Integrating Screening, Surveillance, and Service: The New Jersey Birth Defects Registry

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Mission: To assure that all children with special health needs have access to comprehensive, community based, culturally competent, and family centered care.

Early Identification and Monitoring
Newborn Screening and Genetic Services

Early Intervention Services

Family Centered Care Services
Birth Defects Law

- 1926 - Crippled Children’s Commission appointed
- 1928 - first requirement for reporting of “crippled children” began by identifying children with orthopedic conditions, implemented a system of surveillance and service delivery to children with orthopedic conditions
- 1983 law - reporting of children diagnosed through age 1 with congenital defect(s)
- 1985 - rules adopted
- 2005 – law amended to include severe hyperbilirubinemia
- 2007 – Autism Registry law passed NJ Assembly & Senate
- Purpose of law: establish a birth defects registry...epidemiological surveys...plan for and provide services
- Health Commissioner authorized referral of Birth Defects reports to County Case Management Units
County Care and Treatment Law

- 1922 – PL. 1922, c. 159 Law for county care and treatment (amended several times over the century)
- Section 9:13-7 Expenditures for crippled children and children with cerebral palsy: “The board of chosen freeholders of a county which has no county home and hospital for crippled children and children afflicted with cerebral palsy may appropriate not more than $75,000 each year for the necessary expense incident to the diagnosis and treatment of such children resident in the county under the age of 21 years, including the cost of surgical appliances, support and maintenance of such children, investigation of cases, and necessary traveling expenses incidental to the investigation and transportation of patients to a suitable home or hospital within the State supported by public funds of private charity; provided, however, that the board of chosen freeholders in a county of the first class having a population in excess of 800,000 may appropriate not more than $100,000 each year. ...”
Birth Defects Reporting

- Rules require reporting from hospitals, physicians, dentists, certified nurse midwives, advanced nurse practitioners, medical examiners, and other medical professionals who diagnose birth defects.
- Hospital reporting part of hospital licensing standards.
- Informed consent not required for birth defects.
SCHS Registry

- Two components: Birth Defects and Special Needs
- Statewide surveillance of 113,000 annual births
- Over 8,000 new registrations annually
- Resides in Special Child Health and Early Intervention Services-Title V Program-Children with Special Health Care Needs
- Funded from MCH block funds and CDC cooperative agreement
Quality Assurance - Audits

- Annual audits conducted at every maternity hospital and facility with pediatric beds
- Review 3 months of birth records
- Summation session held with administration and representatives from pediatric disciplines
- Written report provided to each facility
Quality Assurance-Other Methods

-* Linkage to other databases-birth and death files; review all infant death certificates; Universal Billing database
-* Reporting from other health programs, including newborn biochemical and newborn hearing
-* Data indicates 89% children registered appropriately
Quality Assurance - EBC

- NJ implemented EBC in 1995; statewide by 1997
- EBC is a comprehensive database: prenatal history, birth event, maternal history
- Data appended to SCHS Registry
  - improves demographic information
  - identifies state births
  - provides additional information (such as GIS designations)
Newborn Biochemical Screening -
Historical Information

- NJ began screening for PKU in 1964
- Congenital hypothyroidism (1978) and Galactosemia (1982) added
- 1990 - hemoglobinopathies, including sickle cell added
- 2001-2005 major expansion to add more disorders
- Law/rules require screening unless parent objects on religious tenets; informed consent is not required
- Children confirmed with metabolic disorders are registered with SCHS Registry
Newborn Biochemical Screening - Expanded Screening

- New technology, new information, private laboratory inquiry, and public requests, the Newborn Screening Advisory Panel convened in April 2000.
- Panel comprised of medical specialists, nurses, an ethicist, and parents; Department staff not official members.
- Reviewed approximately 30 disorders.
- December 2000 submitted recommendations to Commissioner.
Timeline of Newborn Screening in New Jersey 1960s to 2005

1960
- PKU
- CH
- GAL
- Hgb

1970
- MSUD
- CAH
- BIO
- CF

1980
- MS/MS

1990
- 4FAO
- 2UCD

2000
- 6OA

2001
- 29 HRSA

2002
- NSARC

2003

2004

2005
Newborn Biochemical Screening – Expanded Screening

• 2001
  Maple Syrup Urine Disease, Cystic Fibrosis, Congenital Adrenal Hyperplasia, Biotinidase Deficiency

• 2002
  (4) Fatty Acid Oxidation Disorders
    MCAD, SCAD, LCAD, VLCAD
  (2) Urea Cycle Disorders
    Citrullinemia, Arginosuccinic Acidemia

• 2003
  Six Organic Acidemia Disorders
  Propionic Acidemia, Methylmalonic Acidemia, Isovaleric Acidemia
  3-Methylcrotonyl-CoA Carboxylase Deficiency
  3- Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
  Glutaric Acidemia Type I
Newborn Biochemical Screening – Expanded Screening

2005
Newborn Screening Annual Review Committee convened in March 2005 to discuss adding HRSA recommended non-mandated disorders including:

Fatty Acid Oxidations Disorders
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase (LCHAD) Deficiency
- Trifunctional Protein Deficiency (TFP) Deficiency
- Neonatal Carnitine Palmitoyl Transferase Deficiency – Type II (CPT – II)
- Carnitine/Acylcarnitine Translocase Deficiency
- Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidemia Type 1)
- Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD

Organic Acidemias
- 2-Methylbutyryl-CoA Dehydrogenase Deficiency
- 3-Methylglutaconyl-CoA Hydratase Deficiency
- Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
- Isobutryl CoA Dehydrogenase
- Beta-ketothiolase deficiency
- Mutiple Carboxylase Deficiency
- Malonic Acidemia

Amino Acid Disorders
- Homocystinuria and Hypermethioninemia
- Tyrosinemia, NOTE: Tyrosinemia Type I cannot be detected by MS/MS
Newborn Biochemical Screening – Follow-up Program

- Access to treatment and specialty care centers
- Special Child Health Services provides grant support for:
  - 3 regional metabolic centers (provide diagnosis, treatment, and special metabolic formula)
  - 3 Cystic Fibrosis centers
  - 5 facilities providing pediatric endocrine services
  - 2 biochemical genetics laboratories
  - 5 sickle cell treatment centers

- Pediatric Consultant Groups (Metabolic/Genetic, Endocrine, Pulmonology, Hematology)
- Consultant List is provided to every health care provider who receives referral information
Newborn Hearing Screening

- Newborn Hearing Screening is required by New Jersey law
- 1977 - Original newborn hearing screening law was passed, screening consisted of evaluating the presence of risk factors for hearing loss
- May 2000 - amended rules required phase-in of universal newborn hearing screening
- January 2002 - new legislation supplants 1977 law, UNHS mandated
Newborn Hearing Screening

Universal Newborn Hearing Screening 1-3-6

- Screen all infants by 1 month of age
- Diagnostic evaluation by 3 months of age
- Appropriate, family-centered, culturally competent intervention by 6 months of age
Newborn Hearing Screening

Law mandates:
♦ Universal newborn hearing screening
♦ Testing prior to discharge or by 30 days of age
♦ Hospitals must have protocol to ensure follow-up and parent education
♦ Reporting of all children with any hearing loss to the Special Child Health Services (SCHS) Registry
♦ Establishment of a central registry to provide statistical data, follow-up counseling, intervention and educational services
♦ Insurance coverage of newborn screening
Newborn Hearing Screening

- Hospital-based screening of all infants by 1 month of age
- Hospital follow-up of infants with failed screens, goal is diagnostic evaluation by 3 months of age
- Diagnostician fills out Newborn Hearing Follow-up Report
- Diagnostician fills out SCHS Registry form
- SCHS Registry makes direct referral, within 10 days of receipt, into county-based case management
- Case Managers - single point of access for medical and educational services
- Goal is appropriate, family-centered, culturally competent intervention by 6 months of age
- HRSA UNHS grant supports follow-up activities of 2 staff
- MCH Block grant supports audiologist
Newborn Hearing Screening -
EHDI Surveillance System

- EBC serves to populate the EHDI system
- Through a CDC hearing cooperative agreement, EHDI system was developed
- Vital Statistics has added additional variables to EBC to better monitor hearing screening
- EHDI system to be linked to early intervention data
- Hearing Screening data to be linked to Immunization Registry
Screened before nursery discharge or by 1 month of age
Includes children receiving either outpatient re-screening or diagnostic testing. This data reflects only what has been REPORTED to NJ-EHDI. The true rate may be higher.
Case Management

Purpose

To assist children, age birth through 21 years, to access family centered, coordinated services for children with special health care needs and those at risk for developmental delay.
Case Management - Family Centered Care

- Decentralized, one in each of NJ’s 21 counties
  - reflects uniqueness of local areas
  - better utilization of resources
- Primary focus is medical, but also involved in social aspects of care
- Jointly funded by MCH Block Grant, State funds and the County Boards of Chosen Freeholders
- 1993: case managers began to serve as entry into Early Intervention Services
County Case Management Units

- 13 in County Health Departments
- 4 located in hospitals
- 2 in voluntary non-profit agencies
- 1 in a community nursing agency
- 1 in a regional special needs school district
Case Management - Family Centered Care

Focus

- Primary care provider/other involved professionals contacted in order to coordinate services
- Referrals made for identified needs
- Individual Service Plans developed
- Help families to advocate through various agencies
Case Management Services

- Healthcare resources - medical/dental, developmental, educational, rehabilitative, social-emotional-economic
- Assist families obtaining access to a medical home
- Information and referral to State and Federal programs, such as: Child Evaluation Centers, SSI, NJ FamilyCare (CHIP), Catastrophic Illness in Children Relief Fund program, Division of Developmental Disabilities (DDD), etc.
Early Intervention System

- Special Child Health Services Case Management Unit is the point of access from the Registry and the Social Security Income/Disability Program.
- A service coordinator is assigned to each family referred for early intervention.
- The service coordinator will provide general information about SCHS and Early Intervention, explain the family’s rights, gather basic information about the child and family, and answer the family’s questions.
- Early Intervention is voluntary and requires parental consent for participation.
- Any hearing loss is a presumptive eligible for EI.
Birth Defects Registry – Data Uses

- Surveillance (ex: Accutane, NJ, national, multistate)
- Need assessment (ex: MCH Block, case management, agency grant applications)
- Research (ex: Centers, water-neural tube, infant mortality, accuracy of birth certificates)
- Collaborative projects (ex: AIDS, OPMRDD, folic acid)
- Linkage to services
Research

♦ Centers for Birth Defects Research and Prevention (CDC)
♦ National Down Syndrome Project (Emory Univ.)
♦ World Trade Center – 9/11 Study (NIH)
♦ Accutane
♦ Water contaminants and neural tube defects
♦ Infant mortality - coding and contribution of birth defects
♦ Accuracy of birth defects reporting on electronic birth certificate
♦ Pulse oximetry screening study
Several Programs, 1 Registry

Reporting to SCHS Registry from:
- Newborn Biochemical Screening
- Early Hearing Detection and Intervention
- 64 Birthing Facilities
- Medical professionals
- 21 SCHS County Case Management Units
SCHS Registry - Linking Surveillance to Services

- Families receive letter/information from Registry
- Direct link with local county-based case management units
- Case management coordinates Part C, Early Intervention
- Coordination includes health and social services - federal, state, and local resources
SCHS Registry -
Linking Surveillance to Services

♦ All children reported to SCHS Registry directly referred to county case management unit
♦ County case management units assist families to access family centered, coordinated services for children with special health care needs; attempt contact with every family within 7 days
♦ Case management decentralized; 1 in each 21 counties
♦ Ensure family has a medical home
♦ Contact providers to coordinate services; referrals made for identified needs; Individualized Service Plans developed
Why Does It Work

Law and rules (BDR, County Care & Treatment)
Funding from different sources
- HRSA-MCH Block grant (BDR, Case Mgmt., Specialized Pediatric)
- HRSA-other (Ryan White Title IV)
- CDC (surveillance - BDR)
- State (case mgmt., cleft lip/palate)
- County Freeholders (case mgmt.)
- Dedicated tax (Catastrophic Illness in Children Relief Fund)
Part of an integrated system within Division of Family Health Services
Communication
Data part of the program
Why Does It Work

♦ Integration/partnerships with other agencies (ex: SSA, Medicaid, MCH, WIC, Human Services, Labor, Local Health)

♦ Buy-in from agencies and hospitals

♦ Provides NJ ability to meet challenges

♦ Public involvement/comment
Challenges

- Funding issues
- Confidentiality
- Staffing (internal and external)
- Manual versus electronic reporting
Benefits

- Cost effective and efficient
- Timely identification of children and direct referral to case management/EI evaluation
- Fosters communication/builds partnerships between agencies and departments involved in surveillance and services
- Data available to assist with service planning at the local level
The Bottom Line

- A system has been developed for the early identification of children; law and rules provide the structure.
- Linkage to service encourages reporting.
- Linkage is cost effective and efficient; assures coordinated access to care.
- Strong quality control procedures help to assess the success of the surveillance efforts.
- Work to foster communication and team work.
The Bottom Line

🌟 Thanks for the funding!

♦ HRSA - UNHS
♦ CDC – EHDI & BDR Surveillance
♦ MCH Block Grant
♦ State/County
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