How does utilizing the new counting conditions protocol show the quantitative differences in newborn screening conditions in New England states?

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Definitions

- Newborn screening (NBS) - public health initiative that screens children for treatable congenital conditions (CDC)
- Recommended Uniform Screening Panel (RUSP) - A list of disorders that each state is recommended to screen for (HRSA, 2024)
- Phenotype - an observable trait

Definitions Cont.

- RUSP Core Condition- Laboratory is specifically designed to assess whether a baby has the condition (HRSA, 2025)
- RUSP Secondary Condition- A condition that may be found while screening for a core condition (HRSA, 2025)

Introduction

- States decide:
  - which conditions they will screen for
  - how they will count the conditions they screen for
- Association of Public Health Laboratories (APHL) recognizes the need for harmonization
- Counting differently leads to confusion, inaccurate comparison, and misperceptions of states

Introduction

- Multidisciplinary taskforce offers new uniform counting method:
  - Phenotype- all phenotypes of a condition are counted as one condition
  - Intent- program intends to detect all affected individuals

Methods

- Locate each state NBS panel and record conditions
- Implement new protocol to each state's list
- Compare and visualize differences across states and across protocols

Results

State	Current State Website	New Counting Protocol
CT	79	38
MA	56	28
RI	33	32
VT	33	32
NH	37	37
ME	55	30

Results

CT	MA	RI	VT	NH	ME
Congenital Adrenal Hyperplasia caused by 21-OH	Argininosuccinic Aciduria	Sickle Cell Disease	3-Methylcrotonyl-CoA Carboxylase Deficiency (3 M	Argininemia	3-Hydroxy-3-Methylglutaric Aciduria (HMG)
Primary Congenital Hypothyroidism	β-Ketothiolase deficiency (BKT)	Beta Thalassemia	3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Argininosuccinic Acidemia (ASA)	3-Methylcrotonyl-CoA Carboxylase Deficiency (3M
Argininosuccinic Aciduria	Biotinidase deficiency (BIOT)	Phenylalaine hydroxylase deficiency	Argininosuccinic Aciduria	Biotinidase Deficiency (BIOT)	Argininosuccinic Aciduria (ASA)
Argininiosuccinate synthetase deficiency (Citrullir	Carnitine Uptake Deficiency (CUD)	Maple syrup urine disease	Beta-Ketothiolase Deficiency (BKT)	Carnitine Uptake Deficiency (CUD)	B-Ketothiolase Deficiency (BKT)
Phenylalaine hydroxylase deficiency	Argininiosuccinate synthetase deficiency	Homocystinuria caused by CBS deficiency	Biotinidase deficiency	Argininiosuccinate synthetase deficiency	Biotinidase Deficiency (BIOT)
Homocystinuria caused by CBS deficiency	Congenital Adrenal Hyperplasia caused by 21-OH	Argininiosuccinate synthetase deficiency	Carnitine Uptake Deficiency (CUD)	Methylmalonic Acidemia caused by cobalamin A	Carnitine Uptake Deficiency (CUD)
Maple Syrup Urine Disease	Primary Congenital Hypothyroidism	Argininosuccinic Aciduria	Argininiosuccinate synthetase deficiency	Congenital Adrenal Hyperplasia caused by 21-OH	Argininiosuccinate synthetase deficiency (citrullin
Tyrosinemia Type I	Congenital Toxoplasmosis (TOXO)	Tyrosinemia Type I	Congenital Adrenal Hyperplasia caused by 21-OH	Primary Congenital Hypothyroidism	Galactosemia due to GALT deficiency
Ornithine Transcarbamylase Deficiency (OTC)	Cystic Fibrosis	Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase	Primary Congenital Hypothyroidism	Congenital Toxoplasmosis (TOXO)	Phenylalaine hydroxylase deficiency
Carnitine Uptake Deficiency (CUD)	Galactosemia due to GALT deficiency	Medium-Chain Acyl-CoA Dehydrogenase Deficien	Cystic Fibrosis	Cystic Fibrosis (CF)	Congenital Adrenal Hyperplasia caused by 21-OH
Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase	Glutaric Acidemia Type I (GA 1)	Very Long-Chain Acyl-CoA Dehydrogenase Deficie	Galactosemia due to GALT deficiency	Galactosemia due to GALT deficiency	Primary Congenital Hypothyroidism
Medium-Chain Acyl-CoA Dehydrogenase Deficien	Homocystinuria caused by CBS deficiency	Carnitine Uptake Deficiency	Glutaric Acidemia Type I (GA 1)	Glutaric Acidemia Type I (GA 1)	Cystic Fibrosis
Very Long-Chain Acyl-CoA Dehydrogenase Deficie	3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Isovaleric Acidemia (IVA)	Beta Thalassemia	Other clinically significant variant hemoglobins	Glutaric Acidemia Type I (GA 1)
3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Isovaleric Acidemia (IVA)	Glutaric Acidemia Type I	Sickle Cell Disease	3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Holocarboxylase synthetase deficiency (multi carl
3-Methylcrotonyl-CoA Carboxylase Deficiency (3 M	Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase	3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Holocarboxylase synthetase deficiency (multi carl	Hyperornithinemia Hyperammoninemia	Homocystinuria caused by CBS deficiency
Beta-Ketothiolase Deficiency (BKT)	Maple Syrup Urine Disease	Holocarboxylase synthetase deficiency	Homocystinuria caused by CBS deficiency	Homocitrullinemia Syndrome (HHH)	Isovaleric Acidemia (IVA)
Glutaric Acidemia Type I (GA 1)	Ornithine Transcarbamylase Deficiency (OTC)	methyl malonic acidemia caused by Methylmalon	Isovaleric Acidemia (IVA)	Homocystinuria caused by CBS deficiency	Maple Syrup Urine Disease
Holocarboxylase synthetase deficiency	Phenylalaine hydroxylase deficiency	Methylmalonic Acidemia caused by cobalamin A	Long-chain L-3-OH acyl-CoA dehydrogenase defic	Isovaleric Acidemia (IVA)	methyl malonic acidemia caused by Methylmalon
Isovaleric Acidemia (IVA)	Sickle Cell Disease	3-Methylcrotonyl-CoA Carboxylase Deficiency (3 M	Maple Syrup Urine Disease	Long-chain L-3-OH Acyl-CoA Dehydrogenase Defic	Methylmalonic Acidemia caused by cobalamin A
Methylmalonic Acidemia caused by cobalamin A	Beta thalassemia	Propionic Acidemia	Medium-chain acyl-CoA dehydrogenase deficienc	Maple Syrup Urine Disease (MSUD)	Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase
Sickle Cell Disease	Medium-Chain Acyl-CoA Dehydrogenase Deficien	Beta-Ketothiolase Deficiency	Methylmalonic Acidemia caused by cobalamin A	Medium-chain Acyl-CoA Dehydrogenase Deficienc	Medium-Chain Acyl-CoA Dehydrogenase Deficien
Other clinically significant variant hemoglobins	Very Long-Chain Acyl-CoA Dehydrogenase Deficie	Primary Congenital Hypothyroidism	methyl malonic acidemia caused by Methylmalon	3-Methylcrotonyl-CoA Carboxylase Deficiency(3M	Very Long-Chain Acyl-CoA Dehydrogenase Deficie
Alpha Thalassemia	methyl malonic acidemia caused by Methylmalon	Congenital Adrenal Hyperplasia caused by 21-OH	Mucopolysaccharidosis Type-I (MPS I)	methyl malonic acidemia caused by Methylmalon	Propionic Acidemia (PPA)
Beta Thalassemia	Methylmalonic Acidemia caused by cobalamin A	Cystic Fibrosis	Phenylalaine hydroxylase deficiency	Beta-Ketothiolase Deficiency (BKT)	Beta Thalassemia
methyl malonic acidemia caused by Methylmalon	Propionic Acidemia (PPA)	Pompe Disease	Pompe Disease (POMPE)	Mucopolysaccharidosis Type 1 (MPS1)	Severe Combined Immunodeficiency (SCID)
Propionic Acidemia (PPA)	Severe Combined Immunodeficiency (SCID)	Mucopolysaccharidosis Type-I	Propionic Acidemia (PPA)	Multiple Acyl-CoA Dehydrogenase Deficiency (GA	Sickle Cell Disease
Biotinidase deficiency	Tyrosinemia Type I	Spinal Muscular Atrophy	Severe Combined Immunodeficiency (SCID)	Holocarboxylase synthetase deficiency (multi carl	Tyrosinemia Type I
Galactosemia due to GALT deficiency	Other clinically significant variant hemoglobins	Biotinidase deficiency	Spinal muscular atrophy (SMA)	Pompe Disease (POMPE)	X-linked Adrenoleukpdystrophy (X-ALD)
Congenital cytomegalovirus		Galactosemia due to GALT deficiency	Tyrosinemia type I (TYR I)	Propionic Acidemia (PPA)	Spinal Muscular Atrophy due to deletion of EXON 7 in SMN1 (SMA)
Guanidinoacetate Methyltransferase Deficiency (		Severe Combined Immunodeficiency (SCID)	Very long-chain acyl-CoA dehydrogenase deficien	Phenylalanine hydroxylase deficiency	Other clinically significant variant hemoglobins
Infantile Krabbe Disease		X-Linked Adrenoleukodystrophy	X-linked adrenoleukodystrophy (X-ALD)	Severe Combined Immunodeficiency (SCID)	
Mucopolysaccharidosis Type-I (MPS I)		Other clinically significant variant hemoglobins	Other clinically significant variant hemoglobins	Spinal Muscular Atrophy due to deletion of EXON	
Mucopolysaccharidosis Type-II (MPS II)				Tyrosinemia Type 1 (TYR 1)	
Pompe Disease (POMPE)				Very Long-chain Acyl-CoA Dehydrogenase Deficie	
Severe Combined Immunodeficiency (SCID)				X-Linked Adrenoleukodystrophy (X-ALD)	
Spinal Muscular Atrophy due to deletion of EXON				Beta Thalassemia	
X-Linked Adrenoleukodystrophy (ALD)				Sickle Cell Disease	
Cystic Fibrosis					
Total= 38	Total=28	Total = 32	Total = 32	Total = 37	Total = 30

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Discussion

- Uniform counting can correct misperceptions of states
- Allows the focus to shift to quantitative differences across states
- Easier for public to understand and be well informed about conditions states are screening for

Limitations

- Limitations:
  - Intent and publicly available information
  - Lab needs to consider what their intent is and how to distinguish is

Future Directions

- Future Directions:
  - How to explain number change to public seamlessly
  - Advisory Committee on Heritable Disorders in Newborns and Children eliminated
    - Was our road to implementation, further advocacy and education needed

References

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