conditions?

Metabolic specialists for the metabolic conditions, hematologists for the hemoglobin conditions and immunologists for the T-cell related conditions.

Where can I find more information on secondary conditions?

Discretionary Advisory Committee on Heritable Disorders in Newborns and Children
Baby's First Test