



Review of Up and Coming Disorders and the Future of the RUSP

Metachromatic Leukodystrophy

APHL New Disorders Workgroup Winter Webinar Series, Pt 3
February 18, 2021

Welcome



- 2 children diagnosed with juvenile MLD in 1995
 - Lindy diagnosed at 14 in 1995, 6-year diagnostic odyssey, docs gave us 4-5 years ... she's 40 now
 - Darcee diagnosed at 10, experimental bone marrow transplant
- MLD Foundation launched in 20 years ago ... 2001



We C.A.R.E.™ ... Compassion for families, increasing Awareness, influencing & funding Research, and promoting Education

- Global impact with families, researchers, biopharma, regulators, and policy

Metachromatic Leukodystrophy



	Incidence - Births			Prevalence - Alive			
	USA	More Developed Countries	Global	USA	More Developed Countries	Global	lifespan
Late Infantile (50-60%)	63	190	2,000	380	1,150	12,000	7
Juvenile (20-30%)	28	90	900	540	1,650	3,600	20
Adult (10-20%)	23	70	730	1,120	3,500	37,000	50
Total	114	350	3,630	2,030	6,300	52,600	

© 2021 MLD Foundation. Please contact us for further details and assumptions. "More Developed Countries" as defined by the United Nations. ver 2021-01, some numbers rounded.

- Autosomal recessive – 300+ mutations
 - A handful are common ... expect 50-60% high confidence guidance
 - Sequencing is tier 3 of current MLD flow
- Incidence 1:40,000 ... carrier prevalence 1 in 100

MLD Therapy status



- **Stem cell transplant** ... today's standard, 4-5 US expert centers
- **Gene therapy (Orchard)**
 - approved in EU 12/20 ... results extraordinary
 - studied by comparing untreated older siblings
 - coming to the US ... IND discussions in process
 - multiple biopharma in this space
- **Enzyme replacement therapy** in clinical trial

Efficacy



Early diagnosis empowers families & optimizes outcomes ...

Early Diagnosis = Normal Life!

Pre- symptomatic

Efficacy



Early diagnosis empowers families & optimizes outcomes ...

Early Diagnosis = Normal Life!

*Pre-**progression***

Pilot Status



- Deidentified pilot started in 2016 ... U Wa. – Gelb lab
 - ms/ms sulfatide first tier
- Identified pilots
 - Germany (2020)
 - ScreenPlus – New York (Q1 2021)
 - Italy/EU
 - other sites

RANSIP Overview

RUSP Approval and Newborn Screening Implementation



Mission

To facilitate the preparation of a **RUSP Nomination and Approval ...** representing a **coordinated effort and collaboration** of the MLD and NBS ecosystems.

Further ... to use the **RUSP Nomination and other resources** along with a **coordinated effort** to ... **empower and help constituents of every state and country to implement MLD newborn screening.**

RANSIP Timing

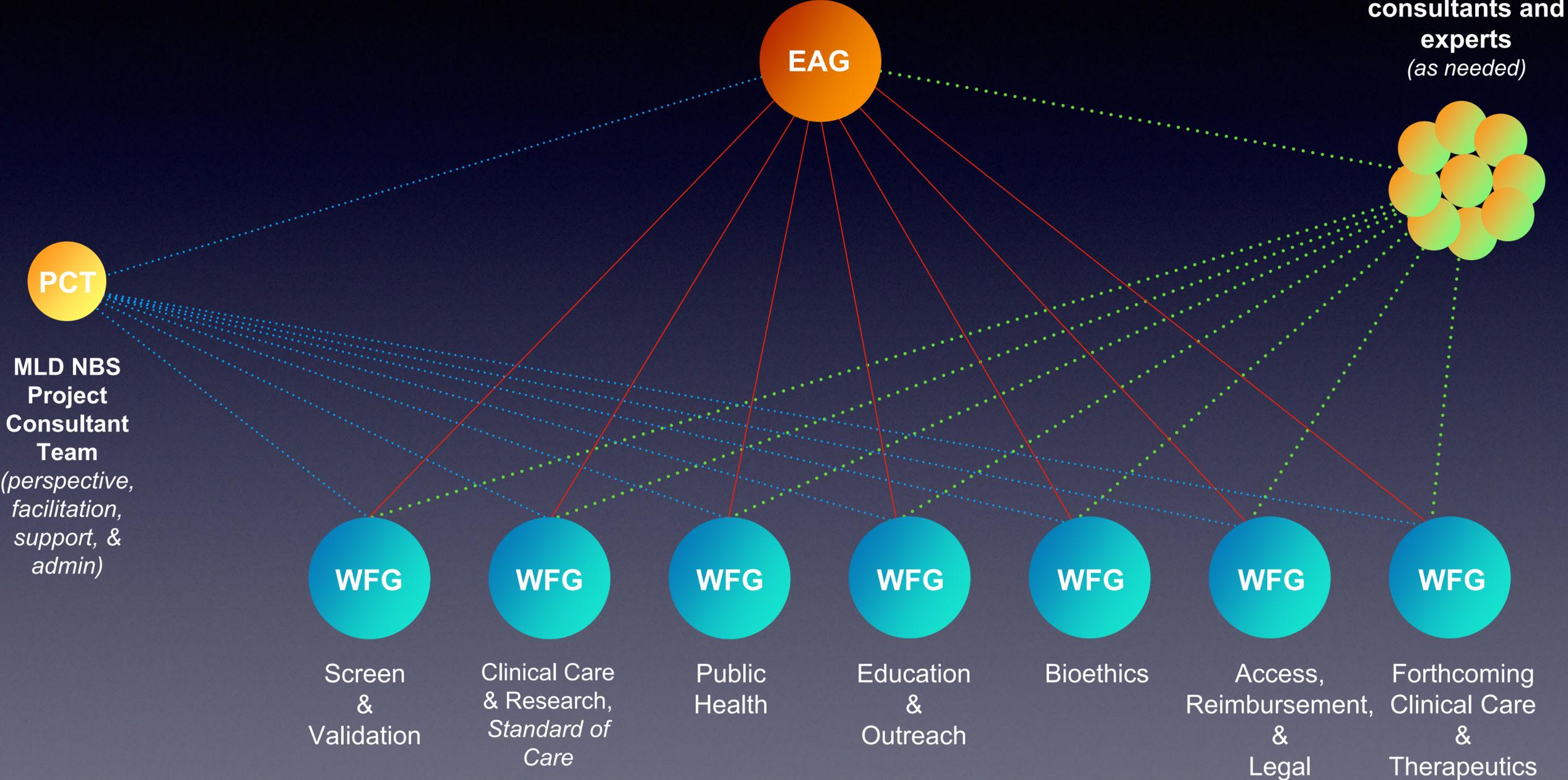


- **2020 ... launched RUSP Approval and Newborn Screening Implementation Program (RANSIP)**
 - Feb. ... formed **MLD Expert Advisory Group**
 - June ... launched **7 Working Focus Groups**
- **2021 ... launching state-by-state implementation**

Organization



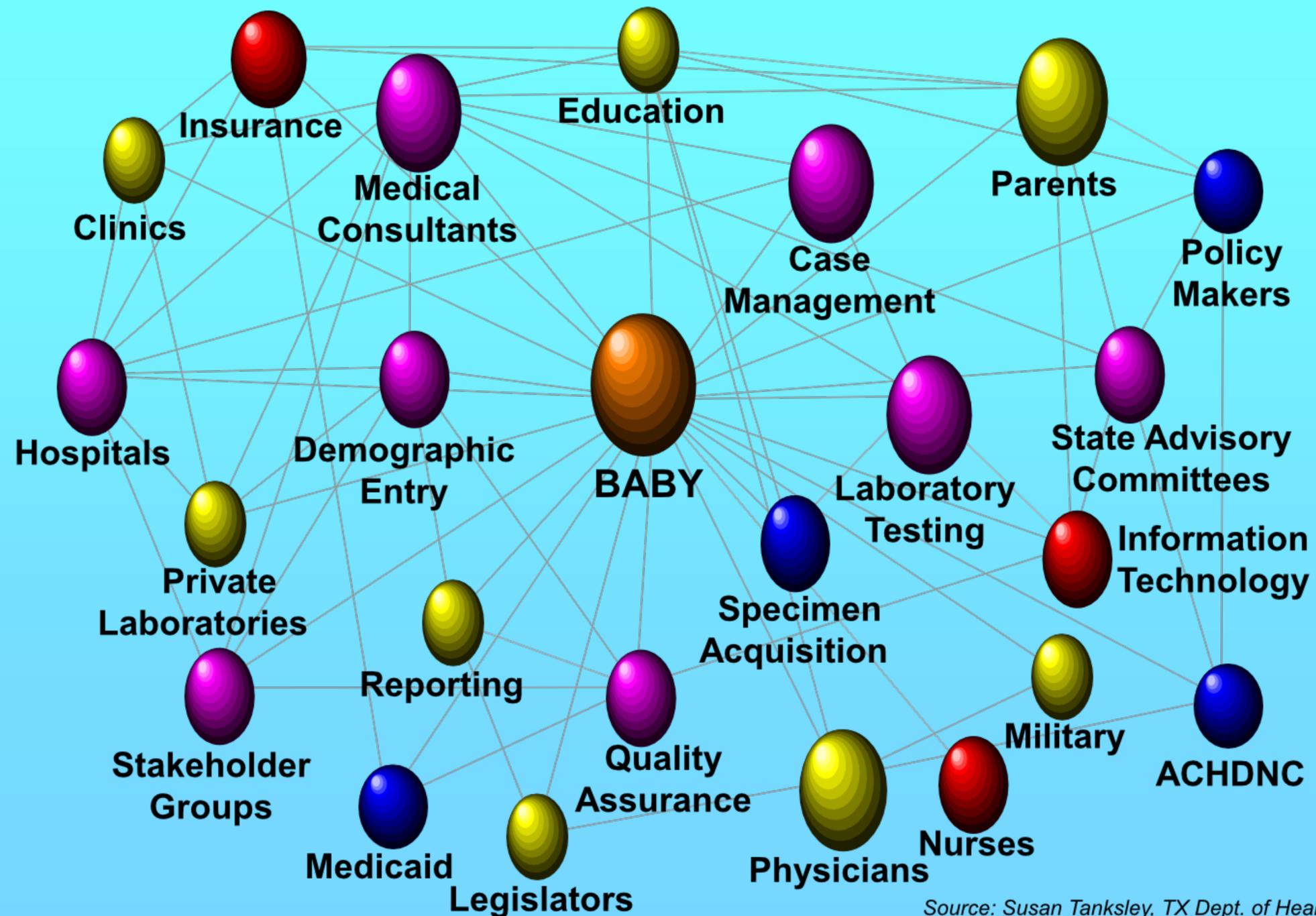
Expert Advisory Group



Working Focus Groups



Newborn Screening Ecosystem

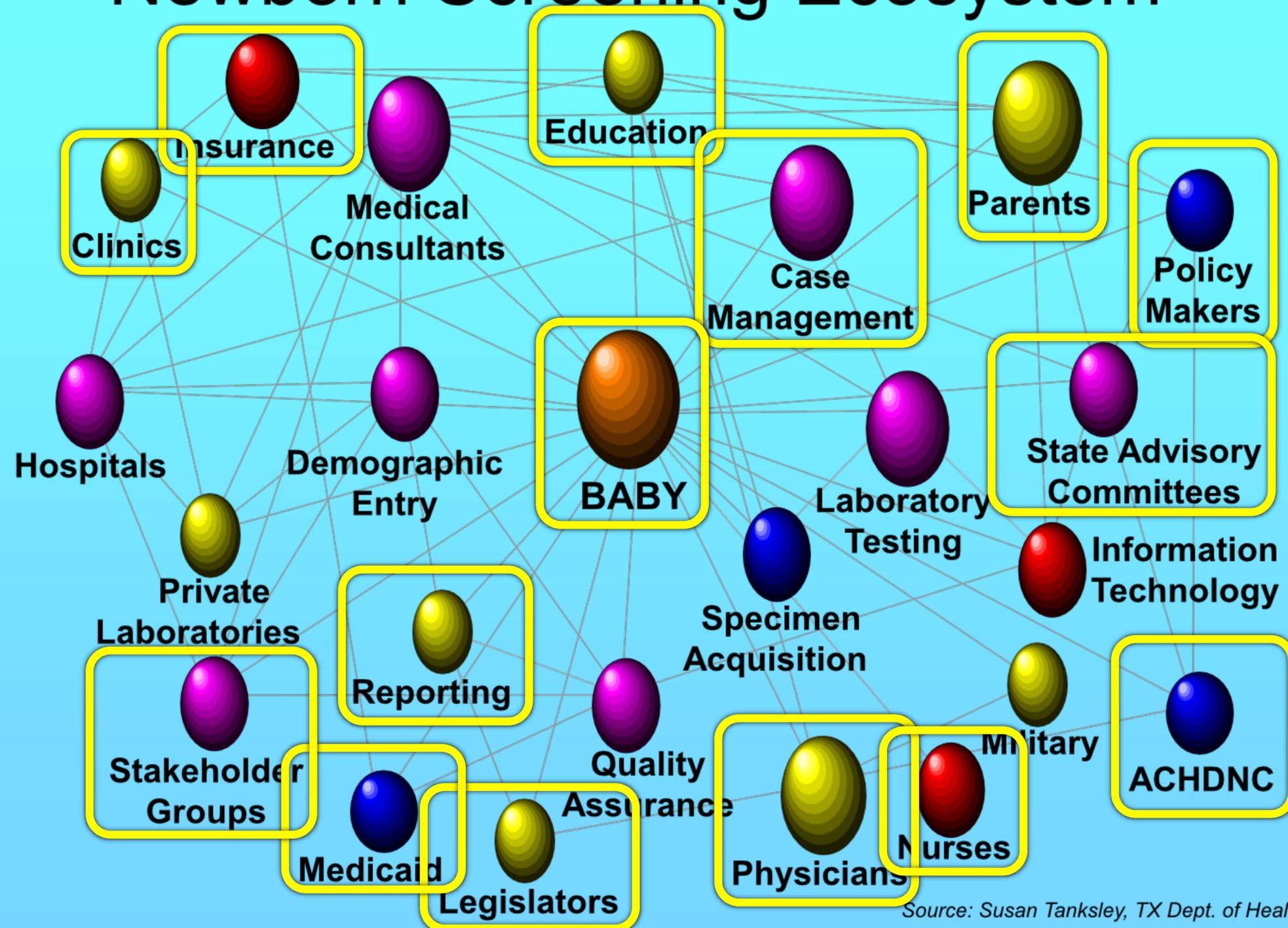


Source: Susan Tanksley, TX Dept. of Health

Advocacy ... we can help!



Newborn Screening Ecosystem



Source: Susan Tanksley, TX Dept. of Health

plus
clinical care,
therapies,
& LTFU...



Discussion



- Role of advocacy in public health
- Policy – RUSP alignment & appropriations
- Value of finding a baby before RUSP Nomination
- Tsunami of new therapies & RUSP nominations
- Long Term Monitoring & Follow Up

We C.A.R.E.™

facilitating **Compassion**

increasing **Awareness**

influencing **Research**

promoting **Education**

... for Metachromatic Leukodystrophy

