

National Newborn Screening Data Initiatives (Dried Blood Spot Screening)

Brad Therrell, Ph.D.

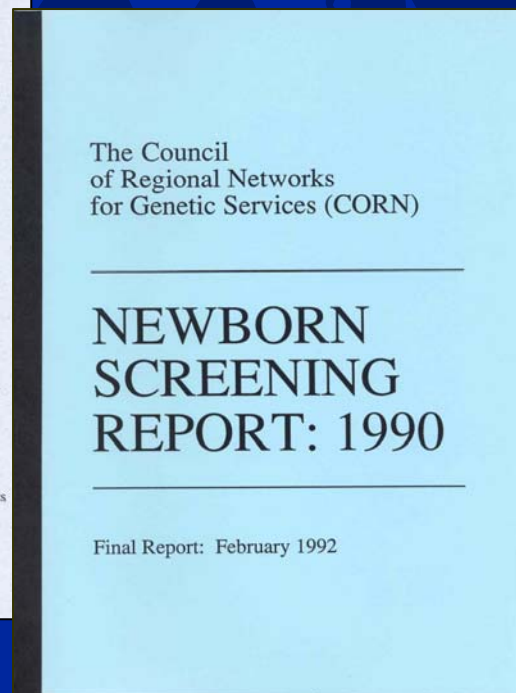
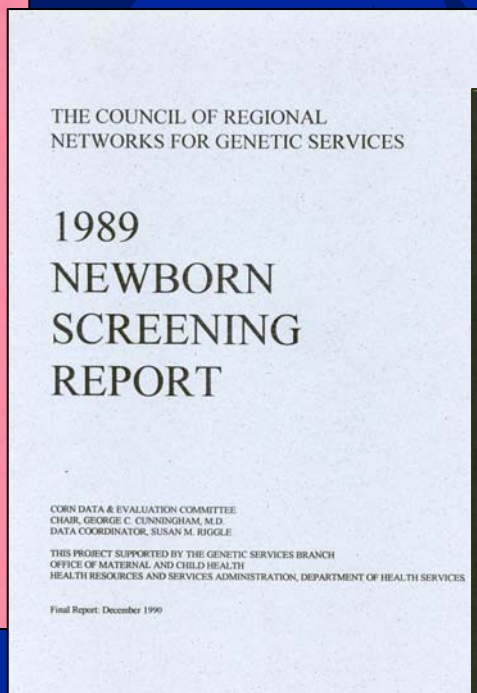
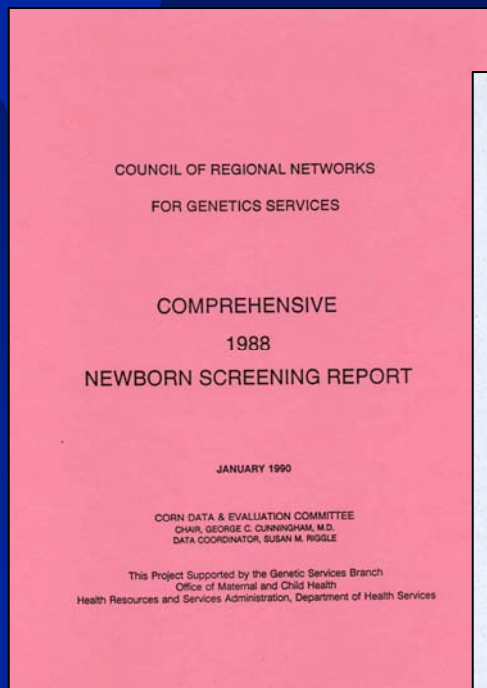
University of Texas Health Science Center at San Antonio

Newborn Screening and Genetics Resource Center

Austin, Texas

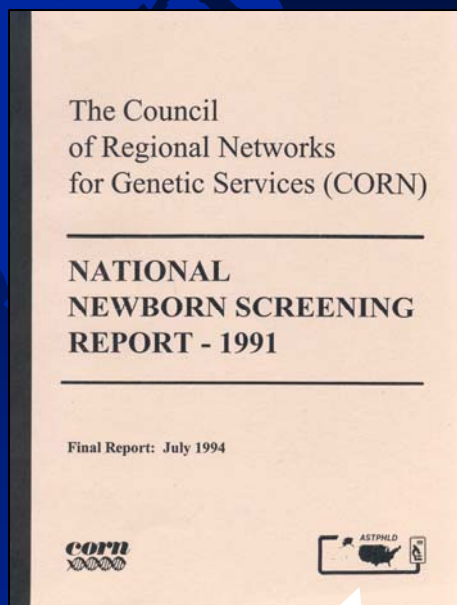
U.S. Newborn Screening Data Report

Initially (1988) a Project of: Council of Regional Networks for Genetic Services (CORN) Data and Evaluation Committee



U.S. Newborn Screening Data Report

Transitioned to CORN Newborn Screening Committee



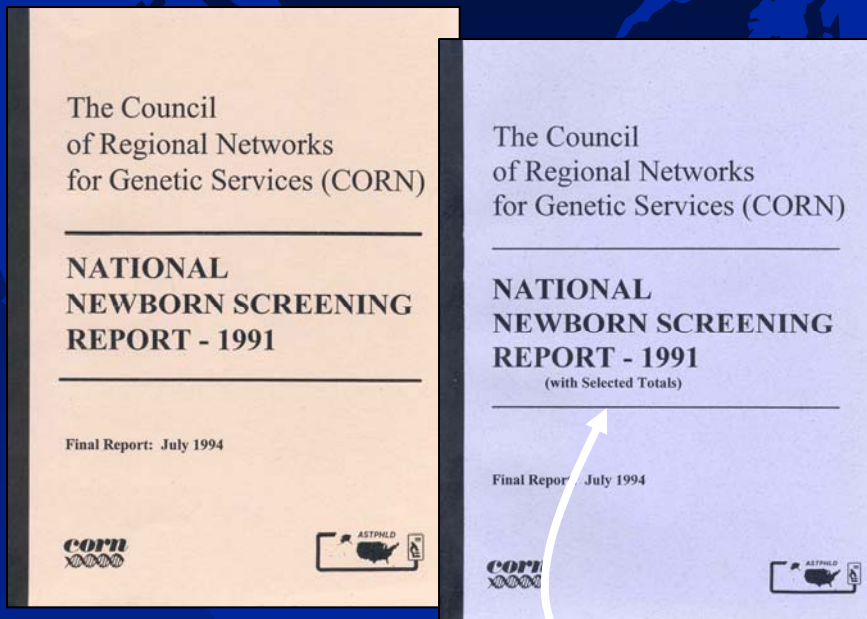
In cooperation with the
ASTPHLD (now APHL)

U.S. Newborn Screening Data Report Collection/Validation Procedure

1. Questionnaire Prepared by Committee
2. Questionnaire Completed by Program (Laboratory and Follow-up)
3. Completed Questionnaire Approved Internally by Laboratory and Follow-up Officials
4. Questionnaire Submitted to Central Collector
5. Data Extracted and Placed in Tables
6. Tables Reviewed (Committee or Submitters)
7. Data Report Published
8. Questionnaire Periodically Reviewed/Updated

U.S. Newborn Screening Data

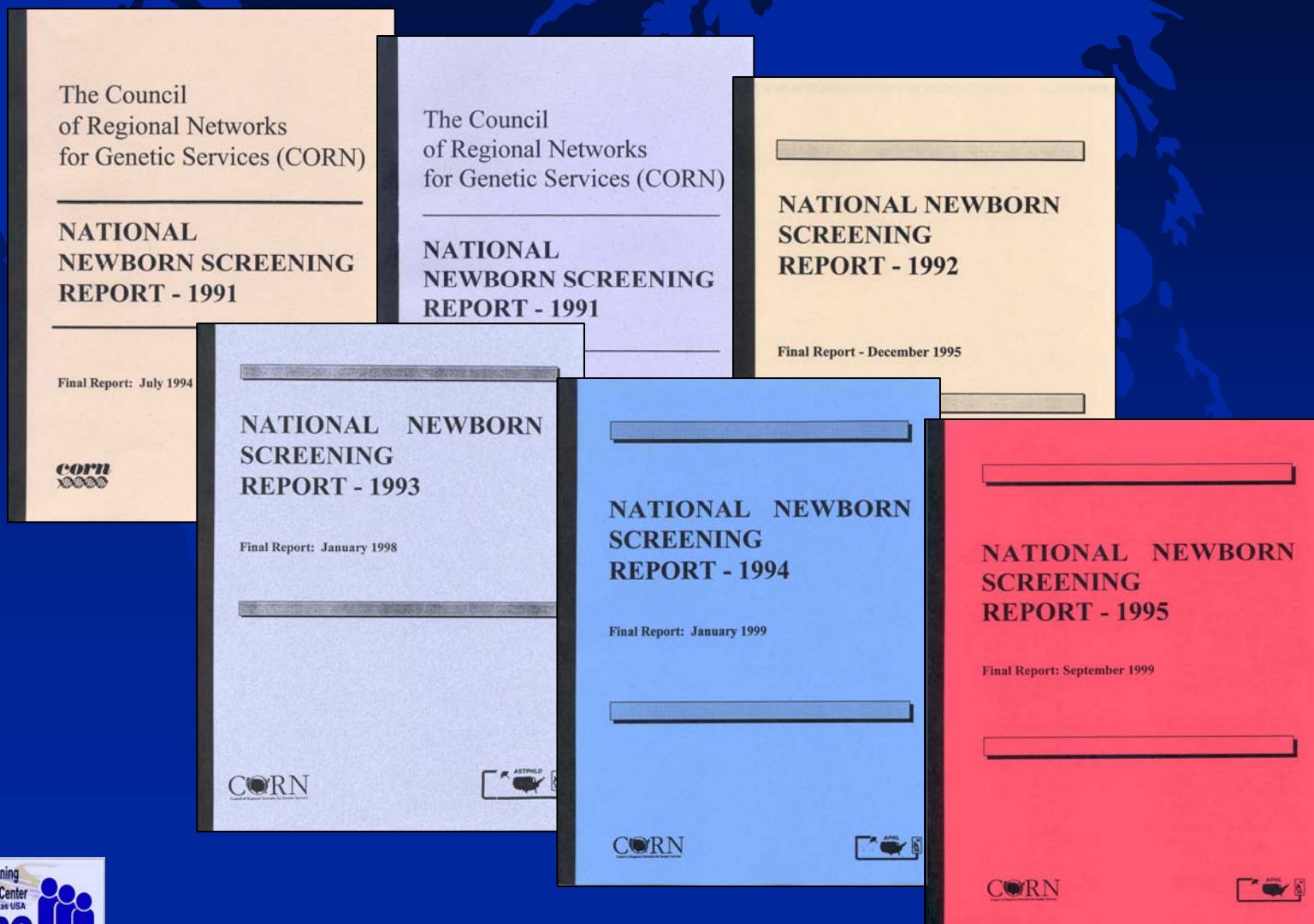
Prepared by: CORN Newborn Screening Committee



Containing
'Selected' Totals

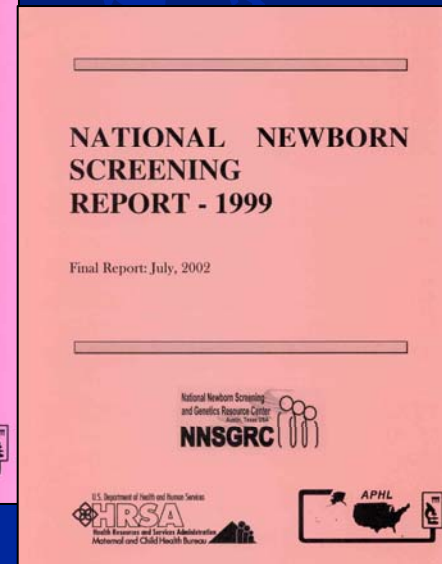
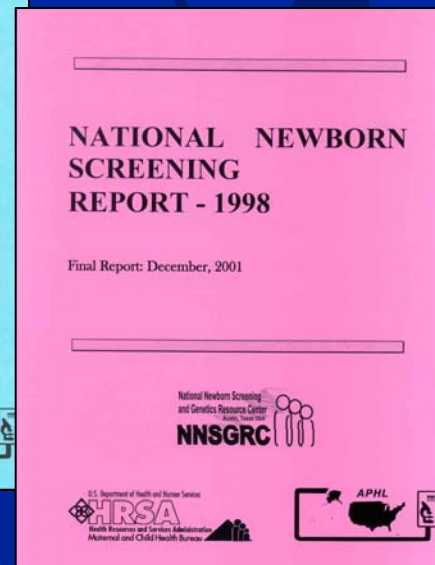
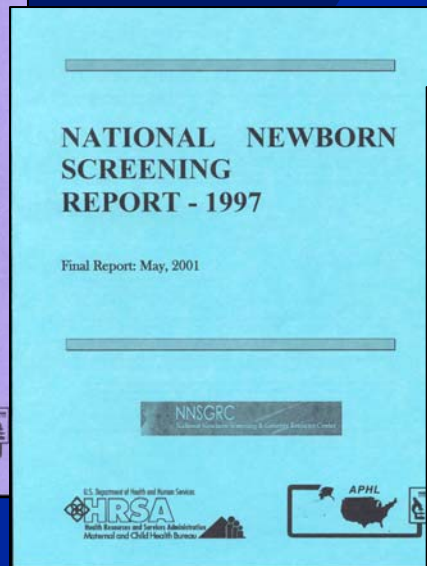
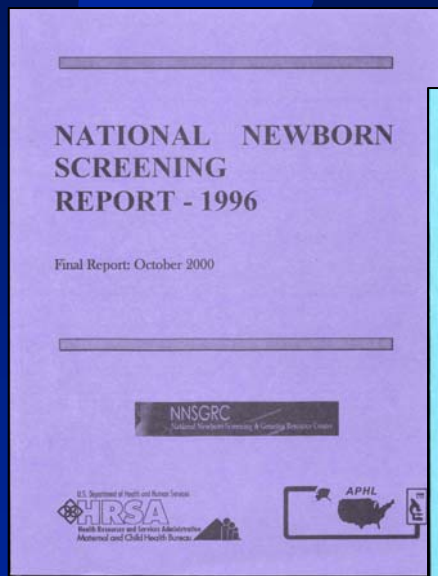
U.S. Newborn Screening Data Report

Prepared by CORN Newborn Screening Committee



U.S. Newborn Screening Data Report

1999 Became a Function of the National Newborn Screening and Genetics Resource Center (NNSGRC) Assisted by Newborn Screening Advisory Committee



U.S. Newborn Screening Data Report

Data Overview

Chapter 1 – Live Birth Statistics

- Births by Occurrence (Obtained from National Center for Health Statistics – including subdivisions by race/ethnicity.
- Asian births – Subdivided into 5 groups – NCHS
- Hispanic births – Subdivided into 6 groups – NCHS

U.S. Newborn Screening Data Report

Data Overview

Chapter 2 – Overview of Programs

- Summary Table of Disorders Screened
- Summary of Laboratories Screening Within States
- Basic Listing of Components of Follow-up
- Summary of Ages at Time of Initial Testing
- Summary of Fees Charged and Components Included
- Criteria for 2nd Screens
- Miscellaneous Information About Specimens (Received, unacceptable, time stored, policy or not, computer info, type education provided)

U.S. Newborn Screening Data Report

Data Overview

Chapters 3-13 Summary Data by Condition

- Program Definitions for Conditions (including variants)
- Laboratory Procedure and Definition of Not Normal
- Initial Screening Results – # screened, # not normal, # lost to follow-up, # diagnosed, # variants diagnosed (# significant; # not significant)
- Second Screening Results – same as above – subdivided by required, discretionary (timing problem), repeat (result of abn. first)
- Diagnosed Cases Subdivided by Race, Sex, Hispanic Ethnicity
- Days from Birth to Treatment
- Summation Data (from beginning of screening)

National Newborn Screening Information System (NNSIS)



National Newborn Screening Information System

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Welcome to the National Newborn Screening Information System (NNSIS) database.

The database is hosted by the **National Newborn Screening and Genetics Resource Center (NNSGRC)** and is designed to provide a secure, Internet based, real-time, information collection and reporting system for capturing state and territorial newborn screening information.

The system uses existing reporting requirements specified in the former National Newborn Screening Annual Report.

-
- » **Obtain a Report**
 - » **Enter Individual or Annual Case Information**
 - » **Contact NNSGRC**
-

Welcome to our Newborn Screening Data System

NNSIS Reports



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Disorder reports

- » [Overview of Disorder Specific Reports](#)
- » [Biotinidase](#)
- » [Congenital Adrenal Hyperplasia \(CAH\)](#)
- » [Cystic Fibrosis \(CF\)](#)
- » [Galactosemia \(GAL\)](#)
- » [Glucose 6 phosphate dehydrogenase \(G-6-PD\)](#)
- » [Homocystinuria \(HCY\)](#)
- » [Hemoglobinopathies](#)
- » [Hyperphenylalaninemia \(PKU\)](#)
- » [Hypothyroidism \(CH\)](#)
- » [Maple Syrup Urine Disease \(MSUD\)](#)
- » [Toxoplasmosis](#)
- » [HIV-Antibodies \(HIV-1\)](#)
- » [Amino Acid Disorders other than PKU, MSUD, HCY](#)
- » [Fatty Acid Oxidation Disorders](#)
- » [Organic Acid Disorders](#)

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NNSIS Reports – Reports for a Condition



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Report Year: 2001

Reports for Congenital Adrenal Hyperplasia (CAH)

- » Descriptive Overview
- » Summation of Cumulative Case Information
- » Laboratory Testing
- » Initial Screening Results
- » Second Screen Results
- » Clinical Observations Prior to Receipt of Screening Report
- » Clinical Observations Subsequent to Normal Screening Result
- » Total Cases Detected

Cases subdivided by ethnicity/sex for

- » Classical salt wasting congenital adrenal hyperplasia (CAH)
- » Classical simple virilizing congenital adrenal hyperplasia (CAH)
- » Non-classical congenital adrenal hyperplasia (CAH)
- » Unclassified congenital adrenal hyperplasia (CAH)
- » CAH other than 21-OH

Cases subdivided by days from birth to physician notification for

- » Classical salt wasting congenital adrenal hyperplasia (CAH)
- » Classical simple virilizing congenital adrenal hyperplasia (CAH)

Cases subdivided by days from birth to treatment for

- » Classical salt wasting congenital adrenal hyperplasia (CAH)
- » Classical simple virilizing congenital adrenal hyperplasia (CAH)

NNSIS Reports - Births

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[Back](#) Home > Reports menu > **National births report**

Report Year: 2001

US Live Birth Statistics by Occurrence (Final) Classified by Race (2001)

State	White	African American	Native American	Asian/Pacific Islander	Total	Hispanic	Non-Hispanic	Last update
Alabama	40,181	18,950	174	461	59,766	2,280	57,486	4/12/2004
Alaska	6,300	441	2,538	628	9,907	1,523	8,384	4/12/2004
Arizona	75,497	2,782	5,342	2,136	85,757	37,851	47,906	4/12/2004
Arkansas	28,544	6,976	294	487	36,301	2,804	33,497	4/12/2004
California	429,023	33,764	2,911	62,841	528,539	265,122	263,417	4/12/2004
Colorado	61,159	2,970	640	2,331	67,100	19,729	47,371	4/12/2004
Connecticut	36,095	5,148	160	1,776	43,179	7,544	35,635	4/12/2004
Delaware	8,342	2,654	29	335	11,360	1,104	10,256	4/12/2004
District of Columbia	6,097	8,352	21	567	15,037	1,482	13,555	4/12/2004
Florida	152,259	47,307	1,234	5,191	205,991	50,125	155,866	4/12/2004
Georgia	86,097	44,142	278	3,885	134,402	17,294	117,108	4/12/2004
Hawaii	3,826	531	185	12,585	17,127	2,313	14,814	4/12/2004
Idaho	19,455	87	329	290	20,161	3,054	17,107	4/12/2004
Illinois	139,830	32,897	256	8,103	181,086	40,985	140,101	4/12/2004
Indiana	75,634	9,649	169	1,258	86,710	6,370	80,340	4/12/2004
Iowa	35,309	1,287	351	809	37,756	2,521	35,235	4/12/2004

NNSIS Reports – Program Overview



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Report Year:

State:

Select the relevant state.

Program Overview for Texas in 2001 As of 01/11/2005

Live Birth Statistics

Total	Race				Ethnicity	
	White	Af. American	Am. Indian	Asia/Pacific Is	Hispanic	Non-Hispanic
370,482	316,858	40,956	851	11,817	177,621	192,861

Disorders Reported - Date Begun and Confirmed Cases

Individual disorder	Date begun	Total cases since screening began through 2001	Confirmed cases 2001 **
Classical salt wasting congenital adrenal hyperplasia (CAH)	1/1/1989	203	16 <small>last up date 12/17/2004</small>
Classical simple virilizing congenital adrenal hyperplasia (CAH)	1/1/1989	81	9 <small>last up date 12/17/2004</small>
Non-classical congenital adrenal hyperplasia (CAH)	1/1/1989	119	16 <small>last up date 12/17/2004</small>
Unclassified congenital adrenal hyperplasia (CAH)	1/1/1989	35	0
CAH other than 21-OH	1/1/1989	18	1 <small>last up date 12/17/2004</small>
Classical galactosemia (GAL)	1/1/1979	111	8 <small>last up date 12/17/2004</small>
Galactosemia variant	1/1/1979	127	20 <small>last up date 12/17/2004</small>

NNSIS Reports – Age When Screened



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Report Year: ▼

Infant's age at time of initial testing in the U.S. in 2001

State/ Territory	0-12 Hours (optional)	13-24 Hours (optional)	Day 1 (Total of first 2)	Day 2	Day 3	Day 4	Day 5	Day 6	Day 7	Over 7 Days	Unknown	Total Tested	Last update
Alabama	0	0	0	0	0	0	0	0	0	0	0	0	10/10/2003
Alaska	591	1,863	2,454	4,263	1,670	606	160	230	104	217	137	9,841	10/10/2003
Arizona	1,978	11,598	13,576	46,331	14,519	4,206	903	394	19	899	2,310	83,157	2/23/2004
Arkansas	100	258	358	2,205	3,554	5,006	5,946	5,606	3,946	10,337	0	36,958	12/30/2004
California	8,000	156,048	164,048	282,347	48,290	15,242	8,627	4,755	1,929	4,995	915	531,148	10/10/2003
Colorado	0	0	0	0	0	0	0	0	0	0	0	0	10/13/2003
Connecticut	0	0	0	0	0	0	0	0	0	0	44,892	44,892	10/13/2003
Delaware	137	631	768	7,422	1,920	390	868	25	21	27	0	11,441	10/14/2003
District of Columbia	0	0	0	0	0	0	0	0	0	0	0	0	10/15/2003
Florida	0	0	0	0	0	0	0	0	0	0	0	0	10/15/2003
Georgia	0	0	0	0	0	0	0	0	0	0	0	0	10/16/2003
Hawaii	42	1,747	1,789	12,466	2,198	385	30	29	12	131	5	17,045	10/17/2003
Idaho	1,103	3,923	5,026	8,510	3,495	1,436	301	161	106	326	0	19,361	12/8/2004
Illinois	152	0	152	0	0	0	0	0	0	0	2,068	2,220	10/22/2003
Indiana	1,834	5,306	7,140	45,261	25,805	2,906	1,504	888	600	1,011	1,167	86,282	11/10/2004
Iowa	52	210	262	30,946	4,919	827	241	66	24	233	26	37,544	10/23/2003
Kansas	675	685	1,360	27,662	6,838	1,002	396	664	246	351	2	38,521	10/29/2003

NNSIS Reports – General Information



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Laboratory Specimen Information for Newborn Screening in the U.S. in 2001

State/ Territory	Total number of specimens received	Number of specimens unacceptable	% of samples unacceptable	Length of time specimens kept	Written policy for storage and disposal of specimens	Type of computer evaluation	Type of education provided	Last update
Alabama	181,488	21622	11.91%	3 mo at 28°C	Yes	Yes Neometrics software	Yes continuing education provided by QA division and QA Prog	10/27/2003
Alaska	17,833	331	1.9%	3 yr	Yes Testing lab stores 1 yr at room temp, returned to AK for 2 yrs storage, and then destroyed	Yes Weekly matches of birth reports to specimens	Yes Practitioners manual, telephone consultation, videos, and presentations	10/30/2003
Arizona	147,028	1367	0.92%	3 months	No AZ unwritten policy is to store samples at room temp. for 3 months and then autoclave for disposal.	No	Yes lab staff do QA and train submitters; health educator developed educational materials and provided in-service.	2/23/2004
Arkansas	37,894	204	0.54%	One year	Yes Samples stored in a ziplock bag with dessicant @ -20°C.	Yes Queries are compiled by lab reporting system for type and total number of rejections results by test	Yes Inservice training for Local Country Health units and hospital staff.	12/30/2004



Some National Data Challenges

- Data reporting, except for Title V, not mandated nor a defined program responsibility (NY not participating in NNSIS)
- National data set has not been mandated (currently depends on committee approach)
- Data definitions are inconsistent from program to program
- Staff shortages/turnover affect data entry (time and understanding)
- Data quality difficult to validate (depends on program)
- Maintaining data on border babies or others moving into jurisdiction
- Data on military births
- Automated downloads from program and laboratory

Thank You

The image features the words "Thank You" in a large, white, 3D block font. The text is positioned centrally and appears to be floating above a dark blue silhouette of a world map. The map shows the outlines of continents in a slightly lighter shade of blue. The entire scene is set against a solid, dark blue background. The lighting on the 3D text creates a sense of depth and volume.