Genetics and Newborn Screening Advisory Council Statistics

Updated January 13, 2021	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**	**209,645**
Number of newborns screened	212,504	211,179	214,348	217,293	222,167	222,124	220,687	221,042	217,025	208,814
Percentage screened	99.45%	98.97%	99.34%	98.81%	99.06%	98.71%	98.71%	99.79%	98.64%	99.60%
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)	*Began following Transit Time %>3days in 2015*				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	259,745
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	44	48	57	126	104	103	101	113	99	74
Partial galt referred	42	58	69	*Stopped referring partial galts to Referral Centers in December 2013*						
Partial galt obtained repeat	*Tgal testing - 12/2013*		6	69	91	61	40	44	53	50
Sickle Cell Disease	258	230	259	248	258	266	260	264	239	250
MS/MS	193	192	155	194	204	213	296	315	288	279
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	19
Borderlines	60	35	42	18	20	20	32	31	18	21
SCID	*SCID started 10/1/12*	4	29	22	30	14	18	25	14	83
SCID Early/Inconclusive		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	198
X-ALD	*X-ALD screening began 5/1/18*							13	11	22
X-ALD Borderline								8	3	*Stopped following*
MPS I	*MPS I screening began 2/3/20*									26
Pompe	*Pompe screening began 2/3/20*									136
SMA	*SMA screening began 4/27/20*									14
Presumptive Pos	1,217	1,161	1,220	1,329	1,258	1,291	1,422	1,443	1,370	1,541
Borderline	6,720	6,708	4,699	5,659	6,049	6,460	6,444	5,583	5,546	6,928
TOTAL	**7,937**	**7,869**	**5,919**	**6,988**	**7,307**	**7,751**	**7,866**	**7,026**	**6,916**	**8,469**
Hearing Refers	6,535	6,865	8,045	8,121	8,355	9,357	9,146	8,582	9,453	9,463
Pulse Oximetry/CCHD Fails	*Data not collected before 7/2014*			43	95	141	177	134	153	195

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CONFIRMED POSITIVES:										
Congenital Hypothyroidism	102	67	78	78	80	54	70	69	64	63
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	26
Sickle Cell Hb S/S	120	124	131	125	144	130	130	133	102	102
Sickle Cell Hb S/C	84	62	78	62	63	71	66	68	58	64
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	37
MS/MS secondary disorders	0	0	0	19	16	19	12	20	22	14
Biotinidase Deficiency	6	3	3	4	0	0	4	3	6	3
partial	14	14	25	5	1	9	9	20	9	7
Cystic Fibrosis 2 Mutations	22	24	28	23	27	32	24	32	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	5	4	11	6
SCID & x-linked	0	0	1	3	3	4	1	1	1	3
Idiopathic T-cell Lymphopenia	*Began using classification in September 2019.*								0	1
Leaky SCID-Omenn	*Began using classifications in November 2015.*				0	0	0	0	0	0
Syndrome w/ decreased T-cells					0	3	5	2	1	0
Secondary T-cell Lymphopenia					1	2	7	4	1	10
Variant	0	2	0	2	0	0	0	0	*Stopped using Sep 2019.*	
Primary Immunodeficiency	0	1	3	1	0	*Stopped using classifications in November 2015.*				
Non-SCID (ie, Chylothorax)	0	1	4	4	3					
X-ALD								13	10	16
Pompe										20
MPS I										0
SMA	*SMA screening began 4/27/20*									14
TOTAL	**489**	**436**	**495**	**450**	**453**	**417**	**459**	**496**	**422**	**433**
Hearing loss ID'd thru NBS	**269**	**335**	**301**	**308**	**305**	**318**	**288**	**305**	**257**	**274**
CCHD - cardiac	*Added to panel in December 2013. No data collected before July 2014.*			4	9	24	16	13	8	13
CCHD - secondary cardiac				5	23	26	23	27	14	22
CCHD - non-cardiac/other				20	50	50	49	47	79	98
CCHD - no disease found				10	33	35	18	45	43	62
TOTAL				**9**	**32**	**50**	**39**	**40**	**22**	**35**
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