NATIONAL ASSOCIATION OF Community Health Centers®

Newborn Screening: An Opportunity for Next-Generation Data and Care Coordination

Julia Skapik, MD, MPH, FAMIA

CMIO

National Association of Community Health Centers

NACHC's STRATEGIC PILLARS

1

Equity and Social Justice

Center everything we do in a renewed commitment to equity and social justice

2

Empowered Infrastructure

Strengthen and reinforce the infrastructure for leading and coordinating the Community Health Center Movement, notably consumer boards and NACHC itself

3

Skilled and Mission-driven Workforce

Develop a highly skilled, adaptive, and mission-driven workforce reflecting the communities served

4

Reliable and Sustainable Funding

Secure reliable and sustainable funding to meet increasing demands for Community Health Center services

5

Improved Care Models

Update and improve care models to meet the evolving needs of the communities served

6

Supportive Partnerships

Cultivate new and strengthen existing mutually beneficial partnerships to advance the shared mission of improving community health

To learn more about NACHC's Strategic Pillars visit https://www.nachc.org/about/about-nachc/

THE NACHC MISSION

America's Voice for Community Health Care

The National Association of Community Health Centers (NACHC) was founded in 1971 to promote efficient, high quality, comprehensive health care that is accessible, culturally and linguistically competent, community directed, and patient centered for all.

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* Disclosure: Julia is also the volunteer Board Chair of HealthLevel7 International, an international health IT standards development organization (SDO).

Agenda

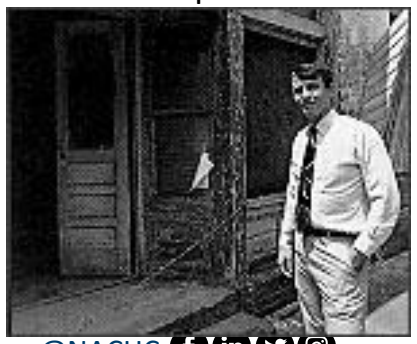
- Current state of health center newborn screening: lack of closing the data loop
- Four components to future success in newborn interoperability and care coordination: trust, standards, policies, implementation issues
- FHIR and the future of unlocking data for patient care and public health
- Operational and implementation success in the FHIR-enabled future

THE COMMUNITY HEALTH CENTERS:
AN OUTGROWTH OF THE CIVIL RIGHTS MOVEMENT

Dr. Robert Smith formed the Southern branch of the Medical Committee for Civil Rights (MCCR) in 1963 to protest the American Medical Association (AMA), which allowed southern medical societies to remain segregated and often kept Black physicians from being employed at hospitals.

Health centers were created to provide culturally competent healthcare in healthcare access deserts, a practice which continues today.

Originally intending to pursue a cardiology practice, Dr. James Hotz somehow found himself practicing family medicine in southwest Georgia instead. Below: one of the first sites of a clinic he helped establish.



HEALTH CENTERS

FIVE ESSENTIAL ELEMENTS

1. Located in **high-need areas.**
2. Provide **comprehensive** health and wraparound services (including enabling services).
3. **Open to all** residents, regardless of insurance or ability to pay, with sliding scale fee based on income.
4. Nonprofits, governed by **community boards**, to assure responsiveness to local needs.
5. **Follow performance and accountability requirements** regarding their administrative, clinical, and financial operations.

TODAY

Community Health Centers are the most comprehensive, wide-spread and effective primary care providers.
No patient is turned away.

14K Delivery Sites

1,487 Health Centers

31.5+M people served (1 in 11)

400K Veterans

1.3M Homeless People

8.6M Children

3.3M Elderly Patients

1 in 5 **uninsured**

1 in 5 **rural residents**

1 in 3 **people living in poverty**

Newborn Screening Requires Care and Data Coordination

- Linkage of newborn and maternal data is not the norm in the industry
- Diffusion of accountability for quality of care rests across outpatient obstetric team, inpatient obstetric team, inpatient pediatric team and outpatient pediatric team
- Handoffs of care and data transmission are problematic – infrastructure for this work requires design, testing and maintenance

What about the EHR?

- Theoretically, a certified EHR has required standards for the coding and transmission of patient and lab data
- In reality, EHRs do not adequately support interoperability of even required data consistently. A concerted effort is required to ensure data is sent and received in a valid way consistently
- Patient data fragmentation and data loss result in clinical care gaps and risk to the patient of harm: **data quality accountability = patient safety**

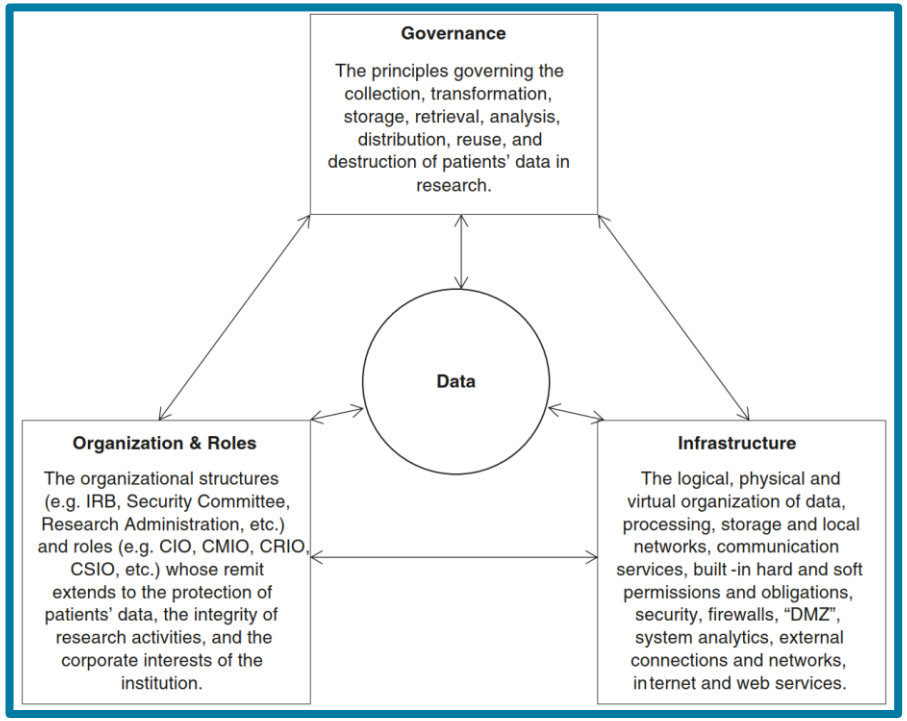
Health centers and newborn screening

- Health centers primarily serve patients with health disparities and barriers to access
- Health centers report receiving newborn screening via:
  - Fax
  - Mail
  - Portal?
- Health center primary OB and pediatrics providers report they do not know if patient has been contacted by hospital or consistently receive results

Trust and Data Governance

Two levels of trust:

- Patient and care team
  - Patients need time and a supportive environment to engage a trust-based relationship and decision-making process
- Organizational and systematic
  - Organizations need to agree on the rules of data exchange, data use and reuse, privacy, security and quality



Privacy, Security and Consent

- Existing systems could create a workflow supporting maternal and family engagement, secure screening data interoperability and reporting and consent
- Engaging families, care teams and their health data systems in pregnancy could create a digital trail to allow data to flow back and forth to and from public health and primary and inpatient obstetric and pediatric teams
- Families could use their own devices and systems to authorize data transfer and engagement

USCDI Draft v5 Summary of Data Classes and Data Elements

Allergies and Intolerances
- Substance (Medication)
- Substance (Drug Class)
- Substance (Non-Medication)
- Reaction

Care Team Members
- Care Team Member Name
- Care Team Member Identifier
- Care Team Member Role
- Care Team Member Location
- Care Team Member Telecom

Clinical Notes
- Consultation Note
- Discharge Summary Note
- Emergency Department Note
- History & Physical
- Operative Note
- Procedure Note
- Progress Note

Clinical Tests
- Clinical Test
- Clinical Test Result/Report

Diagnostic Imaging
- Diagnostic Imaging Test
- Diagnostic Imaging Report

Encounter Information
- Encounter Type
- Encounter Identifier
- Encounter Diagnosis
- Encounter Time
- Encounter Location
- Encounter Disposition

Facility Information
- Facility Identifier
- Facility Type
- Facility Name

Goals and Preferences
- Patient Goals
- SDOH Goals
- Treatment Intervention Preference
- Care Experience Preference

Health Insurance Information
- Coverage Status
- Coverage Type
- Relationship to Subscriber
- Member Identifier
- Subscriber Identifier
- Group Identifier
- Payer Identifier

Health Status Assessments
- Health Concerns
- Functional Status
- Disability Status
- Mental/Cognitive Status
- Pregnancy Status
- Alcohol Use
- Substance Use
- Physical Activity
- SDOH Assessment
- Smoking Status

Immunizations
- Immunizations
- Lot Number

Laboratory
- Tests
- Values/Results
- Specimen Type
- Result Status
- Result Unit of Measure
- Result Reference Range
- Test Kit Unique Device Identifier
- Result Interpretation
- Specimen Source Site
- Specimen Identifier
- Specimen Condition Acceptability

Medical Devices
- Unique Device Identifier - Implantable

Medications
- Medications
- Dose
- Dose Unit of Measure
- Route
- Indication
- Fill Status
- Medication Instructions
- Medication Adherence

Observations
- Advance Directive Observation
- Sex Parameter for Clinical Use

Orders
- Orders

Patient Demographics/ Information
- First Name
- Last Name
- Middle Name (Including middle initial)
- Name Suffix
- Previous Name
- Name to Use
- Pronoun
- Date of Birth
- Date of Death

Patient Demographics /Information (cont)
- Race
- Ethnicity
- Tribal Affiliation
- Sex
- Sexual Orientation
- Gender Identity
- Preferred Language
- Interpreter Needed
- Current Address
- Previous Address
- Phone Number
- Phone Number Type
- Email Address
- Related Person's Name
- Relationship Type
- Occupation
- Occupation Industry

Patient Summary and Plan
- Assessment and Plan of Treatment

Problems
- Problems
- SDOH Problems/Health Concerns
- Date of Diagnosis
- Date of Resolution

Procedures
- Procedures
- Performance Time
- SDOH Interventions
- Reason for Referral

Provenance
- Author
- Author Role
- Author Time Stamp
- Author Organization

Vital Signs
- Systolic Blood Pressure
- Diastolic Blood Pressure
- Average Blood Pressure
- Heart Rate
- Respiratory Rate
- Body Temperature
- Body Height
- Body Weight
- Pulse Oximetry
- Inhaled Oxygen Concentration
- BMI Percentile (2 - 20 years)
- Weight-for-length Percentile (Birth - 24 Months)
- Head Occipital-frontal Circumference Percentile (Birth- 36 Months)

USCDI United States Core Data for Interoperability

United States Core Data for Interoperability

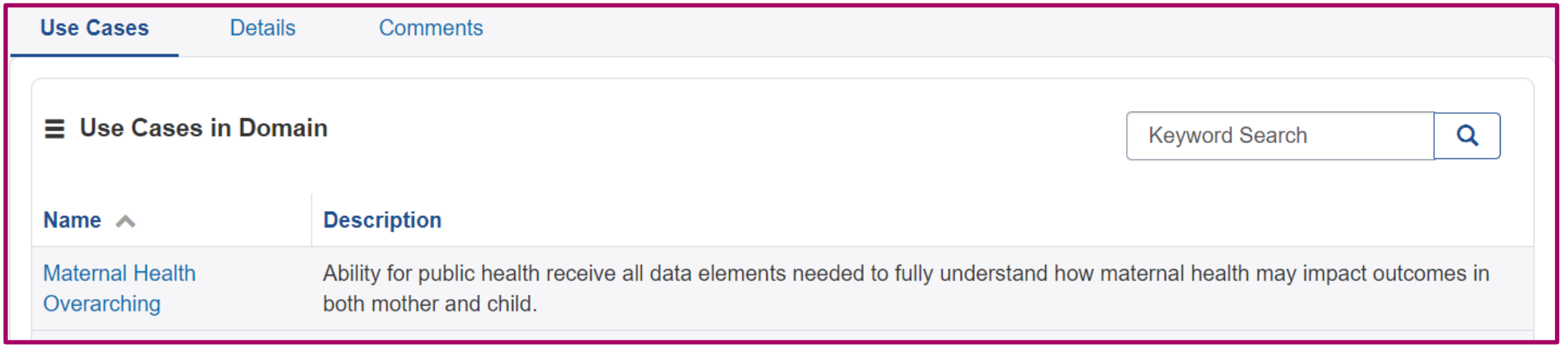
Draft Version 5 | January 2024

US Core Data for Interoperability (USCDI)

NATIONAL ASSOC
Community Heal

@NACHC

USCDI+ Maternal Health



- USCDI+ program is an extension of USCDI program
- Currently it is unclear if and how ONC may require implementation, testing or certification of the USCDI+ domains
- Genomic information can be more difficult to standardize in the field
- **USCDI+ contains maternal health data elements AND infant data–> newborn screening data could be added**

https://uscdiplus.healthit.gov/uscdi?id=uscdi_record&table=x_g_sshh_uscdi_domain&sys_id=75df78228745b95098e5edb90cbb3528&view=sp

The time for FHIR is NOW:
Fast Healthcare Interoperability Resources

Designed for and excels at:

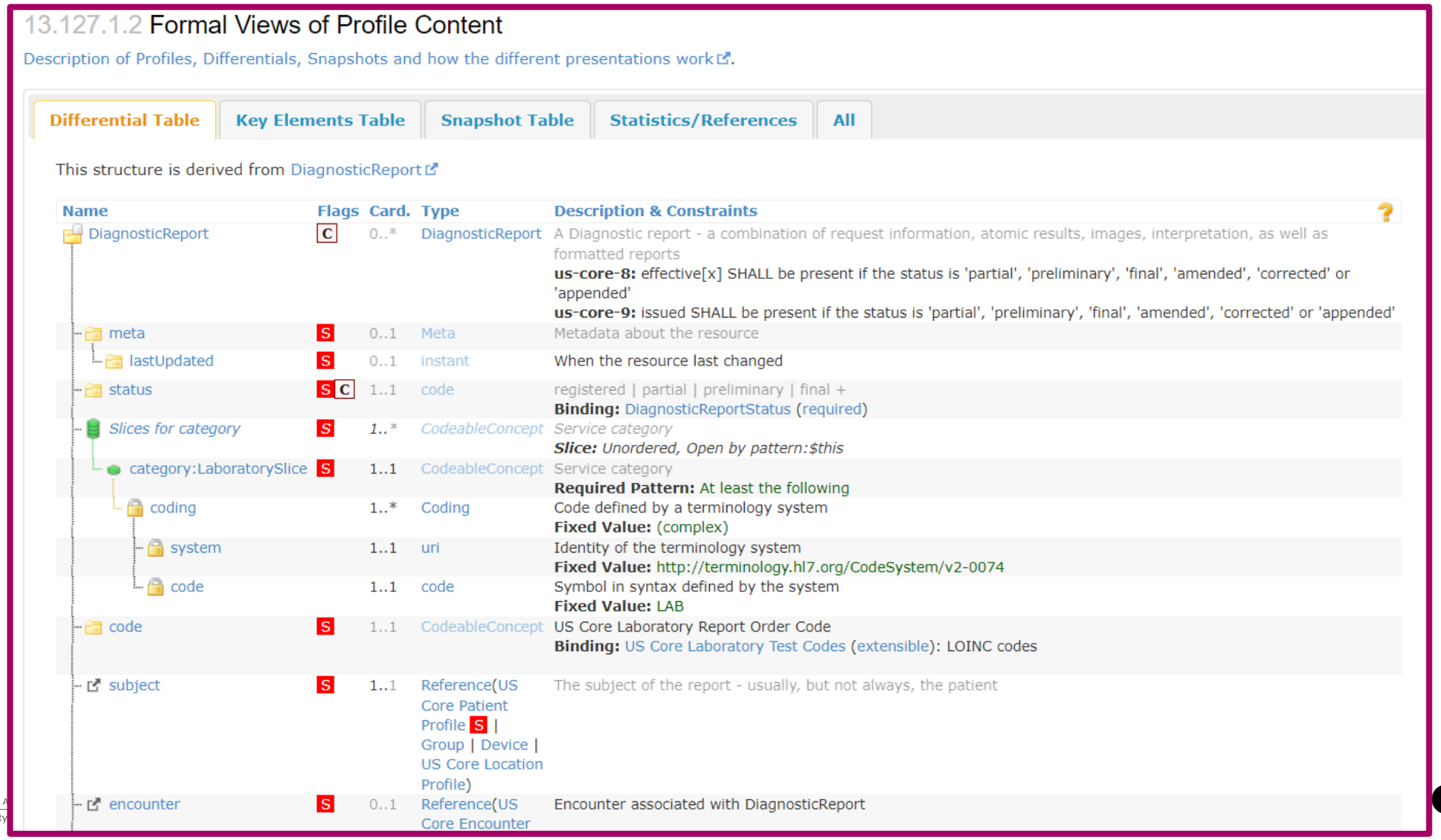
- **Ease of data extraction**
- **Exchange through API**
- **Use of services**
- **Easy for programmers to use,**
  - **even without health IT (HIT) expertise**
- **Open source (free to use)**
- **Required FHIR API available to certified products Dec 31, 2022**

HL7® FHIR®

The Office of the National Coordinator for Health Information Technology

FHIR Laboratory Reporting Profile:

https://www.hl7.org/fhir/us/core/StructureDefinition-us-core-diagnosticreport-lab.html



13.127.1.2 Formal Views of Profile Content

Description of Profiles, Differentials, Snapshots and how the different presentations work.

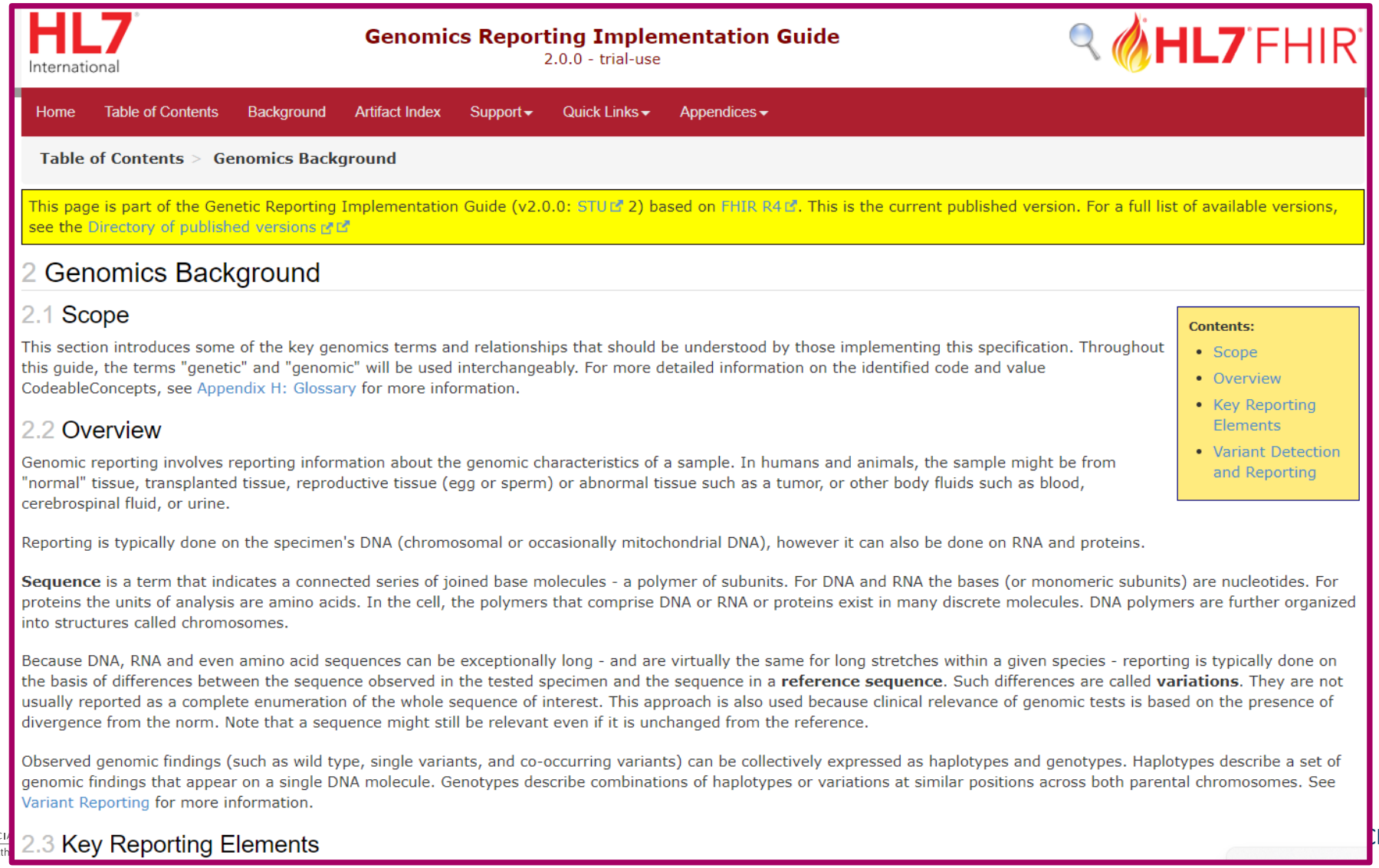
Differential Table | Key Elements Table | Snapshot Table | Statistics/References | All

This structure is derived from DiagnosticReport

Name	Flags	Card.	Type	Description & Constraints
DiagnosticReport	C	0..*	DiagnosticReport	A Diagnostic report - a combination of request information, atomic results, images, interpretation, as well as formatted reports **us-core-8:** effective[x] SHALL be present if the status is 'partial', 'preliminary', 'final', 'amended', 'corrected' or 'appended' **us-core-9:** issued SHALL be present if the status is 'partial', 'preliminary', 'final', 'amended', 'corrected' or 'appended'
meta	S	0..1	Meta	Metadata about the resource
lastUpdated	S	0..1	instant	When the resource last changed
status	S C	1..1	code	registered | partial | preliminary | final + **Binding:** DiagnosticReportStatus (required)
Slices for category	S	*1..**	*CodeableConcept*	*Service category* ***Slice:*** *Unordered, Open by pattern:$this*
category:LaboratorySlice	S	1..1	CodeableConcept	Service category **Required Pattern:** At least the following
coding		1..*	Coding	Code defined by a terminology system **Fixed Value:** (complex)
system		1..1	uri	Identity of the terminology system **Fixed Value:** http://terminology.hl7.org/CodeSystem/v2-0074
code		1..1	code	Symbol in syntax defined by the system **Fixed Value:** LAB
code	S	1..1	CodeableConcept	US Core Laboratory Report Order Code **Binding:** US Core Laboratory Test Codes (extensible): LOINC codes
subject	S	1..1	Reference(US Core Patient Profile S | Group | Device | US Core Location Profile)	The subject of the report - usually, but not always, the patient
encounter	S	0..1	Reference(US Core Encounter	Encounter associated with DiagnosticReport

Clinical Genomics Implementation Guide:

https://hl7.org/fhir/uv/genomics-reporting/



HL7 International

Genomics Reporting Implementation Guide
2.0.0 - trial-use

HL7 FHIR

Home | Table of Contents | Background | Artifact Index | Support | Quick Links | Appendices

Table of Contents > Genomics Background

This page is part of the Genetic Reporting Implementation Guide (v2.0.0: STU 2) based on FHIR R4. This is the current published version. For a full list of available versions, see the Directory of published versions

2 Genomics Background

2.1 Scope

This section introduces some of the key genomics terms and relationships that should be understood by those implementing this specification. Throughout this guide, the terms "genetic" and "genomic" will be used interchangeably. For more detailed information on the identified code and value CodeableConcepts, see Appendix H: Glossary for more information.

Contents:
- Scope
- Overview
- Key Reporting Elements
- Variant Detection and Reporting

2.2 Overview

Genomic reporting involves reporting information about the genomic characteristics of a sample. In humans and animals, the sample might be from "normal" tissue, transplanted tissue, reproductive tissue (egg or sperm) or abnormal tissue such as a tumor, or other body fluids such as blood, cerebrospinal fluid, or urine.

Reporting is typically done on the specimen's DNA (chromosomal or occasionally mitochondrial DNA), however it can also be done on RNA and proteins.

Sequence is a term that indicates a connected series of joined base molecules - a polymer of subunits. For DNA and RNA the bases (or monomeric subunits) are nucleotides. For proteins the units of analysis are amino acids. In the cell, the polymers that comprise DNA or RNA or proteins exist in many discrete molecules. DNA polymers are further organized into structures called chromosomes.

Because DNA, RNA and even amino acid sequences can be exceptionally long - and are virtually the same for long stretches within a given species - reporting is typically done on the basis of differences between the sequence observed in the tested specimen and the sequence in a **reference sequence**. Such differences are called **variations**. They are not usually reported as a complete enumeration of the whole sequence of interest. This approach is also used because clinical relevance of genomic tests is based on the presence of divergence from the norm. Note that a sequence might still be relevant even if it is unchanged from the reference.

Observed genomic findings (such as wild type, single variants, and co-occurring variants) can be collectively expressed as haplotypes and genotypes. Haplotypes describe a set of genomic findings that appear on a single DNA molecule. Genotypes describe combinations of haplotypes or variations at similar positions across both parental chromosomes. See Variant Reporting for more information.

2.3 Key Reporting Elements

HELIOS FHIR Accelerator:

https://confluence.hl7.org/display/PH/Helios+FHIR+Accelerator+for+Public+Health+Home

Helios FHIR Accelerator for Public Health Home

Created by Joshua Procious, last modified by Craig Newman on Feb 06, 2024

About Helios

The Helios FHIR Accelerator is an alliance of Government, Private Sector, and Philanthropic partners that aims to ensure public health needs are at the forefront as the FHIR standard evolves and is implemented nationwide. Helios focuses on extending and adopting existing HL7 specifications in ways that are scalable, adaptable, sustainable, and suitable for public health. Participants in Helios provide the resources and technical support needed for public health practitioners and their trading partners to develop, test, and adopt more efficient, FHIR-based ways of accessing and exchanging data nationwide. Membership is open to state, tribal, local, territorial and Federal Public Health agencies, private sector partners and other groups interested in the equitable and effective use of data for the advancement of public health.

HELIOS

The Foundations of Helios

Helios is helping public health to align with and benefit from the widespread standardization and transformation that is happening around digital health data. The Accelerator operates with three major goals in mind.

Focus on Impact

Prioritize use cases that complement what exists today and that will have an impact in their communities

Multi-Sector Alliance

Create diverse teams to work together to tackle challenges and explore new opportunities to advance interoperability

Align Efforts

Align with current FHIR activities to promote more flexible and effective data in public health and beyond

HELIOS FHIR Accelerator:

https://confluence.hl7.org/display/PH/Helios+FHIR+Accelerator+for+Public+Health+Home

Query for Supplemental Information: Newborn Screening

Note: All query workflow scenarios require successful completion of the *Match Patient and Obtain Demographic and Contact Information* workflow, above, so that the individual's Patient.id can be included in FHIR searches.

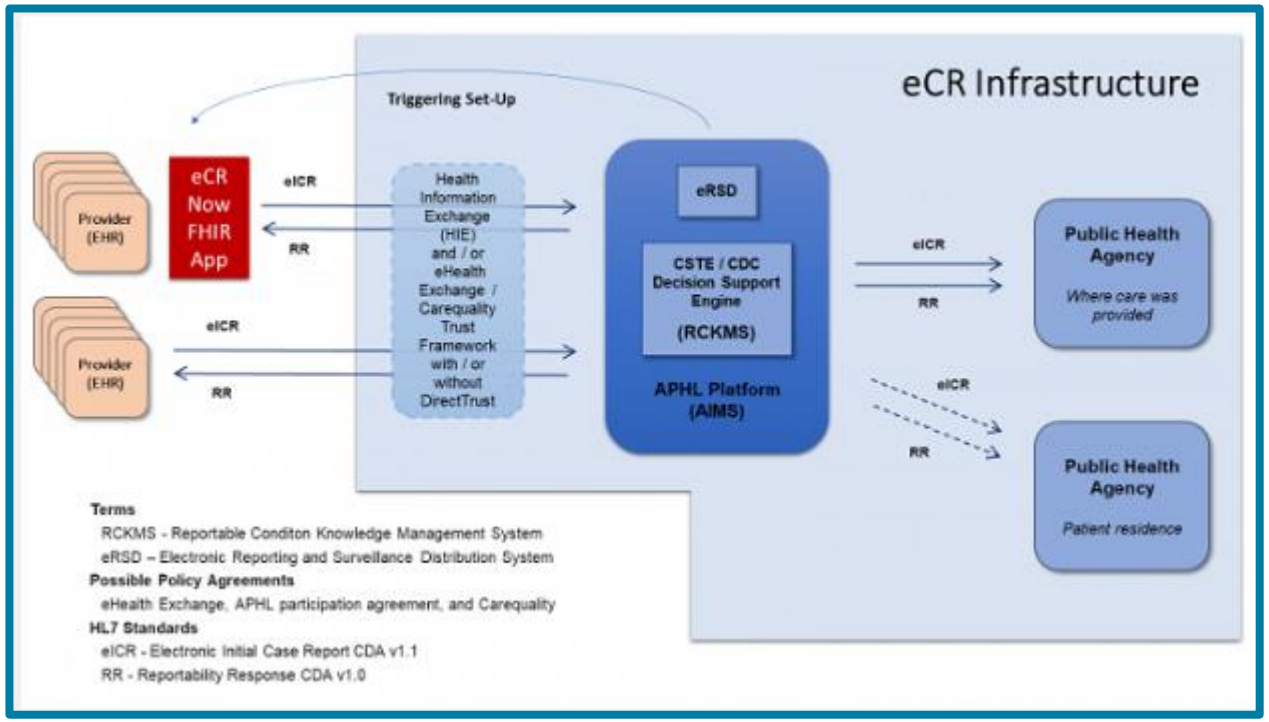
Scenario 1: Newborn Screening

- An investigator at a Newborn Screening (NBS) program queries the birthing facility's EHR for a particular newborn's screening information
- ***Action***: The investigator submits FHIR Observation searches to the EHR for screening information captured shortly after the time of birth
- ***Preconditions***:
  - The Newborn Screening (NBS) program has become aware of an infant born at a FHIR-accessible birthing facility, through an electronic notification or other means
  - The NBS has authorization to query the EHR for patient clinical information
  - The investigator or their system knows the LOINC codes associated with the desired screening information (see query content, below)
- ***Success Criteria***: The EHR returns FHIR Resources containing the desired information. (see "Sample Query Content")

Electronic Case Reporting: eCR Now

https://ecr.aimsplatform.org/general/ecr-now-fhir-app

- Automation of electronic case reporting has been very successful for reducing reporting burden
- Linkage to a bidirectional, queriable database is key to supporting patient care– it is not an optional use case
- Expanding these programs to newborn screening could improve data transmission and close gaps



Health equity and newborn screening

- Cost of genomic and other newborn screening is a major barrier to access – 90% of health center patients are at <200% of federal poverty level, majority minority pop
- Lack of access to digital information and devices, health literacy and language challenges, lack of digital skills and unmet health-related social needs can exacerbate existing health disparities as we move to electronic data approaches
- Equity first approaches should engage these communities to understand preferences, goals and how they need access

OPPORTUNITIES FOR DATA AND SYSTEMS TO SUPPORT NEWBORN SCREENING

Standardized newborn screening templates and data elements

- Requires national standardization of patient, lab, genomic data model
- Could be pushed into regulatory frameworks like USCDI+

Electronic submission of screening data

- Would depend on the infrastructure for a screening registry
- Could be done in the model of electronic case reporting or IZ Gateway by CDC

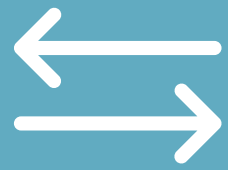
Data support for care teams

- Bidirectional push of this data to primary care teams is key
- Need to record patient consent and communication of results to families
- Maternal-fetal record linkages

Integration of patient-facing access and consent

- Patients can utilize apps to consent to data sharing, find opportunities for research participation and view their results



QUESTIONS?

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THANK YOU!

NATIONAL ASSOCIATION OF
Community Health Centers®

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