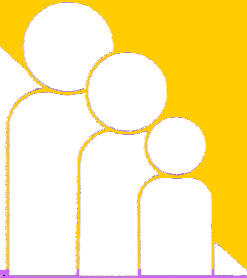




Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Peter C. van Dyck, M.D., M.P.H.
Maternal and Child Health Bureau
January 13, 2005

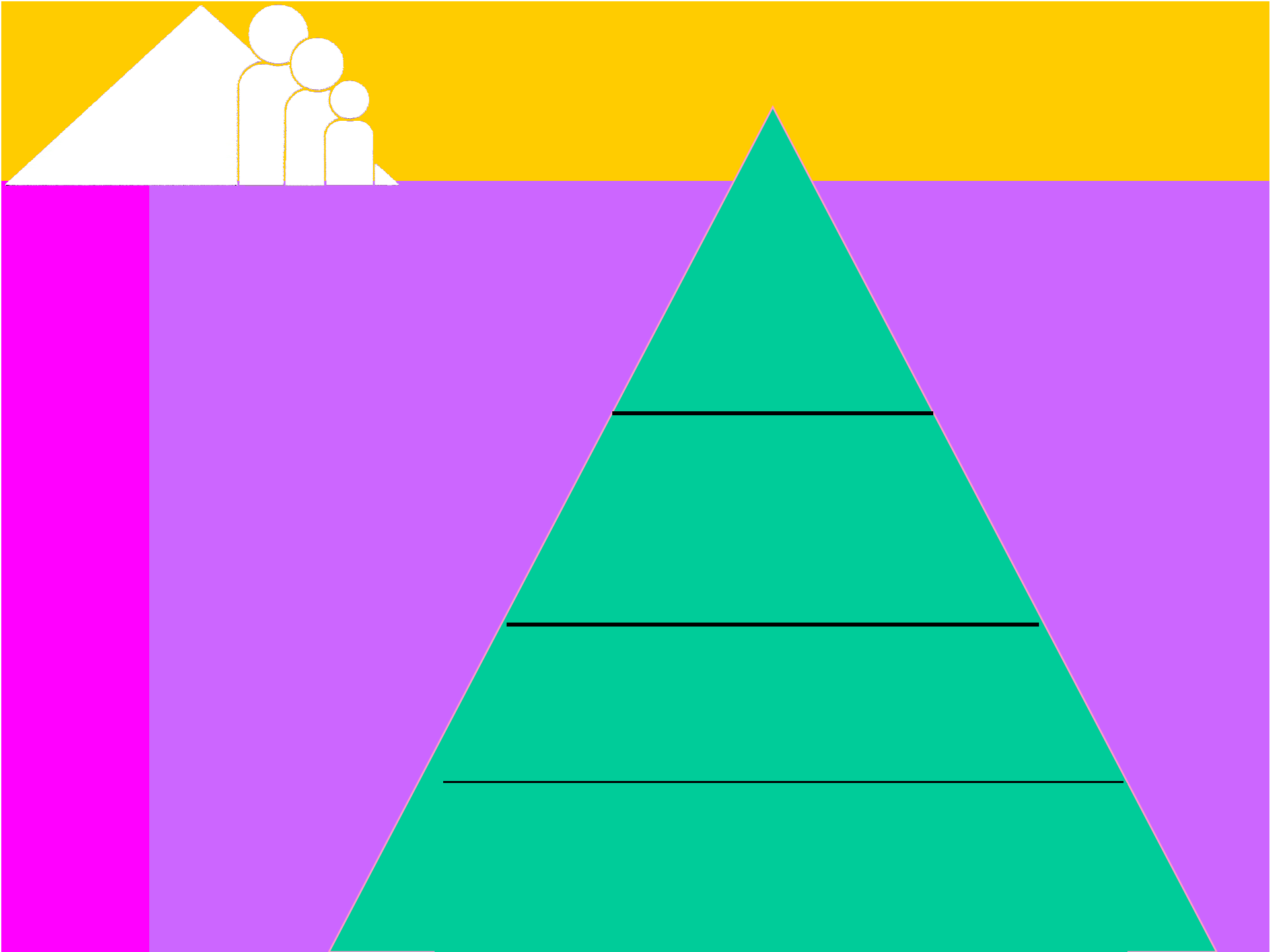
JANUARY 2005



Newborn Screening

“Each year 4 million U.S. infants are screened to detect conditions that threaten their life and long-term health...”







Newborn Screening Program Policy Issues

- Infrastructure Services
 - Policy development
 - Laboratory standards
 - Cut-off values
 - Case definitions
 - Testing strategies
 - Turn around time
 - Nomenclature
 - Report forms

* Items in bold have been discussed or will be at this meeting



Newborn Screening Program Policy Issues

- Infrastructure Services
 - Policy development(Cont)
 - Policies for use and storage of residual blood spots
 - Follow-up protocols
 - **Panel of screening conditions**
 - Informed decision making/consent/dissent



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - Informatics infrastructure
 - **Tracking and surveillance**
 - Definitions of principles and core functions
 - Defining data dictionary
 - **Short and long term tracking capabilities**
 - Knowledge base
 - Evaluation



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - **Integrated information systems development**
 - Integration of public health and personal health services data
 - Provider access to newborn screening results
 - Identification of medical home
 - Privacy/confidentiality
 - Linkage of clinically useful data
 - Child health profile
 - Continuous quality improvement



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - Training on genetics, diseases, treatments and technologies
 - Health care professionals
 - Primary and subspecialty care
 - Public health professionals
 - Laboratories



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - Research
 - Developing new therapies
 - Developing new testing platforms and strategies
 - Evaluation
 - **Cost effectiveness evaluation**
 - Pilot testing of technology
 - Clinical validity and utility
 - Health outcomes evaluation



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - Quality assurance
 - Policy development for laboratory
 - Data standards
 - Case definitions
 - Cut-off definitions
 - Policy development for programs



Newborn Screening Program Policy Issues

- Infrastructure Services(Cont)
 - Distance communication
 - Reimbursement
 - Legal issues
 - Infrastructure
 - **Financing of newborn screening system infrastructure**



Newborn Screening Program Policy Issues

- Population based services
 - Education
 - Health care and public health professionals(hospital, obstetric, and pediatric care)
 - General public
 - Parents



Newborn Screening Program Policy Issues

- Enabling services
 - Access to therapies and supplemental therapies and services (OT/PT, early intervention, Title V, CSHCN)
 - Reimbursement
 - Nutrition support
 - Family support activities
 - Advocacy
 - Educational workshops



Newborn Screening Program Policy Issues

- Direct services
 - Genetic counseling
 - Treatment
 - Subspecialty
 - Nutritionist
 - Other necessary treatment and services for comprehensive care



Contact

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