Enhancing CCHD screening by linking with birth defect surveillance

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Challenge for CCHD screening: make it work, make it quickly, make it better

Editorial

Newborn Screening for Critical Congenital Heart Disease: Essential Public Health Roles for Birth Defects Monitoring Programs

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Newborn screening for critical congenital heart defects, added in September 2011 to the Recommended Uniform Screening Panel in the United States, is a new public health priority and has particular relevance for state birth defects surveillance programs. In this commentary, we review the background to potential involvement by birth defects programs with screening, and detail key questions that these programs can evaluate: (1) health outcomes after newborn screening among affected children; (2) missed primary targets of screening (i.e., affected children who were not screened or had false-negative screens); (3) burden and screening accuracy for secondary targets; (4) the role of altitude, sociodemographic characteristics, and other special circumstances; (5) the contribution of prenatal and clinical diagnoses before newborn screening; and (6) costs and service utilization. To address these issues, monitoring programs will need to pay particular attention to: (1) data sources and quality; (2) timeliness; (3) long-term follow-up for comprehensive outcomes; (4) reporting standards; and (5) state and national program coordination. Although some aspects of involvement with these screening programs will require new partnerships and paradigm shifts in birth defects program operations, the visibility of these screening programs among stakeholders will also provide birth defects programs with new opportunities to demonstrate their usefulness. *Birth Defects Research (Part A) 00:000–000,*

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Challenge for CCHD screening: make it work, make it quickly, make it better

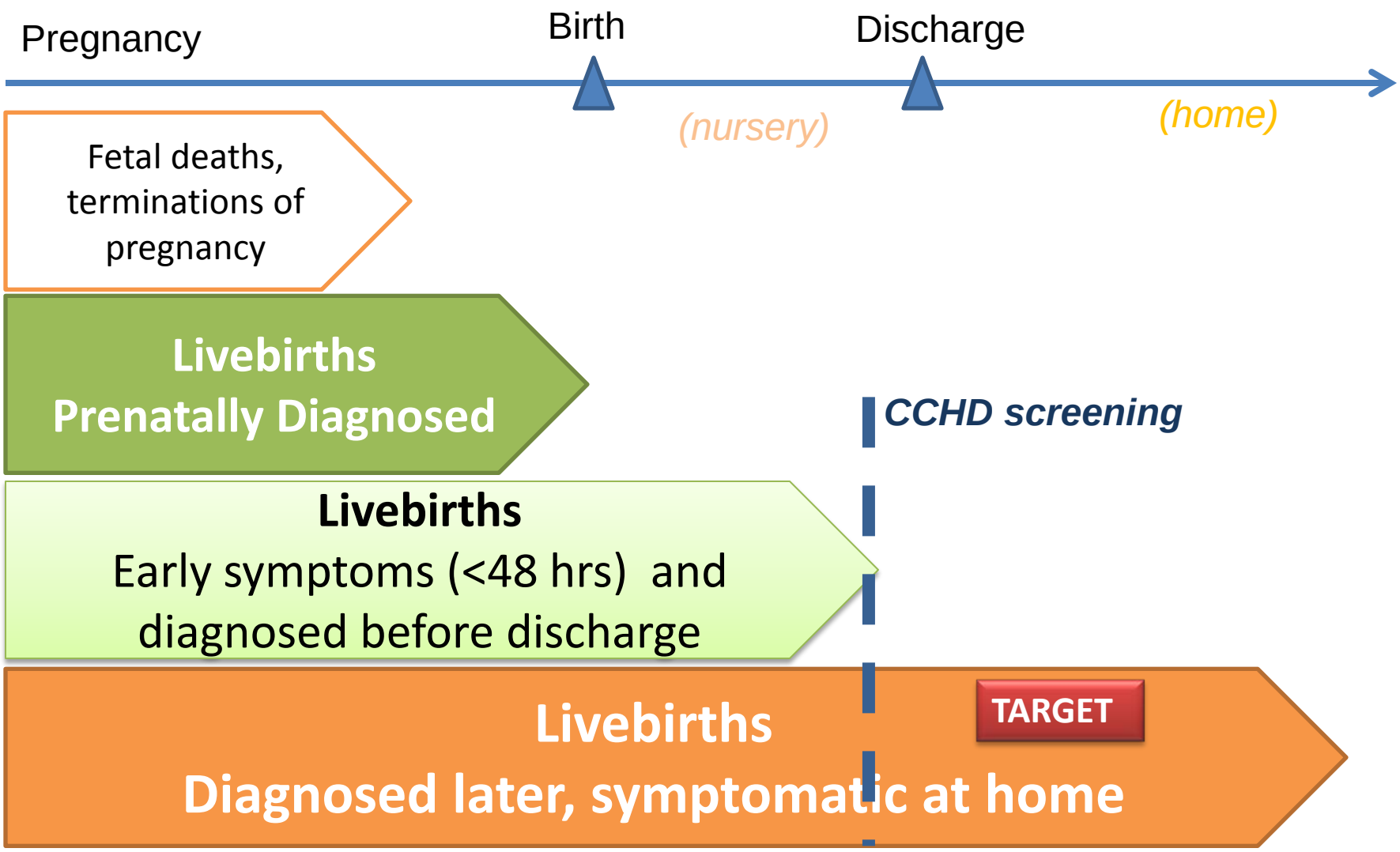
1. What is the problem ?
   – Measure and qualify

2. Is the solution working?
   - Measure and qualify

Birth defect surveillance program:

how can a partnership help?

Newborn screening and CCHD cohorts: Qualifying and Quantifying the Target Population



Basic Question # 1 for CCHD screening: What is the magnitude of the problem

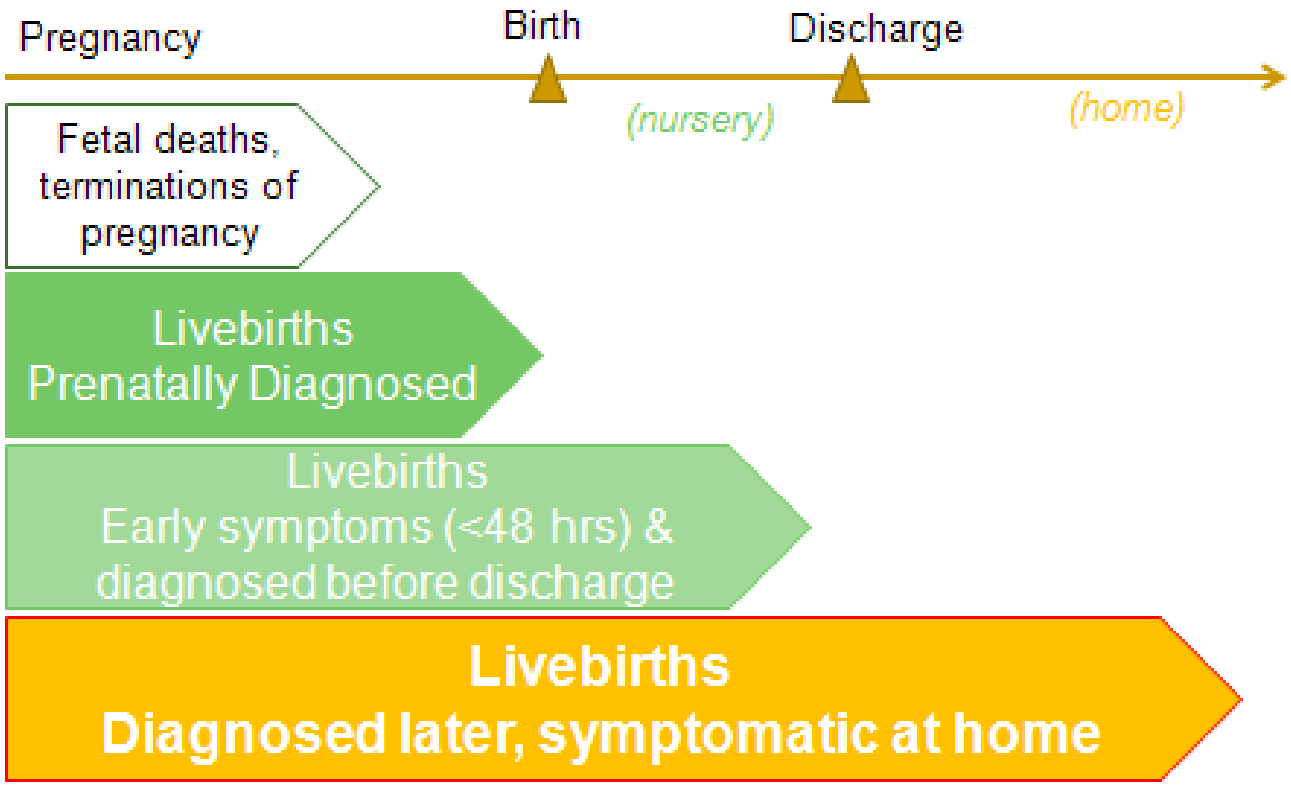
- Assess size, distribution, variation
  - How many CCHDs (birth prevalence) ?
  - Who stands to benefit (target cohort) ?
  - Is the target cohort changing (prenatal dx, length of stay) ?
  - What are the outcomes (mortality, morbidity)?
- When to find out?
  - Best before starting, but also needs tracking: helps interpret results, ongoing QI
  - We can find out! CCHD screening better positioned compared to other types of newborn screening (e.g., metabolic)

Evaluating the occurrence and impact of CCHD: assess recent cohort, trends, CCHD subgroups

Critical congenital heart defects	Number	Rate/10k	Expected/year
Restricted Set ('seven' primary)	745	12.2	**64**
d-Transposition of great arteries	148	2.4	
Tetralogy of Fallot	226	3.7	
Truncus arteriosus	39	0.6	
Hypoplastic Left Heart Syndrome	182	3.0	
Pulmonary valve atresia w/ intact septum	62	1.0	
Tricuspid valve atresia (/stenosis)	80	1.3	
Total anomalous pulmonary venous return	67	1.1	
Extended set	697	11.4	**59**
Aortic valve stenosis	233	3.8	
Coarctation of the Aorta	517	8.4	
Interrupted aortic arch (all types)	148	2.4	
Double outlet right ventricle	96	1.6	
Single ventricle, all	38	0.6	
Total	**1442**	**23.5**	**123**

Note: figures are for live births in Utah, 1999 – 2010 (n=612,789; ~ 50k/year)

Estimating the magnitude of CCHD locally: provides expectations, benchmarks, but don't forget change



Estimating the magnitude of CCHD locally: provides expectations, benchmarks, but don't forget change

Tool to estimate local situation with own data

				Total CCHD Rate (per 10,000)			
				10	**12**	**15**	**20**
			Percent total Rate				
PNDx	58%	Terminated	**20%**	2	2.4	3	4
		Not terminated	**38%**	3.8	4.56	5.7	7.6
No PnDx (born undx)	42%	Diagnosed early	**31%**	3.1	3.72	4.65	6.2
		Diagnosed late	**11%**	1.1	1.32	1.65	2.2
		Total	**100%**	10	12	15	20
			PO sensitivity				
		Rate CCHD dx earlier	**70%**	0.77	0.924	1.16	1.54
			90%	0.99	1.188	1.49	1.98
			PO sensitivity				
% of all CCHD livebirths diagnosed earlier by PO			**70%**	10%	10%	9.6%	10%
			90%	12%	12%	12.4%	12%
		Total births/year	PO sensitivity	Cases diagnosed earlier by pulse oximetry			
Small US state		53,000	Total cases born	42	51	64	85
			Total born undx	22	27	33	45
			No. infant deaths	11	13	16	21
			Dx earlier by PO (Sens. 70%)	4	5	6	8
			Dx earlier by PO (Sens. 90%)	5	6	8	10

Estimating the magnitude of CCHD locally: provides expectations, benchmarks, but don't forget change

Tool to estimate local situation with own data

Baseline 1-year mortality for CCHD			25%				
		Total births/year	If infant mortality in early vs. late dx is decreased by	Increase in number of babies surviving infancy because of PO			
Small US state		50,000	10%	1	1	2	2
			30%	3	4	5	6
			50%	5	6	8	11
			80%	8	10	13	17
Midsize US state		100,000	10%	2	2	3	4
			30%	6	7	9	12
			50%	10	12	15	20
			80%	16	19	24	32
US		4,000,000	10%	80	96	120	160
			30%	240	288	360	480
			50%	400	480	600	800
			80%	640	768	960	1,280

Birth Defect Surveillance: The Tools of the Utah Birth Defect Network

- Population-based
- Most major birth defects
  - Includes all major CHDs
- Case ascertainment
  - Active, trained abstractors (UDOH)
  - Over 100 data sources; prenatal, postnatal
- Authority via Utah Dept Health rule
  - Data kept, stored, owned by Dept of Health
- Curated, analyzed in partnership with Univ Utah
- Emphasis on completeness, accuracy, timeliness

We have data on the magnitude of the problem: How is the solution working ?

1. What is the problem ?
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Birth defect surveillance program:

how can a partnership help?

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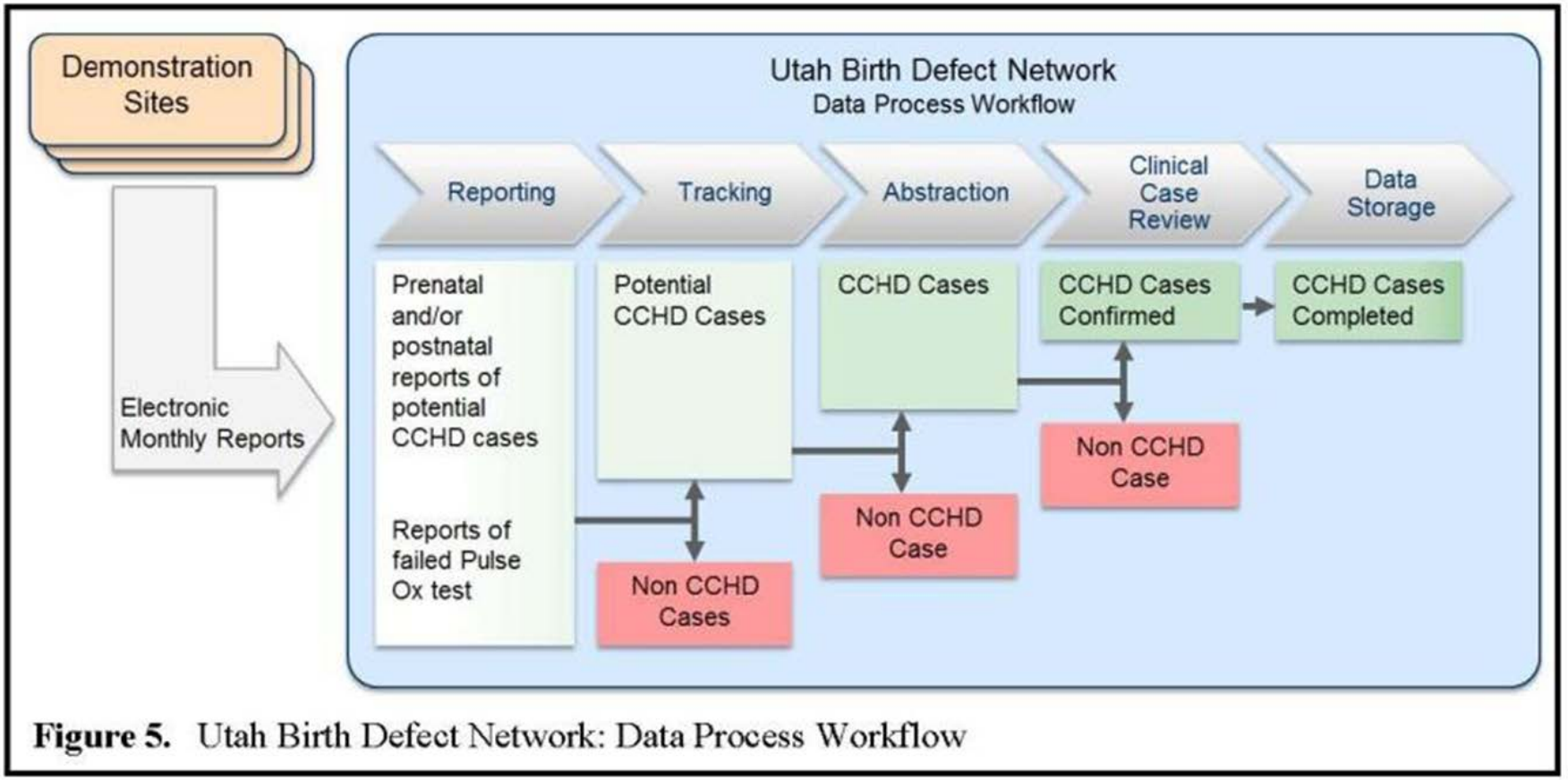
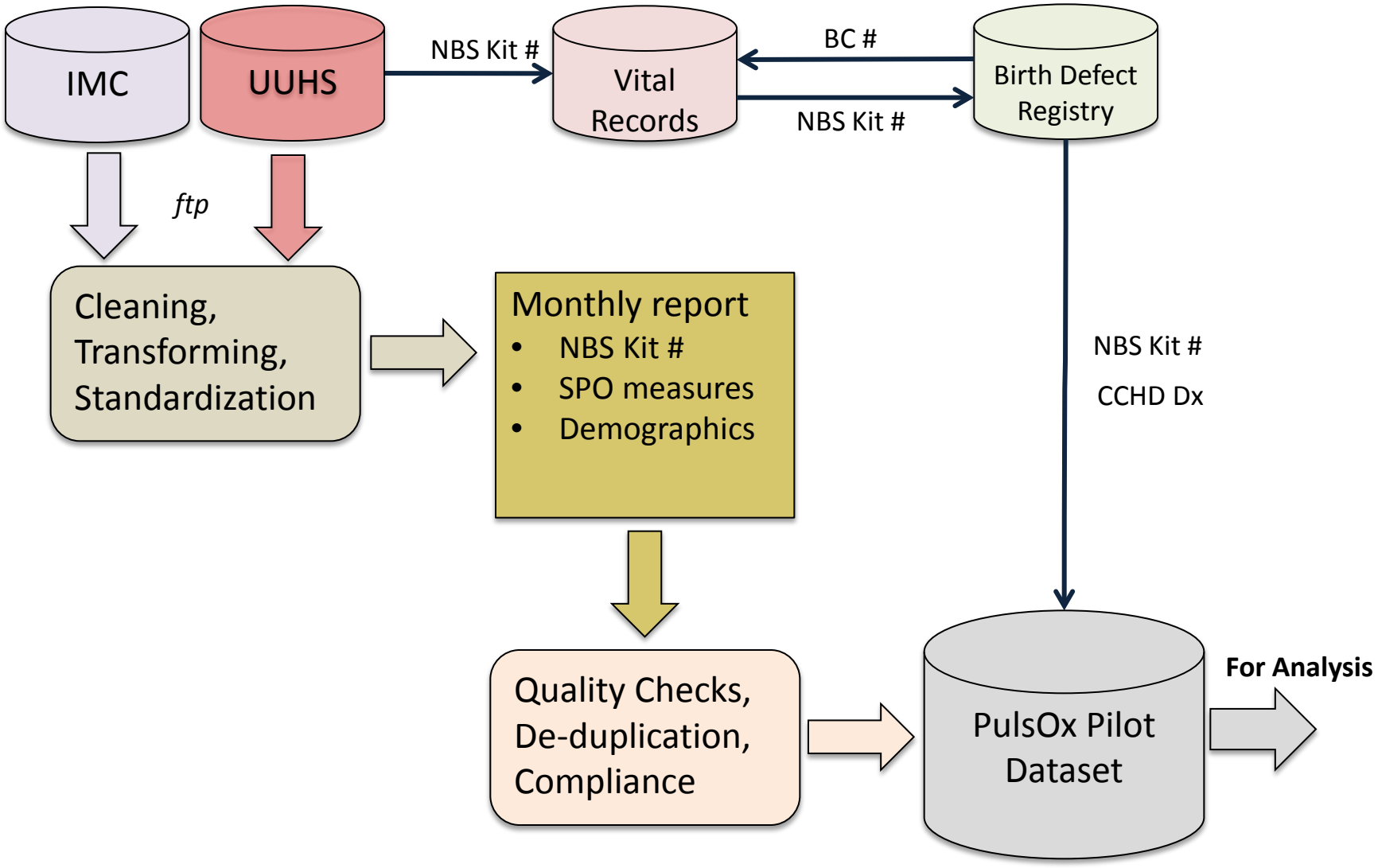


Figure 5. Utah Birth Defect Network: Data Process Workflow

	True CCHD	Other disease	Unaffected
Screen +	True Pos (found)	?	False Pos
Screen -	False Neg (missed)	?	True Neg

Data Flow for CCHD Pilot
Technical analysis, then match with BD surveillance



	True CCHD	Other disease	Unaffected
Screen +	True Pos (found)	?	False Pos
Screen -	False Neg (missed)	?	True Neg

- **Challenges**
  - Authority to review all failed pulse ox (not only CHD)
  - Faster reports, quality of CCHD data (every case counts)
  - Matching screening data with cases (different data flows)
  - More clinical case review: potential cases, failed pulse ox (not necessarily CCHDs)
- **What helps**
  - Authority: reporting rule for review of failed pulse ox
  - Speed: champions at sites, fast track review centrally
  - Matching IDs: MRN, demographics, NBS kit #, BC #
  - Close partnership NBS & BD: talk the talk, but walk the walk

Birth defect surveillance for CCHD screening: Promoting Completeness, Accuracy, Timeliness

Elements of surveillance	Comment
Population –based	Monitor entire population, decrease potential biases
Active case ascertainment	Promotes completeness, accuracy, possibly timeliness; easier to add to data collection (e.g., day of discharge)
Multiple sources	Promotes accuracy and completeness (e.g., ped cardiology, birthing centers, pathology)
Verbatim descriptions, not only codes	Improves accuracy and completeness: codes may be inaccurate or inefficient in describing phenotype
Clinical case review	Accuracy and detail: type of CHD (not always easy), presence of extra-cardiac anomalies or syndromes, but requires clinical expertise
Timely analysis and dissemination	Use the data for action: timely dissemination to those who need to know, in appropriate format and content (public, hospitals, health officials, professionals, etc.)

Making CCHD screening work quickly and well: additional data and lessons learned (and still learning)

- What else is being done in Utah pilot project
  - Nomograms (normal values) at altitude
  - Time and motion study
  - Cost evaluation
  - Eventually, evaluation of changes in outcomes
- Some lessons learned in pilot project
  - Crucial to have integrated team: be humble, get all the skills in the room from wherever they are available
  - Goodwill goes only so far: plan for a sustainable system (resources, public health authority, data feeds, QI/QC)
  - Can/should be a win-win: better NBS, better BD surveillance
  - It can be done

Thank you

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