Northwest Regional Newborn Screening Program

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What is Newborn Screening?

- Began in 1960's
- State-based public health program
- Practice of testing every newborn for harmful or fatal conditions not otherwise apparent
- 2006: Federal Health Resources and Services Administration convened experts to address state-to-state variation and develop core conditions for "panel"

Ten Great Public Health Achievements --- US, 2001-2010

"Improvements in technology & endorsement of a uniform newborn-screening panel of diseases have led to earlier life-saving treatment and intervention for at least 3,400 additional newborns each year with selected genetic and endocrine disorders….By April 2011, all states reported screening for at least 26 disorders on an expanded and standardized uniform panel."

-- *Centers for Disease Control and Prevention*.

Overview

- Statutory authority
- Purpose / process of newborn screening
- Overview of regional program
- National and Oregon screening panel
- Funding

Oregon Newborn Screening Statute

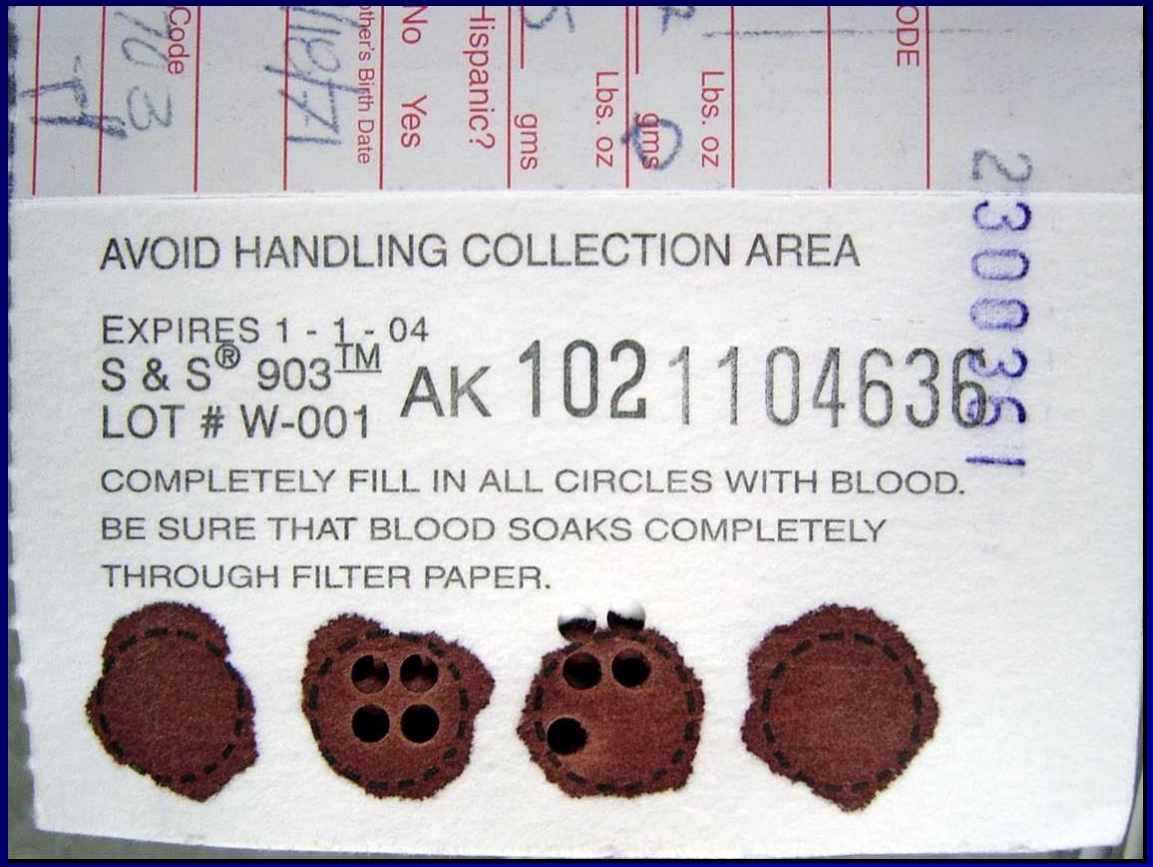
- 433.285 (1)…in the interest of public health and the prevention of mental retardation, every infant, shall be given tests approved by the Oregon Health Authority for the detection of the disease of phenylketonuria and other metabolic diseases.
- (2) The authority by rule shall specify the diseases for which infants shall be tested…the persons responsible for submitting the specimens, the methods of testing and the manner of payment of the fees.

Purpose of screening: To identify asymptomatic infants who need prompt treatment to prevent severe disability or death

Characteristics of Screened Disorders

- All infants are screened for 40+ disorders
- Untreated, cause serious disability/ death
- Can be prevented or controlled if detected before disease develops
- Most have a genetic basis
- Many are result of defective enzyme in a key metabolic pathway

Samples submitted by hospitals, midwives, physicians



Oregon Public Health Laboratory

- Communicable disease testing to support public health
- Newborn Screening
- Regulation of clinical and environmental laboratories

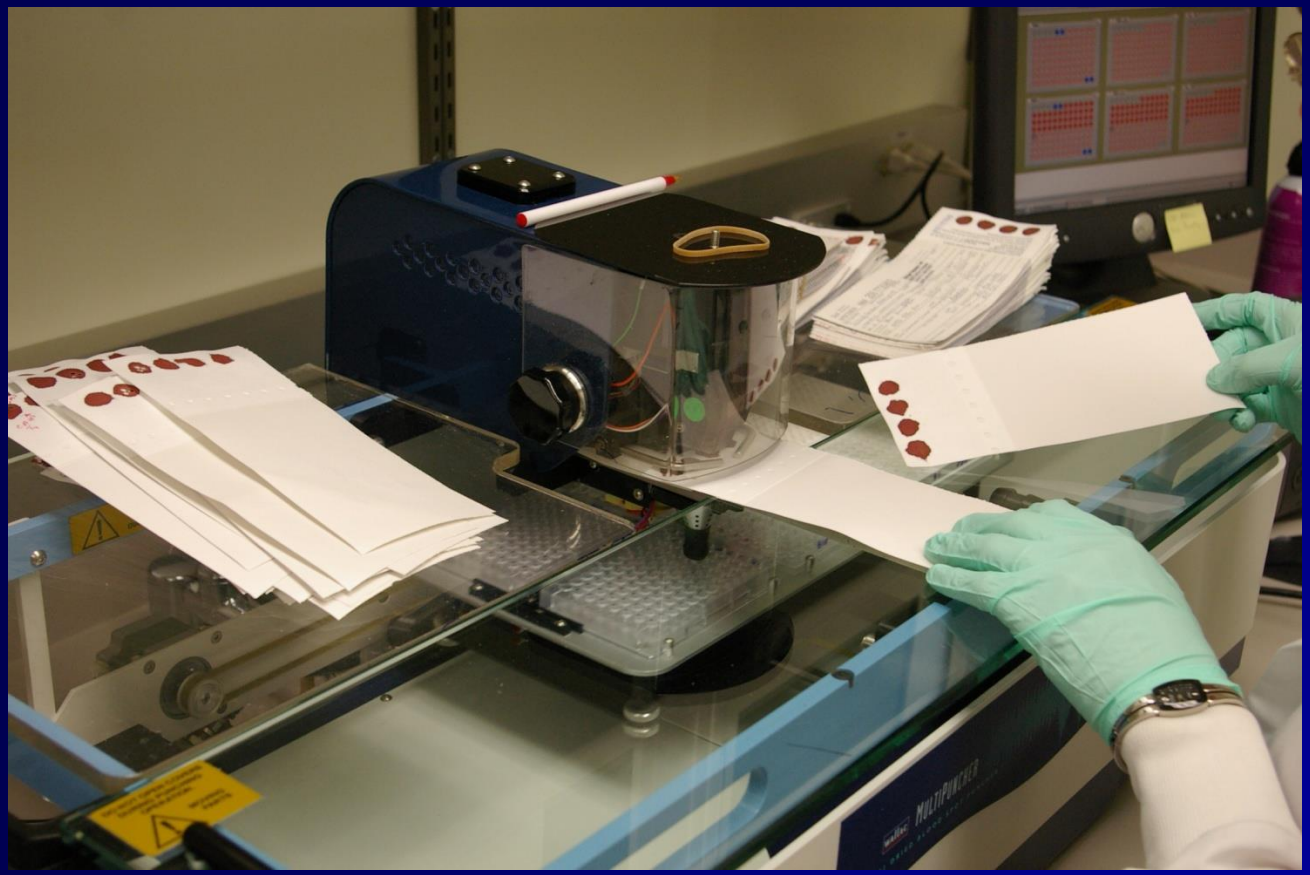
Newborn Screening Program

- Practitioner education
- Parent information
- Laboratory testing
- Tracking and follow-up
- Medical consultation (OHSU)

Newborn Screening Laboratory

Oregon Health Authority

Uniform Standards for Specimen Collection, Processing, Testing



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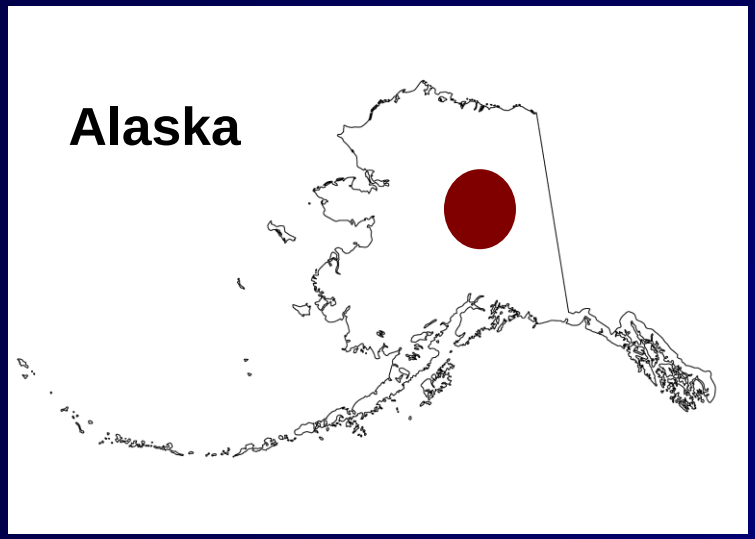

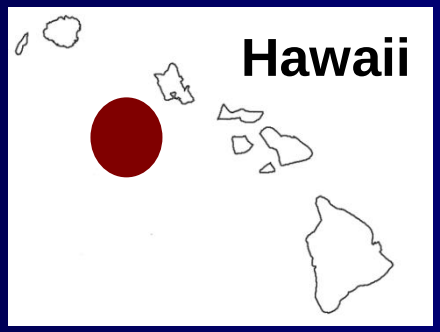

National Rec'd Uniform Screening Panel (RUSP)

Disorder nominated: Secretary's Advisory Committee Heritable Disorders in Newborns & Children

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Evidence-based review of scientific data regarding efficacy of screening and treatment

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SACHDNC recommends addition of disorder to RUSP to Secretary of US DHHS

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Secretary accepts recommendation; adds disorder to RUSP

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State NBS programs add disorder to screening panels when feasible

US DHHS Rec'd Uniform Screening Panel Core Conditions (N=29)

- Metabolic disorder
  - Organic acid condition
  - Fatty acid oxidation disorder
  - Amino acid disorder (e.g. PKU)
- Endocrine disorder (e.g. hypothyroidism)
- Hemoglobin disorder (e.g. sickle cell)
- Other disorders (e.g. cystic fibrosis)

NW Regional Newborn Screening Program
Disorders Detected 1962 - March 2015

States	AK	HI	ID	NV	OR	MT	DE	MISC	NM	
Years Screened	1975-1983 1987-2015	1997-2015	1976-2015	1978-2014	1962-2015	1975-1985	1992-1998	1991-2015	2007-2015	**1962-March 2015**
Disorders										
PKU	25	6	98	64	189	7	8	14	3	**414**
CH	139	124	223	322	708	20	12	54	113	**1715**
GAL	3	2	15	16	35	2	2	2	2	**79**
Biotinidase	2	4	6	13	16	0	3	2	1	**47**
CAH	51	13	18	18	48	0	0	7	15	**170**
SS DIS	9	12	4	156	42	0	55	17	10	**305**
Amino Acids	4	6	1	7	8	0	0	1	4	**31**
Urea Cycle	1	3	4	5	10	0	0	0	0	**23**
FAOs	23	22	37	5	69	0	0	5	15	**176**
Organic Acids	20	12	15	23	30	0	0	2	14	**116**
Cystic Fibrosis	12	6	44	24	92	0	0	5	26	**209**
SCID	0	0	0	0	2	0	0	0	0	**2**
TOTAL	**289**	**210**	**465**	**653**	**1249**	**29**	**80**	**109**	**203**	**3,287**
Infants Screened	365,707	324,659	791,135	926,917	2,074,612	165,342	72,181	111,018	216,289	**5,047,860**

Frequency: 1 in 1,536 births

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Newborn Screening Program Funding

Supported by fees paid by practitioners:

- For Oregon practitioners, newborn screening test kits purchased after May 1, 2014:
  - $32 per one-specimen kit; or
  - $64 per two-specimen kit; or
  - $64 per three-specimen kit (neonatal intensive care unit / special baby care unit use only).
- Other states pay through contracts with OHA.

Questions?

Oregon list of disorders (1)

(1) Cystic fibrosis (CF).

(2) Endocrine disorders:

(a) Congenital hypothyroidism (CH);

(b) Congenital adrenal hyperplasia (CAH).

(3) Galactosemia (GALT).

(4) Hemoglobin disorders:

(a) Sickle cell disease (Hb S/S);

(b) Sickle cell/beta thalassemia (Hb S/A); and

(c) Sickle cell/hemoglobin C disease (Hb S/C).

Oregon list of disorders (2)

(5) Metabolic disorders:

(a) Amino acid disorders:

(A) Homocystinuria (HCY);

(B) Phenylketonuria (PKU); and

(C) Tyrosinemia (TYR).

(b) Biotinidase deficiency;

(c) Fatty acid oxidation disorders:

(A) Carnitine uptake defect (CUD);

(B) Carnitine/acylcarnitine translocase deficiency (CT);

(C) Carnitine palmitoyl transferase deficiency (CPT)

(D) Glutaric acidemia, Type II (GA-II);

(E) Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);

(F) Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);

(G) Short-chain acyl-CoA dehydrogenase deficiency (SCAD);

(H) Trifunctional protein deficiency (TFP); and

(I) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).

Oregon list of disorders (3)

(d) Organic acid disorders:

(A) Beta-ketothiolase deficiency (BKT);

(B) Glutaric acidemia, Type I (GA-I);

(C) Isobutryl-CoA dehydrogenase deficiency (IBG);

(D) Isovaleric acidemia (IVA);

(E) Malonic aciduria (MAL);

(F) Maple syrup urine disease (MSUD);

(G) Methylmalonic acidemia (MMA);

(H) Propionic acidemia (PA);

(I) 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency (2M3HBA);

(J) 2-Methylbutyryl CoA dehydrogenase deficiency (2MBG);

(K) 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG);

(L) 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC);

(M) 3-methylglutaconyl-CoA hydratase deficiency (3MGA); and

(N) Multiple carboxylase deficiency (MCD).

Oregon list of disorders (4)

(e) Urea Cycle Disorders:

(A) Arginase deficiency (ARG);

(B) Argininosuccinate lyase deficiency (ASA); and

(C) Citrullinemia, Type I (CIT I).

(6) Other disorders as defined by Oregon Health Authority.

(7) Severe combined immunodeficiencies (SCID).