

## APPENDIX 1: Newborn screening fact sheet validation

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
<b>Endocrine Disorders</b>					
Congenital adrenal hyperplasia	Maria I. New, MD Cornell University New York, NY	1	1	1	1
	Phyllis Speiser, MD New York Univ. Med Center Schneider Children's Hospital Long Island Jewish Health System New York, NY	3	3	3	1
Congenital hypothyroidism	Phyllis Speiser, MD New York Univ. Med Center Schneider Children's Hospital Long Island Jewish Health System New York, NY	1	1	1	1
	Marvin Mitchell, MD New England Newborn Screening Program University of Massachusetts Medical School Jamaica Plain, MA	1	1	1	1
Type 1 diabetes mellitus (IDDM)	Marian Rewers, MD University of Colorado School of Medicine Denver, CO		1	1	1
	William Tamborlane, MD Yale University New Haven, CT	1	2	2	1
	Charles Stanley, MD Children's Hospital of Philadelphia Philadelphia, PA	1	2	2	2
<b>Carbohydrate Disorders</b>					
Classic galactosemia (GALT deficiency)	Louis B. Elsas, MD University of Miami Miami, FL	4	4	4	4
	Gerard Berry, MD Jefferson Medical College Philadelphia, PA	3	2	1	3
Galactokinase deficiency	Louis B. Elsas, MD University of Miami Miami, FL	4	4	4	4
	Gerard Berry, MD Jefferson Medical College Philadelphia, PA	4	2	2	4
Galactose epimerase deficiency	Louis B. Elsas, MD University of Miami Miami, FL	4	4	4	4
	Gerard Berry, MD Jefferson Medical College Philadelphia, PA	4	2	2	4
Congenital disorder of glycosylation type 1b	Marc Patterson, MD, FRACP Columbia University New York, NY	4	4	4	4
	Donna Krasnewich, MD, PhD National Human Genome Research Institute Bethesda, MD	1	4	1	2
<b>Primary Immunodeficiencies</b>					
Adenosine deaminase Deficiency	Rebecca Buckley, MD Duke University Medical Center Durham, NC	2	N/A	1	1
	Jennifer Puck, MD National Human Genome Research Institute Bethesda, MD	2	N/A	2	2

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Severe combined Immunodeficiency	Rebecca Buckley, MD Duke University Medical Center Durham, NC	1	N/A	1	1
	Jennifer Puck, MD National Human Genome Research Institute Bethesda, MD	1	N/A	1	1
<b>Other Genetic and Non-Genetic Conditions</b>					
$\alpha$ -1-antitrypsin deficiency	Diane Cox, PhD University of Alberta Edmonton, Alberta, Canada	1	1		
Biliary atresia	Deborah K. Freese, MD Mayo Clinic College of Medicine Rochester, MN	2	3	2	3
	Ronald J. Sokol, MD University of Colorado School of Medicine Denver, CO	2	3	3	3
Biotinidase deficiency	Barry Wolf, MD, PhD Connecticut Children's Medical Center Hartford, CT	2	2	2	2
	E. Regula Baumgartner, MD University Children's Hospital Basel, Switzerland	2	1	1	2
	Matthias Baumgartner, MD University Children's Hospital Zurich, Switzerland	2	1	1	2
Cystic fibrosis	Phillip Farrell, MD, PhD University of Wisconsin Madison, WI	1	1	2	3
	Garry R. Cutting, MD Johns Hopkins University School of Medicine Baltimore, MD	1	3		2
Duchenne (DMD)/Becker muscular dystrophy (BMD)	Jon A. Wolff, MD University of Wisconsin Madison, WI	2	2	2	2
	R. Rodney Howell, MD University of Miami Miami, FL	1	2	2	1
Familial hypercholesterolemia (heterozygote)	Joseph P. McConnell, PhD Mayo Clinic College of Medicine Rochester, MN	2	2	1	2
	David Wilcken, MD Prince of Wales Hospital Randwick, NSW, Australia	1	1	1	1
Fragile X syndrome	Stephen Warren, PhD Emory University Atlanta, GA	1	N/A	1	1
	W. Ted Brown, MD, PhD New York State Institute for Basic Research Staten Island, NY	2	2	2	3
Hearing Loss	Cynthia C. Morton, PhD Brigham and Women's Hospital Harvard Medical School Boston, MA	1	1	2	2
	Richard Smith, MD University of Iowa Medical School Iowa City, IA	1	1	1	1

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Hyperbilirubinemia (kernicterus)	Jeffery Maisels, MD William Beaumont Hospital Royal Oak, MI	3	3	3	3
	Vinod Bhutani, MD Children's Hospital of Philadelphia Philadelphia, PA	3	3	3	3
Neuroblastoma	Garrett Brodeur, MD Children's Hospital of Philadelphia Philadelphia, PA	1	1	1	1
	Eizo Hiyama, MD Hiroshima University Hiroshima, Japan and Hiroshi Naruse, MD Quality Control Center for Mass Screening Tokyo, Japan	2	3	2	3
Smith-Lemli–Opitz syndrome	Robert Steiner, MD Oregon Health Science University Portland, OR	1	2	2	2
	Mira Irons, MD Children's Hospital Harvard Medical School Boston, MA	1	1	1	3
	Richard I. Kelley, MD, PhD Johns Hopkins Medical Institution Baltimore, MD	4	2	2	1
Turner syndrome	Virginia P. Sybert, MD Univ. of Washington Seattle, WA	3/4	3/4	3/4	3/4
	Ron G Rosenfeld, MD Lucille Packard Foundation for Children Palo Alto, CA	1	3	3	2
Wilson disease	Benjamin Shneider, MD New York University Medical School New York, NY	3	3	2	2
	Sihoun Haun, MD, PhD Mayo Clinic College of Medicine Rochester, MN	1	2	2	1
X-Linked Adrenoleukodystrophy	Hugo Moser, MD Kennedy Krieger Institute Johns Hopkins University Baltimore, MD	2	2	2	2-3
	Robert Steiner, MD Oregon Health Science University Portland, OR	2	2	2	3
<b>Amino Acid Disorders</b>					
Argininemia	Stephen D. Cederbaum, MD Mental Retardation Research Center, UCLA Los Angeles, CA	3	3	3	3
	Mendel Tuchman, MD Children's National Medical Center Washington, DC	4	4	4	4
Argininosuccinic acidemia	Stephen D. Cederbaum, MD Mental Retardation Research Center, UCLA Los Angeles, CA	1	3	1	3
	Mendel Tuchman, MD Children's National Medical Center Washington, DC	3	3	3	3

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Defects of bipterin cofactor biosynthesis	Nenad Blau, PhD University Children's Hospital University of Zurich Zurich, Switzerland	2	2	2	3
	Harvey Levy, MD Harvard Medical School Boston, MA	2	2	2	2
Defects of bipterin cofactor regeneration	Nenad Blau, PhD University Children's Hospital University of Zurich Zurich, Switzerland	2	2	2	3
	Harvey Levy, MD Harvard Medical School Boston, MA	3	2	2	4
Carbamylphosphate synthetase deficiency	Mendel Tuchman, MD Children's National Medical Center Washington, DC	3	3	3	3
	Mark L. Batshaw, MD Children's National Medical Center George Washington University Washington, DC	3	3	3	3
Citrullinemia(arginosuccinate synthase deficiency)	Mendel Tuchman, MD Children's National Medical Center Washington, DC	3	3	3	3
	Mark L. Batshaw, MD Children's National Medical Center George Washington University Washington, DC	3	3	3	3
Citrullinemia type II (citrin deficiency)	Mendel Tuchman, MD Children's National Medical Center Washington, DC	3	3		3
	Toshihiro Ohura, MD Tohoku University School of Medicine Sendai, Japan	3	2	2	3
	Mark L. Batshaw, MD Children's National Medical Center George Washington University Washington, DC	3	3	3	3
Homocystinuria(cystathionine $\beta$ -synthase deficiency)	S. Harvey Mudd, MD NIH/NIMH Bethesda, MD	1	1	1	4
	Vivian Shih, MD Harvard Medical School Boston, MA	1		3	3
Hypermethioninemia(MAT 1/III deficiency)	S. Harvey Mudd, MD NIH/NIMH Bethesda, MD	1	1	1	4
	Vivian Shih, MD Harvard Medical School Boston, MA	1	1	1	4
Maple syrup (urine) disease	Louis B. Elsas, MD University of Miami Miami, FL	3	3	1	3
	Vivian Shih, MD Harvard Medical School Boston, MA	1	1	1	4

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Ornithine transcarbamylase deficiency	Mendel Tuchman, MD Children's National Medical Center Washington, DC	3	3	3	3
	Mark L. Batshaw, MD Children's National Medical Center George Washington University Washington, DC	3	3	3	3
Phenylketonuria (phenylalanine hydroxylase deficiency)	Nenad Blau, PhD University Children's Hospital University of Zurich Zurich, Switzerland	2	2	2	2
	Harvey Levy, MD Harvard Medical School Boston, MA	2	2	2	2
	Vivian Shih, MD Harvard Medical School Boston, MA	1	1	2	4
Tyrosinemia type I (hepatorenal tyrosinemia)	C. Ronald Scott, MD University of Washington Seattle, WA	2	3	1	2
	Grant Mitchell, MD Hospital Sainte-Justine Montreal, Quebec, Canada	2	2/3	1	2
Tyrosinemia type II (oculocutaneous tyrosinemia)	C. Ronald Scott, MD University of Washington Seattle, WA	2	3	2	2
	Grant Mitchell, MD Hospital Sainte-Justine Montreal, Quebec, Canada	2	4	2	2
Tyrosinemia type III	C. Ronald Scott, MD University of Washington Seattle, WA	3	3	3	4
	Grant Mitchell, MD Hospital Sainte-Justine Montreal, Quebec, Canada	4	4 (sensitivity) 1 (technical)	4	4
<b>Fatty Acid Oxidation Defects</b>					
Carnitine: acylcarnitine translocase deficiency	Nicola Longo, MD, PhD University of Utah Salt Lake City, UT	2	2	1	2
	Charles Stanley, MD Children's Hospital of Philadelphia Philadelphia, PA	3	3	2	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	3	3	2	4
Carnitine palmitoyltransferase I deficiency (CPT1a)	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	3	4	3	4
	Cary Harding, MD Oregon Health Sciences University Portland, OR				
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	4	4	4	4

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Carnitine palmitoyltransferase II deficiency	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	2	4	4	3
	Georgirene D. Vladutiu, PhD Children's Hospital Buffalo, NY	4	2	4	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	2	3	2	4
Carnitine uptake deficiency(Systemic)	Nicola Longo, MD, PhD University of Utah Salt Lake City, UT	1	1	1	1
	Charles Stanley, MD Children's Hospital of Philadelphia Philadelphia, PA	4	3	3	4
Dienoyl-CoA reductase deficiency	Gerard Vockley, MD, PhD Children's Hospital Pittsburgh University of Pittsburgh Pittsburgh, PA	4	4	4	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	4	4	4	4
Glutaric acidemia type II	Stephen I. Goodman, MD University of Colorado Health Science Center Denver, CO	4	4	2	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	3	3	3	4
	William J. Rhead, MD, PhD Medical College of Wisconsin Madison, WI	2	2	2	4
Long-chain 3-OH acyl-CoA dehydrogenase deficiency	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	3	3	3	3
	Arnold Strauss, MD Vanderbilt University School of Medicine Nashville, TN	2	3	3	2
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	3	2	2	3
Medium-chain acyl-CoA dehydrogenase deficiency	Arnold Strauss, MD Vanderbilt University School of Medicine Nashville, TN	2	2	2	2
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	2	1	1	1
Medium/short-chain 3-OH acyl-CoA dehydrogenase deficiency	Arnold Strauss, MD Vanderbilt University School of Medicine Nashville, TN	4	4	4	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	4	4	4	4
Medium-chain ketoacyl-CoA thiolase deficiency	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	4	4	4	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	4	4	4	4

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Short-chain acyl-CoA dehydrogenase deficiency	Gerard Vockley, MD, PhD Children's Hospital Pittsburgh University of Pittsburgh Pittsburgh, PA	2	1	1	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	4	3	2	4
	Dietrich Matern, MD Mayo Clinic College of Medicine Rochester, MN	2	1	1	2
Trifunctional protein deficiency	Arnold Strauss, MD Vanderbilt University School of Medicine Nashville, TN	3	3	3	3
	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	4	4	4	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	3	2	2	3
Very long-chain acyl-CoA dehydrogenase deficiency	Arnold Strauss, MD Vanderbilt University School of Medicine Nashville, TN	2	2	2	2
	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	3	3	3	4
	Piero Rinaldo, MD, PhD Mayo Clinic College of Medicine Rochester MN	3	2	2	3
<b>Organic Acidurias</b>					
2-methylbutyryl-CoA dehydrogenase deficiency	Gerard Vockley, MD, PhD Children's Hospital Pittsburgh University of Pittsburgh Pittsburgh, PA	1	1	1	4
	Dietrich Matern, MD Mayo Clinic College of Medicine Rochester, MN	2	1	1	2
2-methyl 3-hydroxybutyric-aciduria	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	4	4	4	4
	Dietrich Matern, MD Mayo Clinic College of Medicine Rochester, MN	3	4	3	3
	Regina Ensenuer, MD Von Haunersches Kinderspital Ludwig-Maximilians-University Munich, Germany	4	4	4	4
3-hydroxy 3-methyl glutaric aciduria (HMG)	Pinar Ozand, MD, PhD King Faisal Specialist Hospital and Research Centre Riyadh, Saudi Arabia	4	1	1	1
	Grant Mitchell, MD Hospital Sainte-Justine Montreal, Quebec, Canada	2	4	2	3
3-Methylglutaconic Aciduria (Type 1-hydrotase deficiency)	Robert Steiner, MD Oregon Health University Portland, OR	2	2	2	2
	Richard I. Kelley, MD, PhD Johns Hopkins Medical Institution Baltimore, MD	4	2	2	4

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
3-methylcrotonyl-CoA carboxylase deficiency	Matthias Baumgartner, MD University Children's Hospital Zurich, Switzerland	2	1	2	4
	Richard I. Kelley, MD, PhD Johns Hopkins Medical Institution Baltimore, MD	4	2	2	4
β-ketothiolase deficiency	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	4	4	4	4
	Toshiyuki Fukao, MD Gifu University School of Medicine Gifu, Japan	3	3	3	3
Glutaric acidemia type 1	Stephen I. Goodman, MD University of Colorado Health Science Center Denver, CO	2	2	2	3
	Pinar Ozand, MD, PhD King Faisal Specialist Hospital and Research Centre Riyadh, Saudi Arabia	2	2	2	3
Isobutyryl-CoA dehydrogenase Deficiency	Gerard Vockley, MD, PhD Children's Hospital Pittsburgh University of Pittsburgh Pittsburgh, PA	3	1	1	4
	Dietrich Matern, MD Mayo Clinic College of Medicine Rochester, MN	2	2	1	3
Isovaleric acidemia	Gerard Vockley, MD, PhD Children's Hospital Pittsburgh University of Pittsburgh Pittsburgh, PA	1	1	1	3
	Dietrich Matern, MD Mayo Clinic College of Medicine Rochester, MN	1	1	1	1
	Regina Ensenuer, MD Von Haunersches Kinderspital Ludwig-Maximilians-University Munich, Germany	1	1	1	3
Malonic acidemia	Michael Bennett, PhD Children's Hospital of Philadelphia Philadelphia, PA	4	4	4	4
	Pinar Ozand, MD, PhD King Faisal Specialist Hospital and Research Centre Riyadh, Saudi Arabia	4	4	4	4
Methylmalonic acidemia (CblA,B)	David Rosenblatt, MD McGill University Montreal, Quebec, CA	4	4	4	4
	William Nyhan, MD, PhD University of California, San Diego La Jolla, CA	2	1	1	2
Methylmalonic acidemia (Cbl C,D)	David Rosenblatt, MD McGill University Montreal, Quebec, CA	4	4	4	4
	William Nyhan, MD, PhD University of California, San Diego La Jolla, CA	2	1	1	2

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
Methylmalonic acidemia (MUTase deficiency)	David Rosenblatt, MD McGill University Montreal, Quebec, CA	4	4	4	
	William Nyhan, MD, PhD University of California, San Diego La Jolla, CA	2	1	1	2
Holocarboxylase synthetase deficiency	Barry Wolf, MD, PhD Connecticut Children's Medical Center Hartford, CT	3	3	3	3
	E. Regula Baumgartner, MD University Children's Hospital Basel, Switzerland	2	2	2	2
	Matthias Baumgartner, MD University Children's Hospital Zurich, Switzerland	2	2	2	2
Propionyl-CoA carboxylase deficiency	Pinar Ozand, MD, PhD King Faisal Specialist Hospital and Research Centre Riyadh, Saudi Arabia	3	1	1	1
	William Nyhan, MD, PhD University of California, San Diego La Jolla, CA	2	1	1	2
<b>Hematology/Hemoglobinopathies</b>					
Sickle cell anemia (Hb SS disease)	Carolyn Hoppe, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
	Elliott Vichinsky, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
Hemoglobin SC	Carolyn Hoppe, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
	Elliott Vichinsky, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
Hemoglobin S/beta-thalassemia (Hb S $\beta$ -thal)	Carolyn Hoppe, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
	Elliott Vichinsky, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
Variant hemoglobinopathies (including HbE)	Carolyn Hoppe, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
	Elliott Vichinsky, MD Children's Hospital Oakland Oakland, CA	1	2	1	1
Glucose-6-phosphate dehydrogenase deficiency (G6PD)	Ernest Beutler, MD Scripps Research Institute La Jolla, CA	3	1	2	4
	Carolyn Hoppe, MD Children's Hospital Oakland Oakland, CA	2	2	1	4

CONDITION	VALIDATED BY	EVIDENCE LEVELS (1-4)			
		Condition	Test	Diagnosis	Treatment
<b>Creatine Metabolism Disorders</b>					
Guanidinoacetate methyltransferase deficiency (GAMT)	William O'Brien, PhD Baylor College of Medicine Dallas, TX	4	4	4	4
	Robert Steiner, MD Oregon Health Science University Portland, OR	4	4	4	4
Arginine:glycine amidinotransferase deficiency (AGAT)	William O'Brien, PhD Baylor College of Medicine Dallas, TX	4	4	4	4
	Robert Steiner, MD Oregon Health Science University Portland, OR	4	4	4	4
Creatine transporter defect	William O'Brien, PhD Baylor College of Medicine Dallas, TX	4	4	4	4
	Robert Steiner, MD Oregon Health Science University Portland, OR	4	4	4	4
<b>Lysosomal Storage Disorders</b>					
Fabry disease	Gregory A. Grabowski, MD Cincinnati Children's Hospital Medical Center Cincinnati, OH	2	3	3	1
	Robert J. Desnick, MD, PhD Mount Sinai Medical Center New York, NY	2	3	4	1
Krabbe disease	Gregory A. Grabowski, MD Cincinnati Children's Hospital Medical Center Cincinnati, OH	3	3	3	4
Hurler, Scheie, Hurler-Scheie (MPS I)	Gregory A. Grabowski, MD Cincinnati Children's Hospital Medical Center Cincinnati, OH	3	3	4	2
Pompe disease (glycogen storage disease type II)	Gregory A. Grabowski, MD Cincinnati Children's Hospital Medical Center Cincinnati, OH	4	3	3	3/4
	R. Rodney Howell, MD University of Miami Miami, FL	1	4	1	4