Using NewSTEPs Data to Assess Disparities in Newborn Screening

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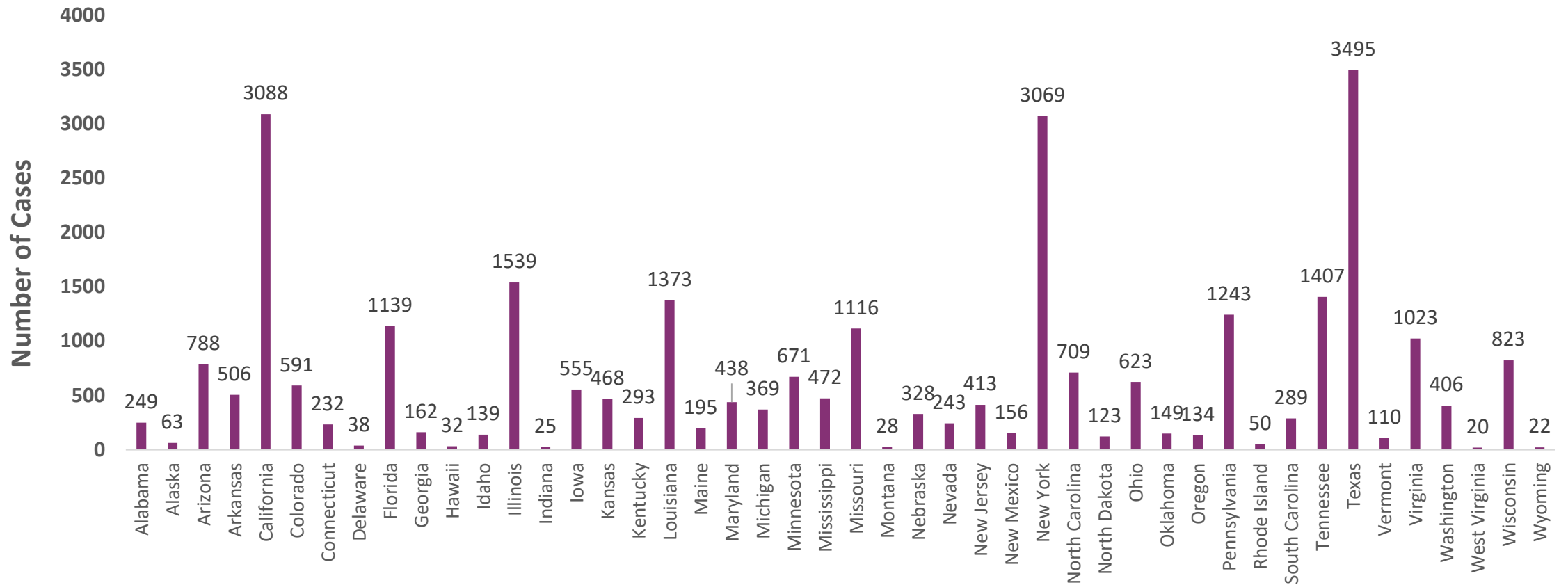
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Data Set Description
(Cases in NewSTEPs from ~2011 to mid-2021)

DRIED BLOOD SPOT DISORDERS ONLY

Total Cases Per State (N=29,404)

Note: case entry for states varies across years as a result of time of signed MOU, case entry mechanisms.

TIME CRITICAL CONDITIONS (N = 3,904)	TIME SENSITIVE CONDITIONS (N = 25,500)	
3-Hydroxy-3-methylglutaric aciduria	3-methylcrotonyl-CoA carboxylase deficiency	Spinal Muscular Atrophy
Argininosuccinic aciduria	Biotinidase deficiency	Tyrosinemia Type 1
β-ketothiolase deficiency	Carnitine uptake/transport defect	X-linked ALD
Citrullinemia type I	Congenital Hypothyroidism	(All secondary conditions)
Classic Galactosemia	Critical Congenital Heart Disease	
Congenital Adrenal Hyperplasia	Cystic Fibrosis	
Glutaric aciduria type I	Hearing Loss	
Holocarboxylase synthase deficiency	Homocystinuria	
Isovaleric academia	Methylmalonic acidemia (cobalamin disorders)	
Long chain L-3-hydroxyacyl-CoA dehydrogenase deficiency	MPS I	
Maple Syrup Urine Disease	Phenylketonuria	
Medium chain acyl-CoA dehydrogenase deficiency	Pompe disease	
Methylmalonic acidemia (methylmalonyl-CoA-mutase)	S,S disease (Sickle Cell Anemia)	
Propionic academia	S, beta-thalassemia	
Trifunctional protein deficiency	S, C disease	
Very Long chain acyl-CoA dehydrogenase deficiency	Severe Combined Immunodeficiency	

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Race/Ethnicity (N=29,404)

Category	Count	Percentage
Asian	1279	4.3%
Black or African American	6685	22.7%
Hawaiian/Pacific Islander	43	0.15%
Native American	154	0.52%
White	11037	37.5%
Mixed	352	1.2%

Category	Count	Percentage
Hispanic/Latino/Spanish	4158	14.14%
Not Hispanic/Latino/Spanish	13629	46%
Not Reported	11617	39.5%

- 39 Different Race Groupings Reported
- 9,854 Cases With No Race Reported (33.5% of cases have no race/ethnicity provided)
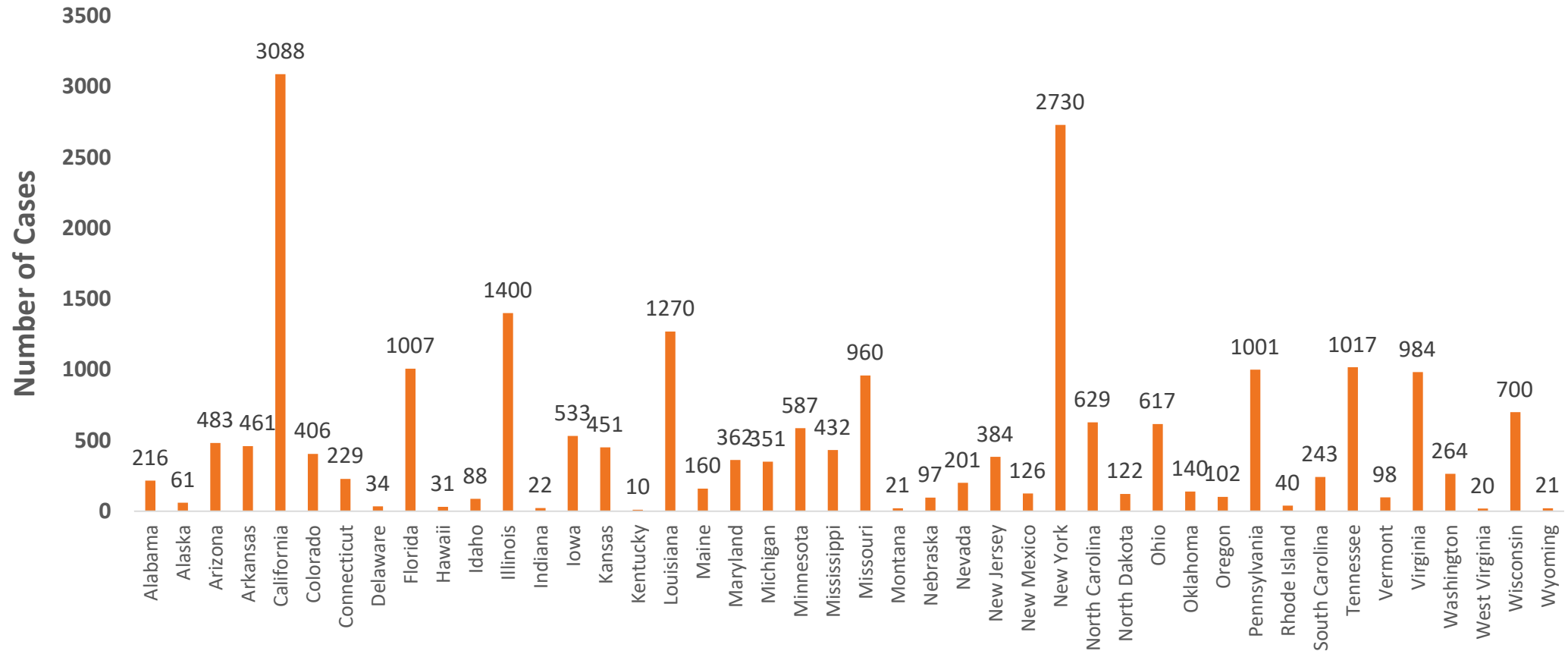
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Timeliness Data
(N=22,199)

PERFORMED ON THOSE CASES REPORTED TO BE DETECTED BY THE INITIAL SPECIMEN ONLY

Initial Specimen Cases Per State (N=22,199)

Number of Cases

State	Cases
Alabama	216
Alaska	61
Arizona	483
Arkansas	461
California	3088
Colorado	406
Connecticut	229
Delaware	34
Florida	1007
Hawaii	31
Idaho	88
Illinois	1400
Indiana	22
Iowa	533
Kansas	451
Kentucky	10
Louisiana	1270
Maine	160
Maryland	362
Michigan	351
Minnesota	587
Mississippi	432
Missouri	960
Montana	21
Nebraska	97
Nevada	201
New Jersey	384
New Mexico	126
New York	2730
North Carolina	629
North Dakota	122
Ohio	617
Oklahoma	140
Oregon	102
Pennsylvania	1001
Rhode Island	40
South Carolina	243
Tennessee	1017
Vermont	98
Virginia	984
Washington	264
West Virginia	20
Wisconsin	700
Wyoming	21

Note: case entry for states varies across years

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Missing Timeliness Data

Category	Number of Cases with No Data	Percentage of Cases with No Data	Number of Outliers*
Birth to Specimen Collection	243	1.1%	202
Birth to Receipt	386	1.8%	187
Birth to Report	597	2.7%	15
Birth to Diagnosis	1897	8.5%	45
Birth to Intervention	3429	15.4%	49

*Outliers defined as follows:
- Birth to Specimen Collection: values of 0 or z-score 2.5 (values above 525 hours)
- Birth to Receipt: values of 0 or >14 days
- Birth to Report: values of 0 or z-score 2.5 (values above 373 days)
- Birth to Diagnosis Confirmation: values of 0 or z-score 2.5 (values above 2000 days)
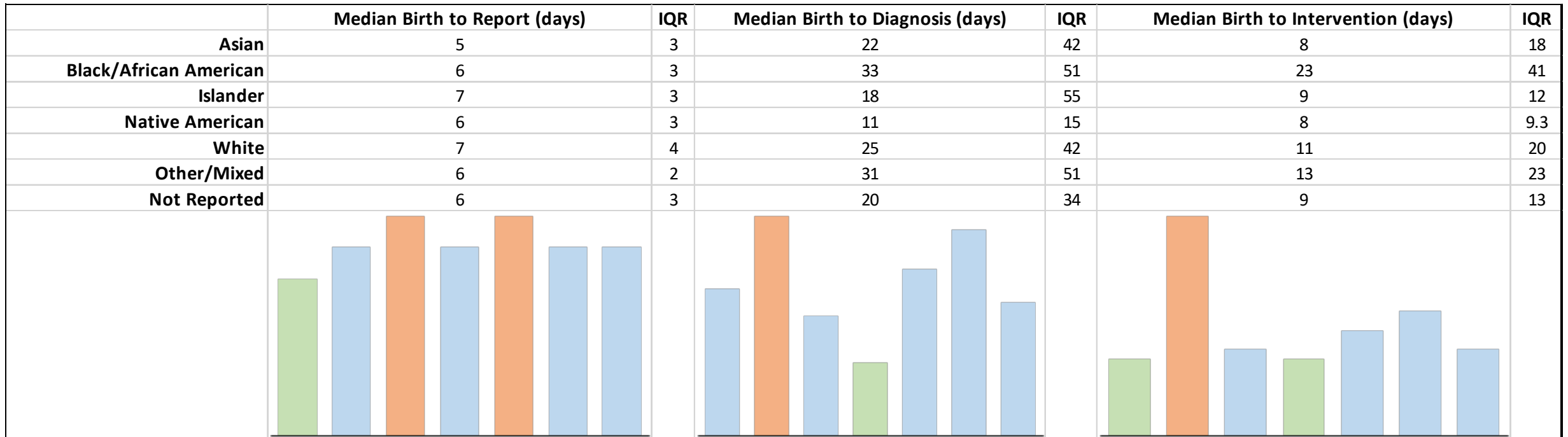- Birth to Intervention: values of 0

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Timeliness Data (By Race)

RACE CONDENSED INTO 7 CATEGORIES

Overarching Timeliness by Race

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
Asian	5	3	22	42	8	18
Black/African American	6	3	33	51	23	41
Islander	7	3	18	55	9	12
Native American	6	3	11	15	8	9.3
White	7	4	25	42	11	20
Other/Mixed	6	2	31	51	13	23
Not Reported	6	3	20	34	9	13

Note: Birth to Specimen Collection and Birth to Receipt were similar and not statistically significant

IQR = Interquartile Range

Timeliness by Race: Birth to Diagnosis

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	*						
Not Reported	*	ns					
Asian	*	ns	*				
White	*	ns	*	ns			
Other/Mixed	*	ns	*	*	*		
Black/African American	*	ns	*	*	*	ns	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

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Timeliness by Race: Birth to Intervention

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	ns	ns					
Asian	ns	ns	*				
White	ns	ns	*	ns			
Other/Mixed	*	ns	*	*	*		
Black/African American	*	*	*	*	*	*	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

Overarching Timeliness by Race Condensed

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
White	6	3	20	34	9	13
Other/Mixed	6	3	31	50	19	38
Not Reported	7	4	25	42	11	20

Note: Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant

Timeliness by Condensed Race: Birth to Diagnosis & Intervention

BIRTH TO DIAGNOSIS	White	Other	Not Reported
White			
Other	*		
Not Reported	*	*	

BIRTH TO INTERVENTION	White	Other	Not Reported
White			
Other	*		
Not Reported	*	*	

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Timeliness Data

TIME CRITICAL DISEASES BY RACE

Timeliness by Race (Time Critical)

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median (IQR) Birth to Intervention (days)	IQR
Asian (N = 129)	5	2	13.5	28	5	3
Black/African American (N = 230)	5	3	17	26	6	6
Islander (N = 6)	5	0.75	5	3	6	10.25
Native American (N = 23)	5	4	13	8.5	7	9.5
White (N = 1591)	5	3	14	28	6	5
Other/Mixed (N = 50)	6	3	17	33	6.6	7.5
Not Reported (N = 874)	5	2	17	34	5	4

Note: Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant

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Timeliness by Race (Time Critical): Birth to Diagnosis

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	ns	ns					
Asian	ns	ns	ns				
White	ns	ns	*	ns			
Other/Mixed	ns	ns	ns	ns	ns		
Black/African American	ns	ns	ns	ns	ns	ns	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

Timeliness by Race (Time Critical): Birth to Intervention

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	*	ns					
Asian	ns	ns	ns				
White	ns	ns	*	ns			
Other/Mixed	ns	ns	*	ns	ns		
Black/African American	ns	ns	*	*	ns	ns	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

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Timeliness Data

TIME SENSITIVE DISEASES BY RACE

Timeliness by Race (Time Sensitive)

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
Asian (N = 788)	5	3	24	44	9	22
Black/African American (N = 4861)	6	3	34	51	25	43
Islander (N = 20)	7	2.75	29	54	10	11.5
Native American (N = 94)	6	3	11	16.5	9	14.25
White (N = 7037)	6	3	21	35	10	15
Other/Mixed (N = 261)	6	2	34	52.5	15	25.5
Not Reported (N = 5790)	7	5	26	42	13	23

Note: *Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant*

Timeliness by Race (Time Sensitive): Birth to Diagnosis

	Native American	Islander	Not Reported	Asian	White	Other/ Mixed	Black/African American
Native American							
Islander	*						
Not Reported	*	ns					
Asian	*	ns	*				
White	*	ns	*	ns			
Other/Mixed	*	ns	*	*	*		
Black/African American	*	ns	*	*	*	ns	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

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Timeliness by Race (Time Sensitive): Birth to Intervention

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	*	ns					
Asian	ns	ns	*				
White	ns	ns	*	ns			
Other/Mixed	*	ns	*	*	*		
Black/African American	*	*	*	*	*	*	

Asterisks (*) indicate significant differences ($p < 0.05$) between reported racial categories.
Conducted by Kruskal-Walls/post-hoc Dunn.

ns = not significant

Timeliness Data

SPECIFIC DISEASE BY RACE

Cystic Fibrosis Timeliness

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
Asian (N = 49)	5	2	71.5	176.5	36	27.5
Black/African American (N = 186)	7	6	41	62	21	24
Islander (N = 5)	7	4	19	20	16	7
Native American (N = 14)	8	7	22	18	15	13
White (N = 2446)	7	5	29	42	17	18
Other/Mixed (N = 98)	5	2.5	50	81	29	47
Not Reported (N = 1263)	8	7	39	52	22	21

Note: *Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant*

Timeliness by Race (Cystic Fibrosis): Birth to Diagnosis

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	*	ns					
Asian	*	*	*				
White	ns	ns	*	*			
Other/Mixed	ns	ns	*	*	*		
Black/African American	*	ns	ns	*	*	ns	

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Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

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Timeliness by Race (Cystic Fibrosis): Birth to Intervention

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	ns	ns					
Asian	*	*	*				
White	ns	ns	*	*			
Other/Mixed	*	*	*	ns	*		
Black/African American	ns	ns	ns	*	*	*	

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Hemoglobinopathy Timeliness

Hemoglobinopathies include sickling and other Hgbs, including Hgb C, D, E, etc. Trait is excluded.

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
Asian (N = 214)	6	4	44	80	33.5	57.5
Black/African American (N = 3687)	6	3	36	51.5	31	46
Native American (N = 7)	8	3	9	17	30.5	33
White (N = 1621)	6	3	39	64.25	29	39
Other/Mixed (N = 35)	6	2.75	59	36	21.5	36.5
Not Reported (N = 109)	9	4	36	43	27	35

Note: *Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant*

Timeliness by Race (Hgbs): Birth to Diagnosis

	Native American	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American						
Not Reported	*					
Asian	*	*				
White	*	ns	ns			
Other/Mixed	*	ns	ns	ns		
Black/African American	*	*	*	ns	*	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories. Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

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Timeliness by Race (Hgbs): Birth to Intervention

	Native American	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American						
Not Reported	ns					
Asian	ns	ns				
White	ns	ns	ns			
Other/Mixed	ns	ns	ns	ns		
Black/African American	ns	ns	ns	ns	ns	

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Congenital Hypothyroidism Timeliness

	Median Birth to Report (days)	IQR	Median Birth to Diagnosis (days)	IQR	Median Birth to Intervention (days)	IQR
Asian (N = 382)	5	3	8.5	14	7	9
Black/African American (N = 589)	6	3	21	34.75	9	14
Islander (N = 9)	7	1	10	47	8.5	3
Native American (N = 61)	6	3	8	6	7	3.5
White (N = 2749)	6	2	11	21.5	7	9
Other/Mixed (N = 73)	6	2.5	13	21.5	9	11
Not Reported (N = 2004)	6	3	8	13	6	5

Note: *Birth to Specimen Collection and Birth to Receipt medians were similar and not statistically significant*

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Timeliness by Race (CH): Birth to Diagnosis

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	ns	ns					
Asian	ns	ns	ns				
White	ns	ns	*	*			
Other/Mixed	*	ns	*	*	ns		
Black/African American	*	ns	*	*	*	*	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

Timeliness by Race (CH): Birth to Intervention

	Native American	Islander	Not Reported	Asian	White	Other/Mixed	Black/African American
Native American							
Islander	ns						
Not Reported	*	ns					
Asian	ns	ns	*				
White	ns	ns	*	ns			
Other/Mixed	ns	ns	*	*	ns		
Black/African American	*	ns	*	*	*	ns	

Asterisks (*) indicate significant differences (p < 0.05) between reported racial categories.
Conducted by Kruskal-Wallis/post-hoc Dunn.

ns = not significant

Limitations

Data Completeness:
- Missing race/ethnicity data for a significant number (~33%) of cases
- Some states did not report which screen identified the case, so none of their data was used in timeliness analyses

Data Integrity/Definitions:
- Unknown origin of reported race/ethnicity data (NBS kit, birth defects, vital records, etc.)
  - Likely all of the above
- Unclear whether race/ethnicity represents the baby or the mother
- Likely differing use of Diagnosis and Intervention Date definitions

Race as Measure:
- Acknowledge that race is a social construct and that self-reported race does not adequately characterize the lived experience, and does not take into account impacts of socioeconomic status, geography, etc.

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Overall Conclusions

- There appear to be variations in post-analytical timeliness correlated with reported Race
  - Most notable in Time-Sensitive conditions
- Better race and ethnicity data are needed for further analyses
  - Cannot address what we cannot assess
- Harmonized diagnosis and intervention date definition use amongst programs is necessary for more robust comparisons

THANK YOU

- Amy Gaviglio
- Jelili Ojodu
- Sari Edelman
- HRSA Maternal and Child Health Bureau
- NewSTEPs Steering Committee
- The 53 U.S. Newborn Screening Programs that strengthen the system and serve the most vulnerable amongst us.

Contact us: newsteps@aphl.org or sikha.singh@aphl.org