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

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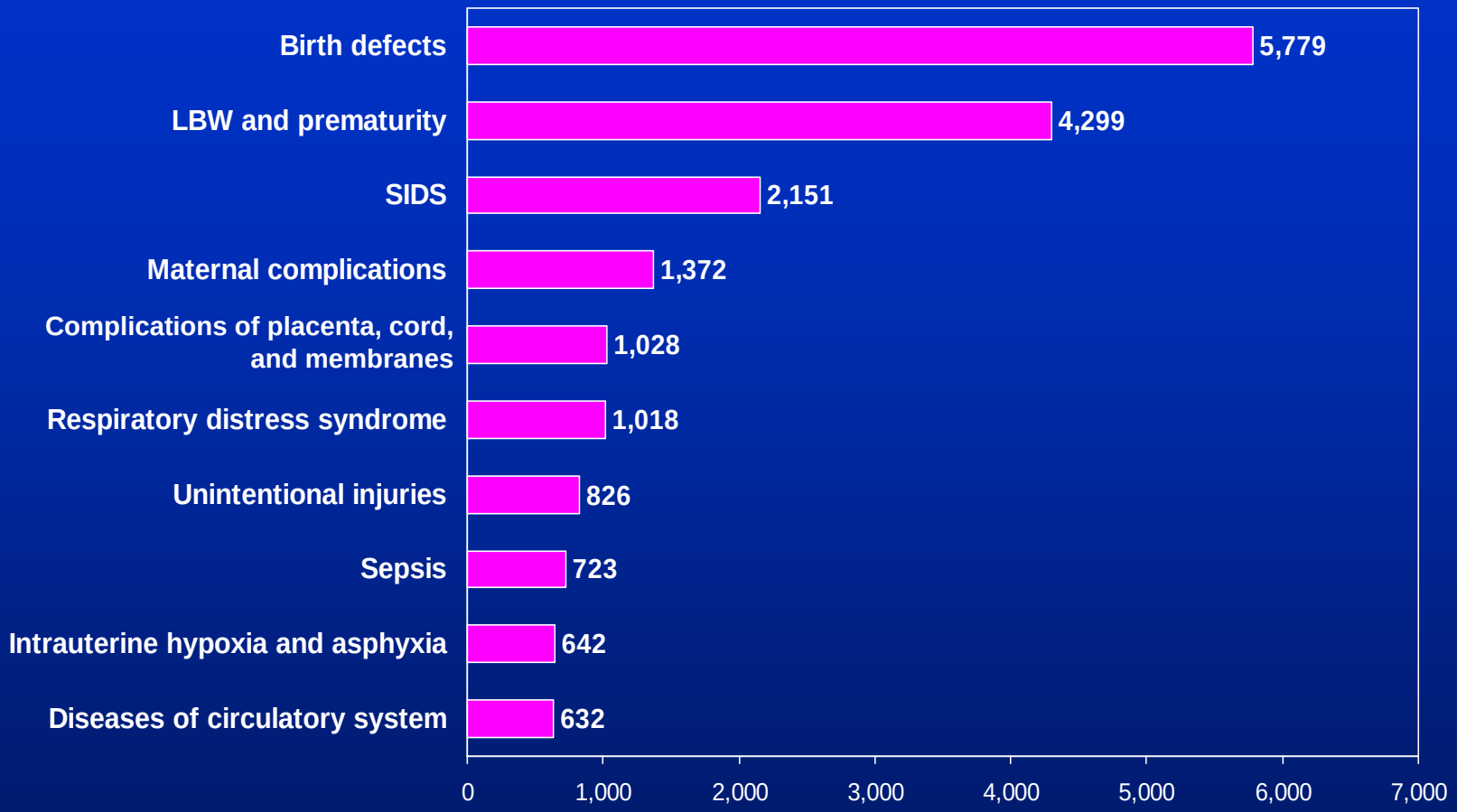
Public Health Importance of Birth Defects

- 1 in every 33 babies is born with a major birth defect each year
- 30% of admissions to pediatric hospitals
- 17 most significant birth defects: $6 billion annually
- Leading cause of infant mortality
- Some causes entirely preventable

Birth Defects: Impact

Ten Leading Causes of Infant Deaths — United States, 2000*



* n = 18,470; other causes = 9,513

Source: Centers for Disease Control and Prevention/National Center for Health Statistics, 1999-2000 National Vital Statistics System, mortality (unlinked file)

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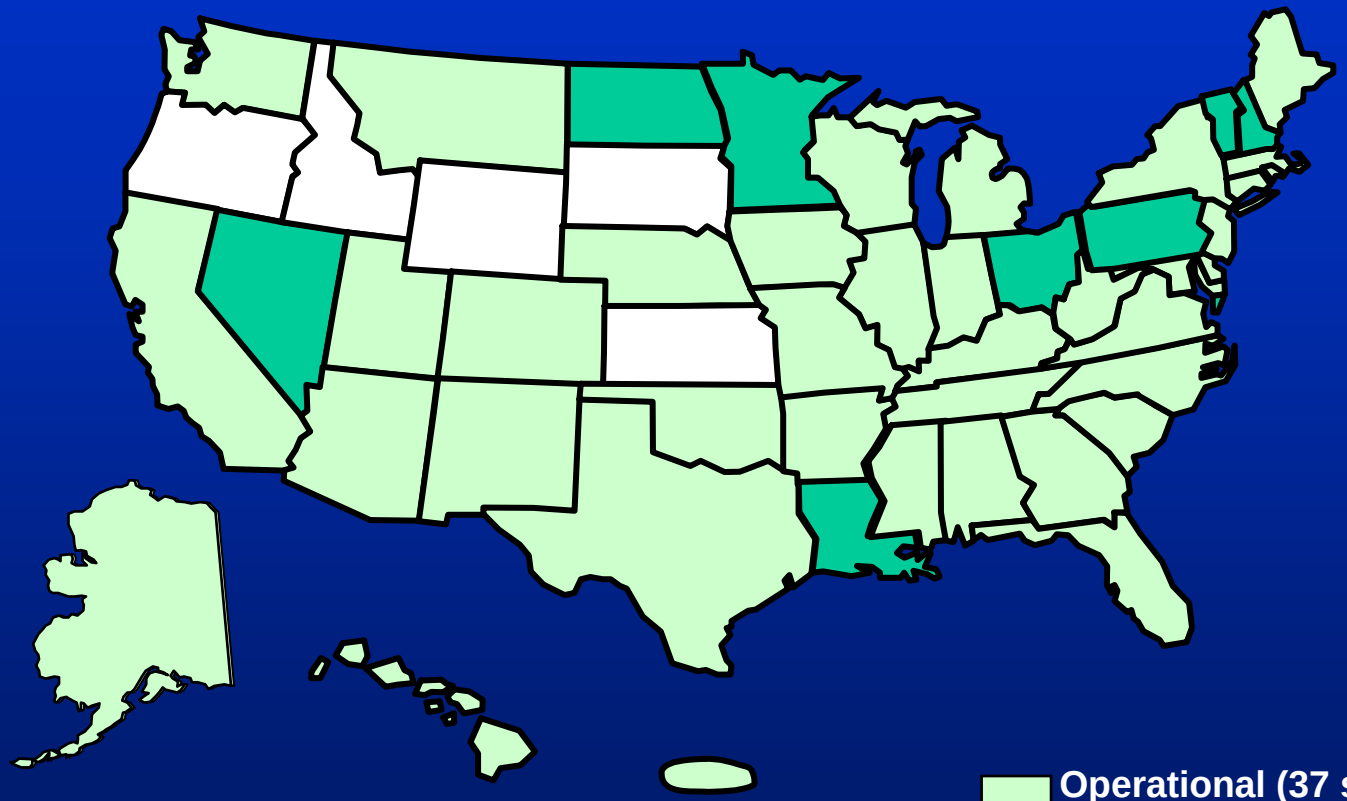
History of Birth Defects Surveillance

1960's	International Interest due to Thalidomide
1968	Metropolitan Atlanta Congenital Defects Program started at CDC
1974	3 State programs
1980's	Epidemiologic research and State surveillance programs
2004	38 operational and 9 planning programs

Current State Activities in Birth Defects Surveillance



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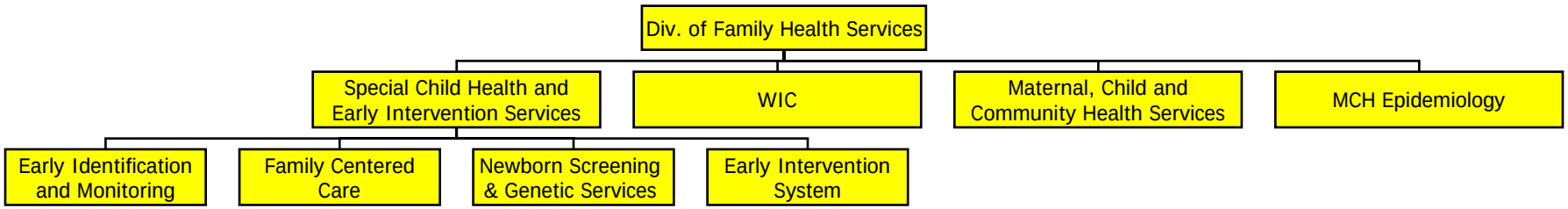
Referral and Intervention Activities State Birth Defects Programs

- CDC surveyed state BD programs concerning their ability to refer children to services
- 26 - system in place for referring children to services
  - 10 mandated by state law
  - In 2003: 62,551 children identified with bd; 17,189 were referred
- 17 – planning or considering a system
- 3 - no plan for a system

Referral and Intervention Activities
Conditions for Referrals

- All birth defects (8) or specific birth defects, e.g., NTDs, clefts (13)
- Developmental disabilities (DD), high risk for DD, or risk factors for developmental delay (7)
- Hearing loss (8)
- Metabolic (or newborn) Screening Disorders (10)
- Other factors, e.g. medically high-risk infants, clinical genetic services, specialty care and resources (8)

Family Health Services



Birth Defects and Special Needs - Historical Information

- Beginning in late 1800's, NJ began commitment to children with special health care needs
- 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
- Federal initiatives, such as Medicaid, Education of the Handicapped, SSI Disabled Children's Program, and block granting of the Title V funds, impact NJ programs

Birth Defects Law

- Due to environmental concerns, a population based surveillance system was needed
- 1983 law - reporting of children diagnosed through age 1 with congenital defect(s)
- 1985 - rules adopted
- Purpose of law: establish a birth defects registry...epidemiological surveys...plan for and provide services
- *Commissioner can give access to records to other agencies*

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 114,000 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 coop 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
- Reporting from other health programs, including newborn biochemical and newborn hearing
- Data indicates 80-90% 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
  - improve 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
- Law/rules require newborns to be screened unless parent objects on religious tenets; informed consent is not required

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

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-Methycrotonyl-CoA Carboxylase Deficiency
  3- Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
  Glutaric Acidemia Type I

Newborn Biochemical Screening – Expanded Screening

- **2005**

Panel reconvening in March to discuss additional 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-Methylgutaconyl-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

- Inborn Errors of Metabolism (IEM) Laboratory, Division of Public Health and Environmental Laboratories
- Newborn Screening and Genetic Services Follow-up Program, Division of Family Health Services

Inborn Errors Of Metabolism (IEM) Laboratory

- Specimen receipt and testing
- Expedite retesting on unsatisfactory specimen
- Report results to hospitals and health care provider
- Quality control

Newborn Biochemical Screening – Follow-up Program

- Makes recommendations for retesting and evaluation
- Case follow-up to final disposition
- Provide information to parents, practitioners, and health care providers
- Registers with SCHS Registry

Newborn Biochemical Screening – Follow-up Program

- Access to treatment and specialty care centers
- Special Child Health Services provides grant support for:
  - Special metabolic formula, medical and nutritional assessment, treatment and care management
  - 3 regional metabolic center
  - 3 Cystic Fibrosis centers
  - 5 pediatric endocrine centers
  - 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 Biochemical Screening – Status

- Of the 113,404 newborns screened in NJ for FY 04, 6,395 were referred for follow-up, 3458 are abnormal results, 2937 sickle cell trait.
- Program follows newborns with abnormal screens to final case disposition
- Final data not yet available for FY04, 271 cases confirmed.

Newborn Biochemical Screening - Quality Assurance

- Monitor time to treatment
- Conduct visits to hospitals for review of screening procedures
- Participate in grand rounds, conferences, other meetings regarding newborn biochemical screening
- Plan to initiate review of medical records for compliance
- Children confirmed with metabolic disorders are registered with SCHS Registry

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 testing

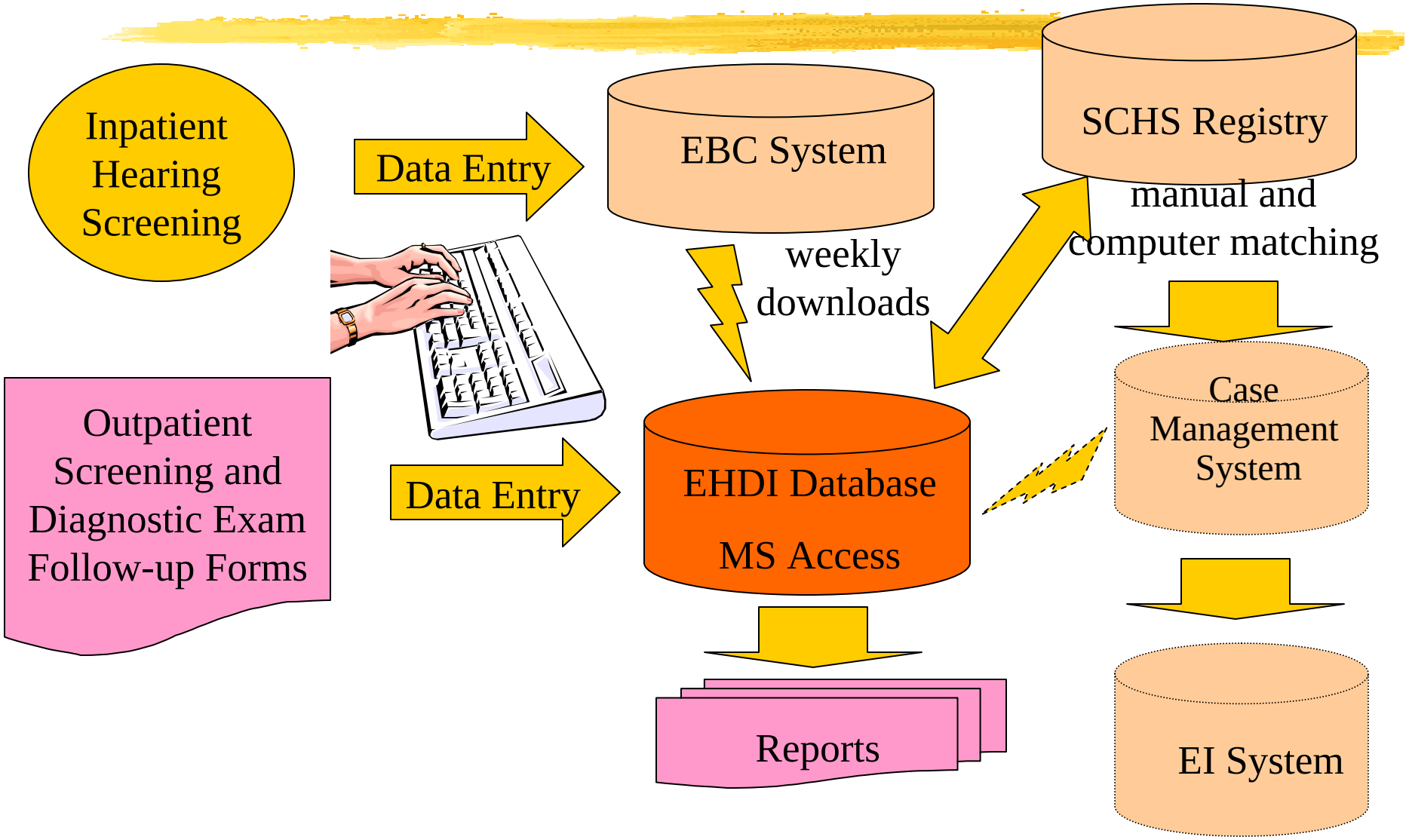
Newborn Hearing Screening

- 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

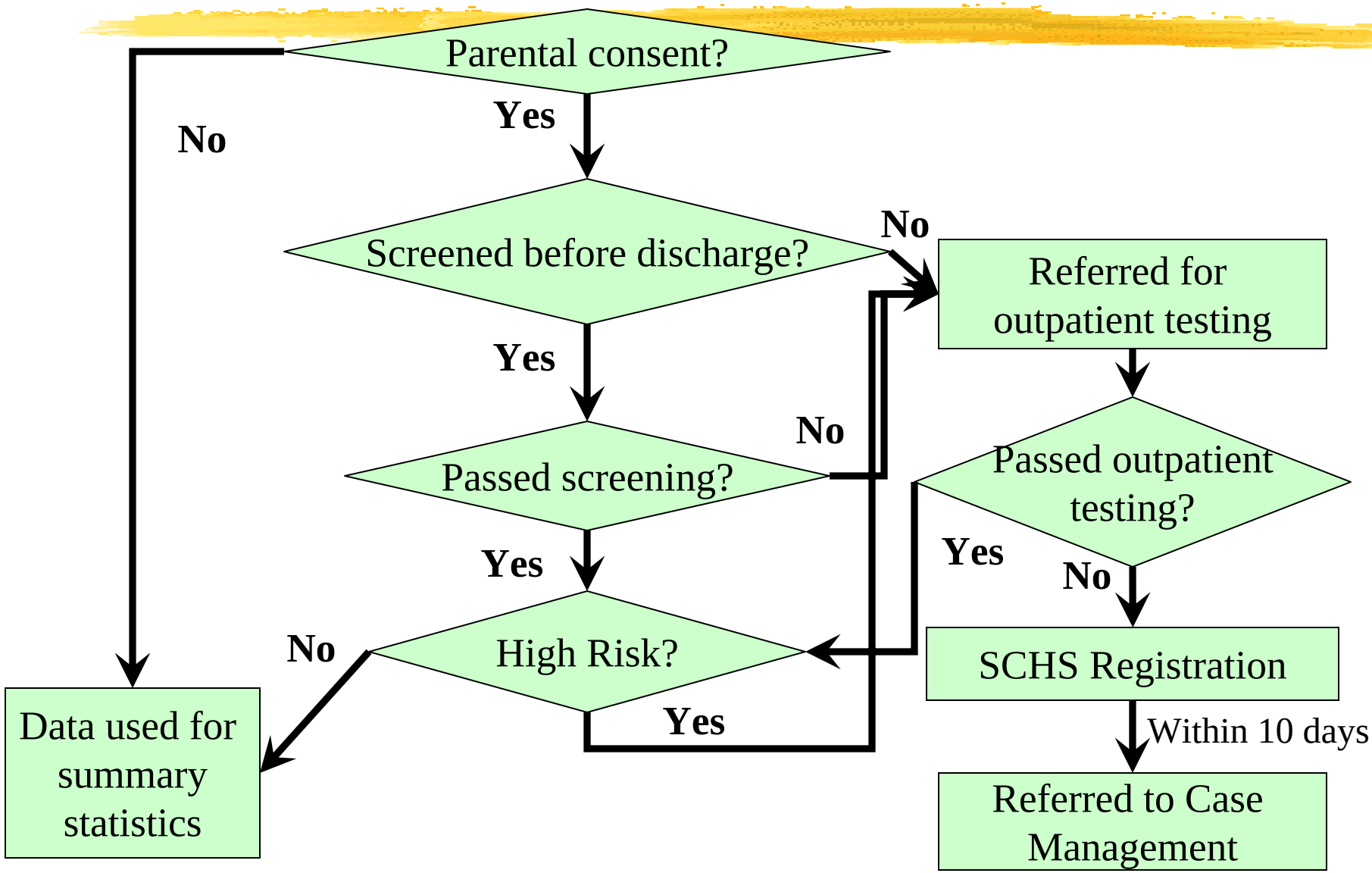
Newborn Hearing Screening - EHDI Surveillance System

- 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

Database


Inpatient Hearing Screening
Data Entry
EBC System
SCHS Registry
manual and computer matching
weekly downloads
Outpatient Screening and Diagnostic Exam Follow-up Forms
Data Entry
EHDI Database
MS Access
Case Management System
Reports
EI System

Screening Process – Flow Chart



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
- Serve as single point of access into a variety of services - medical, mental health, educational, financial
- Jointly funded by MCH Block Grant, State funds and County Freeholders
- 1993: case managers began to serve as entry into EIP services

Case Management - Family Centered Care

- SCHS Registry refers children to case management within 10 days of receipt of registration
- > 90% of case load from direct referrals from the SCHS Registry
- Contact with families within 7 days of referral

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.

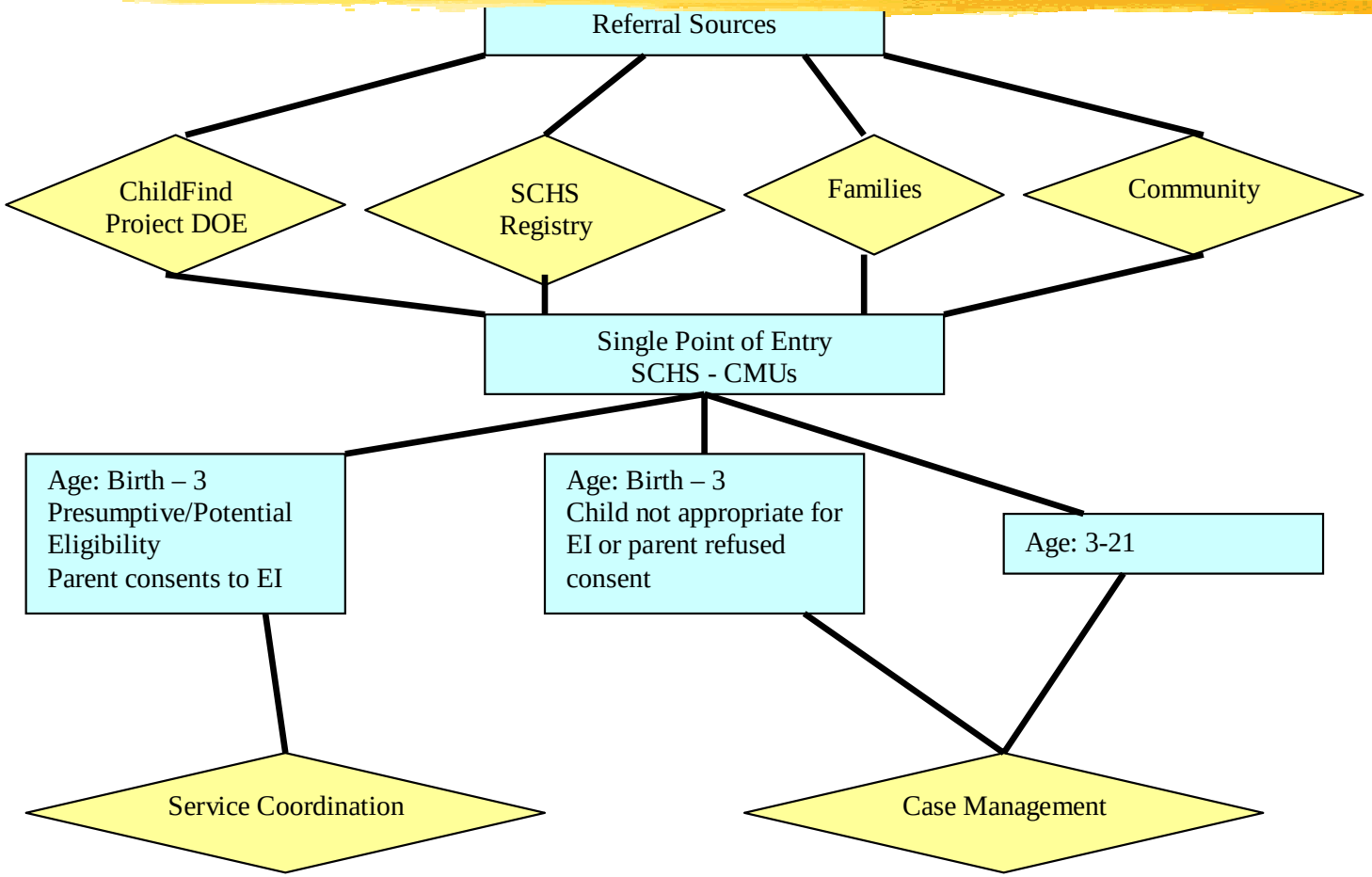
Case Management Services

- Assistance with obtaining hearing aids and prosthetic devices
- Assistance with obtaining drugs for asthma and cystic fibrosis
- Assistance with transition to adulthood, helping families navigate between systems
- Advocacy for child/parent dealing with multiple systems
- Rehabilitation resources
- Parent-to-Parent support and/or referral to support groups

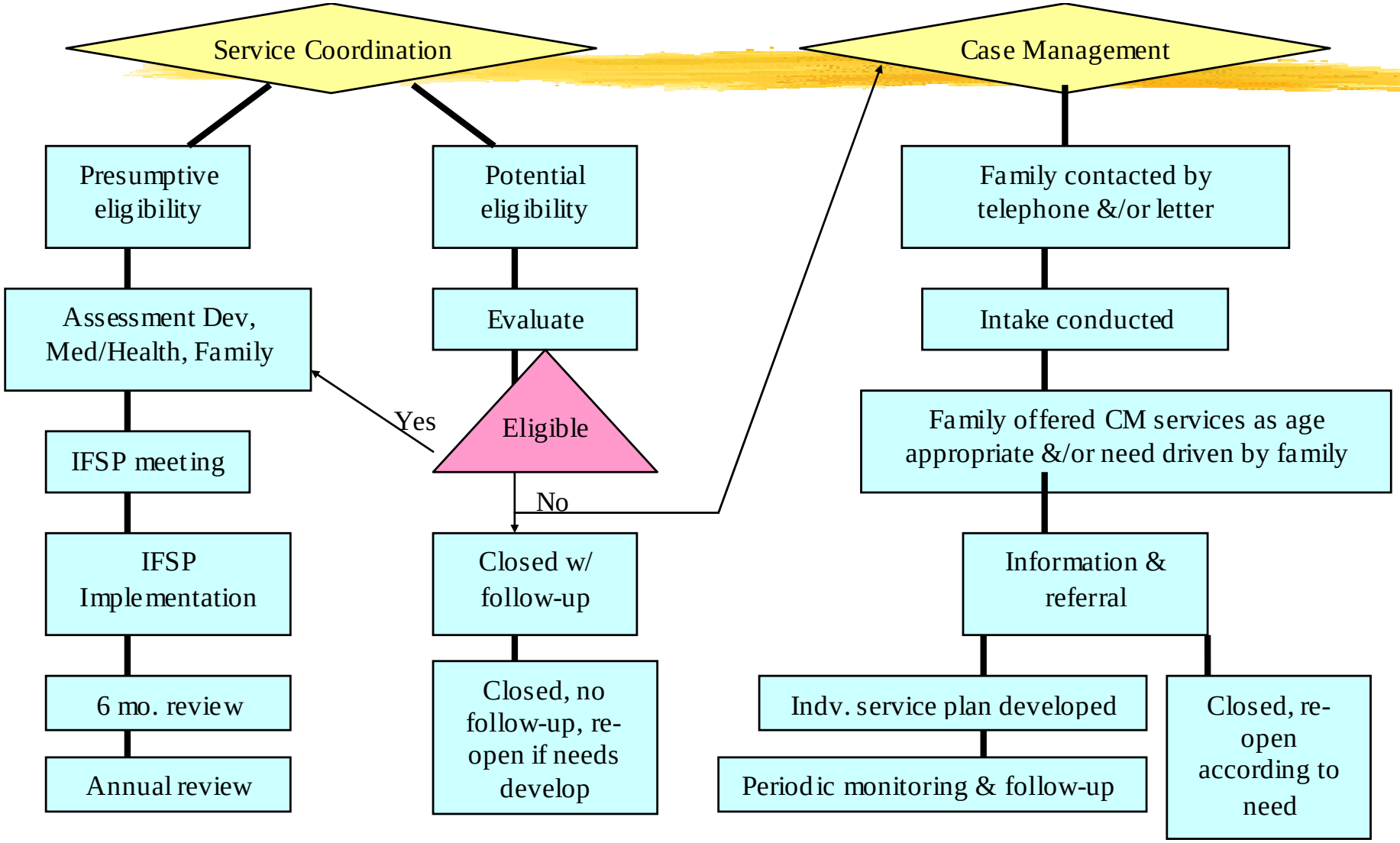
Early Intervention System

- Special Child Health Services Case Management Unit is single point of access.
- 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

Case Management/EI Process



Case Management/EI Process



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

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, UNHS, NBS)
- Funding from different sources
  - MCH Block grant (BDR, Case Mgmt., EHDI)
  - HRSA (UNHS)
  - CDC (surveillance - BD & EHDI)
  - State (case mgmt.)
  - County Freeholders (case mgmt.)
  - Hospital (fee for blood spot kit - NBS)
- 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: SSI, Medicaid, MCH, WIC, Human Services, Labor)
- Buy-in from agencies and hospitals
- Provides NJ ability to meet challenges
- Public involvement (ex: rule readoption, DHSS/governor referrals, parents-EIS/case management)

Challenges

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

Benefits

- Cost effective and efficient
- Timely identification of children and direct referral to case management/EIP evaluation
- Fosters communication/builds partnerships between agencies and departments involved in surveillance and services
- Data available to answer public concerns

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

For More Information

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