Newborn Screening Use Case Update: Moving Newborn Screening Into the Electronic Age

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Overview

- Updates from HITSP, PHII, IHE
  - The HITSP Newborn Screening Interoperability Specification will be completed in January 2010
  - The APHL has approved the Newborn Screening HL7 Implementation Guide prepared by PHII
  - IHE has finalized their Newborn Screening White Paper
- HITSP Requirements Design Standards Selection (RDSS)
- Inspection Testing of the Draft Interoperability Specification
  - Re-use of constructs from other Use Cases
- Anticipated Use of SNOMED and LOINC
- Filter Paper Data Capture of lab test ordering information
- Considerations for ACHDNC

Requirements Design Standards Selection

- The Requirements Design Standards Selection (RDSS) Document is the first milestone in the HITSP development process and provides specific solutions for each aspect of the original use case that remains fixed
- The RDSS will be open for public comment Sept 18 – Oct 16
- A key feature of the RDSS is the listing of the Data Requirements for each Information Exchange. The community will:
  - need to review the data captured on the dried blood spot filter paper as part of the test ordering data requirements
  - need to review the separate data requirements for newborn hearing screening
  - need to review the data requirements for newborn screening lab reports

Interoperability Specification Inspection Testing

- Draft Interoperability Specification (IS) based on RDSS document - completed by Oct 30, 2009
- Inspection testing and public comments - due Dec 4, 2009
- Final Interoperability Specification - expected completion Jan 2010
- Key components of IS will make extensive re-use of material from other use cases (e.g. EHR-Lab).

Anticipated Use of SNOMED and LOINC

- Migration from ICD9-CM to SNOMED coded problem lists and required use of LOINC coded lab reports is anticipated to be part of certification criteria for EHR under ARRA and implementation for NBS is part of the use case.
- SNOMED coded problem lists will be a requirement by 2015 and will begin use soon
- Use of LOINC codes to report both analytes (measures) and genetic test interpretation by condition using the methods developed for the Personalized Healthcare Use Case and electronic reporting of genetic testing
- Newborn Screening Codes project at NLM will maintain the SNOMED and LOINC codes required for newborn screening test ordering, result reporting, and some follow-up activities

NBS Laboratory Test Order Data Fields

- Rational for Data Collection and Analysis
  - The data will help identify an inclusive set of requirements for lab test ordering that will work for all states
  - The goal is not to produce one standard national form or terminology but to assure that all state requirements are included
- Methods
  - Filter paper forms from 50 states and DC were scanned and all fields tabulated with identification of synonyms and value sets
  - Cluster analysis created three groups of fields that were most prevalent, frequently occurring, and used in only a few states
- Next Steps
  - Data Verification

Newborn Screening (NBS) Data Field Analysis - Explanation

Data Elements were plotted by two sets of values:

NBF: Newborn Factors (y-axis)

-Indicates potential volume of data for each data element
-Calculated by summing the number of live births in the states where the data element is found, based on the National Newborn Screening and Genetics Resource Center (NNSGRC) 2006 data set.

Sample calculation: if the baby's middle name is found in NY (10 births) MD (5 births) and CA (15 births) the NBF factor would be 30.

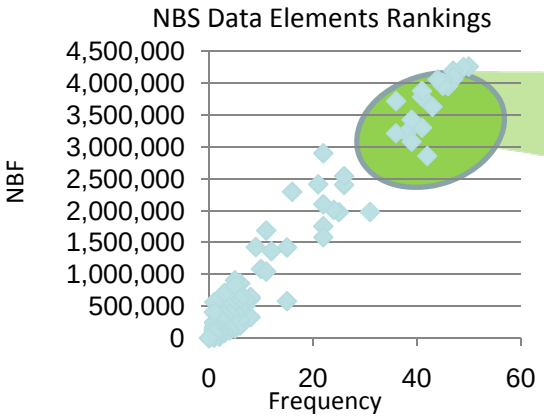
Frequency (x-axis)

-Indicates degree of usage as a key data element among states
-Measures of the occurrence of the data element among the states,

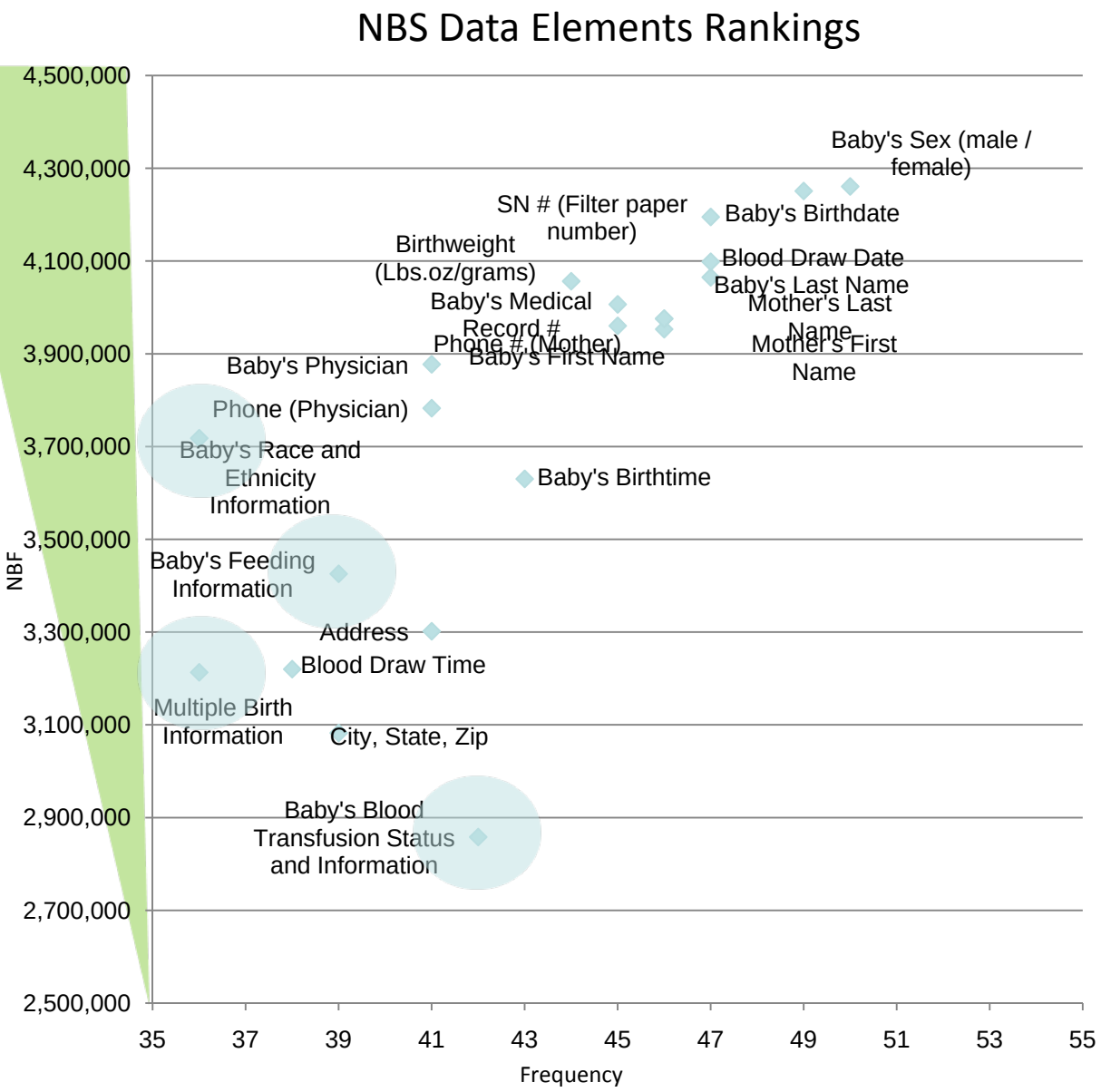
Elements appearing in the top right quadrant are the most prevalent, with a common occurrence among states and a potentially very large volume of data nationally. Conversely elements occurring in the lower left are relatively uncommon and have a lower potential for data volume.

Elements centered on a blue circle are representations of several data elements that are part of the same category.

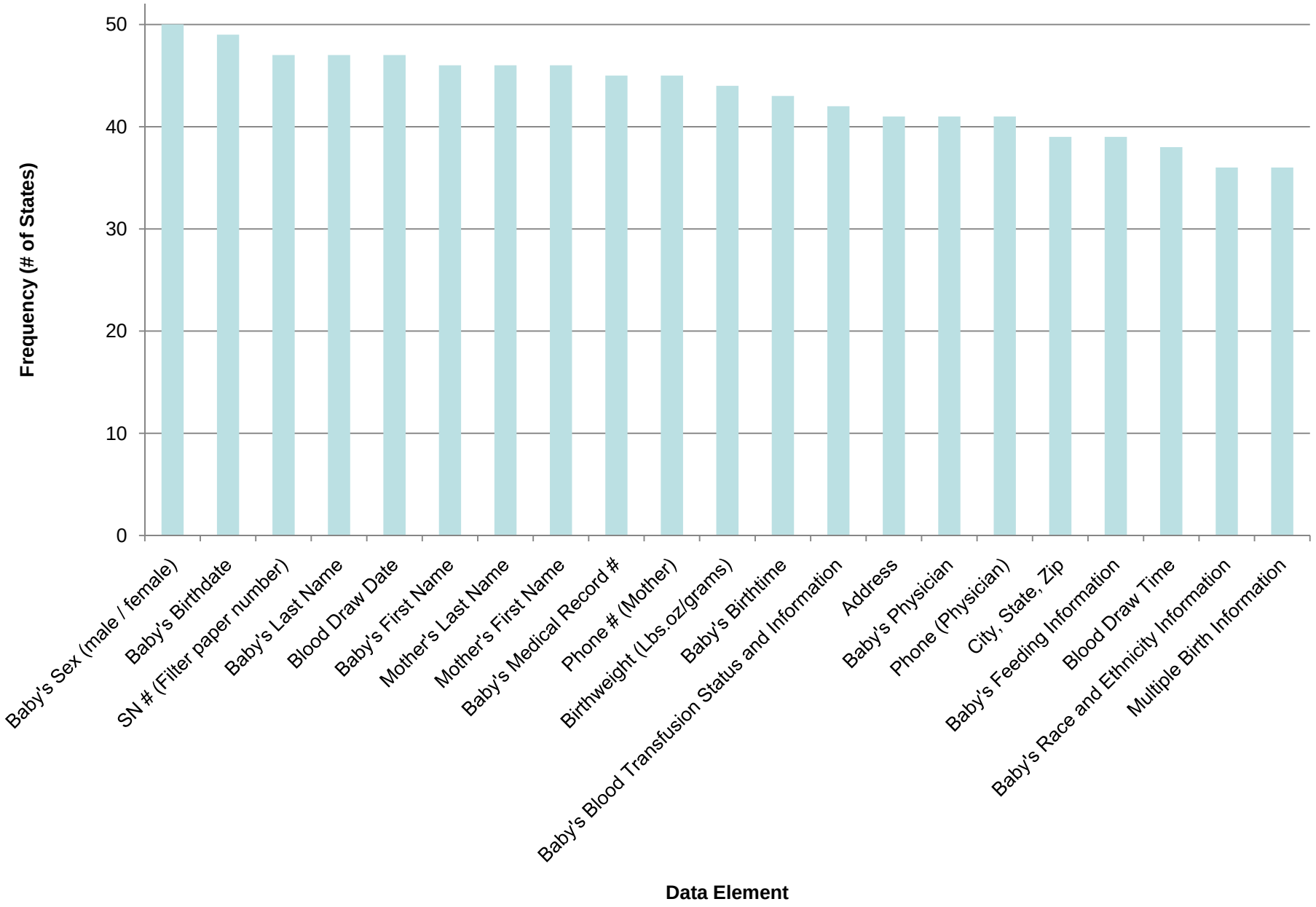
Most Prevalent Newborn Data Elements (1/2)



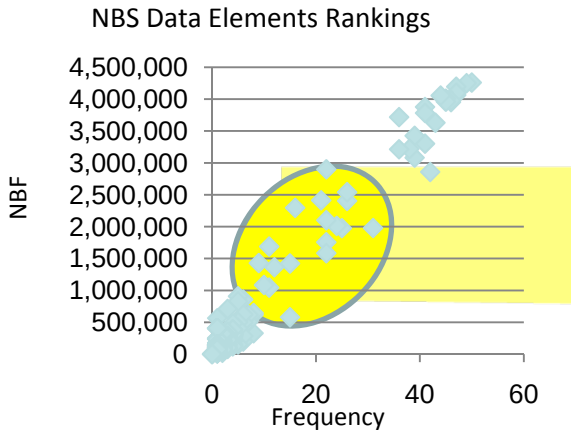
Data Element	Frequency	NBF
Baby's Sex (male / female)	50	4,260,839
Baby's Birthdate	49	4,250,964
SN # (Filter paper number)	47	4,194,948
Baby's Last Name	47	4,064,958
Blood Draw Date	47	4,098,438
Baby's First Name	46	3,953,028
Mother's Last Name	46	3,975,780
Mother's First Name	46	3,975,780
Baby's Medical Record #	45	4,006,540
Phone # (Mother)	45	3,960,265
Birthweight (Lbs.oz/grams)	44	4,056,419
Baby's Birthtime	43	3,630,119
Baby's Blood Transfusion Status and Information	42	2,857,901
Address	41	3,301,714
Baby's Physician	41	3,877,282
Phone (Physician)	41	3,782,594
City, State, Zip	39	3,082,293
Baby's Feeding Information	39	3,425,516
Blood Draw Time	38	3,219,910
Baby's Race and Ethnicity Information	36	3,717,878
Multiple Birth Information	36	3,213,023



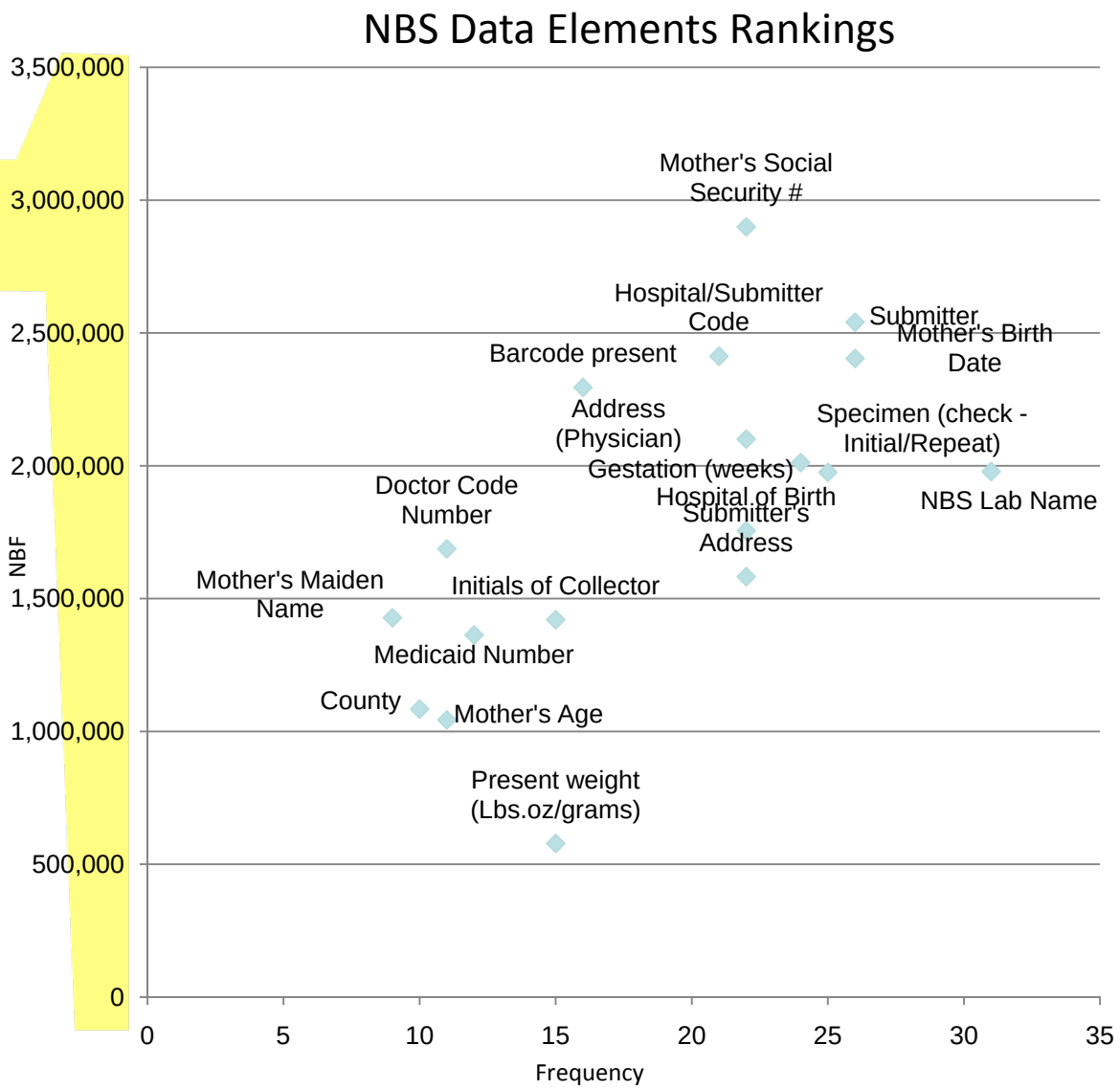
Most Frequently Occurring Data Elements



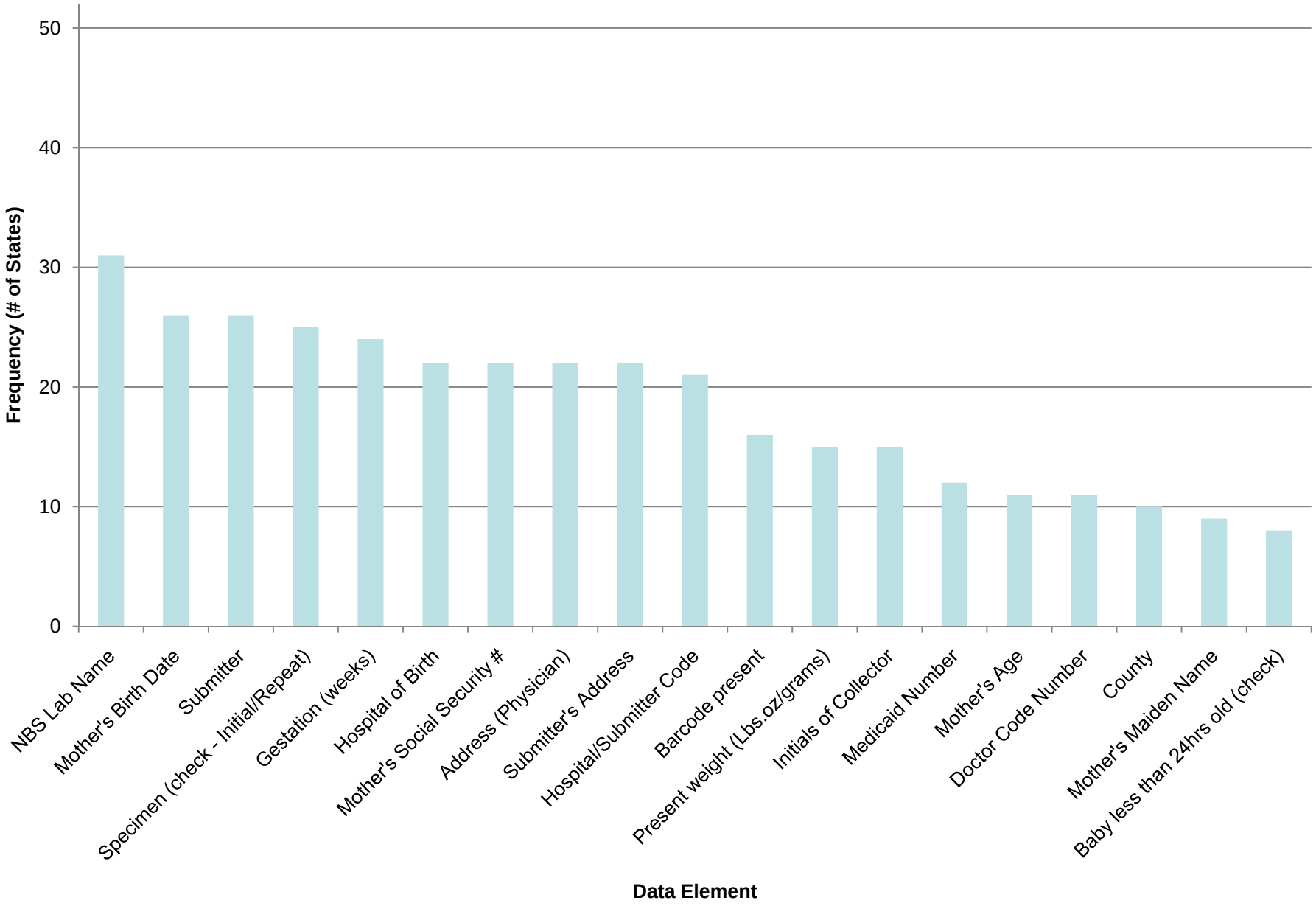
Most Prevalent Newborn Data Elements (2/2)



Data Element	Frequency	NBF
NBS Lab Name	31	1,978,266
Mother's Birth Date	26	2,403,759
Submitter	26	2,540,253
Specimen (check - Initial/Repeat)	25	1,975,183
Gestation (weeks)	24	2,011,845
Hospital of Birth	22	1,755,440
Mother's Social Security #	22	2,899,004
Address (Physician)	22	2,100,411
Submitter's Address	22	1,582,596
Hospital/Submitter Code	21	2,412,277
Barcode present	16	2,294,882
Present weight (Lbs.oz/grams)	15	578,449
Initials of Collector	15	1,420,650
Medicaid Number	12	1,363,063
Mother's Age	11	1,043,495
Doctor Code Number	11	1,687,363
County	10	1,084,177
Mother's Maiden Name	9	1,427,863



Often Occurring Data Elements



Considerations for ACHDNC

- Participate in public comments on the Requirements Design Standards Selection Document RDSS – Due Oct 16, 2009
- Participate in the inspection testing (public comment) on the Interoperability Specification – Due Dec 4, 2009
- Propose a mechanism to assign identifier numbers to all newborn screening laboratories (not all have CLIA numbers)
- Endorse implementation of the Interoperability Specification at the next ACHDNC Meeting Jan 21-22, 2010
- Review NLM Newborn Screening Codes using SNOMED and LOINC – On-going