

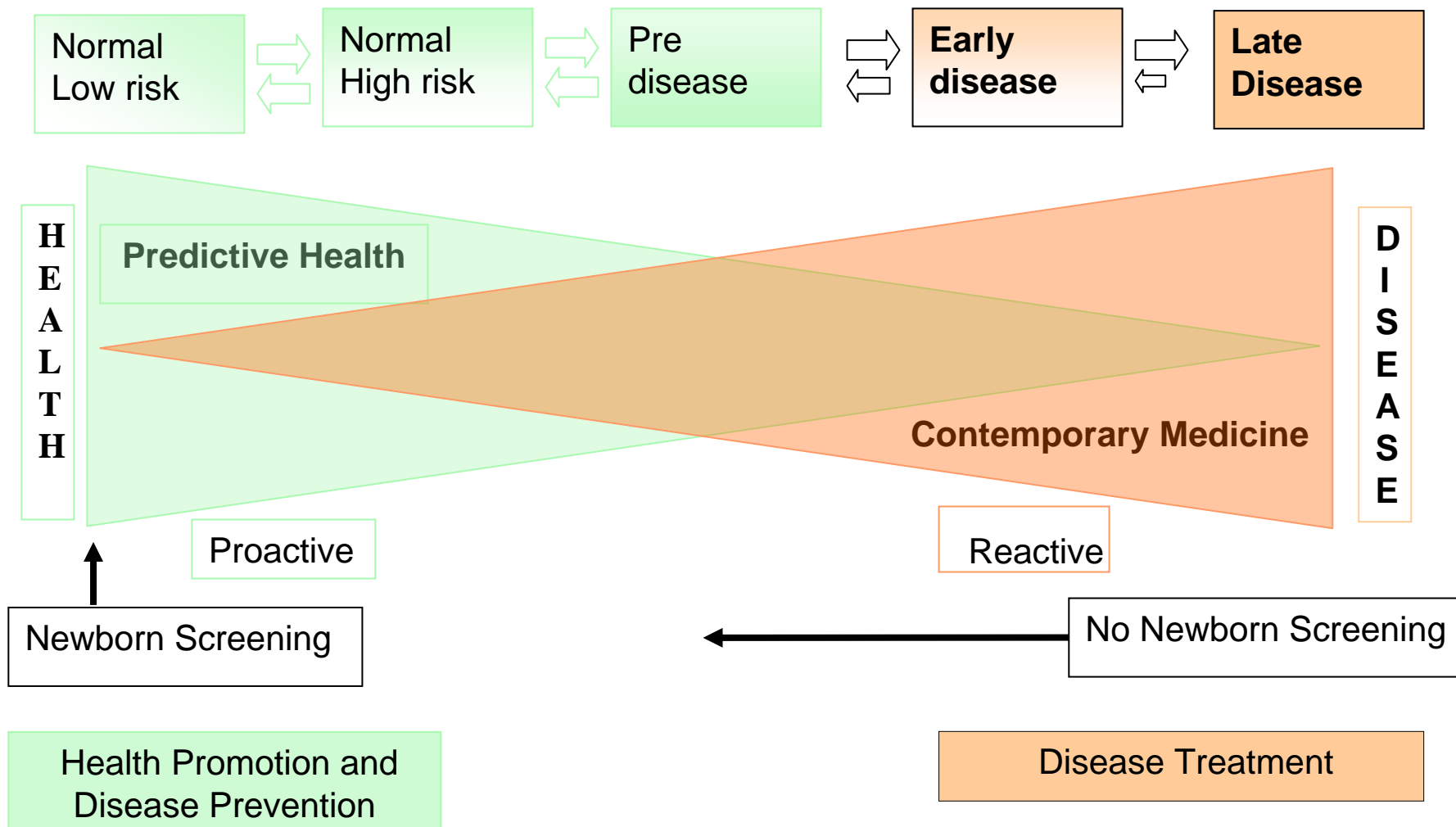


# SERGG/Region 3

## Long Term Follow-Up, An Overview

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# The Health/Disease Continuum & Nutrition Intervention for NBS



# HRSA/ACMG Uniform Panel (MS/MS)

## Amino Acid Disorders

## Fatty Oxidation Disorders

## Organic Acid Disorders

U  
N  
I  
F  
O  
R  
M

P  
A  
N  
E  
L

Phenylketonuria  
MSUD  
Homocystinuria  
Tyrosinemia type I  
Argininosuccinic acidemia  
Citrullinemia type I

MCAD deficiency  
VLCAD deficiency  
LCHAD deficiency  
TFP deficiency  
Carnitine uptake defect

Isovaleric acidemia  
Glutaric acidemia type I  
HMG deficiency  
3MCC deficiency  
BKT deficiency  
Multiple carboxylase deficiency  
Methylmalonic acidemia (MUT)  
Methylmalonic acidemia (Cbl A,B)  
Propionic acidemia

S  
E  
C  
O  
N  
D  
A  
R  
Y

T  
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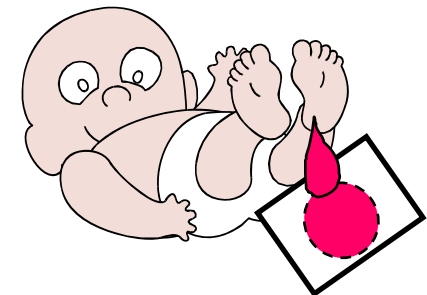
Hyperphenylalaninemia  
Tyrosinemia type II  
Biopterin defects (Bios)  
Tyrosinemia type III  
Biopterin (Reg)  
Argininemia  
Hypermethioninemia  
Citrullinemia type II

M/SCHAD deficiency  
SCAD deficiency  
MCKAT deficiency  
CPT-I deficiency  
Glutaric acidemia type II  
CACT deficiency  
Dienoyl red. deficiency  
CPT-II deficiency

Methylmalonic acidemia (Cbl A,B)  
2M3HBA deficiency  
IBG deficiency  
2MBCAD deficiency  
Methylglutaconic acidemia  
Malonic acidemia

**20 Primary targets**

**22 “Secondary” targets**



# The Growing Need for tracking Long-Term Follow-up



- Inequities among states in their long-term management of disorders
- Expansion of diagnosis with expanded NBS panel
- Lack of information, training, varying resources, inequitable access to care
- Poor transitioning throughout life cycle.
- Poor care coordination
- Lack of data tracking to build evidence

# Outline of Region 3 Project



- Region 3 vision of long term follow-up
- Project activities planned

# A long-term f/u program must:



- Integrate the public health infrastructure and private sector (public-private partnership)
- Incorporate medical home and support services, transitioning, emergency preparedness issues
- Provide timely management and treatment services
- Develop data systems for monitoring outcomes and quality improvements
- Utilize robust communications to facilitate and maintain access to services
- Remain flexible to accommodate rapid technology and knowledge change

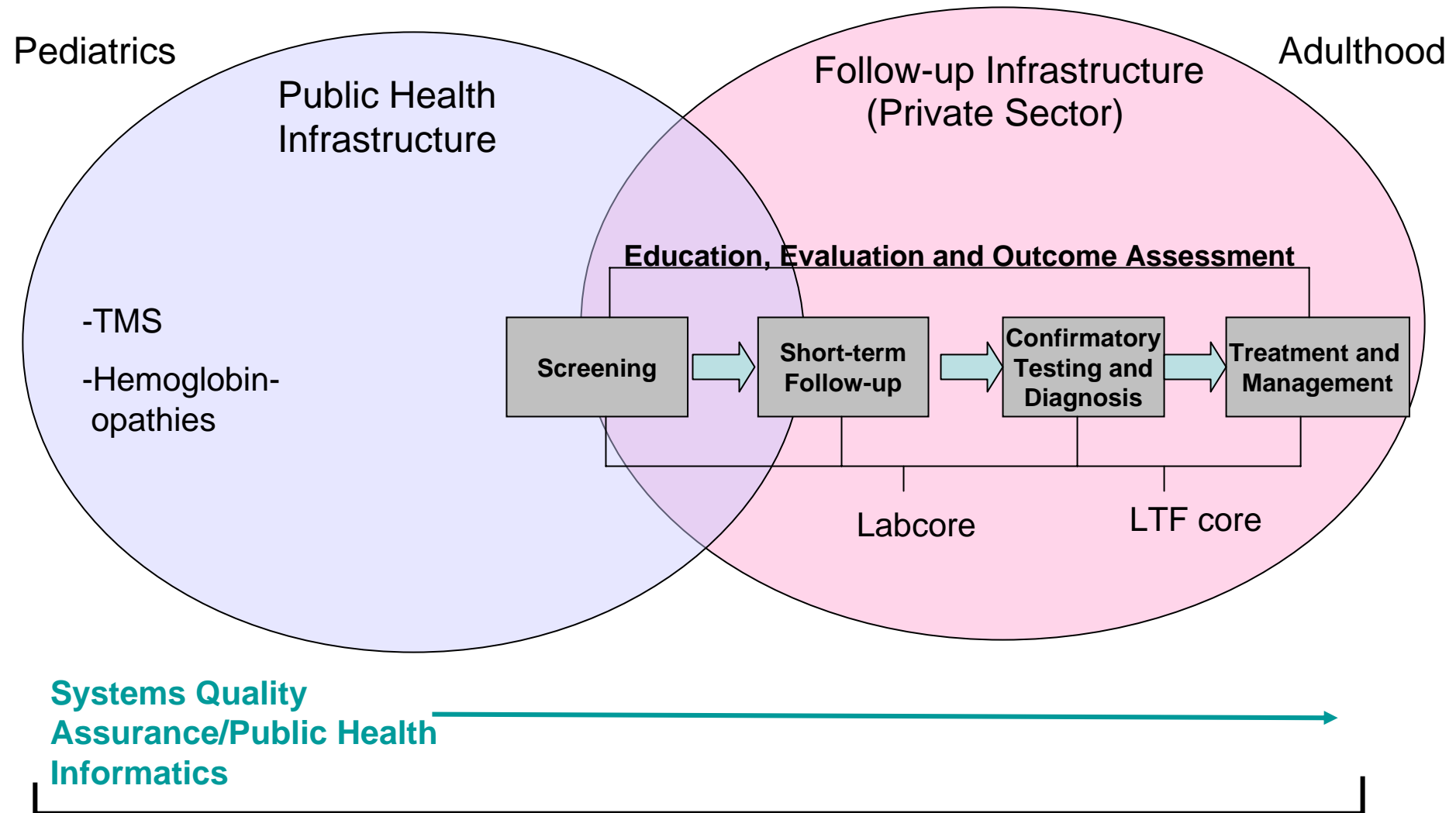
# Goals



Construct an information system to:

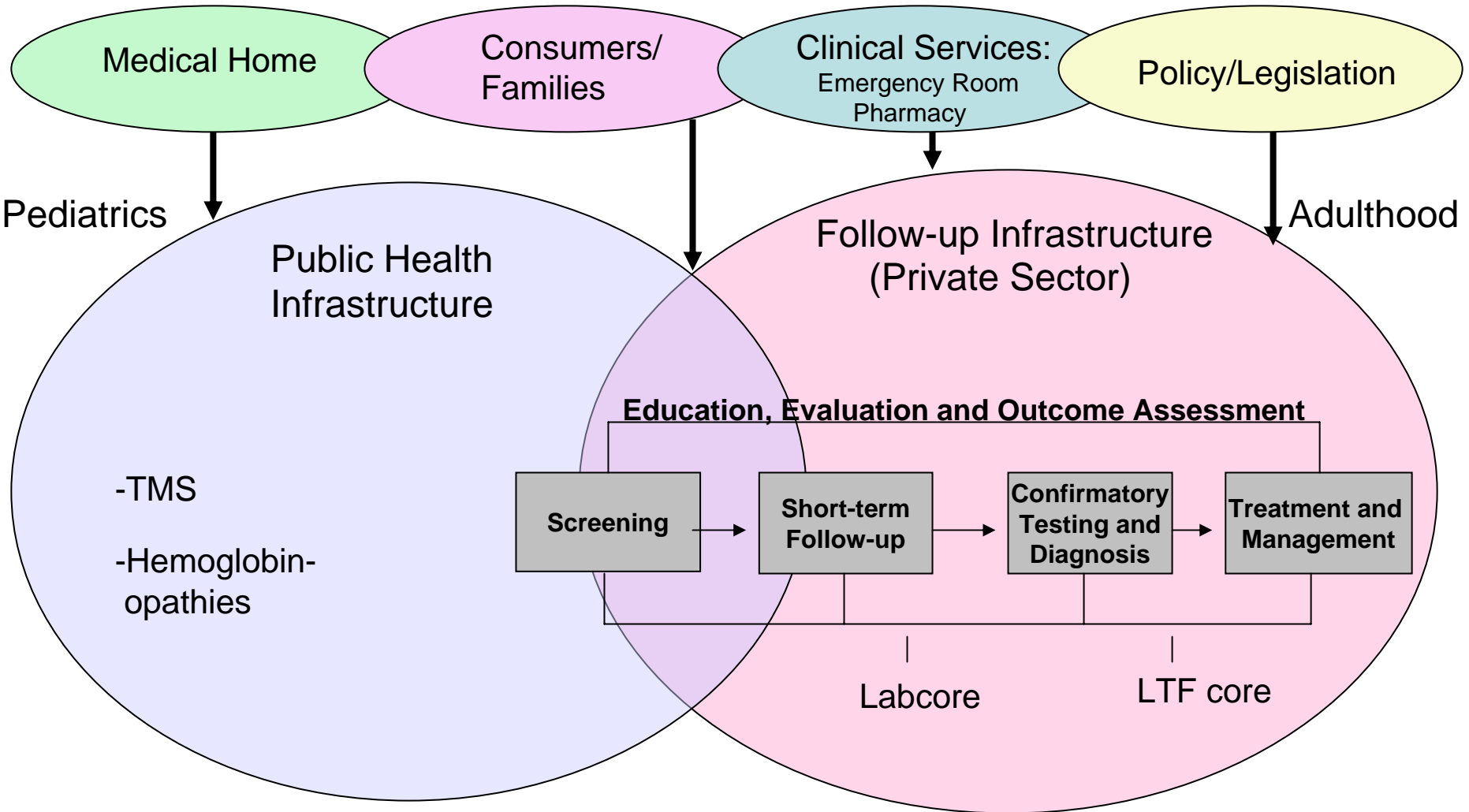
- Provide care coordination, and
- Address stakeholders information needs, and
- Create data linkages, and
- Build evidence for standards of practice and infrastructure for research

# Long-term Follow-up for Patients and Families-Region III





# Long-term Follow-up for Patients and Families-Region III



**Systems Quality**  
**Assurance/Public Health**  
**Informatics**

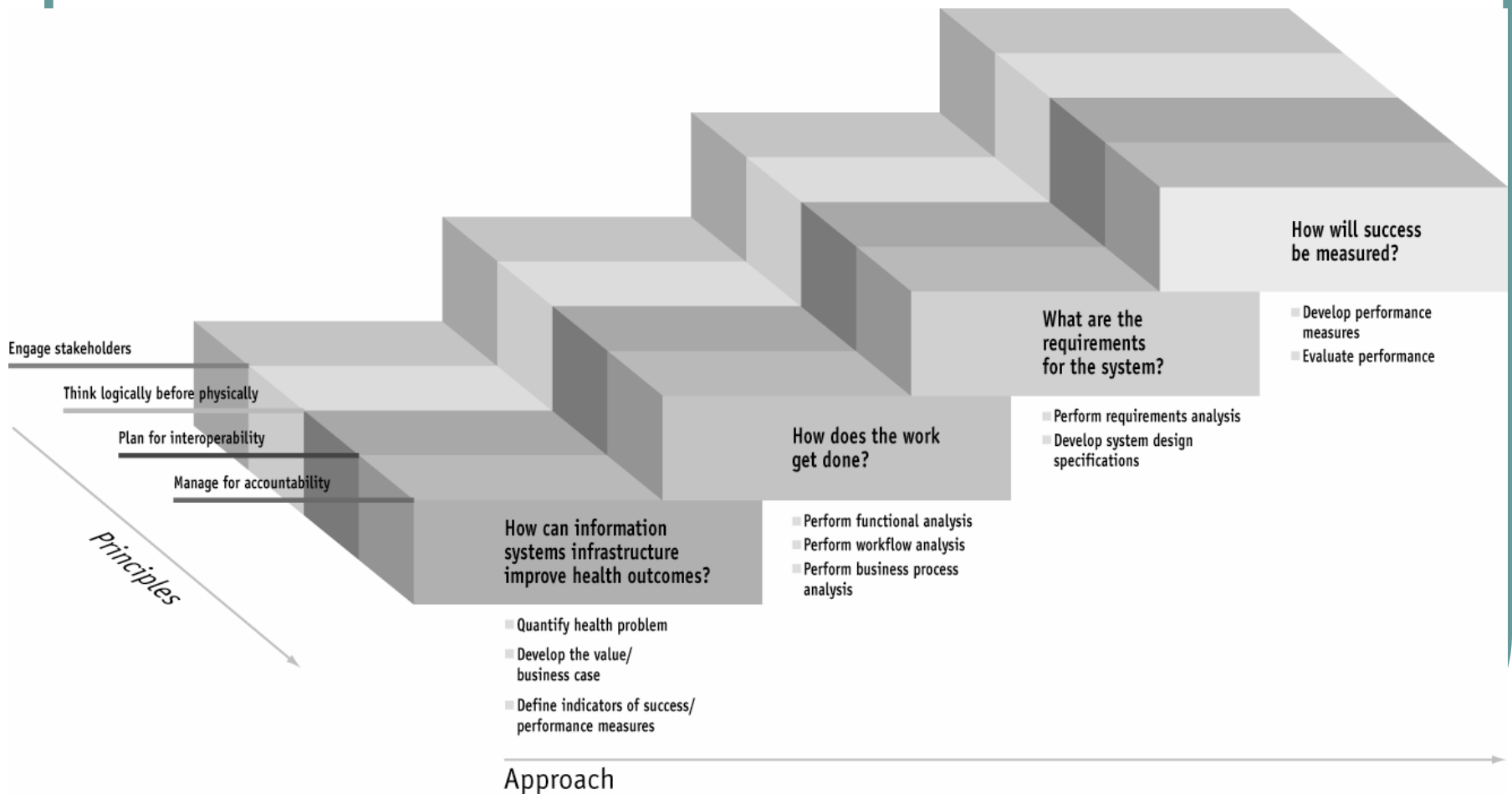


# Planned Project Activities



1. **Capacity and performance evaluation of long-term follow-up systems**
  - **Using a phased approach in partnership with Public Health Informatics Institute (PHII) the region will determine, engage, and utilize stakeholder groups to ascertain an information system usable by practitioners, patients, state public health departments, and consumer groups.**

# The PHII Approach



# Activity 1: 5-year plan



- **Year 1:** Charter the project and recruit participants.
- **Year 2:** Core workgroup convened to ascertain the essential functions of the information system.
- **Year 3:** System design and build contract.
- **Year 1 & 4:** Screen-positive cases and identified information entered and tabulated by condition.
- **Year 5:** All states and territories within Region 3 using the information system + system evaluation strategies

# Planned Project Activities



- 2. Development of a regional information system for all analytes of positive screens and treatment protocols.**

Planned Activities: Nutrition care and medical management plans will be gathered and compiled. Results will be shared and used to assist in development of best practice models for medical management and nutrition protocols.

# Activity 2 5-year plan



- **Year 1:** Contact and survey of NBS clinicians from each Region 3 state and territory to ascertain information about management strategies and care plans. Simultaneous committee formation of clinicians and dietitians and three external experts to begin planning for nutritional plan guidelines.
- **Year 2:** Comparison of Region 3 with Mountain States RC management strategies and care plans. Survey of regional current nutrition care plans.
- **Year 3:** Best practice models developed for both management and nutrition plans.
- **Year 4:** Both models piloted and evaluated.
- **Year 5:** Both models disseminated and applied within the region.

# Planned Project Activities



## 3. **Positive-screen outcome analysis to improve management practices and decrease inequities in Region 3.**

Management guidelines may be improved if they take a more formal, evidence-based approach.

### Planned Activities:

- Literature review and library creation to catalogue articles and research related to disorders detected by TMS.
- Health professionals, 4-5 per topic, will be asked to form a regional journal club.
- Implementation and evaluation of a family-focused resource available in a multi-media format to permit communication beyond the typical clinical setting.

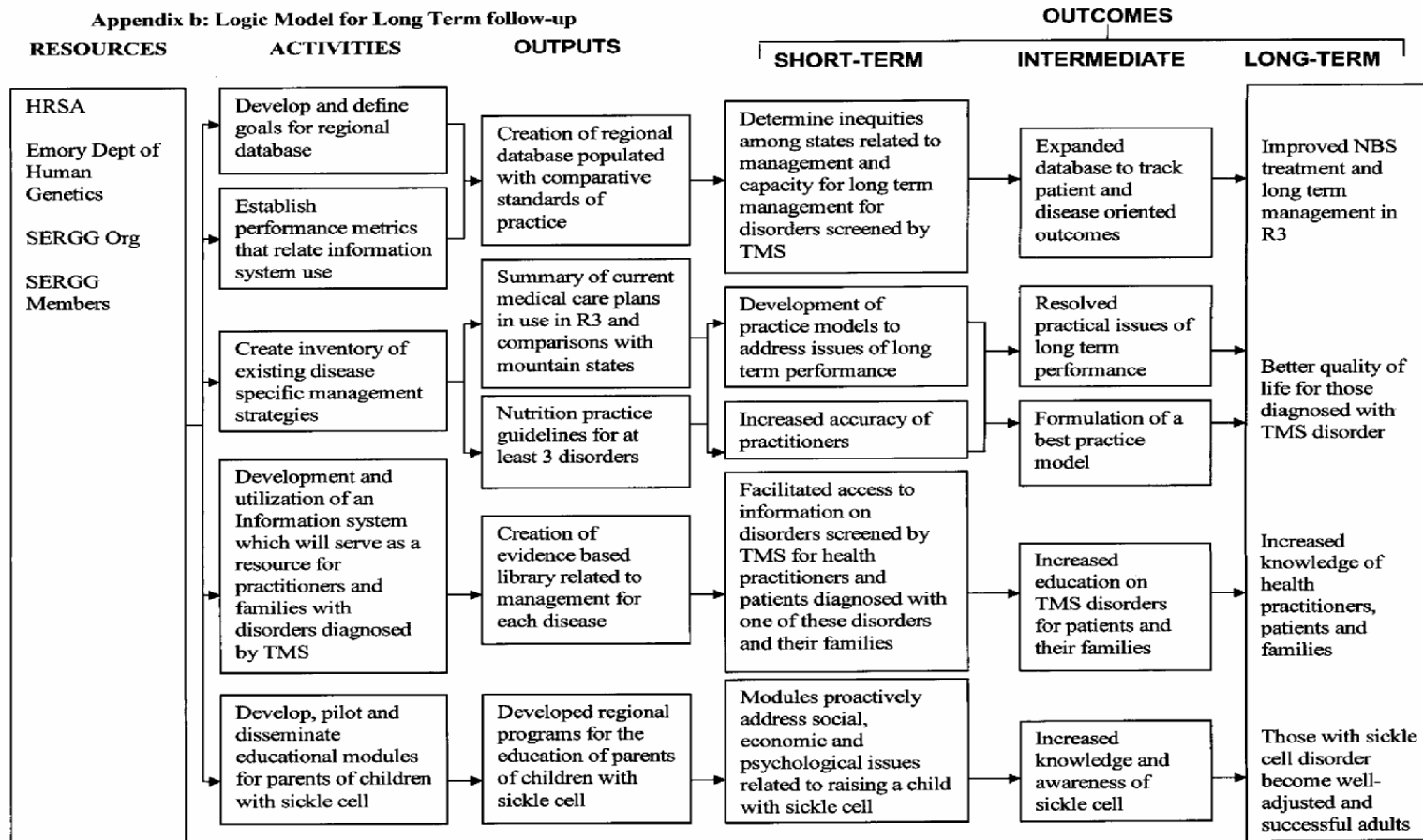
# Activity 3 5-year plan



- **Year 1:** Conduct literature review. Families and health care professionals invited to participate in semi-structured interviews.
- **Year 2:** Library of literature formed. Links and other information developed and available to patients, families, and practitioners.
- **Year 3:** Journal club of experts formed. Information system log and content analysis, ongoing through end of project.
- **Years 4-5:** Action model developed and shared inter-regionally and nationally.



# Overarching Outcomes of the Region's Long-Term Follow-Up Activities



# Outcomes



- Identification and initiation of components of NBS information system as a resource for improving care coordination
- Data Tracking System
  - Including:
    - Positive cases
    - Analyte biomarkers, screening, diagnosis, and management
    - Evidence based library:
      - Regional journal club
      - Partnerships with organizations such as GMDI and ADA
- Management Protocols
  - Medical and nutrition management