Genetic Services State Plan for Indiana

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**ACRONYM DICTIONARY**
ACRONYMS UTILIZED IN THE TEXT OF THE REPORT

**BPR**: Birth Problems Registry

**CDC**: Centers for Disease Control and Prevention

**CHDR**: Child Health Data Repository

**CME**: Continuing Medical Education

**CORN**: Council of Regional Networks for Genetic Services

**CSHCS**: Children’s Special Health Care Services

**FSSA**: Family and Social Services Administration

**HNPCC/FAP**: Hereditary Nonpolyposis Colorectal Cancer/Familial Adenomatous Polyposis

**HPSAs**: Health Professional Shortage Areas

**IBDSS**: Indiana Birth Defect Surveillance System

**ICD-9**: International Classification of Disease, 9th revision

**ICD-10**: International Classification of Disease, 10th revision

**IGAC**: Indiana Genetics Advisory Committee

**IPIN**: Indiana Parent Information Network

**ISDH**: Indiana State Department of Health

**IU**: Indiana University

**IUBRC**: Indiana University- Bowen Research Center

**IUCB**: Indiana University- Center for Bioethics

**IUSOM**: Indiana University School of Medicine

**MCHS**: Maternal and Child Health Services

**MUPs**: Medically Underserved Populations

**NBS**: Newborn Screening Program

**OMPP**: Office of Medicaid Policy and Planning

**PKU**: Phenylketonuria

**UNHS**: Universal Newborn Hearing Screening
1.0 GENETIC SERVICES STATE PLAN

Rapid advances in technology and discoveries fueled by the Human Genome project present unique challenges to health policy leaders and decision makers in Indiana. Genetic research has recently catalogued the Human Genome, and almost daily, new findings increase knowledge of the risks associated with human disease and chronic ailments that may someday help treat or ultimately prevent difficult to cure disorders such as diabetes, heart disease, or perhaps even cancer.

Current statistics regarding genetic disorders in the nation and state also raise important policy concerns. Each year in the United States, about 150,000 birth defects occur, resulting in approximately 6,200 deaths. Birth defects are the leading cause of infant mortality accounting for more than one in five infant deaths. Cost estimates for the families and the nation range up to eight billion dollars annually (Pew Environmental Health Commission, 1999; Waitzman, Scheffler and Romano, 1996), and according to Indiana State Department of Health (ISDH) estimates, approximately $160 million for Indiana residents. In Indiana, in 1999, 85,489 births were recorded with an estimated 3,420 birth defects and 671 infant deaths, 21 percent of which were linked with birth defects (March of Dimes, na.) For those individuals who survive, service systems must respond to service needs for survival and quality of life services. The effects of genetic disorders can be far-reaching and sometimes devastating to the families. The existence of a genetic condition indicates an increased risk for other family members and in future pregnancies. Individuals affected usually have long-term health, social service and education needs, resulting in out-of-pocket expenses for families, third-party payers and/or public funds.

To address these important public health and policy concerns, the Indiana State Department of Health (ISDH) was awarded federal funds through the Maternal and Child Block Grant (Genetics Planning Grant - CDFA # 93.110A) to support development of an information and data infrastructure to guide and assist public health professionals with the identification of infants and children born with birth defects. The ultimate goal of that grant is the reduction of mortality and morbidity associated with the more common genetic disorders. Six additional focus areas were identified by the grant as interim outcome objectives. Those areas included 1) establishing a statewide Indiana Genetics Advisory Committee, 2) conducting a Needs Assessment of Genetic Services in Indiana, 3) addressing confidentiality issues regarding data access and use, 4) developing comprehensive data standards for linking data between state and federal agencies, 5) ascertaining the feasibility of linking various public health information systems 6) and developing an Indiana State Genetics Plan.

This report details the recommendations that have grown from the grant activities listed above. The Indiana Genetics Advisory Committee (IGAC) was established and divided into subcommittees (See Figure 1.1) that prepared interim and long-range goals and objectives (See Figure 1.2) in response to problems identified in the Needs Assessment process. To support those recommendations, components of the statewide needs assessment are reviewed in this plan. This State Plan is divided into four sections. Following this introduction, a description of the process for developing the plan is included along with highlights from the
needs assessment report. The full report can be accessed through the Indiana State Department of Health’s Maternal and Child Health Services (MCHS) website index page at http://www.in.gov/isdh/ programs/mch. Section 3.0 details the specific goals, objectives and action steps developed from seven subcommittees of the IGAC. Section 4.0 includes a final overview and summary.

**Figure 1.1:**
Indiana Genetics Advisory Committee (IGAC)

**Subcommittees:**
- Data
- Education
- Ethical & Legal Issues/Policy Development
- Laboratory
- Public Health & Standards
- Reimbursement for Genetic Services
- Screening

**Figure 1.2:**
Goals of Genetic Services State Plan

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<th>Interim:</th>
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<td>- Reduce mortality and morbidity associated with common genetic/congenital disorders.</td>
<td>- Establish Statewide Genetics Advisory Committee</td>
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<td>- Conduct statewide needs assessment</td>
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<td>- Address confidentiality issues</td>
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<td>- Develop standards for linking data</td>
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<td>- Develop one-year and five-year objectives</td>
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2.0 DEVELOPMENT OF A STATE PLAN

The development of a plan to create, staff and implement new services and improve existing ones ideally should grow from consensus among decision makers, providers, planners and consumers. This development of the Indiana State Genetic Services Plan was based on the assumption that decision makers needed accurate data to identify the level of the problem(s), the nature of existing services, and the perceptions of need and preferences for services among consumers and providers. The IGAC was formed in October of 2000, identifying approximately forty individuals with expertise or interest in genetic services. The committee was divided into seven subcommittee interest areas to review data, reports, and to evaluate recommendations from various planning consultants (Subcommittee Membership is listed in Appendix A).

The Indiana University Bowen Research Center (IUBRC) was selected to prepare a variety of needs assessment components for subcommittee review and to participate with subcommittees in the development of final recommendations for the state plan. The project was conducted under the leadership of Dr. Karen Harlow, Associate Professor of Public and Urban Affairs with substantial assistance of Ms. Kimberly Sundblad as Project Director and Dr. Michael Przybylski, statistical consultant. Graduate students from the Masters of Health Administration Program and the Masters of Public Health Program, Angelique Nelson, Steve McCoy, Brenda Simpson, Tess Weathers, and Scott Wooldridge participated in various components of the project. Project Director for Phase I was Ms. Carolyn Muegge.

The IUBRC team began the project with the goal of applying three different needs assessment models to the data gathering process. A gap method was utilized to identify problems and match service provision patterns to those problems. A marketing approach was utilized through key informant surveys, provider surveys and the use of other forums conducted by state agencies. From these two activities, a needs assessment report was prepared for review by the subcommittees who applied the third method, that of decision-analysis. Decision analysis involves the evaluation and ranking of various data elements into specific recommendations and plans. The needs assessment process also included benchmarking Indiana’s genetics legislation with that of other states and providing detailed information that addressed the core public health functions of assessment, policy development and assurance. Survey instruments were developed with the guidelines provided by the State Assessment Tool: Comparing State Genetic Services to the CORN Guidelines for Clinical Genetic Services for the Public’s Heath (Emory University). The remainder of this section contains a summary of the various components of the needs assessment that were reviewed by the subcommittees. The full report can be accessed through the Indiana State Department of Health- Maternal and Child Health index page at http://www.in.gov/isdh/programs/mch.

2.1 A Gap Analysis

Gap Analysis involves identifying prevalence levels of various problems in the state and the resources available to respond to those problems. It provides basic prevalence and supply
data to address both the assessment and policy development guidelines from the Council of Regional Networks for Genetic Services (CORN) instrument. Surveys of existing data identified the following patterns.

- Data on genetic conditions are expensive and difficult to collect in the general population.
- Newborn screening programs can be effective in identifying some specific genetic conditions, but cannot identify all conditions.
- Birth outcomes monitoring systems (that cover longer periods of time and include service provision issues) have been effective in some states, but have not yet been fully attempted in Indiana.
- Under current Indiana monitoring systems, many types of genetic problems are probably under-reported.

General Data Issues

- Population-based estimates of most genetic conditions are prohibitive in cost.
- National data sets containing information on prevalence cannot be accurately generalized to the state or local level.
- State and local databases of incidence of genetic problems may suffer from reporting inconsistencies and can result in undercount when they include only officially reported cases.

Even with these caveats, the data sources used in this report represent some of the best available information on genetic conditions for planners and decision makers.

National Estimates


- Learning disability 1.8%
- Mental retardation 0.3%
- Cerebral palsy 0.1%
- Down syndrome 0.05%

All others are even less common.

March of Dimes Estimates of Birth Defects for 2000 shows that the most common birth defects occur in nearly one percent of births (i.e. structural defects of the heart or circulatory system: 87 per 10,000), but most are rare (i.e. Down syndrome: 11.1 per 10,000 or phenylketonuria (PKU): 0.8 per 10,000).

Indiana Estimates

Congenital Anomalies (Structural Birth Defects) Compared to Other States:

- It has been shown that the exclusive use of birth certificate data is inadequate to document incidence prevalence of congenital anomalies. However, birth certificate
data are still the only means used to identify the incidence of congenital anomalies in Indiana.

- Unsurprisingly, Indiana has a lower reported incidence of most categories of congenital anomalies.
- Of the seven classes of congenital anomalies studied, Indiana had the lowest rate among comparison states four times and was second lowest once.

**Metabolic Birth Defects:**

- Indiana’s 1995-99 incidence of PKU (0.83 per 10,000 births) was very similar to the March of Dimes estimate for 2000 (0.8 per 10,000)
- Indiana’s 1995-99 incidence of galactosemia was 6.85 per 10,000 births
- Indiana’s 1995-99 incidence of hypothyroidism was 3.78 per 10,000 births

**Screening for Hearing:**

- In the second half of 2000, 97% of Indiana newborns were screened for hearing loss.
- Of those screened, 12.7% failed to pass either an initial or a follow-up screen.
- The National Health Interview Survey reports a 1.26% rate of hearing impairment in persons less than 18 years old.

**Screening for Controlled Substances:**

- In 2000, 2,282 Indiana newborns received meconium screens. During the year, the criteria for requiring a screen were expanded.
- Of those screened, 17% tested positive for at least one type of drug.
- Screening methods have been revised to reduce concern about false negatives.
- Hospital reporting and compliance has been a problem. Only 65% of Indiana hospitals participated in 2000.

**Developmental disabilities:**

- No systematic screening exists in Indiana.
- Certified developmental delay is one criterion for the “First Steps” program and the children so qualifying represented 3.38% of all children 2 and 3 years of age in Indiana as of January 1, 2001.

**Conclusions:**

- It is difficult to assess the true incidence of birth defects in Indiana because of limitations in the surveillance systems.
- The Indiana process is “passive” in that it accepts data generated beyond its control without direct verification. A more “active” system is preferable where employees of the department are directly involved in collecting and verifying the data.
- The screening process for some metabolic disorders is more active and comprehensive but covers relatively few children (about 100 per year come onto the registry).
- Screenings for hearing disorders and exposure to drugs (as detected in a meconium screening) seem to be much less developed.
Understanding these prevalence levels of problems and difficulties with specific data sets leads to a better sense of need and demand in the state. However, a Gap analysis must also look at the supply of services that exist to meet that need/demand in the community. The strength of health service providers for a given population or county, availability, and distribution of health care professionals developmental service providers, licensed physicians, type of education activities, and genetic services offered by private/publicly funded genetic clinics throughout the state was analyzed by several methods. These included the Health Professional Shortage Area (HPSA), Medically Underserved Populations (MUP) method, and the Council of Regional Networks for Genetic Services (CORN) specified criteria. Data were collected from clinic sites via telephone surveys, and various data sets were provided by the Indiana State Department of Health (ISDH) and the Family and Social Service Administration (FSSA). To understand the distribution of other health service providers across the state, data sets provided by the Indiana State Family and Social Service Administration (FSSA) were prepared for geographical analysis in the Arcview GIS (V 3.2) software package. The data set comprised over 3,400 cases of service providers that are routinely tracked by FSSA and categorized as those who provide services for developmentally delayed, speech delayed, or counseling services. The number of categories of professional providers was condensed from 38 (N=38) into 12 (N=12) similar sub-classifications for clarity and ease of analysis.

These 12 sub-classifications are as follows: Speech and Hearing [Speech and Hearing Therapists], Alternative Therapy [Massage Therapists], Developmental Services [Developmental /Educational Associate, Developmental Assistant, Developmental/ Educational Specialist], Transportation [Ambulance, Bus, Common and Non-Common Transports], Medical Physicians [ENT, Surgeons, etc.], Language Interpreters, Nurses [Licensed Practical Nurses (LPN), Registered Nurses (RN)], Allied Service Providers [Physical Therapists (PT), Occupational Therapists (OT), OT Assistants, PT Assistants, Rehabilitation Services], Counseling Services [Marriage and Family, Psychiatrist, Psychologist, Social Worker-MSW], Registered Dietician with a specialty in genetics, Service Coordination [Service Coordinator, Service Coordination Associate], and vision specialists. Telephone surveys and FSSA data identified whether these professionals were available at specific clinic sites and those findings were overlaid on maps of the state.

For appropriate assessment, decision makers must understand the nature of the state’s population and socioeconomic characteristics. Geographically Indiana ranks 38th in size (area) and is one of the top twenty in population in the U.S., with 5,900,000. Indiana currently ranks as the 28th fastest growing state in the nation since 1990. This pattern is due to in-migration or more persons moving into the state than moving out, which is a significant reversal of the out-migration experienced in the 1980s. This recent in-migration (27%) and more births (73%) than deaths, has resulted in relatively rapid population growth for the state of Indiana in the 1990s. Of the 92 counties in Indiana, the ten largest in population are Allen, Elkhart, Hamilton, Lake, Madison, Marion, Porter, St. Joseph, Tippecanoe, and Vanderburgh. Elkhart, Hamilton, Johnson, Madison, and Vanderburgh counties expect to see increases in population by the year 2020. Counties projected to decline in population include Delaware,
Grant, and Vigo. Indiana has a largely metropolitan populace (64% urban, 36% rural) that is still experiencing greater growth in the urban areas with rural inhabitants relocating to the more centralized “big city” setting.

More than 87 percent of the population is white with the largest minority group as African American, 8.4 percent. Almost 26 percent of the population is under 18 years of age and 12.4 percent are over 65. Slightly less than 10 percent live at or below poverty level, but for children the rate is somewhat higher, 14.8 percent (U.S. Census Bureau, Quickfacts, census.gov, 2002). Almost 11 percent have no access to a primary care provider, and 52 percent of Indiana counties are designated as all or part medically underserved areas (MUAs). More than 57 percent are designated all or part health profession shortage areas (HPSAs) (HRSA, Stateprofiles, hrsa.gov/1999). The health care system in Indiana appears to mirror the demographics of the population, with the more targeted and specialty health care services offered in the larger metropolitan areas with the rural areas limited to the basic health care providers.

Several patterns were observed in mapping of the service delivery structure in the state. The prevalence rates of various chromosomal anomalies verified that the heaviest concentrations of some problems with high rates were not in the urban areas. The majority of high prevalence rate areas were distant from services and previously mentioned providers, as noted when the networks and providers mapped over the prevalence data (See Figures 2.1 and 2.2). A high rate in a sparsely settled area did not necessarily translate into adequate raw numbers of clients to support a wide array of services. Nonetheless, the coverage available to some areas of the state did appear to present access challenges and results in lengthy traveling distances to obtain necessary services. The observed patterns can best be summarized as follows:

- Primary services are limited in the rural counties.
- Resources are shared between the smaller counties and the mid size counties.
- Rural areas are marginalized with lack of service provider types.
- Educational activities are the state’s strong point, as most sites offer the required services.
- Staffing is inconsistently spread across the state, with the Indianapolis area approaching the goals and expectations identified in the CORN standards.
- A cost-benefit study may be necessary to determine the most effective and efficient model to organize and improve access in more remote areas of the state.
- Incentive programs may be necessary to attract new genetics counselors to the state and to enhance more equitable distribution of their services.
- A crucial resource available to existing practitioners is the education program “Genetics and Your Practice.” This resource may need broader marketing to increase knowledge of its availability.
Figure 2.1: Service Networks in Indiana; Part I

GY = Gary Nexus
FW = Fort Wayne Nexus
CFPD = Center for Prenatal Diagnostics
Aegis = Aegis Network
Cylle = Clarksville Nexus

Rate of Congenital Anomalies 1995-99 (Deviation from Average)
-1 - 0 Std. Dev.
Mean
0 - 1 Std. Dev.
1 - 2 Std. Dev.
2 - 3 Std. Dev.
> 3 Std. Dev.
Figure 2.2: Service Networks in Indiana; Part I
IU/Riley & Indianapolis Non-Network Sites

IU/Riley & St. Vincent
South Bend
Terre Haute
Jeffersonville
Evansville
Bloomington

Rate of Congenital Anomalies
-1 - 0 Std. Dev.
Mean
0 - 1 Std. Dev.
1 - 2 Std. Dev.
2.2 Market Analysis—Perceptions

The market analysis portion of the needs assessment provided feedback from key informants involved in provision or consumption of genetic services across the state, consumers in statewide forums and physician providers. Various survey formats were used to collect data that apply both to assessment and policy development activities in public health. Questions concerning the assurance function of public health were also included in the survey format.

Key Informant Survey--A survey of 37 key informants was completed to assess opinions and perceptions of access, quality and availability of genetic services in the state.

- A majority of respondents described access to services in the state as fair.
- Location of services, cost reimbursement, and lack of knowledge of the providers and consumers of services were ranked as the most significant barriers in the state.
- Although a large majority ranked existing service quality as excellent, 81 percent reported an inadequate supply of physicians, clinical providers and genetics counselors to meet the needs of citizens with birth defects and genetics conditions.
- A substantial majority favored the use of written and verbal communication such as brochures, pamphlets, follow-up phone calls and letters to convey information to patients.
- The group of respondents indicated that grants, state and federal money to support services and programs and expansion of services covered by both private and public payers would be very helpful.
- Financial support for clients was identified as a need as was the need for expanded educational programs both for consumers and providers.

First Step Early Intervention Forums--Perceptions of community residents and families impacted by disabilities were obtained through the utilization of a focus group/forum activity sponsored by the Bureau of Child Development, Division of Families and Children, Family and Social Service Administration (FSSA) for its First Steps Early Intervention system. Between February 7, 2000 and June 30, 2000 a total of 17 forums were held across the state focusing on issues of concern for families, children and communities in identifying need, developing services and providing access to community services for disabilities. More than 300 individuals participated in 17 forums across the state. The four most common issues for families identified in the 17 forums were:

1. Families knowing about and accessing resources when needed (ranked as one of the top two concerns in 11 of the 17 forums). The commentary surrounding this issue focused on knowledge about resources and how to access them and on actual availability (service is offered).
2. Families advocate by exercising their rights in choosing goals, services and supports (ranked as one of the top nine of 17 forums). Families who are facing the consequences of disabilities and illnesses, many of which are associated with genetic origins, desire accurate and understandable information about these processes in order to appropriately advocate for their child or family member’s needs and service regimen.

3. Families express understanding of their child’s disability (ranked as one of the top five of the 17 forums). Specifically mentioned were knowledge of the medical condition, prognosis, needs support groups, etc. with emphasis on knowledge about specific diagnoses and long-term implications.

4. Families are connected to other families and natural community supports for emotional support. Families impacted by the disease or disability consequences desire knowledge about networks of support from other families, communities, agencies, schools, etc.

Other family themes focused on maintaining positive and nurturing relationships with affected individuals, ability to maintain normal family activities and routines, ability to participate in community settings and activities, families remaining together, understanding transitions that occur throughout the life of individuals affected with disease/disability, and expectations for satisfactory services.

Other key concerns identified specifically for children focused on goals of independence for individuals with disabling conditions, attaining developmental skills, and participation in community activities. Community concerns focused on the need for communities to welcome and fully include individuals with disabilities and their families through childcare, transportation, retail, housing and employment support. Participants identified the need for communities to provide resources regardless of age, income, insurance status and self-help status.

Provider Surveys—An electronic survey was e-mailed to Indiana family practice physicians. Response was received from 71 physicians in 29 counties. Ninety percent specialize in family medicine, and almost two-fifths graduated since 1990. Almost three-quarters of the respondents were male. The majority reported their practice panel size as 2501-5000 (39%) or 1001-2500 (26%).

The questions in the survey focused on newborn screening, adult screening, access issues and educational needs. The following were the key findings of the survey:

- Twenty-one percent (21%) of the physicians deal with problems associated with diseases identified through newborn screens at least once per month or more.

- Almost three-fifths have very high or high confidence in the analytical validity of test results from labs, but twenty-one percent (21%) prefer retesting if tests are positive.
• Adults with family histories of genetic-related problems are the most likely to request genetic screening and appear to be better informed about the process than other groups. Expectant parents also demonstrate some interest in the process, but over half are unfamiliar with the process.

• A substantial majority of physicians report fears of potential problems with privacy/confidentiality of findings from genetic screens. Three-quarters believe the information may interfere with insurability, and three-fifths report fears of employment discrimination for individuals. The potential for other forms of discrimination are also noted by the respondents, but no more than one-third of the physicians focus on these categories.

• Few physicians integrate routine genetic testing for adult onset problems linked with genetic problems but a substantial minority utilizes such tests when family history indicates the need or when the patient specifically requests such a screen. Illnesses most frequently screened for genetic bases due to family history or patient requests were hereditary nonpolyposis colorectal cancer/familial adenomatous polyposis (HNPCC/FAP) (colon cancer); BRCA1 and BRCA2 for breast and ovarian cancer; hemachromatosis and Huntington’s disease. Maternal serum screens during pregnancy are routinely performed by more than forty percent (40%) of the respondents.

• More than four-fifths indicate they have access to genetic counseling or consulting services. Most often, the providers of these services are medical geneticists.

• More than half indicate services are within a 30-minute drive. However, more than one-quarter must drive one to three hours for services.

• Although more than half had made no genetic referrals during the past year, forty-seven percent (47%) had made from 1-5.

• Many physicians are unaware of the length of time their patients must wait for a referral service or appointment. Among those who were familiar with wait time, one-third indicated a wait of one week or less, and slightly more than one-fifth indicated waits of one month or more.

• The most frequently mentioned barrier to accessing health and community services was lack of insurance, 73.2%, with lack of understanding of importance of the services second with 60.7 percent (60.7%). More than half also identified inability to afford services even when insured as a barrier.

• When asked to rank the top two concerns out of a lengthier list, the lack of community understanding of importance ranked first, 41.1 percent, and the lack of insurance ranked second, 39.3 percent. Inadequate availability or number of providers was ranked as third in concern, 25 percent.
The most frequently mentioned information resources or support needed was accurate, accessible web-based information, 84.2 percent, and Continuing Medical Education (CME) short courses at professional meetings, 80.7 percent. Physicians also supported better educational resources to share with families, 68.4 percent.

Common themes permeated the three different data bases developed to identify knowledge, perceptions and attitudes although the participant characteristics differed from group to group. Although respondents demonstrated strong faith and confidence in individuals currently providing specialty services, most providers, consumers and physicians believed that additional educational training is necessary to improve other providers’ skills and to expand the number of trained providers in the state. Financial support was identified as a key factor in developing a better educated/trained workforce. Insurability and costs of services also appeared as concerns for both providers and consumers. Both key informants and physicians viewed lack of education in the community about the importance of genetic testing and services as a major barrier to developing an adequate support infrastructure. The need for more written materials to provide to families was a key request of physicians along with more continuing education training for practicing physicians and more modules or courses integrated into medical school training. Similar wishes and concerns were voiced by participants in the First Steps forums for better information from physicians to enable families to advocate appropriately for their members.

Some gaps in knowledge among physicians appeared to exist concerning specific diagnostic tests related to adult onset problems. Although diagnostic testing to identify the specific diseases is considered a standard procedure, the use of the genetic screen associated with some of these diseases does not appear to be as common. Without better data to support the genetic linkage with adult onset disease and clarify the environmental interaction with the genetic characteristics, these valuable diagnostic tools may be overlooked in treating disorders.

Findings from the gap analysis and market analysis steps identified common themes across four areas: data issues, access issues, educational issues and privacy/confidentiality concerns. Data concerns can be summarized as follows:

1. The current surveillance systems in Indiana probably result in undercounting of genetic-based birth defects. Comparison to active rather than passive systems indicates Indiana’s rates to be substantially below other states. Since no demographic or population characteristics of the state would justify this finding, the assumption is that the differences are an artifact of passive versus active reporting and surveillance. IUBRC recommended to IGAC subcommittees that they examine the possibility of supporting legislation that would require and fund an active system that would improve understanding of the actual burden of disease and disability in the state. Iowa and Georgia models currently provide the best models for replication.

2. Data concerning the linkages of adult onset genetic diseases and environmental or behavioral risks are lacking. The IGAC might explore the possibility of
supporting a specialized module on the Behavioral Risk Factor Surveillance System, a data source collected yearly in the state and partially supported by Centers for Disease Control and Prevention (CDC). This module might obtain specific genetic history information for the respondent that could then be linked to behavioral risk activities that are already identified in the survey. Tracked over time, these important data elements would greatly enhance understanding of applied genomics and lead to better service and educational solutions to problems.

3. As the applied genomics field expands with improving technology, an Oversight Committee may be necessary to evaluate the role of public health agencies in performing or supporting tests whose validity and utility must be constantly monitored. The legal, ethical and social issue of producing diagnostic data for problems where no known remedy exists is an ongoing challenge in the field. Indiana experts should play a dominant role in guiding state and local policies and in national debates concerning this important policy issue.

Access concerns expressed by experts, community residents and physicians focused on the following themes.

4. Lack of insurance and inadequate insurance are major barriers to fully accessing the new information technologies that genetic analysis can produce. IGAC members might explore strategies to involve the insurance industry as well as public, nonprofit and for profit providers in supporting better diagnostic data and services.

5. Rural access to diagnostics and service provision is a challenge in Indiana. Prevalence maps indicate that higher rates are not necessarily associated with urban locations, but small numbers make the market reality for provision of services a major challenge. IGAC members should explore funding sources for additional mobile services from existing networks to increase access for underserved areas in the state.

6. To assist in capacity building for support services as well as diagnostic and clinical services, IGAC members should explore possible partnerships with FSSA First Step Councils. These councils are in place for local planning and ongoing capacity assessment. Participation in these local capacity building networks is an efficient and reasonable approach to enhancing local service provision and addressing access issues. These local councils provide a unique opportunity to explore public and private partnerships at the local level to solve local problems and decrease barriers.

Educational concerns identified in the study focused both on physician training and on community knowledge.
7. Although existing providers are viewed as providing excellent services, the perception remains both in the community and among physicians that providers are inadequately trained to meet the needs of individuals with genetic-based diseases. Additional classes in medical school are supported as are the development of CME credit courses linked to professional meetings or CME web-based course that providers can access easily.

8. Community forums and provider surveys both identified lack of understanding in the community about the nature and importance of these problems as a major barrier to provision of appropriate services. The development of educational campaigns that inform the public about new advances and technologies and the importance of risky behavioral activities that interact with genetic predispositions are identified as major needs.

9. Physicians identified better teaching materials for patients as a major need and this perception was confirmed by participants in community forums who wished for better information about the diagnoses and the long-term impact of disease to assist them in advocating for their family members.

Concerns with confidentiality and the adverse impact of diagnostic information on the lives of individuals permeated the different survey groups and forums.

10. As funding and political support is sought for strengthening Indiana’s surveillance activities, stringent privacy guidelines must be developed and enforced to protect individuals from adverse consequences in insurability, employability and social participation.

The Needs Assessment report prepared in September 2001 was presented to the IGAC and its various subcommittees to complete the third step of the assessment and plan development process. Subcommittees reviewed and weighed the findings and analysis of the Bowen Research Center consultant team and prepared subcommittee reports selecting goals, objectives and action steps that needed to be implemented over the next five years. These were submitted to the Bowen Research Center consultant team who, with the assistance of ISDH staff, assimilated subcommittee recommendations into the plan outlined in Section 3.0 of this report.

BIBLIOGRAPHY

Emory University, http://www.cc.emory.edu/PEDIATRICS/corn/corn.html

HRSA, Stateprofiles, hrsa.gov/1999


U.S. Census Bureau, Quickfacts, census.gov, 2002
**3.0 GENETIC SERVICES PLAN FOR INDIANA**

**GOAL 1:** Incorporate Vital Statistics, Birth Defects, Immunization, Newborn Screening (including Newborn Hearing Screening) and Children Requiring Special Health Care Needs into Child Health Data Repository (CHDR)

*dates dependent upon funding.

**Objectives:**

1.1 Provide data feed from Vital Statistics to Child Health Data Repository (CHDR) by 4/1/03.

1.2 Provide data feed from Newborn Screening Database (includes newborn hearing screening) to CHDR by 2/1/04.

1.3 Provide data feed from Children with Special Health Care Services to CHDR by 12/1/06.

1.4 Provide data feed from Indiana Birth defects Surveillance System to CHDR by 9/1/05.

1.5 Provide data feed from Indiana Immunization Registry to the CHDR by 5/30/06.

**Action Steps:**

- Partner with staff at ISDH currently working on agency-wide integration efforts.

- Hire contractual data systems analyst and develop work plan.

- Direct analyst to incorporate data elements of given databases identified through Genetics Planning Grant into CHDR.

- Establish required data connectivity.

- Ensure compatibility between integrated child health data system and new electronic Indiana Birth and Death Certificate System (operational by 2005) through oversight of Epidemiology Data Director.

- Explore feasibility of data feed from First Steps Program to the CHDR, considering necessary legislative changes required.
GOAL 2: Support Indiana Birth Defect Surveillance System (IBDSS) to improve quality and completeness of data so that it can be effective tool to estimate service needs.

Objective 2.1: Develop and implement a tracking system to verify case identification and reporting to IBDSS by professionals by 1/1/04.

Action Steps:

- Obtain hospital discharge data from the Indiana Hospital and Heath Association and compare with information reported to IBDSS by practitioners.
- Develop and implement protocols to obtain alternate data sources such as electronic medical records.
- Educate midwives regarding IBDSS to ensure infants born at home are identified and served.
- Educate families and the community-at-large about the importance of data collection.

Objective 2.2: Implement a pilot study to determine the accuracy of the IBDSS data by comparison with medical record abstraction by 6/1/04.

Action Steps:

- Develop a protocol for abstraction.
- Identify and train abstractors.
- Obtain medical record abstractors.
- Compare data to IBDSS data.

Objective 2.3: Implement a method to determine the timeliness of the IBDSS reporting and dissemination of service information to families by 7/1/03.

Action Steps:

- Abstraction project in Year 3 will also collect and analyze data to determine time between first diagnosis and first report to IBDSS.
- Devise protocol to determine time lapse from entry into IBDSS and when physician mailed information.
• Protocols will be developed and implemented to increase timeliness of reporting and information distribution.

• Once IBDSS data proven accurate, the IGAC ethics subcommittee, including individuals who represent the perspective of families and consumers of genetic services, will evaluate the appropriateness of distributing information directly to families and implement if deemed feasible.

**Objective 2.4**: Obtain resources for long-term sustenance and support of the IBDSS by 6/1/05.

**Action Steps**:

• Create state position or contract for coordinator of IBDSS who will also direct birth defects prevention activities.

• Research availability of ongoing state, federal, or private funding sources to support this position.

• Write for grant support, if necessary.

• Under IBDSS, establish a consumer advisory committee composed of families with children with special needs.

• Consider seeking legislative appropriation for these activities.

**GOAL 3**: Infants and children identified with special health needs will receive appropriate intervention and services.

**Objective 3.1**: Develop a statewide follow-up and tracking system by which all children identified with hearing loss, where the etiology is not clear, receive comprehensive genetic evaluation and the parents receive appropriate information and counseling (by 1/1/04).

**Action Steps**:

• Develop guidelines for genetic testing and evaluation of infants and children identified with hearing loss.

• Develop educational material to provide appropriate explanations of the genetics of hearing loss as well as benefits of getting genetic testing and evaluation.
• Develop program protocol to identify and track infants and children who receive genetic testing and services.

• Identify available clinic sites for genetic testing and evaluation.

• Provide educational materials to physicians.

**Objective 3.2:** In conjunction with families of children with special needs, develop a system to ensure that all children identified through newborn screening receive appropriate and early intervention, and other services for children with special health care needs by 1/1/03.

**Action Steps:**

• Utilization of services will be tracked through database comparison with First Steps and Children’s Special Health Care Services (CSHCS).

• Physicians who follow children with a positive newborn screen will be surveyed to determine their referral practices for early intervention services.

• Educational activities will be targeted to physicians to increase referral and use of these services.

• Outreach will be provided to midwives regarding newborn screening to ensure infants born at home are screened.

• New physicians identified through newborn screening will be provided with information on early intervention and services for children with special health care needs.

• Verification of referral for support services will be added to confirmed positive newborn screen follow-up.

• Families with children with positive heel-stick screens who receive care out of state will be surveyed regarding awareness of services in Indiana and provided information.

**Objective 3.3:** In conjunction with families of children with special needs, implement a follow-up system for children with birth defects that will ensure the children are receiving appropriate early intervention and services by 3/15/03.
**Objective 3.4:** Implement a system to improve awareness of health care providers regarding early intervention and support services for children with special health care needs by 3/15/03.

**Action Steps:**

- Provide educational information for practitioners on early intervention and other services for children with special health care needs through general mailings to the state professional societies.

**Objective 3.5:** In conjunction with families of children with special needs, implement a system to ensure awareness of families regarding available early intervention and support services for children with special health care needs by 1/15/03.

**Action Steps**

- Assemble educational materials appropriate for families with review from the Indiana Parent Information Network (IPIN).

- Educational Materials will be distributed through health care providers and physicians to distribute to families as appropriate.

- Educational materials will be provided to relevant support groups throughout Indiana for distribution to families.
GOAL 4: Families and medical professionals will have appropriate knowledge about genetics and care of children with special health care needs.

Objective 4.1: Working with families, professionals and the community, develop the infrastructure to promote the medical home concept in Indiana by 8/1/03.

*Dependent on external funding.

**Action Steps:**

- Convene “Medical Home Task Force” to ensure coordination of education and promotion activities and the most efficient use of resources.

- Distribute the “Medical Passport” to families identified through newborn screening, the IBDSS and CSHCS program.

- Assist in promoting educational forums among health care professionals regarding the medical home concept.

- Develop a website through ISDH to support the medical home concept with links to information on genetics, birth defects, newborn screening, early intervention, and children with special health care needs.

- Market website to professionals and families.

Objective 4.2: Develop a physician education medical home campaign to promote health-care services for children with special health care needs by 6/1/04

* dependent on external funding.

**Action Steps:**

- Convene Medical Home Task Force with other medical home trainers in Indiana.

- Identify sites for professional trainings.

- Contract with Unified Training Systems of Indiana to give presentations based on American Academy of Pediatrics Medical Home Training Curricula.

- Medical home educational materials for physicians will also be developed and distributed to physicians who care for children with confirmed positive newborn heel-stick screen, failed newborn hearing screen, or birth defects as identified through IBDSS.
• Distribute information to physicians through mailings from professional societies.

**Objective 4.3:** In conjunction with families of children with special needs, develop a distribution system for medical home education to target families of children with special health care needs by 10/1/03.

**Action Steps:**

• Develop educational materials on medical homes for families.

• Families who have child with positive newborn hearing or heel-stick screen will receive medical home educational information directly from ISDH.

• Physicians who report to the IBDSS will receive medical home education information to distribute to families.

• Workshops will be presented in partnership with IPIN for families on the medical home concept to ensure they receive proper services through medical home.

**Objective 4.4:** Provide support and curriculum material to maintain a quality information environment concerning genetics and related services in Indiana (ongoing as staff and funding permit).

**Action Steps:**

• Construct a website for state listing genetics providers (counselors and physicians) with correct contact information (with provider permission).

• Construct website with links to genetics material for families.

• IGAC should work with state to recommend curriculum minimum requirements for academic institutions for education on genetics.

• Create roll-o-dex cards with website addresses and other contact information for basic genetics data, services, and resources as part of Genetics and Your Practice materials.

• Create topic specific PowerPoint presentations available to genetics specialists to present to lay and professional groups as an expansion of the Genetics and Your Practice presentations.

• Provide genetics materials appropriate to the populations to be served including appropriate language translations.
• Develop an education plan/curricula for improving the awareness of adult conditions (two per year) related to genetics.

**GOAL 5:** Improve reimbursement and financing for genetics services in Indiana.

**Objective 5.1:** Negotiate with Medicaid/Medicare and insurance companies to improve reimbursement for genetic services by 1/6/2007.

**Action Steps:**

- Work with state and national groups to create new International Classification of Disease, 10th revision (ICD-10) and CPT codes to cover genetic conditions not currently in the system.

- Monitor and report on state and national legislative issues regarding reimbursement for genetics direct services and grant support for Genetics Programming.

- Gain representation on appropriate Office of Medicaid Policy and Planning (OMPP) advisory boards, Child Health Policy Board’s Children With Special Needs advisory committee, Medicaid managed care organizations, and other organizations that make recommendations on chronic health issues.

- Provide education to service providers regarding appropriate billing procedures for genetic services.

**GOAL 6:** Establish an infrastructure to expand the integration of genetics into public health, to process genetics issues and to disseminate consensus or policy decisions regarding genetics in public health.

**Objective 6.1:** Establish a standing committee of Newborn Screening Program (NBS) Advisory Board and IGAC subcommittee members with relevant expertise including consumers of genetic services, to evaluate emerging science and to advise when specific genetic tests or prevention measures should be employed by 6/1/03.

**Action Steps:**

- Develop a consensus report or protocols to promote the proper use of IBDSS and other genetics data.
• IGAC screening subcommittee will develop a consensus paper regarding predictive and diagnostic screening for IGAC to adopt.

• IGAC screening and/or ethics committee will review individual diagnostic and predictive screens and develop a consensus report regarding use recommendations.

**Objective 6.2:** ISDH Genetics Disease Program Staff will integrate genetics information and education within existing ISDH programs (ongoing for next five years).

**Action Steps**

• Increase emphasis of ISDH’s Genetic Disease Program in adult onset disease, including improved evaluation, prevention and amelioration efforts.

• Increase coordination between genetics services and the normally distinct programmatic areas of chronic disease and environmental and occupational health and safety.

**Objective 6.3:** IGAC/ISDH will build partnerships with other entities focused on genetics and ethics issues and develop Memoranda of Understanding with them on an ongoing basis.

**Action Steps:**

• IGAC/ISDH will develop a collaboration with Indiana University Center for Bioethics (IUCB) and others (*detailed below) to develop and disseminate consensus documents on genetics policy to advise the legislature and ISDH.

• Ethics subcommittee will be expanded to include representatives from a variety of expert ethics resources in Indiana (*detailed below).

• IGAC will review draft templates for different types of genetic research created by IUCB and give input.

• Develop collaboration with IUSM and Department of Education to promote genetics as a profession.

*Indiana University School of Medicine (IUSM) Department of Medical and Molecular Genetics, Indiana Genome Initiative, Poynter Center for the Study of Ethics and American Institutions, Indiana University (IU) School of Law, Indiana University Center for Bioethics (IUCB), Bioethics Committee of St. Vincent Hospital and Health Care Services, Indiana University School of Public and Environmental Affairs (SPEA) and Indiana University Department of Public Health, genetic consumer organizations and other such ethics resources.
**Objective 6.4:** IGAC/ISDH will develop and disseminate standards and protocols related to genetics services provided in Indiana through an ongoing process as issues arise.

**Action Steps**

- IGAC will research and develop a method of monitoring for genetic discrimination in health insurance and make recommendations for improving privacy protections.

- IGAC/ISDH will seek to ensure that all established ethical standards are upheld in the state’s genetic disease program, such as informed consent and prenatal screening guidelines.

- Improve and increase accessibility to genetic resources and services by supporting expansion of genetic outreach clinics and using non-traditional counseling/education such as telephone consults and telemedicine, and websites.

**GOAL 7:** Periodically review and evaluate the achievements and appropriateness of the State Plan.

**Objective 7.1:** Conduct periodic review of benchmark achievements and barriers to achievement compared to State Plan objectives, twice yearly throughout life of State Plan.

**Action Steps:**

- The genetics specialist from ISDH, the chair of IGAC, and subcommittee chairs, including consumer representatives, will review six-month progress reports on objectives stated in the plan.

- Evaluative judgments will be based on a combination of structure, process, and outcome measures of success ranging from development of materials and data bases, delivery of programs, staffing, and the broader goal of reducing morbidity and mortality in the state.

- A yearly evaluation report will be provided to IGAC concerning achievement of objectives and identification of barriers that may arise during the time-line. Appropriate adjustments to expectations can then be made by IGAC.
4.0 SUMMARY AND CONCLUSIONS

The decision analysis phase of the needs assessment process yielded recommendations for seven major goals with associated objectives and action steps that address the four themes and ten major areas of concern identified in the needs assessment process. Implementation of the seven goals in a genetics plan for the state will result in substantial coverage of all areas identified in the CORN instrument (See Appendix B) as critical for state genetics planning services.

The seven goals include:

1. Incorporate Vital Statistics, Birth Defects, Immunization, Newborn Screening and Children Requiring Special Health Care Needs into Child Health Data Repository (CHDR).
2. Support IB DDS to improve quality and completeness of data so that it can be an effective tool to estimate service needs.
3. Infants and children identified with special health care needs will receive appropriate intervention and services.
4. Families and medical professionals will have knowledge about genetics and care of children with special health care needs.
5. Improve reimbursement and financing for genetics services in Indiana.
6. Establish an infrastructure to expand the integration of genetics into public health, to process genetics issues and to disseminate consensus or policy decisions regarding genetics in public health.
7. Periodically review and evaluate the achievements and appropriateness of the State Plan.

Each goal is supplemented with specific objectives and expected dates of accomplishments, with a corresponding list of action steps necessary to accomplish the objectives. The time lines involve benchmarks across the next five-year period.

An examination of the plan and its goals and objectives using the guidelines provided by CORN indicates that all expected aspects are included. The state health agency has employed a full-time State Genetics coordinator and expects to refill that currently vacant position. The Advisory Board was established and a State Plan developed. The Plan contains aspects for addressing all three core functions of public health—assessment, policy development and assurance. The objectives and action steps require collaboration with other groups and specifically develop linkages with other state programs. A comprehensive legislative history of genetics-related policies was included in the original needs assessment, and a brief update of recent legislation is included as an appendix to this report (See Appendix C). The availability of specific services, family-based, population-based and clinical laboratory, as well as supply of professional providers, is addressed in the needs assessment report and with specific goals in the State Plan. A goal that addresses funding of services is included in the State Plan as are two goals that focus on improving data availability for documentation of needs and services.
The State Plan and the more detailed needs assessment prepared for this project provide key decision makers with an action plan and basis for building support, both financial and political, for the necessary investments of resources. Public health experts predict that genetics issues will become the overriding concern of public health in the near future. Developing and maintaining accurate assessments of need and prevalence of problems are crucial activities that will contribute to quality of life of Indiana residents.

Carrying out the goals and objectives identified in this plan will create an environment in the state that identifies problems and provides intervention for those needing assistance. Accomplishing those goals will entail support from providers, consumers and legislators, and will require a mix of state and public-private partnerships. The challenges will be difficult in a resource constrained environment. Nonetheless, the commitment must be made to vulnerable populations to provide the necessary screening and intervention services to those in need.
APPENDICES
APPENDIX A:

Indiana Genetics Advisory Committee
(IGAC)

Subcommittee Membership List
# INDIANA GENETICS ADVISORY COMMITTEE

## SUBCOMMITTEE MEMBER LIST

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## ETHICAL & LEGAL ISSUES/POLICY DEVELOPMENT

**CO-CHAIRS:** Kimberly Quaid & Joe Rautenberg

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## LABORATORY

**CHAIR:** Patricia Bader, M.D.

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## PUBLIC HEALTH & STANDARDS

**CHAIR (Interim):** Ronda Brewer

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## REIMBURSEMENT FOR GENETIC SERVICES

**CHAIR:** Fernando Escobar

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## SCREENING

**CHAIR:** Weilin Long

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APPENDIX B:

CORN State Assessment Tool for Indiana-

Comparing the State of Indiana’s Genetics Services to the CORN Guidelines for Clinical Genetic Services for the Public's Health
State Assessment Tool for Indiana
Comparing the State of Indiana’s Genetics Services to the CORN Guidelines for Clinical Genetic Services for the Public's Health

Note: Section I-C addresses the content of the plan for the state genetics program, not whether or not a specific program actually exists.

### I. ORGANIZATION & ADMINISTRATION

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<td>e.</td>
<td>A framework for existing quality assurance measures for clinical and laboratory services</td>
<td>☒</td>
</tr>
<tr>
<td>f.</td>
<td>Other:</td>
<td></td>
</tr>
<tr>
<td>4.</td>
<td>Collaboration with relevant groups</td>
<td></td>
</tr>
<tr>
<td>a.</td>
<td>Consumers</td>
<td>☒</td>
</tr>
<tr>
<td>b.</td>
<td>Researchers</td>
<td>☒</td>
</tr>
<tr>
<td>c.</td>
<td>Genetics Services Providers</td>
<td>☒</td>
</tr>
<tr>
<td>d.</td>
<td>Teachers</td>
<td>☒</td>
</tr>
<tr>
<td>e.</td>
<td>Other:</td>
<td></td>
</tr>
<tr>
<td>E.</td>
<td>Linkages with Other State Programs</td>
<td></td>
</tr>
<tr>
<td>1.</td>
<td>Chronic Disease</td>
<td>☒</td>
</tr>
<tr>
<td>2.</td>
<td>Epidemiology</td>
<td>☒</td>
</tr>
<tr>
<td>3.</td>
<td>Other: First Steps, CSHCS, NBS</td>
<td>☒</td>
</tr>
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II. SERVICES

A. Family Based Services

<table>
<thead>
<tr>
<th></th>
<th>Centrally Coordinated</th>
<th>Available Regionally (in state)</th>
<th>Available in Select Communities</th>
<th>Coordinated/Available in Other States</th>
<th>Not Coordinated with Other States/Not Available</th>
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<tbody>
<tr>
<td>1.</td>
<td>General Genetic Clinics</td>
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<tr>
<td>2.</td>
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<tr>
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<td>✗</td>
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</tr>
<tr>
<td></td>
<td>b. Cystic Fibrosis</td>
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<td>✗</td>
<td>✗</td>
<td>✗</td>
</tr>
<tr>
<td></td>
<td>c. Other:</td>
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<td>4.</td>
<td>Prenatal Genetics Clinics</td>
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B. Population Based Services

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<th>Centrally Coordinated</th>
<th>Available Regionally (in state)</th>
<th>Available in Select Communities</th>
<th>Coordinated/Available in Other States</th>
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<tbody>
<tr>
<td>1.</td>
<td>Prenatal Screening</td>
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<td>✗</td>
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<tr>
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<td></td>
<td>b. AMA</td>
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<tr>
<td></td>
<td>c. Family Hx</td>
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<td>✗</td>
<td>✗</td>
</tr>
<tr>
<td></td>
<td>d. Carrier screening for targeted populations</td>
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<td>✗</td>
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<td>✗</td>
</tr>
<tr>
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<td>e. Maternal Serum</td>
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<tr>
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<td>f. Maternal Age</td>
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<tr>
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<tr>
<td>2.</td>
<td>Newborn Screening</td>
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<td>✗</td>
<td>✗</td>
<td>✗</td>
</tr>
<tr>
<td></td>
<td>a. Lab Services</td>
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<td>✗</td>
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<td></td>
<td>b. Follow Up</td>
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<td>✗</td>
<td>✗</td>
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</tr>
<tr>
<td></td>
<td>c. CH</td>
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<tr>
<td></td>
<td>d. PKU</td>
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<tr>
<td></td>
<td>e. Hemoglobinopathie</td>
<td>✗</td>
<td>✗</td>
<td>✗</td>
<td>✗</td>
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<td></td>
<td>f. CAH</td>
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<tr>
<td>3.</td>
<td>Childhood Screening</td>
<td>✗</td>
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<tr>
<td>4.</td>
<td>Adult Screening</td>
<td>✗</td>
<td>✗</td>
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</tr>
</tbody>
</table>
C. Clinical Laboratory Services | Available In-State | Coordinated with Out-of-State Lab | Not Available/Not Coordinated
--- | --- | --- | ---
1. Cytogenetics | ☒ | | 
2. Biochemical Genetics | ☒ | | 
3. Molecular Genetics | ☒ | | 

D. Modes of Service Delivery | Check all that apply
--- | ---
1. Genetics Unit of State Health Department | ☒
2. Large, comprehensive genetics center | ☒
3. Genetics unit of a comprehensive managed care facility | ☒
4. Resident genetics unit within a primary health care facility | ☒
5. Resident board certified genetic counselor and/or PhD medical geneticist with periodic visits by a board certified MD Medical Geneticist | ☒
6. Periodic visits by a board certified genetic counselor, medical geneticist, or other staff with local coordinators at outreach clinics | ☒
7. Genetics clinics in the private sector conducted by trained MD geneticists | ☒
8. Board certified genetic counselor and/or PhD medical geneticist within single disease/medical specialty setting | ☒
9. Other: | 

E. Genetics Professionals | Centrally Coordinated | Available Regionally | Available in Select Communities | Coordinate d/Available in Other States | Not Coordinated/Not Available
--- | --- | --- | --- | --- | ---
1. Clinical Geneticist | ☒ | ☒ | ☒ | | 
2. PhD Medical Geneticist | | | | | 
3. Genetic Counselor | ☒ | ☒ | | | 
4. Clinical Cytogeneticist | ☒ | ☒ | ☒ | | 
5. Clinical Biochemical Geneticist | ☒ | | | | 
6. Clinical Molecular Geneticist | ☒ | | | | 
7. Cytogenetic Technologist | ☒ | ☒ | ☒ | | 
8. Genetics Nurse | | | | | 
9. Advance Practice Nurse in Genetics | | | | | 
10. Perinatologist/Obstetrician | ☒ | ☒ | ☒ | | 
11. Dietician | ☒ | | | | 
12. Other: | | | | |
### III. FUNDING OF SERVICES

<p>| | | |</p>
<table>
<thead>
<tr>
<th></th>
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<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>A.</td>
<td>Medicaid, Medicare reimbursement</td>
<td>☒</td>
</tr>
<tr>
<td>B.</td>
<td>Third Party Carriers reimbursement</td>
<td>☒</td>
</tr>
<tr>
<td>C.</td>
<td>Newborn Screening Surcharge</td>
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</tr>
<tr>
<td>D.</td>
<td>State General Funds</td>
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</tr>
<tr>
<td>E.</td>
<td>Federal Title V (MCH Block Grant)</td>
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</tr>
<tr>
<td>F.</td>
<td>Other State and/or Federal Grants</td>
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</tr>
<tr>
<td>G.</td>
<td>Specific Disease-Related Organizations</td>
<td></td>
</tr>
<tr>
<td>I.</td>
<td>Other:</td>
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</table>

### IV. DOCUMENTATION OF NEEDS & SERVICES

<table>
<thead>
<tr>
<th></th>
<th>Data Sources</th>
<th>Available; Utilized</th>
<th>Available; Not Utilized</th>
<th>Not Available</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>State level clinical genetics database</td>
<td>☒</td>
<td></td>
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</tr>
<tr>
<td>2.</td>
<td>Newborn screening database</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>4.</td>
<td>Statewide hospital discharge data</td>
<td></td>
<td>☒</td>
<td></td>
</tr>
<tr>
<td>5.</td>
<td>Medicaid/Medicare eligibility, claims, provider datasets</td>
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<td>☒</td>
<td></td>
</tr>
<tr>
<td>6.</td>
<td>Local/Statewide/Regional cytogenetics registry</td>
<td></td>
<td>☒</td>
<td></td>
</tr>
<tr>
<td>7.</td>
<td>Local/Statewide/Regional birth defects registry</td>
<td>☒</td>
<td></td>
<td></td>
</tr>
<tr>
<td>8.</td>
<td>Local/Statewide/Regional population based cancer/tumor registry</td>
<td></td>
<td>☒</td>
<td></td>
</tr>
<tr>
<td>9.</td>
<td>Directory of genetic service providers and referral sources</td>
<td>☒</td>
<td></td>
<td></td>
</tr>
<tr>
<td>10.</td>
<td>Cytogenetics laboratory databases collected by ACT</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11.</td>
<td>Federal census data</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>12.</td>
<td>Special surveys and projects:</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>a. Pregnancy Risk Assessment Monitoring System (PRAMS)</td>
<td></td>
<td></td>
<td>☒</td>
</tr>
<tr>
<td></td>
<td>b. National Maternal &amp; Infant Health Survey</td>
<td></td>
<td></td>
<td>☒</td>
</tr>
<tr>
<td></td>
<td>c. Behavioral Risk Factor Surveillance System (BRFSS)</td>
<td></td>
<td>☒</td>
<td></td>
</tr>
<tr>
<td></td>
<td>d. National Survey of Family Growth</td>
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<td>☒</td>
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<tr>
<td>13.</td>
<td>Other:</td>
<td></td>
<td></td>
<td>☒</td>
</tr>
<tr>
<td>B.</td>
<td>Data Linkages</td>
<td>Linkage in Place</td>
<td>Planned Linkage</td>
<td>Linkages Being Considered</td>
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<tr>
<td>----</td>
<td>------------------------------------------------------------------------------</td>
<td>------------------</td>
<td>-----------------</td>
<td>---------------------------</td>
</tr>
<tr>
<td>1.</td>
<td>Birth &amp; death for all deaths up to six years of age</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>2.</td>
<td>Birth defects &amp; tumor registry for all pediatric cancer cases</td>
<td>☐</td>
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</tr>
<tr>
<td>3.</td>
<td>Birth defects registry records with vital statistics</td>
<td>☒</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>4.</td>
<td>Inpatient hospital discharge records with birth certificates</td>
<td>☒</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>5.</td>
<td>Newborn screening records with birth certificates</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>6.</td>
<td>MSAFP/AFAFP/Triple Screen with vital statistics</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>7.</td>
<td>Statewide clinical genetics services database and birth/fetal death certificates (in form of numerator/denominator ratio)</td>
<td>☐</td>
<td>☐</td>
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</tr>
<tr>
<td>8.</td>
<td>System for direct referral from clinical genetics to early intervention services for infants &lt; 3 years of age; Children with Special Health Care Needs (CSHCN); Supplemental Social Insurance (SSI); etc.</td>
<td>☐</td>
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</tr>
<tr>
<td>9.</td>
<td>Other: Universal Newborn Hearing Screening /Birth Certificate</td>
<td>☐</td>
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</tr>
</tbody>
</table>

**Note:** An alternative way of categorizing services is according to their level of prevention (primary, secondary, or tertiary). Below is a framework for assessing services in this manner.
### V. PREVENTION

#### A. Primary Prevention Programs

1. *Folic Acid Education*  
   - In Place: ☒  
   - Planned: ☐

2. *Teratogen Information Services*  
   - In Place: ☒  
   - Planned: ☐

3. *Other: Prenatal Substance Use Prevention Program*  
   - In Place: ☒  
   - Planned: ☐

4. *Other: Baby First Right From the Start*  
   - In Place: ☒  
   - Planned: ☐

5. *Other:*  
   - In Place: ☐  
   - Planned: ☐

#### B. Secondary Prevention Programs

- **Readily available to all state residents**: ☐
- **Readily available to most state residents**: ☒
- **Readily available to some state residents**: ☐
- **Not readily available to state residents**: ☐

1. **Prenatal Screening**
2. **Newborn Screening**
3. **Childhood Screening**
4. **Adult Screening**

#### C. Tertiary Prevention Programs

- **Check all that apply**

1. *Educational and other special services for individuals with special needs*  
   - In Place: ☒  

2. *Appropriate management of genetic disorders*  
   - In Place: ☒  

3. *Access to medical devices*  
   - In Place: ☒  

4. *Referral of families to support groups; or facilitation of contact with similarly affected families*  
   - In Place: ☒  

5. *Other:*  
   - In Place: ☐  

APPENDIX C:

Indiana Legislative Activity Update
Indiana Legislative Activity Update

In 1991 a bill was introduced which subsequently resulted in the amendment of the Indiana Code, IC 12-17-15, Chapter 15 entitled “Infants and Toddlers With Disabilities Program” (First Steps). Public Law 34-1991 established the Comprehensive Early Childhood Grant Program, known as Step Ahead (1). The vision of this program was to provide statewide, comprehensive, uninterrupted delivery of services for children (birth to thirteen years of age) in Indiana, ensuring accessibility, affordability and quality (1). The Implementation of Comprehensive Plan was prepared by Developmental Disabilities Task Force and addressed in Public Law 245-1997, Section 1. The Commission on Mental Retardation and Developmental Disabilities (MRDD) was established by Public Law 78-1994, which was subsequently amended with Public Law 272-1999, extending the life of the commission to January 1, 2005 (1).

The most recent activity in Indiana, which specifically addresses birth defects, occurred in 2002, Senate Bill 0139 – Birth Defects Registry was introduced to the 112th General Assembly of the Indiana Senate on January 7th (2). The bill required the Indiana State Department of Health to: 1) inform parents of children with birth problems about physicians and local community resources; and 2) report to the legislature council any birth problem trends identified through the data collected. The bill was subsequently passed by the Senate and the Indiana House of Representatives. Governor O’Bannon signed the bill on March 14th. This bill for an Act amended the Indiana Code concerning health (IC 16-38-4 Chapter 4. Birth Problems Registry) and is effective as of July 1, 2002 (2).

Bibliography: