

Genetics and Newborn Screening Advisory Council Statistics

Updated December 31, 2020	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020
LIVE BIRTHS	213,669	213,374	215,770	219,905	224,273	225,018	223,579	221,508	220,010	209,186
Number of newborns screened	212,504	211,179	214,348	217,293	222,167	222,124	220,687	221,042	217,025	212,032
Percentage screened	99.45%	98.97%	99.34%	98.81%	99.06%	98.71%	98.71%	99.79%	98.64%	101.36%
SW Unsatisfactory Specimen Rate	1.92%	1.85%	2.36%	1.60%	1.21%	1.17%	1.21%	1.75%	1.92%	1.97%
Hospital Unsat Rate	1.66%	1.55%	2.08%	1.34%	1.05%	1.01%	1.02%	1.49%	1.68%	1.74%
SW Transit time (%>3days)	<i>Began following Transit Time %>3days in 2015</i>				24.25%	22.96%	22.58%	20.91%	21.10%	21.28%
Hospital Transit (%>3days)					22.99%	21.80%	21.25%	19.54%	19.68%	19.69%
# of specimens submitted	265,194	263,234	268,046	267,706	273,999	278,113	273,571	272,403	270,717	258,421
PRESUMPTIVE POSITIVES and BORDERLINE REFERRALS:										
Congenital Hypothyroidism	143	131	151	162	146	118	136	131	137	157
TSH Borderline	1,438	870	818	1,328	1,878	1,841	2,199	1,552	1,854	3,441
Congenital Adrenal Hyperplasia	16	23	20	25	22	18	21	17	21	16
CAH >24 hrs >2500g	104	138	181	175	172	153	103	105	115	115
CAH <24 hrs >2500g - Early	150	184	242	241	199	152	125	118	178	161
CAH <2500g Age-N/A - Low wt	42	46	42	52	36	46	27	27	34	27
Galactosemia	86	103	126	126	104	103	101	113	99	74
Partial galt obtained repeat	<i>Tgal testing - 12/2013</i>		6	69	91	61	40	44	54	54
Sickle Cell Disease	258	230	259	248	258	266	260	264	239	249
MS/MS	193	192	155	194	204	213	296	315	291	268
MS/MS Borderline	2,522	2,530	2,455	3,004	2,737	2,834	2,604	2,563	2,257	2,020
Biotinidase Deficiency	24	20	35	13	3	12	14	30	25	18
Borderlines	60	35	42	18	20	20	32	31	18	20
SCID		4	29	22	30	14	18	25	14	83
SCID Early/Inconclusive	<i>SCID started 10/1/12</i>	65	304	288	418	351	317	322	267	71
SCID Borderline		3	26	18	13	12	7	12	4	146
Cystic Fbrosis - 2 Mutations	33	29	34	30	22	40	26	40	40	32
Cystic Fibrosis - 1 Mutations	464	426	480	509	469	507	550	495	496	433
Ultra-Hi IRT/No mutations in NICU	473	531	518	383	379	596	695	673	683	678
Partial Unsatisfactory	91	96	65	83	106	394	295	128	80	192
X-ALD	<i>X-ALD screening began 5/1/18</i>							13	11	20
X-ALD Borderline								8	3	<i>Stopped following 2/2020</i>
MPS I	<i>MPS I screening began 2/3/20</i>									
Pompe	<i>Pompe screening began 2/3/20</i>									
SMA	<i>SMA screening began 4/27/20</i>									
Presumptive Pos	1,217	1,161	1,289	1,329	1,258	1,291	1,422	1,443	1,373	1,526
Borderline	6,720	6,708	4,699	5,659	6,049	6,460	6,444	5,583	5,547	6,925
TOTAL	7,937	7,869	5,988	6,988	7,307	7,751	7,866	7,026	6,920	8,451
Hearing Refers	6,535	6,865	8,045	8,121	8,355	9,357	9,146	8,582	9,453	9,351
Pulse Oximetry/CCHD Fails	<i>Data not collected before 7/2014</i>			43	95	141	177	134	153	182
<p style="text-align: center;">Presumptive positive screening results are referred to CMS Referral Centers for confirmatory testing and diagnostic evaluation. Borderlines are followed by CMS Newborn Screening Follow-Up Program through repeat screening. Transit Rate: Beginning 2015. Transit Rate is measured by %>3 days.</p>										

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LIVE BIRTHS	213,669	213,374	215,770	219,905	224,273	225,018	223,579	221,508	220,010	209,186
CONFIRMED POSITIVES:										
Congenital Hypothyroidism	102	67	78	78	80	54	70	69	64	51
CH diagnosed thru BDL	32	13	7	4	5	2	14	9	4	5
Congenital Adrenal Hyperplasia	8	13	9	10	12	6	13	7	9	7
Galactosemia (G/G)	2	4	3	4	5	4	4	3	0	3
Variant	26	38	49	42	35	34	43	48	43	24
Sickle Cell Hb S/S	120	124	131	125	144	130	130	133	102	86
Sickle Cell Hb S/C	84	62	78	62	63	71	66	68	58	51
Sickle Cell Hb S/A	9	11	15	7	7	9	14	11	11	1
MS/MS (including PKU)	46	41	45	37	37	34	26	33	32	35
MS/MS secondary disorders	0	0	0	19	16	19	12	20	22	6
Biotinidase Deficiency	6	3	3	4	0	0	4	3	6	3
partial	14	14	25	5	1	9	9	20	7	6
Cystic Fibrosis 2 Mutations	22	24	28	23	27	32	24	31	32	23
Cystic Fibrosis 1 Mutations	11	10	8	10	8	1	12	16	6	8
Cystic Fibrosis High IRT; 0 mutations	0	0	0	2	0	0	1	0	0	0
CFTR Metabolic Syndrome	7	8	8	8	6	3	4	3	11	5
SCID & x-linked	0	0	1	3	3	4	1	1	1	2
Idiopathic T-cell Lymphopenia	<i>Began using classification in September 2019.</i>								0	1
Leaky SCID-Omenn	<i>Began using classifications in November 2015.</i>				0	0	0	0	0	0
Syndrome w/ decreased T-cells					0	3	5	2	1	0
Secondary T-penia					1	2	7	4	1	8
Variant	0	2	0	2	0	0	0	0	<i>Stopped using Sep 2019.</i>	
Primary Immunodeficiency	0	1	3	1	0	<i>Stopped using classifications in November 2015.</i>				
Non-SCID (ie, Chylothorax)	0	1	4	4	3					
X-ALD	<i>X-ALD screening began 5/1/18</i>							11	9	10
Pompe	<i>Pompe screening began 2/3/20</i>									15
MPS I	<i>MPS I screening began 2/3/20</i>									0
SMA	<i>SMA screening began 4/27/20</i>									14
TOTAL	489	436	495	450	453	417	459	492	419	364
Hearing loss ID'd thru NBS	269	335	301	308	305	318	288	305	257	215
CCHD - cardiac	<i>Added to panel in December 2013. No data collected before July 2014.</i>			4	9	24	16	13	8	3
CCHD - secondary cardiac				5	23	26	23	27	14	15
CCHD - non-cardiac/other				20	50	50	49	47	79	48
CCHD - no disease found				10	33	35	18	45	43	33
TOTAL				9	32	50	39	40	22	18
Physician Requests (CMS)	12,307	7,801	7,186	6,130	4,901	1,111	1,642	1,804	1,855	1,912
FNSR Registered Users	2,327	2,956	2,712	4,133	4,357	6,110	6,948	6,356	2,694	4,004