

Genetics and Newborn Screening Advisory Council Statistics

***Updated June 30, 2020***	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020
<b>LIVE BIRTHS</b>	<b>213,669</b>	<b>213,374</b>	<b>215,770</b>	<b>219,905</b>	<b>224,273</b>	<b>225,018</b>	<b>223,579</b>	<b>221,508</b>	<b>220,010</b>	<b>108,630</b>
Number of newborns screened	212,504	211,179	214,348	217,293	222,167	222,124	220,687	221,042	217,025	103,730
Percentage screened	99.45%	98.97%	99.34%	98.81%	99.06%	98.71%	98.71%	99.79%	98.64%	95.49%
SW Unsatisfactory Specimen Rate	1.92%	1.85%	2.36%	1.60%	1.21%	1.17%	1.21%	1.75%	1.92%	2.18%
Hospital Unsat Rate	1.66%	1.55%	2.08%	1.34%	1.05%	1.01%	1.02%	1.49%	1.68%	1.96%
SW Transit time (>3days)	<i>Began following Transit Time &gt;3days in 2015</i>				24.25%	22.96%	22.58%	20.91%	21.10%	20.69%
Hospital Transit (>3days)					22.99%	21.80%	21.25%	19.54%	19.68%	19.15%
# of specimens submitted	265,194	263,234	268,046	267,706	273,999	278,113	273,571	272,403	270,717	127,321
<b>PRESUMPTIVE POSITIVES and BORDERLINE REFERRALS:</b>										
<b>Congenital Hypothyroidism</b>	143	131	151	162	146	118	136	131	137	67
TSH Borderline	1,438	870	818	1,328	1,878	1,841	2,199	1,552	1,854	1,471
<b>Congenital Adrenal Hyperplasia</b>	16	23	20	25	22	18	21	17	21	10
CAH >24 hrs >2500g	104	138	181	175	172	153	103	105	115	64
CAH <24 hrs >2500g - Early	150	184	242	241	199	152	125	118	178	88
CAH <2500g Age-N/A - Low wt	42	46	42	52	36	46	27	27	34	13
<b>Galactosemia</b>	44	48	57	126	104	103	101	113	99	37
Partial galt referred	42	58	69	<i>Stopped referring partial galts to Referral Centers in December 2013</i>						
Partial galt obtained repeat	<i>Tgal testing - 12/2013</i>		6	69	91	61	40	44	54	18
<b>Sickle Cell Disease</b>	258	230	259	248	258	266	260	264	239	127
<b>MS/MS</b>	193	192	155	194	204	213	296	315	290	122
MS/MS Borderline	2,522	2,530	2,455	3,004	2,737	2,834	2,604	2,563	2,257	949
<b>Biotinidase Deficiency</b>	24	20	35	13	3	12	14	30	25	10
Borderlines	60	35	42	18	20	20	32	31	18	10
<b>SCID</b>	<i>SCID started 10/1/12</i>	4	29	22	30	14	18	25	14	53
SCID Early/Inconclusive		65	304	288	418	351	317	322	267	71
SCID Borderline		3	26	18	13	12	7	12	4	61
<b>Cystic Fbrosis - 2 Mutations</b>	33	29	34	30	22	40	26	40	40	18
<b>Cystic Fibrosis - 1 Mutations</b>	464	426	480	509	469	507	550	495	496	224
Ultra-Hi IRT/No mutations in NICU	473	531	518	383	379	596	695	673	683	319
Partial Unsatisfactory	91	96	65	83	106	394	295	128	80	52
<b>X-ALD</b>	<i>X-ALD screening began 5/1/18</i>							13	11	5
X-ALD Borderline								8	3	0
<b>MPS I</b>	<i>MPS I screening began 2/3/20</i>									12
<b>Pompe</b>	<i>Pompe screening began 2/3/20</i>									70
<b>SMA</b>	<i>SMA screening began 4/27/20</i>									6
<b>Presumptive Pos</b>	1,217	1,161	1,220	1,329	1,258	1,291	1,422	1,443	1,372	761
<b>Borderline</b>	6,720	6,708	4,699	5,659	6,049	6,460	6,444	5,583	5,547	3,116
<b>TOTAL</b>	<b>7,937</b>	<b>7,869</b>	<b>5,919</b>	<b>6,988</b>	<b>7,307</b>	<b>7,751</b>	<b>7,866</b>	<b>7,026</b>	<b>6,919</b>	<b>3,877</b>
<b>Hearing Refers</b>	6,535	6,865	8,045	8,121	8,355	9,357	9,146	8,573	9,388	4,520
<b>Pulse Oximetry/CCHD Fails</b>	<i>Data not collected before 7/2014</i>			43	95	141	177	134	153	89

## Genetics and Newborn Screening Advisory Council Statistics

***Updated June 30, 2020***	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020
<b>LIVE BIRTHS</b>	<b>213,669</b>	<b>213,374</b>	<b>215,770</b>	<b>219,905</b>	<b>224,273</b>	<b>225,018</b>	<b>223,579</b>	<b>221,508</b>	<b>220,010</b>	<b>108,630</b>
<b>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 %&gt;3 days.</b>										

Genetics and Newborn Screening Advisory Council Statistics

***Updated June 30, 2020***	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	
<b>LIVE BIRTHS</b>	<b>213,669</b>	<b>213,374</b>	<b>215,770</b>	<b>219,905</b>	<b>224,273</b>	<b>225,018</b>	<b>223,579</b>	<b>221,508</b>	<b>220,010</b>	<b>108,630</b>	
<b>CONFIRMED POSITIVES:</b>											
<b>Congenital Hypothyroidism</b>	102	67	78	78	80	54	70	69	64	22	
CH diagnosed thru BDL	32	13	7	4	5	2	14	9	4	2	
<b>Congenital Adrenal Hyperplasia</b>	8	13	9	10	12	6	13	7	9	4	
<b>Galactosemia (G/G)</b>	2	4	3	4	5	4	4	3	0	3	
Variant	26	38	49	42	35	34	43	48	43	8	
<b>Sickle Cell Hb S/S</b>	120	124	131	125	144	130	130	133	101	32	
<b>Sickle Cell Hb S/C</b>	84	62	78	62	63	71	66	68	57	14	
<b>Sickle Cell Hb S/A</b>	9	11	15	7	7	9	14	11	11	1	
<b>MS/MS (including PKU)</b>	46	41	45	37	37	34	26	33	32	9	
<b>MS/MS secondary disorders</b>	0	0	0	19	16	19	12	20	22	2	
<b>Biotinidase Deficiency</b>	6	3	3	4	0	0	4	3	6	2	
partial	14	14	25	5	1	9	9	20	9	0	
<b>Cystic Fibrosis 2 Mutations</b>	22	24	28	23	27	32	24	31	29	7	
<b>Cystic Fibrosis 1 Mutations</b>	11	10	8	10	8	1	13	15	6	2	
<b>Cystic Fibrosis High IRT; 0 mutations</b>	0	0	0	2	0	0	0	0	0	0	
<b>CFTR Metabolic Syndrome</b>	7	8	8	8	6	3	4	3	10	2	
<b>SCID &amp; x-linked</b>	0	0	1	3	3	4	1	1	1	1	
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	2	
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						
<b>X-ALD, Male</b>	<i>X-ALD screening began 5/1/18</i>							4	3	0	
<b>X-ALD, Female</b>								6	3	1	
<b>X-ALD, Probable</b>								1	1	1	
<b>X-ALD, Possible</b>								0	1	0	
<b>Zellweger</b>								2	1	0	
<b>Pompe, IOPD</b>	<i>Pompe screening began 2/3/20</i>										2
<b>Pompe, LOPD</b>											0
<b>Pompe, LOPD Probable</b>											1
<b>Pompe, LOPD Possible</b>											3
<b>MPS I, Severe</b>	<i>MPS I screening began 2/3/20</i>										0
<b>MPS I, Attenuated</b>											0
<b>SMA</b>	<i>SMA screening began 4/27/20</i>										5

Genetics and Newborn Screening Advisory Council Statistics

***Updated June 30, 2020***	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020
<b>LIVE BIRTHS</b>	<b>213,669</b>	<b>213,374</b>	<b>215,770</b>	<b>219,905</b>	<b>224,273</b>	<b>225,018</b>	<b>223,579</b>	<b>221,508</b>	<b>220,010</b>	<b>108,630</b>
<b>TOTAL</b>	<b>489</b>	<b>436</b>	<b>495</b>	<b>450</b>	<b>453</b>	<b>417</b>	<b>459</b>	<b>492</b>	<b>415</b>	<b>127</b>
<b>Hearing loss ID'd thru NBS</b>	<b>269</b>	<b>335</b>	<b>301</b>	<b>308</b>	<b>305</b>	<b>318</b>	<b>288</b>	<b>265</b>	<b>194</b>	<b>40</b>
<b>CCHD - cardiac</b>	<i>Added to panel in December 2013. No data collected before July 2014.</i>			4	9	24	16	13	6	0
<b>CCHD - secondary cardiac</b>				5	23	26	23	27	12	2
<b>CCHD - non-cardiac/other</b>				20	50	50	49	47	70	18
<b>CCHD - no disease found</b>				10	33	35	18	45	37	8
<b>TOTAL</b>				<b>9</b>	<b>32</b>	<b>50</b>	<b>39</b>	<b>40</b>	<b>18</b>	<b>2</b>
<b>Physician Requests (CMS)</b>	12,307	7,801	7,186	6,130	4,901	1,111	1,642	1,804	1,855	489
FNSR Registered Users	2,327	2,956	2,712	4,133	4,357	6,110	6,948	6,356	2,694	3,304