State of Illinois
Illinois Department of Public Health

Genetic Program Manual for Local Health Departments

August 2008

Illinois Department of PUBLIC HEALTH

TABLE OF CONTENTS

PREFACE

The Illinois Department of Public Health (IDPH) developed this handbook to facilitate the integration of family health history into all programs currently provided through local public health departments, with emphasis on genetics and the expanding role of prevention. Due to advances such as the human genome project, we can better relate genetics to our family history, environment, personal lifestyle choices and individual behaviors. This explains the 21st century term –-- genomics.

The manual describes the reasons why genetic services should be an essential part of every primary health care and prevention program, and provides recommendations for incorporating the use of the family health history questionnaire (FHHQ) into existing programs. Included in this manual is the FHHQ and detailed rationale that will help identify those clients who can benefit from genetic counseling and/or other services, protocols for making referrals to a genetic center and for establishing an on-site genetic clinic. Also included are resources for clients and public health providers and quarterly reporting instructions for health department grantees.

This manual is periodically updated in an effort to offer the most current suggestions and recommendations. However, if you have any comments or questions, please call the Department's Genetics Program at 217-785-8101.

INTRODUCTION

WHAT ARE GENETIC SERVICES?

The purpose of genetic services is to provide information about a specific condition or family concern by genetic health care professionals—genetic counselor and/or a geneticist. A referral to a clinical genetic center will help the client understand genetic inheritance, family history of a particular concern or condition and, if possible, determine an accurate diagnosis.

There are many components to a clinical genetic evaluation including a physical examination, review of family and medical histories, laboratory and diagnostic testing, treatment, patient education and genetic counseling. Since there are thousands of known genetic disorders, many with overlapping features, an accurate diagnosis is often complicated and should be performed by a health professional with advanced training in genetics. Determining an accurate diagnosis is necessary for genetic counseling of recurrence risks in future offspring and prenatal diagnosis considerations for future pregnancies.

Diagnostic evaluations require comprehensive patient assessment. Many times laboratory testing or other diagnostic technologies are necessary. Some of the more common examples include: chromosome analysis performed by cytogenetic laboratories; metabolic testing by biochemical laboratories; and identification of specific mutations in disease-causing genes by DNA laboratories. Diagnostic tests may involve radiologic imaging (X-ray, computerized tomographic imaging (CT scan), magnetic resonance imaging (MRI), ultrasonography, electroencephalogram (EEG), electrocardiogram (EKG) and hearing or vision tests. The number and variety of screening tests are increasing rapidly. These include newborn metabolic screening, carrier testing, prenatal screening and presymptomatic disease detection.

Accurate diagnosis is important for appropriate treatment and clinical management. Available treatment options involve drug therapy, dietary management, surgical intervention, rehabilitation services and special education.

Genetic counseling provides information and education regarding testing procedures, diagnosis, prognosis, available options, recurrence risks and support services.

In summary, genetic services provided at a clinical genetic center encompass the following:

- **Comprehensive patient evaluation**
- **Laboratory and other clinical testing**
- **Accurate diagnosis by a genetic specialist**
- **Appropriate treatment**
- **Genetic counseling**

HOW COMMON ARE GENETIC DISEASES?

To date, more than 6,000 single-gene genetic diseases have been described. In recent years, it has become more evident that genetic inheritance also plays an important role in a wide range of conditions, including birth defects, chronic diseases (heart disease, diabetes, asthma, etc.), mental illness and cancer. In addition, genetic factors contribute significantly to reproductive failure, as well as to infant and childhood morbidity and mortality. It is estimated that 50 percent to 60 percent of first trimester miscarriages and 7 percent of stillbirths and neonatal deaths are due to chromosomal abnormalities.[1] In the state of Illinois, approximately 5,000 infants are born each year with a significant birth defect, many of which are due to chromosomal or genetic anomalies. In fact, congenital disorders are the leading cause of infant mortality. Furthermore, 30 percent of all pediatric hospitalizations involve a child with a genetic disorder.[2]

In addition, at least 80 percent of the population older than the age of 25 will develop a health condition with a genetic component.[2] Individuals who are affected by, or at risk for, an inherited disorder can be of any age, and represent people of all racial, ethnic, socioeconomic and educational backgrounds. In Illinois, more than 2.5 million people have a disease with genetic or hereditary components. There may be numerous implications that are likely to impact the entire family.

These data provide compelling reasons to include genetics as an integral component of public health services. Providers of public health programs have the opportunity and the obligation to offer genetic services to all citizens of our state.

The challenge for public health professionals at the local health department is to determine which clients should be offered a referral to a genetic center. Obtaining information about health and family history will assist the provider in identifying individuals who might benefit from a genetic consultation. In the sections that follow, information and a family health history questionnaire are provided to aid in the appropriate identification of those clients who could benefit from genetic services.

In summary

- **6,000 single-gene genetic disorders have been identified, with genetic factors also playing a key role in the development of many chronic diseases**
- **50 percent to 60 percent of first trimester miscarriages and 7 percent of stillbirths and neonatal deaths are due to a chromosomal abnormality**
- **5,000 infants are born with a major birth defect annually in Illinois**

- **30 percent of pediatric hospitalizations are due to a genetic disease**
- **80 percent of individuals older than the age of 25 will develop a health condition with a genetic component**
- **2.5 million Illinois residents are affected by a genetic disorder**

Some of the more common genetic disorders are listed below:

Common pediatric genetic disorders by type of genetic inheritance:

- Chromosomal:
  - Down syndrome
  - Trisomy 13
  - Trisomy 18
  - Turner syndrome
- Autosomal recessive:
  - Sickle cell disease
  - Alpha- and beta-thalassemia
  - Cystic fibrosis
  - Inborn errors of metabolism (newborn screening disorders)
- Autosomal dominant:
  - Neurofibromatosis
  - Achondroplasia
  - Marfan syndrome
- X-linked:
  - Duchenne Muscular Dystrophy
  - Fragile-X syndrome
  - Hemophilia
- Multifactorial:
  - Spina bifida
  - Cleft lip/palate
  - Isolated congenital heart defect
  - Asthma

Common/chronic adult diseases with genetic components (usually in a multifactorial inheritance pattern from a genetic predisposition as well as lifestyle choices):

- Heart disease
- Cancer
- Diabetes
- Arthritis
- Alzheimer disease
- Asthma

WHO PROVIDES GENETIC SERVICES?

Genetic centers are usually affiliated with university-based or tertiary care medical facilities. The services are provided by a comprehensive team of health care professionals with specialized training in genetics. This team may include the following:

- **Medical geneticists** are physicians, usually pediatricians or obstetricians, with advanced training in clinical genetics. A geneticist's role is to provide comprehensive clinical evaluation in an attempt to determine a medical diagnosis. This person also provides appropriate medical management, follow-up and counseling to the patient and family.

- **Genetic counselors** are licensed professionals, trained at the master's level in the areas of clinical genetics and psychosocial counseling. They are responsible for gathering pertinent medical, developmental and family histories that assist in risk assessment and diagnosis. Genetic counselors provide education and psychosocial support regarding the diagnosis, prognosis, disease management, recurrence risks, and prenatal diagnosis and management options.

- **Genetic nurse specialists** are registered nurses who have advanced training in clinical genetics. In the clinical setting, these nurses perform some aspects of the physical examination and also may assess risks and provide information to the clients.

- **Public health genetic coordinators** are registered nurses with experience in public health who have received continuing education in genetics. Their primary role is to identify individuals in their health departments through the use of the IDPH family health history questionnaire who would benefit from genetic services. They serve as the liaison between the genetic center, the client and the local health department so that appropriate referrals and medical follow-up are achieved.

- **Registered dietitians** are licensed health professionals who have specialized training in the nutritional aspects of genetic conditions. Since certain metabolic diseases require dietary intervention, dietitians help the family and patient to understand and maintain the necessary diet.

- **Social workers** are allied health professionals who can facilitate referrals to necessary community resources and provide family support.

WHAT IS THE ROLE OF THE LOCAL PUBLIC HEALTH DEPARTMENT IN THE PROVISION OF GENETIC SERVICES?

The Illinois Department of Public Health provides grants for genetic services to local health departments. It is recommended that each local health department have one designated individual who coordinates all referrals and acts as the contact person with the regional or local genetic center. This individual also serves as a resource person and facilitates genetic in-service programs. The coordinator is generally a public health nurse who has received continuing education in genetics.

Staff in all local health department programs will identify those clients and families who may benefit from a genetic consultation by completing the enclosed family health history questionnaire. The completed questionnaire will be reviewed by the genetic coordinator to assess whether a referral is appropriate. The genetic coordinator will ensure that appropriate follow-up services are offered and available to the client through ongoing interaction with the genetic center. Occasionally, it may be necessary for the coordinator to obtain information for the client, such as material about a specific disorder or support group. The genetic coordinator also should pursue further professional development opportunities by participating in educational offerings and facilitate continuing education activities in the area of genetics for other local health department staff.

The role of the local health department in providing genetic services is:

- **to designate a nurse within the health department as the genetic coordinator**
- **to establish a working relationship with the genetic center**
- **to establish screening, referral and follow-up protocols for clients served through all programs in the health department**
- **to attend and provide on-going educational opportunities in clinical genetics for the coordinator and other health department staff**

WHAT IS THE ROLE OF THE GENETIC COORDINATOR?

The genetic coordinator should serve as the point of contact in the health department for genetic, newborn screening (NBS) and sudden infant death syndrome (SIDS) referrals.

The genetic coordinator should ensure that the expanded Family Health History questionnaire is implemented within the health department programs. It is mandatory for each health department to screen clients in at least one program area within the health department. However, the overall goal is to continue to expand the use of the questionnaire to eventually reach all clients, both children and adults, to effectively screen and educate clients about the transmission and effects of inherited conditions and identify healthy lifestyle choices in the role of disease prevention.

Some of the programs that can effectively use the genetic screening tool for family health history are:

- Women, Infant and Children Program (WIC), Family Case Management (FCM), family planning, prenatal clients, sexually transmitted diseases, adverse pregnancy outcome referral system (APORS), oral health, hearing and vision, blood pressure/cholesterol/osteoporosis screenings, breast and cervical cancer screening.
- other programs provided by the health department – think education and prevention.

The genetic coordinator should:

- establish screening, referral and follow-up protocols for clients served through all programs in the health department.
- assist clients with referrals and act as a liaison to genetic centers for consultation.
- establish a working relationship with the local genetic center or satellite clinic.

It is mandatory that the genetic coordinator have educational programs designed to educate clients, local health department (LHD) professional staff and their local community with genetics, NBS, SIDS services and resources. Educate the community and health care professionals that the health department has genetic resources for referral and educational materials available and can assist in obtaining genetic referrals. Some of the areas to include in your educational activities are:

- Health department staff
- Physicians – obstetrics/gynecology (OB/GYN), family practice, dermatologists, opticians
- Hospitals – for current literature for NBS, SIDS, genetic resources
- Clients
- Consumers
- Schools – teachers, nurses

The genetic coordinator is responsible to make sure that quarterly reports are submitted timely and accurately with the following schedule:

First Quarter is due October 31
Second Quarter is due January 31
Third Quarter is due April 30
Fourth Quarter is due July 31

The genetic coordinator should encourage all new nurses and nurses that are involved in the use of the questionnaire to attend educational opportunities provided by the Department, especially the annual "Genetics: A Family Affair Conference" held each year in the spring. The coordinator also should attend each year to remain up to date and to be able to update staff who are unable to attend.

WHAT HAPPENS DURING A GENETIC CONSULTATION?

Once it has been determined that a family and/or client may benefit from genetic services, the public health genetic coordinator will contact the genetic center or satellite clinic where these services are available. Communication between the coordinator and the genetic center will determine whether further evaluation is necessary. If a referral is made, certain information will be required by the genetic center. This information will vary depending on the nature of the consultation, but may include family, pregnancy, medical and developmental histories and records. In addition, a comprehensive physical examination of the client and other family members may be performed. When medical tests or procedures are necessary, they will be explained to the individual or family at the time of the consultation. The combination of all the information obtained through histories, records, tests and procedures are used by the genetics team in making a diagnosis and formulating a management strategy.

When a diagnosis is established, several issues will be discussed including prognosis, information about the condition, appropriate management and treatment options, risks of recurrence for family members and reproductive options available to the family. Recommendations may include additional diagnostic testing and referrals to other medical specialists, social service and support organizations. For some clients, ongoing follow-up services are provided through the clinical genetic center. The center also will provide information to the public health genetic coordinator regarding the diagnosis, prognosis and recommendations made for each client.

In summary, the following may occur at a genetic visit:

- **Collection of family/medical history and records**
- **Physical examination of the client or family**
- **Explanation of tests and procedures**
- **Discussion of diagnosis/prognosis**
- **Discussion of recurrence risks**
- **Discussion of management, treatment and reproductive options**
- **Referrals**
- **Ongoing follow-up by genetic center**
- **Feedback to public health genetic coordinator**

WHAT ARE THE COSTS FOR GENETIC SERVICES?

Costs for genetic consultations vary depending on the type of services provided. Each genetic center has a fee scale, and reimbursement through third-party payers is available for some services. Private insurance, the Illinois Department of Human Services and the Division of Specialized Care for Children at the University of Illinois and the All Kids or Family Care Programs are examples of potential payment resources. Most genetic centers will attempt to work with the family to provide the necessary services. However, if there are any financial concerns, they should be discussed prior to the consultation so that efforts can be made to best accommodate the needs of the family. The Illinois Department of Public Health does not have funding available to assist in the cost of genetic testing.

GUIDELINES FOR GENETIC SERVICES

GENETIC SERVICES FOR CHILDREN

WHY ARE GENETIC SERVICES BENEFICIAL FOR CHILDREN?

Genetic and congenital conditions are not rare. Every year, thousands of parents in Illinois learn that their child has a birth defect, genetic disorder or mental retardation. The purpose of a genetic consultation for these families is to establish a diagnosis and to provide information about the cause of the condition, the expected outcome for the child, possible treatment methods and the chance of the condition occurring again in the family. This information helps the family to provide the day-to-day care for the child and to cope with the emotional aspects of having a child with special health care needs. Genetic professionals can help the family make plans relating to medical care, reproductive options, schooling and support services.

WHO WOULD BENEFIT FROM GENETIC SERVICES?

An infant, child or adolescent with any of the following may benefit from genetic services:

- one or more major birth defects
- confirmed positive newborn screening test
- known chromosomal abnormality
- known genetic condition
- any unusual facial appearance or physical features
- developmental delay, mental retardation or significant learning disabilities
- unexplained abnormal neurological findings, e.g., deafness, seizures, hypotonia
- regression of developmental skills
- unexplained failure to thrive
- abnormal growth (height, weight or head circumference)

For further explanation of these indications, please refer to the rationale for the FHHQ.

HOW CAN GENETIC SERVICES FOR CHILDREN BE IMPLEMENTED?

All children seen in local health departments should be screened in order to assess their need for a genetic referral. Health department staff from all programs that target children should utilize the FHHQ. These programs include:

- **Pediatric Family Case Management**
- **High-Risk Infant Follow-up**
- **Pediatric Primary Care**
- **Early Intervention**
- **Special Supplemental Nutrition Program for Women, Infants and Children (WIC)**
- **Immunization**
- **Oral Health**
- **Hearing and Vision Screening**

GENETIC SERVICES FOR ADULTS

WHY ARE GENETIC SERVICES BENEFICIAL FOR ADULTS?

Genetic services for adults can provide a variety of information, ranging from implications for an individual's personal health to issues relating to future reproductive plans. When common chronic disorders such as heart disease, cancer, diabetes, arthritis, and Alzheimer disease are considered, nearly every family is impacted by a genetic condition in some way. It is estimated that approximately 80 percent of the adult population has a condition with a genetic component.[2] If a genetic condition is present in a family member, assessment of the entire family history is useful to help explain the potential for recurrence. Genetic services for adults provide information regarding diagnosis, early detection, treatment, healthy lifestyle choices, recurrence risks and support services.

WHO WOULD BENEFIT FROM GENETIC SERVICES?

An adult with a family history of any of the following may benefit from genetic services:

- birth defects or other genetic/inherited diseases
- learning problems, developmental delay, mental retardation
- neurological disorders, muscular diseases
- abnormalities of the bones or skin
- disorders of the blood
- vision or hearing loss
- cancers or tumors
- multiple miscarriages, stillbirth or infant death

See FHHQ for additional questions related to women of childbearing age or to women who are currently pregnant.

For further explanation of these indications, please refer to rationale for FHHQ.

GENETIC SERVICES FOR WOMEN

WHY ARE GENETIC SERVICES BENEFICIAL FOR WOMEN?

Women of reproductive age can benefit from genetic services for a number of reasons. Preconception genetic counseling helps identify risk factors based on maternal health conditions, possible teratogenic exposures and family history. This allows for appropriate medical management, risk assessment and education in order to optimize future pregnancy outcomes. In addition, all women should receive education regarding the benefits of folic acid supplementation, which reduces the risk for neural tube defects in their offspring. During pregnancy, women who have been identified through preconception counseling or prenatal screening or who have other risk factors may benefit from additional diagnostic testing, such as chorionic villus sampling (CVS) or amniocentesis, to detect chromosome abnormalities and other genetic conditions. Genetic services for women provide information regarding diagnosis, early detection, treatment, recurrence risks and support services.

WHO WOULD BENEFIT FROM GENETIC SERVICES?

Any woman of childbearing age or who is currently pregnant may gain useful information for use in decision making from genetic services if any of the following apply:

- is older than the age of 35 years
- has abnormal prenatal test or screening results
- has diabetes or seizures
- is currently taking prescription medications
- is exposed to cigarettes, alcohol or illicit drugs
- is exposed to harmful substances in the workplace
- is related by blood to her partner

For further explanation of these indications, please refer to FHHQ rationale.

HOW CAN GENETIC SERVICES FOR WOMEN BE IMPLEMENTED?

All women seen in the local health departments should be screened in order to assess their need for a genetic referral. Health department staff from all programs that target women should utilize the questionnaire. These programs include:

- Preconception Screening
- Family Planning
- Prenatal Clinics
- Family Case Management
- Special Supplemental Nutrition Program for Women, Infants and Children (WIC)
- Sexually Transmitted Diseases
- Oral Health
- Hearing and Vision Screening
- Blood Pressure Screening
- Breast and Cervical Cancer Screening

WHO WOULD BENEFIT FROM EDUCATIONAL INFORMATION REGARDING COMMON CHRONIC DISEASES?

For many common chronic adult illnesses, such as heart disease, cancer, diabetes, arthritis, and Alzheimer disease, a genetic center referral will not be necessary ***unless*** the client is interested in an in-depth evaluation, which may include DNA analysis and formal genetic counseling. Since most common chronic illnesses are lifestyle dependent, educating the client about the benefits of healthy lifestyle choices, (regular exercise, weight control, healthy diet and nutrition information) in reducing or delaying the onset of these conditions is of great public health importance. An adult with a personal and/or family history of any of the following may benefit from education and possibly a referral to genetic services:

- early cardiovascular event (heart attack, stroke) or disease (<50 years for men, <60 years for women)
- early onset cancer
- adult onset diabetes
- arthritis
- Alzheimer disease
- obesity

HOW CAN GENETIC SERVICES FOR ADULTS BE MPLEMENTED?

All adults seen in local health departments should be screened in order to assess their need for education or a genetic referral. Health department staff from all programs that target adults should utilize the FHHQ. These programs include:

- Family Case Management
- Special Supplemental Nutrition Program for Women, Infants and Children (WIC)
- Preconception Screening
- Family Planning
- Sexually Transmitted Diseases
- Oral Health
- Hearing and Vision Screening
- Blood Pressure Screening
- Breast and Cervical Cancer Screening

FAMILY HEALTH HISTORY QUESTIONNAIRE (FHHQ)

The following is the revised and expanded family health history questionnaire formerly identified as the genetic screening tool that was developed to be used with all families beginning July 1, 2005. It is recommended that the questionnaire be administered by a staff member, preferably a nurse, during an interview process. Periodically, the information should be reviewed and updated with the client. If the individual responding to the questionnaire is not the client (e.g., mother and child), it is important for the staff member to document the name of the respondent and his or her relationship to the client.

Included with the questionnaire is a detailed explanation of each question. **All questionnaires with a positive indicator or a "yes" answer with family members having a history of a disorder or client concern should be forwarded to the genetic coordinator within the local health department.** The coordinator will review the information and contact the client to arrange a referral with the regional genetic center when indicated and/or requested by the client or provide further information and local resources.

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
EXPANDED GENETIC SCREENING TOOL

Note: Completion of this form is optional. Disclosure of this information is voluntary and there is no penalty for noncompliance.

A positive response should be reviewed by the local health department genetic coordinator and should be referred to a genetic center if they are high risk. See Rationale for details.

Client's Name_____
Birth Date_____ Age____ Sex____
Date of Assessment_____

County of Residence_____
Agency/Program Name_____
Staff Person Completing Form_____

Race/Ethnicity: Client_____ Partner_____
Religion: Jewish Non-Jewish (please circle one)

If respondent is different than client, please indicate name and relationship to client.

For the respondent:Mark an X if you, your partner, your child/children or other family members have or have had any of the following:	You	Your Partner	Your Child/ Children	Other Family Members	Comments
1. Medical problems since birth or birth defect (cleft lip/palate, spina bifida, heart defects, ambiguous genitalia, etc.)					
2. A child with any unusual facial appearance or physical features					
3. Learning problems, developmental delay, mental retardation					
4. A child with regression of developmental skills					
5. A child with abnormal growth (height, weight, or head circumference)					
6. A stillborn child or an early infant death					
7. A child with a confirmed positive newborn screening test					
8. Two or more first trimester miscarriages					
9. You and your partner are blood relatives					
10. Chromosomal abnormalities (Down syndrome, trisomy 13, Turner Syndrome, etc.)					
11. Neurological or muscular diseases (muscular dystrophies, Huntington's disease, etc.)					
12. Disorders of the blood (sickle cell disease, thalassemia, hemophilia, factor V Leiden, etc.)					
13. Vision loss (not glasses) or hearing loss at an early age					
14. Abnormalities of the bones or skin (brittle bones, bone deformities, unusual birth marks)					
15. Other genetic diseases (cystic fibrosis, polycystic kidney disease, Tay Sachs disease, newborn screening disorder, i.e., PKU)					
*16. A. Breast, ovarian, colon cancer, history of colon polyps or skin cancer prior to age 50 B. An evaluation and/or diagnosis for any cancer conditions					
*17. A. A heart attack, stroke or sudden early death (women <60 yrs; men <50 yrs) B. An evaluation and/or diagnosis for a heart condition C. High blood pressure and/or high cholesterol					
**18. Diabetes (Type 1, Type 2 or Maturity Onset Diabetes of the Young (MODY)					
**19. Asthma					
**20. Arthritis					

*Clients with a positive response regarding questions #16 and #17 should only be referred if they are high risk or have a significant concern. Please see rationale for details.

**At this time, clients with a positive response regarding questions #18, #19 and #20, do not need a genetic referral but may be offered educational materials. Please see rationale for details.

ADDITIONAL QUESTIONS FOR WOMEN: **For any prenatal case, a positive response requires an immediate referral to the genetic coordinator to your local genetic center, except for questions 5 and 6.**

Mark an X if any of the following apply to the respondent	Yes	No	Comments
1. Are you over the age of 35 years?			
2. Do you have diabetes or a history of gestational diabetes?			
3. Do you have seizures or being treated for seizures?			
4. Are you currently taking prescription drugs?			
5. Are you currently taking vitamins containing folic acid?			
6. Do you have concerns about exposures to cigarette smoking?			
7. Do you have concerns about alcohol consumption or drug use?			
8. Do you have concerns about exposures to harmful substances in the workplace?			
9. If you are pregnant, have you had chicken pox, rubella or CMV during this pregnancy?			
10. If you are pregnant, have you had an abnormal 1st trimester or maternal serum multiple marker screen?			
11. If you are pregnant, have you had an abnormal finding on an ultrasound examination?			
12. Have you had abnormal results from a CVS or an amniocentesis?			

If pregnant: Name of OB_____ LMP_____ EDC____ Gravida_____ Para ____

Does the client wish to receive further information about any answers that triggered a yes response? Yes or No Would they be interested in seeing a Genetic Counselor? Yes or No
Can client be contacted by Genetic Case Manager Yes or No Are they already being followed by a Geneticist/Genetic Counselor? Yes or No Condition or Diagnosis: _____
Client prefers to be contacted by Phone or Mail Phone Number: Address:

Disposition by Genetic Coordinator

No referral indicated _____ REFERRAL INDICATED _____ REFUSED: Date: __/__/__ If being followed by geneticist, indicate provider:	Referral Made: Date__/__/__ Appointment Kept____ Missed____ Referred to: _____ Diagnosis: _____ Follow-Up Attempts: Indicate: Phone (P), Mail (M) Home Visit (HV) 1st Attempt: Date: __/__/__ 2nd Attempt: Date: __/__/__ 3rd Attempt: Date: __/__/__ Additional Comments/Informational handouts provided:

RATIONALE FOR FAMILY HEALTH HISTORY QUESTIONNAIRE

For questions #16 and #17, only clients identified as high risk or those with a significant concern should be referred to a genetic center. At this time, clients with a positive response regarding the chronic disease questions #18, #19 and #20 do not need a referral to a genetic center.

Racial/Ethnic background and religion

Certain genetic conditions are more prevalent among specific ethnic groups. The following table lists examples of ethnic groups, the conditions for which they are at increased risk and screening tests that can identify a carrier. In the case of clients, for example, of Eastern European (Ashkenazi) Jewish descent, religious affiliation is significant because this ethnic and religious group represents an isolated population in which certain recessive disease-causing genes are present in higher frequencies than in other populations. If a client has concerns about his or her risk, a referral to a genetic center can be made. For each of these disorders, there is carrier testing available in order to identify at-risk individual and couples. Once an at-risk couple is identified, prenatal diagnosis is available.

Racial/Ethnic Group	Prevalent Disorders	Laboratory Tests
African American,	Hemoglobinopathies:	Hemoglobin electrophoresis;
Caribbean, Latin American, Asian, Mediterranean	Sickle cell disease alpha/beta-thalassemia, SC disease	CBC; hematology evaluation
Ashkenazi Jewish Eastern European descent	Tay Sachs disease	Enzyme levels/DNA analysis
	Gaucher disease, type I	DNA analysis
	Cystic fibrosis	DNA analysis
	Familial dysautonia	DNA analysis
	Canavan disease	DNA analysis
	Glycogen storage disease, Ia	DNA analysis
	Niemann-Pick, type A	DNA analysis
	Fanconi anemia, group C	DNA analysis
	Bloom syndrome	DNA analysis
	Mucolipidosis, type IV	DNA analysis
Mediterranean (Greek, Italian)	Cystic fibrosis	DNA analysis
Northern European Caucasian	Cystic fibrosis	DNA analysis

French Canadian	Tay Sachs disease	Enzyme levels/DNA analysis

1. Medical problems since birth or birth defect (e.g., cleft lip/palate, spina bifida, heart defects, ambiguous genitalia, etc.)

Birth defects may have multiple causes. Therefore, it is important to identify any underlying cause(s) of a birth defect in a family in order to provide an accurate risk assessment for future pregnancies and other family members. Some areas to consider when a birth defect is identified in a family are:

- Does the affected individual have additional birth defects, mental retardation or other medical problems?
- Does the affected individual have a known diagnosis or is the cause of the birth defect known?
- Who provided the medical diagnosis for the affected individual?
- Are medical records or access to medical records available?
- Are there other affected family members?

2. A child with any unusual facial appearance or physical features

Many disorders are characterized by a specific pattern of facial features or birth defects. For instance, children with Down syndrome have a typical facial appearance that makes them recognizable. There are hundreds of other syndromes that each have their own unique set of features. An evaluation by a clinical geneticist may help in establishing a diagnosis.

3. Learning problems, developmental delay, mental retardation

Genetic conditions can be associated with learning problems or mental retardation. In addition, the presence of developmental delay may be an indicator of an underlying genetic disorder. There are both genetic and non-genetic causes of mental retardation. Fragile X is one of the most common genetic disorders for mental retardation. Recent developments have included autism and autism spectrum disorders. The following five items are termed "red flags" for developmental delay, mental retardation and have been included for autism spectrum disorders:

- No babbling by 12 months
- No pointing or other gesturing by 12 months
- No single words by 16 months
- No two-word phrases by 24 months
- Loss of any language or social skills at any age

If the client has older family members with a history of developmental delay or mental retardation, some helpful ways to assess the degree of an individual's learning problems would be to ask:

- Was the individual in special education classes?
- Can the individual live independently?
- Can the person hold a job?

Other information to elicit from the family would be:

- Does the affected individual have medical problems or birth defects?
- At what age did the family initially become concerned about the individual's development?
- Does the affected individual have a known diagnosis or is the cause of the developmental delay known?
- Who provided the family with the medical diagnosis for the affected individual?
- Are medical records or access to medical records available?
- Are there other affected family members?

It is important to refer any individual who has mental retardation, learning disabilities or autism spectrum disorders (ASDs) including autistic disorder, Rett's disorder, childhood disintegrative disorder, Asperger's disorder, pervasive development disorder-not otherwise specified. Fetal alcohol syndrome is a non-genetic example of developmental delays and should also be referred to a genetic center for a comprehensive evaluation.

4. A child with regression of developmental skills

There are some conditions that are associated with neurological deterioration. For instance, some metabolic disorders present with loss of developmental milestones that were previously attained. When a child displays loss of skills, a genetic evaluation can be useful in establishing a medical reason for the regression. About 25 percent of children with autism will seem to have normal development until about 18 months, after which they will gradually or suddenly:

- stop talking if they had begun to say a few words
- stop waving goodbye
- stop turning their heads when their names are called
- withdraw into a shell and seem more distant and less invested in their surroundings

5. A child with abnormal growth (height, weight or head circumference)

Geneticists become concerned when growth parameters are either above or below the normal range. Some parameters that are evaluated are height, weight and head circumference. Unexplained abnormal growth may be indicative of an underlying medical or genetic condition. Poor weight gain is often attributed to feeding difficulties, chronic diarrhea and chronic vomiting, which are symptoms that may

be indicative of a metabolic condition. In addition to careful follow-up and evaluation with their primary care physician, a genetic evaluation may be beneficial to these families.

6. A stillborn child or an early infant death

Approximately 7 percent of all stillbirths and neonatal deaths are due to chromosomal abnormalities.[1] In addition, there is a tenfold increased risk for congenital anomalies in stillbirths and neonatal deaths as compared to the incidence in liveborns.[1] Depending on the cause of the loss, the risk to future offspring may vary.

When a referral to a genetic center is made, a comprehensive evaluation is necessary to determine the etiology of the loss. An examination of the placenta, autopsy, genetic studies, X-rays and photographs can be helpful in determining the cause; however, if all of this information is not available, a referral can still be made. A review of these medical records, along with family and obstetrical histories, may be beneficial in elucidating the cause of the death and may clarify recurrence risks for future pregnancies.

7. A child with a confirmed positive newborn screening test

Illinois currently screens all newborns for various metabolic, endocrine and hemoglobin disorders. These include biotinidase deficiency, congenital adrenal hyperplasia, galatosemia, hypothyroidism, fatty acid oxidation disorders, organic acid disorders, phenylketonuria (PKU) and other amino acid/urea cycle disorders and hemoglobinopathies, such as sickle cell disease Most of these conditions require immediate medical treatment or dietary management to decrease the chance of illness, mental retardation and other medical complications. It is very important that the child who has been identified as having one of these disorders be followed by a geneticist, an endocrinologist or a hematologist, depending on the specific condition. If the child has been seen by one of these medical specialists, it is important for the family to have and to keep follow-up appointments. Since many of these conditions require special diets, it is helpful to be supportive of the child and the family since dietary management is often difficult and stressful.

8. Two or more first trimester miscarriages

Some genetic causes of miscarriages are single gene disorders, multifactorial conditions and chromosomal anomalies. In 4 percent to 8 percent of couples who experience recurrent first trimester miscarriages, one of the partners carries a balanced chromosomal rearrangement.[1] Such rearrangements can put the couple at increased risk for having a miscarriage or a child with a chromosomal anomaly. A genetic consultation can provide these couples with information regarding their risks and offer blood chromosomal analysis to rule out such a rearrangement. If a chromosomal rearrangement is identified, the couple should be offered the option of prenatal diagnosis to rule out unbalanced

chromosomal rearrangements in the fetus. It is important to realize that, even though a couple may carry a chromosomal rearrangement, they usually are able of having a healthy pregnancy outcome.

9. You and your partner are blood relatives (e.g., first cousins)

Consanguinity is the mating of two individuals who have one or more common ancestors, thus increasing the chances that they inherited some of the same genes. The risk of having a child with a medical condition or adverse pregnancy outcome is greater for consanguineous couples than for two people who are unrelated to each other. The closer the degree of relationship, the higher the risk; however, ***these risks are often overestimated***. The only couples who are at increased risk are those individuals who are first cousins or those who are more closely related. If a couple has questions or concerns regarding risks, referral to a genetic center is appropriate. The center can determine precise risks, possible diseases that could affect their offspring and the availability of carrier or prenatal testing. The most important question to ask is how the individual and his/her partner are related?

10. Chromosomal abnormalities (e.g., Down syndrome, trisomy 18, Turner syndrome, etc.)

Chromosomes are the units of genetic information found in almost every cell in the body that code for physical and mental development. Chromosomes carry the information that code for eye color, hair color and how the body functions. Changes in the number or structure of the chromosomes can lead to various birth defects, mental retardation or other medical conditions. Depending on the type of chromosome abnormality in an affected individual, other family members may be at an increased risk for having children with a chromosomal anomaly.

11. Neurological or muscular diseases (e.g., Huntington disease, muscular dystrophy, etc.)

There are many neurological or muscle diseases that are genetically determined. Some examples are Huntington disease, Duchenne muscular dystrophy, spinal muscular atrophy and ataxia. In addition, many genetic disorders have neurological or muscle involvement, such as some metabolic disorders, Down syndrome and Prader-Willi syndrome. Onset for such conditions can be at birth or later in life and can be progressive.

12. Disorders of the blood (e.g., sickle cell disease/trait, thalassemia, hemophilia, factor V Leiden, etc.)

Hemoglobinopathies are a large group of inherited blood disorders that affect structure, function or production of blood cells. Sickle cell disease is the most common

hemoglobinopathy found in those of African-American or Mediterranean descent. It is important to distinguish between individuals who have sickle cell disease and those who are carriers of sickle cell disease, also known as, sickle cell trait. Individuals with sickle cell trait do not have symptoms of sickle cell disease; however, they are at an increased risk to have a child with the disease if their partner is also a carrier of sickle cell disease, or another hemoglobinopathy. (Combination disorders such as sickle hemoglobin C and sickle-thalassemia disease also have clinical and genetic implications.)

The thalassemias are another group of hemoglobinopathies that are commonly found in people of Mediterranean, Caribbean, Latin American, African, and Asian descent. There are two classes of thalassemia, beta and alpha, determined by which globin chain is involved. *Beta-thalassemia major* produces severe anemia and skeletal changes and requires blood transfusions and iron chelation therapy. Carriers of this condition are said to have *beta-thalassemia minor* and usually do not have symptoms. There are several types of alpha-thalassemia, which can vary clinically from asymptomatic (*alpha-thalassemia minor*) to mild anemia to significant anemia. The most severe form of alpha-thalassemia leads to hydrops fetalis, which results in death. It is important to, not only have an accurate diagnosis, but to again differentiate between disease and carrier status in order to determine risks for the family.

Illinois screens all newborns for hemoglobinopathies. Carrier testing in adults should be performed on individuals at an increased risk, ideally prior to conception. If both partners in a couple are found to be carriers of any hemoglobinopathy, and are at an increased risk for having a child with a hemoglobinopathy, the option of prenatal diagnosis should be discussed.

Another class of blood disorders affects clotting. Hemophilia is an example of a clotting disorder resulting in an insufficiency of clotting factors and an increase in bleeding. Such clotting disorders can be inherited through X-linked, autosomal recessive or autosomal dominant inheritance patterns (see Appendix B). Some symptoms include prolonged bleeding time, easy bruising, heavy nosebleeds or bleeding problems following surgery or dental work.

Clients with a personal or family history of such a clotting disorder or any of the following symptoms should be referred to a genetics center. The thrombophilias are a group of clotting disorders that result in over-clotting and contributes to an increased risk of deep vein thrombosis, venous thromboemboli (i.e. pulmonary emboli), heart attack, stroke, cardiovascular disease, and miscarriage. It is estimated that as man as one in five Americans has some type of thrombophilia. Among the most common of the thrombophilias are factor V Leiden and prothrombin mutations, which are autosomal dominant disorders, affecting up to 9 percent of Caucasian Americans. Mild hyperhomocystenemia is an autosomal recessive disorder affecting as man as 10 percent of Caucasian Americans. Less common thrombophilias include protein C deficiency and protein S deficiency.

13. Vision loss (not glasses or contacts) or hearing loss at an early age

There are many causes for blindness and deafness such as *in utero* infections, trauma or some childhood illnesses. Nearly half of early onset vision loss and 30 percent of all

deafness has an underlying genetic cause that can be inherited through X-linked, autosomal recessive or autosomal dominant inheritance patterns (see Appendix B. Inheritance Patterns, page 55).[1] Some factors, such as early onset and bilateral involvement, indicate that a genetic cause may be more likely. Some questions to consider are:

- When was the vision or hearing loss detected?
- Was a cause of the condition given?

Wearing corrective lenses (glasses or contacts) would not be a condition that would trigger a positive indicator or reason for a referral to a genetic center.

14. Abnormalities of the bones or skin (e.g., frequent fractures, bone deformities, unusual birth marks)

Skeletal dysplasias are a large group of genetic disorders associated with abnormalities in the size and structure of limbs, trunk and skull. They can be associated with short stature, frequent bone fractures and other radiologic findings.[2] Achondroplasia is one of the most common skeletal dysplasias and is characterized by disproportionate short stature, short limbs, a large head, a low nasal bridge and an increased risk for ear infections.

In addition to skeletal disorders, connective tissue disorders and some dermatological conditions can be caused by underlying gene defects. For example, neurofibromatosis is a common genetic disorder that affects skin pigmentation and bone structure, as well as a number of other systems.

15. Other genetic/inherited diseases (e.g., cystic fibrosis, polycystic kidney disease, Tay Sachs disease, phenylketonuria [PKU])

Whenever there is a genetic disorder identified in a family, it is important to verify the diagnosis by reviewing the medical records of the affected family members. There are many different ways in which a genetic disorder can be passed on in a family; therefore, it is critical to document how the affected individual is related to your client. This will enable you to assess the client's individual risk and the risk to his or her offspring. Individuals at risk for being carriers of these disorders should be referred for a genetic consultation to ascertain their carrier status. In addition, if the client is affected, an evaluation at a genetic center should be offered in order to provide the client with the appropriate medical services. For example, individuals with a metabolic condition may

have special dietary restrictions or need diagnostic examinations.

CHRONIC DISEASES:

For questions #16 and #17, only clients identified as high risk or those with a significant concern should be referred to a genetic center. At this time, clients with a positive response regarding the chronic disease questions #18, #19 and #20 do not need a referral to a genetic center. Note: A recommendation for follow-up is suggested for each condition.

The next five questions relate to discovering "red flags" in a family history suggesting a genetic condition or inherited susceptibility to a common or chronic disease. These diseases are rarely caused by a single gene mutation and are often influenced by environmental conditions and lifestyle choices. Early identification of families with an increased risk for these chronic diseases can often improve, delay, or even prevent adverse health outcomes to individual members. Examples of prevention and treatment options may include increased surveillance, lifestyle changes, prophylactic medical measures, surgical intervention or genetic testing. Personal risk factors, such as body mass index, diet, exercise, use of tobacco products and alcohol and screening (behaviors) tests such as mammograms, pap smears, colonoscopies, lipid profile screening, etc. also can be used as an educational component when discussing the risks of these disorders.

The following algorithm can be used to assess various degrees of risk for many chronic diseases by examining the client's family history.

High-risk criteria:

1. Premature disease in a first-degree relative
2. Premature disease in a second-degree relative (coronary artery disease only)
3. Two affected first degree relatives
4. One first degree relative with late or unknown disease onset and an affected second-degree relative with premature disease from the same lineage
5. Two second-degree maternal or paternal relatives with at least one having premature onset of disease
6. Three or more affected maternal or paternal relatives
7. Presence of a "moderate risk" family history on both sides of the pedigree

Recommendations for Intervention:
Genetic counseling is warranted if the client is interested in genetic counseling and risk analysis, a referral to a genetic center is appropriate.

Moderate risk criteria:

1. One first-degree relative with late or unknown onset of disease
2. Two second-degree relatives from the same lineage with late or unknown disease onset

Recommendations for Intervention:
Genetic counseling is not usually warranted but might be appropriate if the client is interested in genetic counseling and risk analysis. The client should be offered the "Everyday Choices for a Healthier Life" pamphlet. This pamphlet was developed by the American Heart Association, American Diabetes Association and American Cancer. The pamphlet is available by phone 866-399-6789 or through the Web site: http://www.everydaychoices.org/downloadables/pdf/englishPHP.pdf .

Average risk criteria:

1. No affected relatives
2. Only one affected second-degree relative from one or both sides of the pedigree
3. No known family history
4. Adopted person with unknown family history

Recommendations for Intervention:
Genetic counseling is not warranted. The client should be offered the "Everyday Choices for a Healthier Life" pamphlet.

[3]

16. Breast, ovarian, colon, skin cancer, history of colon polyps before the age of 50?

Identifying families with a genetic predisposition to cancer is done primarily through assessing the family history. It is estimated that approximately 15 percent of all cancers are caused in part by an inherited factor.[4] Presymptomatic testing is available for some types of cancer, including melanoma, breast, ovarian and colon cancer. It is often difficult to differentiate families with hereditary forms of cancer from those affected with a sporadic cancer. Some common indicators of inherited forms of cancer include multiple family members with the same type or related type of cancer, increase in multi focal

(many tumors in the same area) or bilateral cancer, many primary cancers and cancer affecting several generations. A younger age at onset is more likely an indicator of an inherited form of cancer – breast and colon cancer <age 45-50 years; prostate cancer <45-60 years.

For example, in breast cancer and colorectal cancer, 5 percent to 10 percent of the cases are hereditary in nature. Specific gene mutations have been identified which, if inherited, significantly increase an individual's risk for breast/ovarian cancer. The most common mutations are called BRCA1 and BRCA2. Medical intervention measures can be offered to reduce an individual's risk for breast/ovarian/colorectal cancers and early detection can significantly lower the death rate.
The client should be offered the "Everyday Choices for a Healthier Life" pamphlet.

High or Moderate Risk Referral Indications for Breast and Ovarian Cancer:
Any family that meets the following criteria should be considered high or moderate risk. If they are interested in genetic counseling and risk analysis, a referral to a genetic center is appropriate.

Any personal or family history of:

Breast cancer:

- Diagnosed <age 40
- In >two close relatives<age 50
- Bilateral, first cancer <age 50
- In a male, any age

Breast & ovarian cancer:

- In any woman
- One of each cancer in two close relatives, diagnosed any age

Ovarian cancer:

- In two close relatives, diagnosed any age

In Ashkenazi Jewish families:

Breast cancer:

- Diagnosed <age 50
- Any age if family history of breast and/or ovarian cancer

Ovarian cancer:

- Personal or family history, diagnosed any age

High or Moderate Risk Referral Indications for Colon Cancer and Colon Polyps:
Although the vast majority of colon cancer diagnoses are not inherited, approximately 5 percent of all colon cancers are directly caused by inherited factors. Some of the more recognized hereditary colon cancers include Hereditary Nonpolyposis Colon Cancer (HNPCC), Familial Adenomatous Polyposis (FAP), and Gardner syndrome. [5]

Any family that meets the following criteria should be considered high or moderate risk. If they are interested in genetic counseling and risk analysis, a referral to a genetic center is appropriate.

A person or family history of colorectal cancer (CRC):

- Diagnosed at <35 years
- In more than two close relatives; one diagnosed at <50 years
- In more than three close relatives; diagnosed at any age
- CRC with endometrial cancer in the same female relative
- CRC with endometrial cancer, one of each cancer in two close relatives, diagnosed at any age

High or Moderate Risk Referral Indications for Skin Cancer:
Approximately one in 90 people will develop melanoma sometime during their lifetime. Although the vast majority of cases are due to environmental factors such as over exposure to UV light, 5 percent to 10 percent will develop melanoma due to an inherited autosomal dominant gene. [6]

Any family that meets the following criteria should be considered high or moderate risk and is interested in genetic counseling and risk analysis, a referral to a genetic center is appropriate.

- Melanoma in two or more close relatives
- Personal history of multiple primary melanomas, with or without a positive family history
- Dysplastic nevi are present with a personal or family history of melanomas

17. Experienced a heart attack, stroke or died suddenly at an early age (women <60; men <50 yrs). Evaluation and/or diagnosis for a heart condition? High blood pressure and/or high cholesterol?

A family history with several members who died at an early age may be at risk for cardiovascular disease. Cardiovascular disease (CVD) is the number one killer of men and women in the United States. Several types of CVD are known to have hereditary components, including coronary artery disease (CAD), cardiomyopathies, arrhythmia, clotting disorders and aneurysm. CVD affects one in five Americans and it is common

to see family histories with multiple individuals in successive generations who have experienced a cardiovascular event. CAD is the most common type of heart disease in the U.S. CAD is known to be a multifactorial disease caused by both inherited susceptibility and environmental risk factors. As a multifactorial disease, reducing risk factors such as, smoking, hypertension, high cholesterol, diabetes, obesity and sedentary lifestyle can greatly reduce the risk of heart disease. Many individuals benefit from lifestyle modification and possible drug therapy.

Recommendations for Intervention:

- Clients with histories of early onset CAD and/or family histories that appear to follow an autosomal dominant pattern of inheritance (affected individuals are seen in successive generations) should be considered high risk. High-risk families should be referred for genetic counseling and possible genetic testing to personalize their health care recommendations.
- For clients who do not have a personal history or significant family history of CVD, but who may benefit from information regarding healthy lifestyles, the "Everyday Choices for a Healthier Life" pamphlet.

18. Diabetes

Diabetes affects greater than 17 million Americans. There are several types of diabetes. The most common forms are Type 1 and Type 2. Of all people with diabetes, approximately 90 percent to 95 percent have Type 2 diabetes, which often manifests after 40 years of age. Type 2 diabetes generally results from either insulin resistance or an insulin secretory defect. Type 1 diabetes, which usually has onset in the first two decades of life, is an autoimmune disorder which affects the insulin-producing pancreatic beta-cells, resulting in insulin deficiency. Both of these types involve multiple genes
and are felt to be multifactorial in inheritance. Individuals with multifactorially inherited disorders inherit predisposing, or susceptibility genes, and in the face of other non-genetic or environmental influences, manifest the disease. In the case of Type 1 diabetes, it is linked to a gene in the HLA-region of chromosome six. Type 2 diabetes is associated with a family history of obesity, high blood pressure and high cholesterol, which all have genetic components. Another type of diabetes is Maturity Onset Diabetes of the Young (MODY). MODY is an autosomal dominant disorder with onset at 10-25 years of age.

Recommendations for Intervention:

- For clients who have diabetes and have a significant family history, or clients with a family history with multiple affected relatives in successive generations (an autosomal dominant pattern of diabetes), a referral to a genetic center is appropriate.
- For clients who do not have a personal history or significant family

history of diabetes, but who may benefit from information regarding healthy lifestyles, the "Everyday Choices for a Healthier Life" pamphlet.

19. Asthma

Approximately 15 million Americans have asthma with nearly 5 million children affected. Family clustering of asthma cases indicates a genetic component. There is no clear cause for the airway inflammation seen in asthma, however, it is felt that it is most likely due to combination of genetics factors and environmental triggers, such as allergens, exercise, irritants and viral infections. Although all ages are affected with asthma, onset is most often in childhood. In childhood, more boys are affected than girls, while in adulthood, more women are affected than men. Asthma is seen in all races, however, African Americans have more asthma attacks, are more likely to be hospitalized, and more likely to die from an asthma attack. New research suggests that exposures early in life to environmental triggers may contribute to the development of asthma later in life. Much of the current research is focusing on gene-environmental interactions, given the rising trend in asthma in the last 15 years.

Recommendations for Intervention:

- Clients with a personal history and family history of asthma should be informed about the familial nature of this condition and be encouraged to discuss their personal health concerns with their primary care provider.
- Clients should be directed to the Illinois Department of Public Health (IDPH) Health Fact Sheets regarding asthma for more information, accessible at the IDPH Web site, www.idph.state.il.us under A-Z topics – Asthma.

20. Arthritis

Arthritis and related conditions affect one in three people or approximately 70 million Americans. In 2000, it was estimated that 26% of adults in Illinois, or 2.4 million adults, suffered from some type of arthritis. Although there are more than 100 different types of arthritis and related conditions that affect the joints, muscles and other soft tissues, the three most common are osteoarthritis, fibromyalgia, and rheumatoid arthritis. Clinical researchers are studying several major factors that are thought to contribute to the symptomatic picture of this group of diseases. Genetic factors, the role of inflammation and the immune system in causing joint damage, joint injuries, and lifestyle factors such as weight, diet and exercise, all contribute to the development of arthritis. Early identification and early intervention are the key to successful treatment of most forms of arthritis.

Recommendations for Intervention:

- If a client has a family history of arthritis or an arthritis related condition and is experiencing symptoms (such as joint pain and swelling), a referral

to a primary care provider for evaluation and intervention is warranted.

- For clients with a family history of arthritis or an arthritis related condition, who have not experienced symptoms, but who are seeking more information, direct these clients to the Department's Health Fact Sheets accessible at the IDPH Web site, www.idph.state.il.us under A-Z topics – Arthritis.

ADDITIONAL QUESTIONS FOR WOMEN

The following questions should be used to assess any woman who is of childbearing age or who is currently pregnant.

1. Are you older than 35 years of age?

Although any woman, any age, can have a child with a chromosome abnormality, the risk of such an event increases as a woman ages. Women who will be 35 years old or older at delivery should be offered the option of genetic counseling and prenatal testing, such as chorionic villus sampling or amniocentesis.

2. Do you have diabetes?

Women with diabetes have an increased risk over non-diabetic women for having children with birth defects such as neural tube defects, heart defects and caudal regression (abnormal development of the lower limbs). Preconception management of maternal glucose levels is known to reduce the risks; poor control during early pregnancy is associated with a greater risk. Genetic services can provide these women with more detailed information about their risks and the options for prenatal screening to help identify the associated birth defects.

3. Do you have seizures?

Women with seizure disorders have a greater risk for having children with birth defects than women who do not have seizures. In addition, some medications taken to control seizures may cause an increased risk for certain congenital anomalies. It is important for pregnant women to maintain good seizure control during a pregnancy; therefore, a change in medication or dosage should be made only by a physician. A genetic consultation can provide information regarding the benefits of medication use and seizure control versus the potential risks to the fetus. The options of prenatal screening to help identify the associated birth defects also would be reviewed.

4. Are you currently taking prescription medications?

Some prescription medications can adversely affect the developing fetus. A genetic consultation can provide information regarding the risks associated with a specific agent. Important questions to ask are:

- What is the name of the medication?
- When during the pregnancy was the medication used?
- How often and how much of the medication was taken?
- How was the medication taken (oral, IV, topical, etc.)?

5. Are you taking a vitamin containing folic acid?

The incidence of neural tube defects in infants is reduced when women take folic acid (one of the B vitamins, sometimes called "folate") prior to conception and throughout pregnancy. The U.S. Public Health Service recommends that all women capable of becoming pregnant consume 0.4 milligrams of folic acid each day. Folate can be found in some foods, such as green leafy vegetables, fortified breads and cereals, citrus fruits and liver. Since it is often difficult for women to obtain enough folic acid from their diet, it is recommended that a daily multiple vitamin that contains 0.4 milligrams of folate be taken by all women of childbearing age. However, women who are at an increased risk of having a child with a neural tube defect are advised to take 4 milligrams of folate per day. Women who are considered to be at an increased risk are those who have had a previous pregnancy affected with a neural tube defect. The local health department should educate the client about the benefits of folic acid.

6. Do you have concerns about exposures to cigarette smoking?

Smoking can cause low birth weight, prematurity and pregnancy complications. The client should be educated through the local health department about the risks associated with smoking. Genetic counseling is available for those women who have additional concerns.

7. Do you have concerns about alcohol consumption or drug use?

Because no safe amount of alcohol consumption during a pregnancy can be quantified, it is recommended that pregnant women or those planning a pregnancy, refrain from all alcohol intake.

The more alcohol consumed during a pregnancy, the greater the risk of adverse effects, including cognitive impairment and fetal alcohol syndrome. The use of some illicit drugs also may adversely affect fetal growth and development, as well as pregnancy outcome. The client should be educated through the local health department about the risks associated with alcohol or drug use. Genetic counseling is available for those women who have additional concerns.

8. Do you have concerns about exposures to harmful substances in the workplace?

Maternal exposure to some substances in the workplace may increase risks for congenital anomalies or pregnancy complications. The information needed to provide risk assessment is:

- What are the names of substances involved?
- If a woman is pregnant, what was the timing of the exposure (gestational age)?

- Did the woman have headaches, nausea or dizziness while being exposed?
- Did the woman wear a mask or gloves while working with the agent?
- Was the room/area well-ventilated? A genetic consultation can provide the woman with available information on the potential fetal effects of the maternal exposure in pregnancy and possible ways to reduce such risks.

9. If you are pregnant, have you had any illnesses during this pregnancy?

Maternal illness during a pregnancy can increase the risks for congenital anomalies and pregnancy complications. If a woman has varicella (chicken pox, shingles), cytomegalovirus (CMV), toxoplasmosis or other illness during a pregnancy, it is important to ask:

- When did the exposure occur?
- Was the client only exposed to the illness or did she actually have the illness?
- Was any special testing performed to help diagnose the condition?
- Did she use any medication to treat the illness?

10. If you are pregnant, have you had an abnormal first trimester screen or maternal serum multiple marker screen?

First trimester screening, done at 11 through 13 weeks gestation, incorporates the measurement of the fetal nuchal translucency and the measurement of two biochemical markers, (PAPP-A and free beta hCG). These results are used to identify women who are at an increased risk for carrying a fetus with Down syndrome or trisomy 18. Maternal serum multiple marker screen provides a client with a modified risk for Down syndrome, trisomy 18, or an open neural tube defect or abdominal wall defect based on the level of one or more biochemical markers in the maternal serum. When a woman receives an elevated risk based on this screen, an ultrasound examination should be performed to confirm the gestational age of the pregnancy. Other information to gather includes:

- Has an ultrasound examination been performed and did it confirm the dating of the pregnancy?
- Was the client offered a genetic consultation to discuss the screening result?
- Did the woman have further testing, a detailed ultrasound examination or an amniocentesis?
- Where was the test done and are results available? A referral to a genetic center should be offered if the client has not had genetic counseling and if she desires counseling to explain the test or to have follow-up diagnostic testing.

11. If you are pregnant, have you had an abnormal finding on an ultrasound examination?

If an abnormality is detected during an ultrasound examination, a genetic consultation can provide the woman with information about the finding, what this may mean for the

fetus and if further testing would be helpful. Questions to ask the client are:

- What was the name of the abnormality?
- When was it detected?
- Where was the ultrasound examination done?
- Was the woman offered genetic counseling to discuss the finding?
- What does the woman understand about the condition?
- Was additional testing, such as an amniocentesis, performed?
- Does the woman have questions or concerns about the finding? If there are unresolved questions or concerns, a referral to a genetic center can help address these.

12. Have you had abnormal results from a CVS or an amniocentesis?

An abnormal result from a CVS or amniocentesis procedure could indicate that the fetus has a genetic disorder, chromosome abnormality or a neural tube defect. It is important to ask:

- What is the specific result?
- Has the client been offered genetic counseling regarding the result?
- Does the client have questions or concerns about the results?
- Where was the testing done and are copies of the results available? If the client has not had genetic counseling, or still has questions about the information or result, a referral to a genetic center can help address her concerns.

PROTOCOLS FOR ESTABLISHING GENETIC SERVICES

In order to provide accessible genetic services to clients served by the local health department, a relationship should be established with a genetic center. Provided in this manual is a list of all the genetic centers in the state of Illinois (see Appendix C.1 page 56).

To overcome service barriers, it may be beneficial to establish an on-site genetic clinic. The following issues need to be addressed when establishing an on-site clinic:

- Contact the Illinois Department of Public Health Genetics Program to discuss establishing an on-site clinic.
- Identify a local public health genetic coordinator.
- Identify a local genetic center that will provide services at the health department facility.
- Define the responsibilities of the health department and the genetic center, including issues such as scheduling client appointments, obtaining pertinent medical information and follow-up.
- Discuss billing issues.
- Determine frequency of clinics.
- Review facility issues such as staffing, exam room and equipment availability.

All of the above issues should be clearly delineated in a contract/letter of agreement between the health department and the genetic center. It may not be feasible to have an on-site clinic, nor can all genetic services be provided on-site. Therefore, arrangements should be made for direct referrals to the genetic center under these circumstances.

REFERENCES

1. Robinson, A. and Linden, M. 1993. *The Clinical Genetics Handbook*. Blackwell Scientific Publications: Boston.

2. Rimoin, D., Connor, J.M., Pyeritz, R., eds. 1996. *Emery and Rimoin's Principles and Practices of Medical Genetics*. Churchill Livingstone: New York.

3. Scheuner, M. et. al., 1997. *American Journal of Medical Genetics*, 71:315-324 modified.

4. Schneider, K. 1994. *Counseling About Cancers: Strategies for Genetic Counselors*. Graphic Illusions: Dennisport.

5. University of Texas, MD Anderson Cancer Center Web site: http://cancer.stanfordhospital.com/healthInfo/geneticsAndCancer/types/skin/

6. Stanford University Cancer Center Web site: http://cancer.stanfordhospital.com/healthInfo/geneticsAndCancer/types/skin/

APPENDIX A. GLOSSARY

ABERRATION	Any abnormality of chromosome structure or number.
ACROCENTRIC	A chromosome with the centromere near one end. These chromosomes have satellited short arms that carry the genes for ribosomal RNA.
AGAROSE GEL	A porous, semi-solid material used for many research purposes, among them electrophoresis.
AGENESIS	Absence of an organ resulting from the failure of formation of the precursor of an organ during embryonic development.
ALLELE	Any one of two or more alternate forms of a gene located at the same locus.
ALPHA-FETOPROTEIN	Also known as AFP. A protein of unknown function that is specific to the fetus. It is found in amniotic fluid and maternal serum. Elevated levels of AFP can be indicative of neural tube defects or an abdominal wall defect. Decreased levels in maternal serum may indicate the presence of Down syndrome or other chromosome abnormality.
AMINO ACID	Any one of 20 nitrogenous molecules that are linked in a linear sequence to form proteins. The sequence of amino acids determines the protein formed and its function.
AMNIOCENTESIS	A prenatal diagnostic procedure, performed at 15 weeks gestation or greater, in which fluid is withdrawn from the amniotic sac surrounding the fetus.
AMPLIFICATION	In molecular biology, production of multiple copies of a portion of DNA. a usually massive replication of genetic material and especially of a gene or DNA sequence (as in a polymerase chain reaction).
ANENCEPHALY	A neural tube defect characterized by the partial or complete absence of the cranial vault and only a rudimentary brain.

ANEUPLOID	A chromosome number that is not an exact multiple of the haploid (n) number.
ANOMALY	Abnormal variation in form or structure.
ANTICIPATION	Onset of an inherited disorder at an earlier age, or worsening of symptoms in successive generations.
APLASIA	Absence or irregular structure of tissue or organ.
AUTORADIOGRAPH	Image produced on an X-ray film by a radioactively labeled substance.
AUTOSOME	Any chromosome other than the sex chromosomes.
BANDING	A technique of staining chromosomes in a characteristic pattern of light and dark bands to allow more precise identification of each chromosome.
BASE	One of the five molecules that make up the informational content of DNA and RNA. In DNA, bases pair across the two chains of the double helix: adenine with thymine and guanine with cytosine. RNA is single-stranded and contains uracil instead of thymine.
BASE PAIRS	Pairs of complementary nucleotides forming the DNA double helix.
BRACHYDACTYLY	Abnormally shortened digits.
CARRIER	An individual who has a gene but does not manifest the trait.
CENTROMERE	Heterochromatic constricted region within a chromosome where the chromatids are held together and to which the spindles attach during cell division. This region is also known as the *kinetochore.*
CHORIONIC VILLUS SAMPLING (CVS)	A prenatal diagnostic procedure, performed in the 10th to 12th week of pregnancy, in which fetal tissue is withdrawn from the placenta and studied for genetic abnormalities.

CHROMOSOME	Strands of DNA and protein on which genes are located.
CHROMOSOME BANDS	Patterns of light and dark bands produced by chemical staining of the chromosomes. Each chromosome or fragment of one has its own identifiable pattern of bands seen under the microscope, and the individual bands serve as landmarks for the positions of the genes.
CLINODACTYLY	Crooked finger that is curved inward (sideways), usually the fifth digit.
CLONE	A genetically identical cell population derived from a common ancestor; to clone an organism is to make a genetically identical copy of that organism.
CODOMINANCE	The expression of each of a pair of alleles when present in the heterozygous state.
CODON	A triplet of three bases in a DNA or RNA molecule that specifies a single amino acid.
CONGENITAL	A medical finding, (physical, mental, biochemical, etc.) which is present at birth. Of or relating to a condition that is present at birth, as a result of either heredity or environmental influences: *a congenital heart defect; congenital syphilis.*
CONSANGUINITY	Referring to the members of a couple who are related by sharing a common ancestor; a relationship with a blood relative.
COSMID	A vector used for cloning DNA fragments.
CROSSING OVER	When a section of one chromosome switches places with the same section from the other chromosome of the pair. This occurs when a germ cell makes copies of its chromosomes before dividing and is a normal event in the cell division process.
CYTOGENETICS	A branch of genetics concerned with the study of chromosomes.
DELETION	A chromosome abnormality in which a part of the chromosome is missing.

DEOXYRIBONUCLEIC ACID (DNA)	DNA is a double-stranded, helical molecule that codes for genetic material. Portions of DNA are referred to as genes. Deoxyribonucleic acid; a nucleic acid that consists of two long chains of nucleotides twisted together into a double helix and joined by hydrogen bonds between complementary bases adenine and thymine or cytosine and guanine; it carries the cell's genetic information and hereditary characteristics via its nucleotides and their sequence and is capable of self-replication and RNA synthesis.
DIMERIC INHIBIN-A (DIA)	A biochemical marker, similar to hCG, of placental origin detectable in maternal serum. Combined evaluation of maternal serum DIA with uE3, hCG, AFP, and maternal age has value in predicting risk for fetal chromosome abnormality during pregnancy.
DIPLOID (2N)	Having two complete sets of chromosomes. In humans, the normal diploid chromosome number is 46.
DNA DENATURATION	The separation of DNA into its two strands of nucleotides, for example by exposing it to near-boiling temperatures or to extreme alkaline conditions.
DNA PROBE	A specific portion of single-stranded DNA used to seek out a complementary portion in other single strands of DNA. The probe is usually radioactively labeled so that its location can be easily detected.
DNA SEQUENCING	Determining the order of nucleotide bases of DNA; mapping. This testing method is used in locating genes and mutations.
DOMINANT	Describes a gene that produces an effect whenever it is present; a trait, pattern of inheritance or disorder caused by a gene of this type. Of, relating to, or being an allele that produces the same phenotypic effect whether inherited with a homozygous or heterozygous allele.
DUPLICATION	A chromosome abnormality in which a portion of the chromosome is repeated.

DYSMORPHISM	Abnormality in development of structure, as seen in many syndromes of genetic or environmental etiology.
DYSPHASIA	Developmental abnormality of a tissue; an example is a nevus.
ELECTROPHORESIS	A laboratory procedure method of separating substances, such as DNA fragments or types of hemoglobin, by using an electric field to make them move through a medium, such as a gel, at rates that correspond to their electrical charge and size.
ELSI	An acronym for the Ethical, Legal and Societal Implications Program of the Human Genome Project.
ESTRIOL, UNCONJUGATED (uE3)	A type of estrogen synthesized in the placenta and found in serum of pregnant women. Combined evaluation of maternal serum uE3, hCG AFP, DIA, and maternal age has value in predicting risk for fetal chromosome abnormality during pregnancy.
EXON	The segment of a gene that is represented in the mature mRNA and thus codes for a protein.
EXPANSION	A type of DNA mutation in which there is an addition of multiple copies of trinucleotide repeat sequences during meiosis. These expansions result in gene instability.
EXPRESSIVITY	The extent to which a gene's effects are seen in an individual. If a gene is said to have variable expressivity, then the trait may vary from very mild to severe but it is never completely unexpressed.
FAMILIAL	Refers to a trait or disease which occurs in more members of a family than would be expected by chance.
FETOSCOPY	An imaging technique used in prenatal diagnosis for direct visualization of the fetus by means of a fiber optic endoscope inserted into the amniotic cavity.

FISH	An acronym for fluorescence *in situ* hybridization, a cytogenetic technique used to identify specific chromosomes, or portions thereof, in which DNA probes are tagged with fluorescent dyes.
GAMETE	A male or female reproductive cell (sperm or egg).
GENE	A unit of heredity; a segment of the DNA molecule containing the code for a specific function.
GENE CLONING	A DNA sequence, such as a gene, that is transferred from one organism to another and replicated by genetic engineering techniques.
GENE EXPRESSION	The manifestation of a gene as a specific trait.
GENETIC CODE	The set of instructions that directs the development and functioning of a person; the universal key by which genetic information is recorded and translated in all life-forms.
GENETIC DIAGNOSIS	Cytogenic, biochemical or molecular testing or identification of a clinical phenotype that identifies the patient as having an inherited or genetic condition.
GENETIC ENGINEERING	Altering genetic material to study molecular processes and potentially to correct genetic defects. See recombinant DNA technology.
GENETIC LINKAGE	A method of DNA analysis often used when the gene causing a particular genetic disease has not yet been identified and isolated, but the location of the gene has been narrowed down to a specific region of a chromosome. Examination of the DNA of a family is then conducted to determine which members may be at an increased risk for a genetic disorder based upon the inheritance of neighboring genes known to be inherited with a culprit gene. Laboratory technicians look for variations that consistently appear in the DNA of family members with the disorder. These DNA variations may or may not be related to the genetic disorder. However, if they appear in the DNA of another family member, it can indicate the person's risk of inheriting the disorder.

GENETICS	The study of heredity.
GENOME	The complete DNA sequence of an organism or individual.
GENOMICS	The branch of genetics that studies organisms in terms of their genomes (their full DNA sequences).
GENOTYPE	The genetic constitution of an individual.
HAPLOID (n)	The chromosome number of a normal gamete. In humans n=23.
HEMIZYGOUS	The condition in which only one copy of a gene pair is normally present; therefore, the effect is expressed (e.g., the genes on the X chromosome of the male since there is no counterpart present).
HETEROZYGOTE	An individual who has two different alleles in a specific gene pair at a given locus on a pair of homologous chromosomes.
HOMOLOGOUS	A matching pair of chromosomes, one from each parent.
HOMOZYGOTE	An individual who has two identical alleles in a specific gene pair at a given locus on a pair of homologous chromosomes.
HUMAN CHORIONIC GONADOTROPIN (hCG)	A hormone produced by the cells of the placenta, detectable in maternal plasma and urine. 1) Beta hCG used in second trimester screening of maternal serum hCG, uE3, AFP, DIA, and maternal age, has value in predicting risk for fetal chromosome abnormality during pregnancy. 2) Free-hCG, used in first trimester screening along with PAPP-A, nuchal translucency measurement and maternal age has value in predicting risk for fetal chromosome abnormality during pregnancy.
HUMAN GENOME PROJECT	The scientific mission to "map" the order of bases as they appear in the DNA of human chromosomes. The

	Human Genome Project actually is not one project, but rather many hundreds throughout the world. The objective is to create a directory of the genes that can be used to answer questions such as what specific genes do and how they work.
HYDROCEPHALUS	Abnormal accumulation of cerebrospinal fluid in the ventricles of the brain, usually accompanied by increased pressure on the brain.
IMPRINTING	Differential expression of the maternal and paternal alleles of a gene.
INBORN ERROR OF METABOLISM	An inherited disorder such as a specific enzyme defect blocks a metabolic (biochemical) process, causing illness.
INTRON	A DNA sequence that interrupts the sequences coding for a gene product (exons). After information from genes is transcribed into new strands of RNA, the introns are cut out of the RNA. The function of introns is still being explored.
INVASIVE TROPHOBLAST ANTIGEN (ITA)	A form of hCG produced by embryonic cells during implantation. ITA is the newest of the biochemical markers used in second trimester prenatal screening for chromosome abnormalities. Along with maternal serum AFP, uE3, hCG, DIA, and maternal age, ITA has value in predicting risk for fetal chromosome abnormality during pregnancy.
IN VIVO	In the living organism.
IN VITRO	In the test tube or laboratory.
ISOCHROMOSOME	An abnormal chromosome with duplication of one arm and deletion of the other arm.
KARYOTYPE	A pictorial representation of an individual's chromosome set.
LINKAGE	The tendency of two genes, that are close in proximity to each other on the same chromosome, to be inherited together. Linked genes are used to study the

	transmission of a specific disorder in a family. An association between two or more genes such that the traits they control tend to be inherited together.
LOCUS	The position of a gene on a chromosome. Different forms of the gene (alleles) may occupy the locus.
MACROSOMIA	Excessive growth of prenatal onset, associated with several genetic disorders and sometimes seen in infants of diabetic mothers.
MARKER	A detectable genetic variant, such as one of the ABO blood types. Some markers are found only among individuals with certain diseases and can be used to determine the presence of these diseases.
MEIOSIS	The special type of cell division by which the diploid chromosome number is reduced to a haploid state to form egg or sperm.
MENINGOCELE	Spina bifida with bulging of meninges without involvement of the spinal cord.
MICROCEPHALY	Small head circumference, usually defined as below the third percentile for age, height and weight; associated with mental retardation in most cases.
MITROCHONDRIAL INHERITANCE	The mitochondria are cellular organelles containing DNA that encode genes that may sustain disease causing mutations. Since only maternal mitochondria are inherited, mitochondrial diseases exhibit non-Mendelian inheritance patterns.
MITOSIS	Cell division that results in the formation of two new cells (daughter cells), each with the same chromosome set as the initial or parent cell. The process in cell division by which the nucleus divides, typically in four stages (prophase, metaphase, anaphase, and telophase) resulting in two new nuclei, each of which has exactly the same chromosome and DNA content as the original cell. Also called *indirect nuclear division*, *karyokinesis*, *mitotic division*.

MONOSOMY A condition in which one chromosome of a pair is missing.

MOSAIC	An individual (or tissue) with at least two cell lines differing in genotype (genes) or karyotype (chromosomes).
MULTI FACTORIAL	A trait determined by the interaction of one or more genes and environmental factors.
MUTATION	A permanent change in the genetic material. The process by which such a sudden structural change occurs, either through an alteration in the nucleotide sequence of the DNA coding for a gene or through a change in the physical arrangement of a chromosome.
MYELOMENINGOCELE	Spina bifida with cord and membranes protruding. A congenital defect of the central nervous system in which a sac containing part of the spinal cord and its meninges protrude through a gap in the vertebral column; frequently accompanied by hydrocephalus and mental retardation.
NONDISJUNCTION	Failure of two members of a chromosome pair to separate during cell division so that both are passed on to the same daughter cell. Meiosis in which there is a failure of paired homologous chromosomes to separate; results in an abnormal number of chromosomes in the daughter cells.
NUCHAL TRANSLUCENCY	An ultrasound measurement of the nap of the fetal neck. This first trimester measurement is used in combination evaluation with maternal serum biochemical markers and maternal age in predicting risk for chromosome abnormality during pregnancy.
NUCLEIC ACIDS	DNA and RNA, the molecules that carry genetic information.
NUCLEOTIDE	A building block of DNA or RNA. It includes one base, one phosphate molecule and one sugar molecule (deoxyribose in DNA, ribose in RNA).
ONCOGENES	Genes that can potentially cause cancer. A dominantly acting gene that induces uncontrolled cell growth and proliferation leading to tumor development.

P ARM	The short arm (petite) of a chromosome.
PEDIGREE	A genetic family tree.
PENETRANCE	Proportion of all of the individuals carrying a particular gene that manifests the trait determined by the gene. When penetrance is less than 100 percent in a dominant disorder, some individuals who inherit and transmit the gene do not manifest the disorder themselves.
PHENOTYPE	Individuals with a specific genotype. The frequency, under given environmental conditions, with which a specific genotype is expressed by those individuals that possess it, usually is given as a percentage. The observable characteristics of an individual.
PLEIOTROPY	Multiple phenotypic effects produced by a single mutant gene. The control by a single gene of several distinct and seemingly unrelated phenotypic effects
POINT MUTATION	A change in a single base pair in the DNA.
POLYMORPHISM	A genetic variation with two or more alleles that is maintained in a population so that the frequency of the most common one is not more than 0.99 and the frequency of one of the uncommon ones is maintained at least 0.01.
POLYDACTYLY	Presence of extra (supernumerary) digits.
POLYMERASE CHAIN REACTION (PCR)	PCR is now a common and often indispensable technique used in medical and biological research labs for a variety of applications. These include DNA cloning for sequencing, DNA-based phylogeny, or functional analysis of genes; the diagnosis of hereditary diseases; the identification of genetic fingerprints (used in forensics and paternity testing); and the detection and diagnosis of infectious diseases.
POLYPLOID	Any multiple of the basic haploid chromosome number other than diploid (e.g., 3n=triploid).

PREGNANCY ASSOCIATED PLASMA PROTEIN-A	Also known as PAPP-A, a protein found in maternal serum used in combination with other biochemical markers, ultrasound measurement of the nuchal translucency and maternal age to predict risk of chromosome abnormality during pregnancy.
PREIMPLANTATION DIAGNOSIS	Chromosomal or molecular disease diagnosis performed on an embryo after in vitro fertilization, usually at the eight-cell stage, and prior to uterine implantation.
PREMUTATION	An unstable series of trinucleotide repeat sequences in a gene that, during meiosis in the critical parent, can undergo expansion to a full mutation and thereby cause disease in offspring.
PROBAND	The index case through which a family comes to the attention of genetic professionals.
PROBE	A radioactive, biotinylated or otherwise labeled DNA or RNA sequence used to detect the presence of a complementary sequence by molecular hybridization.
PROTEIN	Large, complex chemicals that have many roles in the structure and functioning of cells.
PURINE	The bases adenine and guanine.
PYRIMIDINE	The bases cytosine and thymine in DNA and cytosine and uracil in RNA.
Q ARM	The long arm of a chromosome.
RECESSIVE	Refers to traits that are outwardly expressed only when both members of the same gene pair are identical. Of or relating to a trait that is expressed only when the determining allele is present in the homozygous condition.
RECOMBINANT DNA	The hybrid DNA produced in the laboratory by joining pieces of DNA from different sources.
RECOMBINANT DNA	Techniques for cutting apart and splicing together pieces

TECHNOLOGY	of DNA from different sources.
REPLICATION	Formation of an exact copy. DNA replication occurs when each strand of DNA acts as a template for a new, complementary strand formed according to base-pairing rules.
RESTRICTION ENZYME	An enzyme that recognizes a specific base sequence (usually four to six base pairs in length) in a double-stranded DNA molecule and cuts both strands of the DNA molecule at every place where this sequence appears.
RESTRICTION ENZYME RECOGNITION SITE	The DNA site where a specific restriction enzyme cuts the DNA molecule.
RESTRICTION FRAGMENT LENGTH POLYMORPHISM (RFLP)	The presence of two or more variants in the size of DNA fragments from a specific region of DNA that has been exposed to a particular restriction enzyme. These fragments differ in length because of an inherited variation in a restriction enzyme recognition site.
RESTRICTION	Fragments of DNA produced by cuts made FRAGMENTS with restriction enzymes.
RETROVIRUS	An RNA-containing virus that replicates by means of an enzyme (reverse transcriptase) which, upon infection of a host cell, makes a strand of DNA that complements the infecting virus strand; the double-stranded DNA produced in this way, then becomes part of the host cell's chromosomal DNA and reproduces along with it, eventually also producing an RNA strand identical to the original virus.
RIBONUCLEIC ACID	A single-stranded nucleic acid that contains the sugar (RNA) ribose. There are many forms of RNA, including messenger RNA, transfer RNA and ribosomal RNA (all involved in protein synthesis).

ROBERTSONIAN TRANSLOCATION	A translocation between two acrocentric chromosomes formed by end to end fusion at the centromere and loss of the short arms.
SEX CHROMOSOME	Chromosome responsible for sex determination. In humans, the X and Y chromosomes.
SEX LINKED	Refers to trait determined by a gene located on the X chromosome.
SOUTHERN BLOTTING	A procedure for transferring DNA fragments from an agarose gel to a filter paper without changing their relative positions.
SPINA BIFIDA	Neural tube defect of the spinal column through which the cord or membranes can protrude.
SPORADIC	Single occurrence of a disorder in a family, either of nongenetic etiology or the first appearance of a genetic disorder in that family.
SYNDACTYLY	Webbing or fusion of adjacent fingers or toes.
SYNDROME	Recognizable pattern of multiple abnormalities, presumed to have the same etiology.
TELOMERE	The extreme end of each chromosome arm.
TERATOGEN	An agent acting adversely on the embryo or fetus prenatally, altering morphology or subsequent function.
TERATOGENESIS	Exogenous induction of structural, functional or developmental abnormalities caused by agents acting during embryological or fetal development.
TORCH	An acronym for the following organisms: toxoplasmosis, rubella, cytomegalovirus and herpes simplex.
TOXIC GAIN OF FUNCTION	A mutation in a gene resulting in the production of an altered protein such that the activity of the new protein causes harm to the organism.
TRANSCRIPTION	The transfer of information from various parts of the

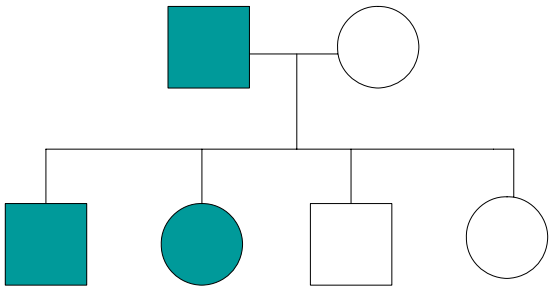
	DNA molecule to new strands of messenger RNA, which then carries this information from the nucleus to the cytoplasm.
TRANSLOCATION	A chromosome abnormality in which a chromosome, or a segment thereof, becomes attached to another chromosome. A translocation is balanced when no chromosome material is lost or gained in the rearrangement.
TRINUCLEOTIDE REPEAT SEQUENCE	A repeating series of three base pairs within a gene.
TRIPLE SCREENING	A prenatal screening test, performed between 15 and 20 weeks gestation, that measures alpha-fetoprotein, unconjugated estriol and human chorionic gonadotrophin. It is used to estimate risks for neural tube defects and Down syndrome in a given pregnancy.
TRIPLOID	Having three complete sets of chromosomes, resulting in 69 chromosomes per cell.
TRISOMY	A condition in which there are three, rather than two, copies of any one chromosome in the same cell. (Down syndrome is trisomy 21).
ULTRASONOGRAPHY	An imaging technique in which high frequency sound waves are used to outline internal body structures.
LEVEL II	An obstetrical ultrasonographic exam of the fetus targeting specific organs.
REAL TIME	Rapid repetitive sequenced images, that, seen through scanning, allows visualization of a moving structure such as the fetal heart, placenta, and fetal movement.
UNIPARENTAL DISOMY	The inheritance of two copies of a chromosome from the same parent instead of the normal situation of one from each parent.
VECTOR	A plasmid, phage or cosmid into which foreign DNA may be inserted for cloning.

WILD TYPE	Normal allele or normal phenotype.
ZYGOTE	A fertilized egg cell produced by the union of an egg and a sperm; the first cell of a new person.
ZYGOTE RESCUE	The spontaneous loss of a chromosome in a trisomic zygote such that the normal chromosome number (46) is restored. (May result in uniparental disomy.)

APPENDIX B. INHERITANCE PATTERNS

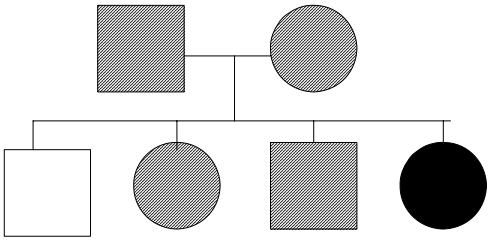
Autosomal Dominant

If one parent has a dominant gene disorder, there is a 50 percent chance that it will be passed to each child. Both sexes can be affected.



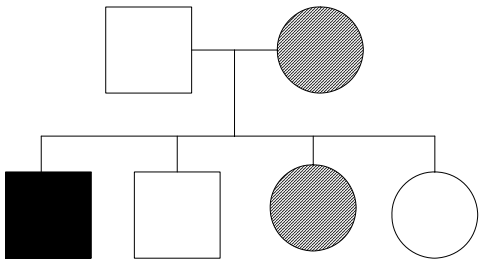
Autosomal Recessive

If both parents are carriers of a recessive disorder, there is a 25 percent chance that a child will be affected, a 50 percent chance that a child will be a carrier and a 25 percent chance that a child will not carry the gene and will be unaffected. Both sexes can be affected.

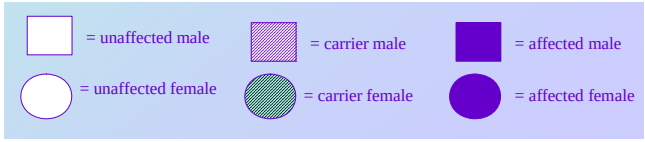


X-linked Recessive

If a woman is a carrier of an X-linked recessive condition, there is a 50 percent chance that a male child would be affected and a 50 percent chance that a female child will be a carrier. If a man has an X-linked recessive condition, all of his daughters will be carriers and none of his sons will be affected.



KEY



APPENDIX C. RESOURCES
APPENDIX C.1 – CLINICAL GENETIC CENTERS

CLINICAL GENETIC CENTERS

CARBONDALE AREA

St. Louis University Phone 314-577-5639
Cardinal Glennon Children's Hospital Fax 314-268-6411
Department of Pediatrics
1465 S. Grand Blvd.
St. Louis, MO 63104-1095

CHAMPAIGN/URBANA AREA

Carle Clinic Phone 217-383-3105
Genetics/Adult Medicine Fax 217-383-4681
602 W. University Ave.
Urbana, IL 61801

CHICAGO AND AREA SUBURBS

Children's Memorial Hospital Phone 773-880-4462
Division of Genetics Fax 773-929-9565
2300 Children's Plaza, Box 59
Chicago, IL 60614

Advocate Christ Hospital and Medical Center Phone 708-684-4395
Department of Pediatrics Fax 708-684-3045
4440 W. 95th St.
Oak Lawn, IL 60453

Evanston Hospital Phone 847-570-2864
Fetal Diagnostic Center Fax 847-570-1846
2650 Ridge Ave.
Evanston, IL 60201

John H. Stroger Hospital of Cook County Phone 312-864-4158
Division of Genetics Fax 312-864-9783
1900 W. Polk St.
Chicago, IL 60612

Reproductive Genetics Institute Phone 773-296-7095
2825 N. Halstead Fax 773-871-5221
Chicago, IL 60657

Loyola University Medical Center
Department of OB/GYN
2160 S. First Ave.
Maywood, IL 60148
Phone 708-216-8167
Fax 708-216-5669

Lutheran General Hospital
Perinatal Center
1875 Dempster St., Suite 340
Park Ridge, IL 60068
Phone 847-723-7705
Fax 847-723-8675

Illinois Teratogen Information Service (ITIS)
680 N. Lake Shore Drive, Suite 1230
Chicago, IL 60611
800-252-4847
Phone 312-981-4360
Fax 312-981-4366
*ITIS Hotline

Rush Medical Center
Section of Genetics
1753 Congress Parkway
Chicago, IL 60612
Phone 312-942-6298
Fax 312-942-2857

The University of Chicago Hospitals
Center for Medical Genetics, MC 2050
5841 S. Maryland Ave.
Chicago, IL 60637
Phone 773-834-9110
Fax 773-834-0556

University of Illinois at Chicago
College of Medicine
Department of Pediatrics
840 S. Wood St., Rm 1245, MC 856
Chicago, IL 60612
Phone 312-355-0732
Fax 312-355-0739

University of Illinois at Chicago
College of Medicine
Maternal Fetal Medicine Center
1801 W. Taylor St., M/C 650
Chicago, IL 60612
Phone 312-996-1391
Fax 312-996-4238

METRO-EAST

St. Louis University
Cardinal Glennon Children's Hospital
Department of Pediatrics
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Phone 314-577-5639
Fax 314-268-6411

Washington University School of Medicine
Division of Medical Genetics
Department of Pediatrics
1 Children's Plaza
St. Louis, MO 63110
Phone 314-454-6093
Fax 314-454-2075

PEORIA AREA

University of Illinois
College of Medicine at Peoria
Department of Pediatric Genetic Services
420 N.E. Glen Oak Ave.
Peoria, IL 61637
Phone 309-655-4242
Fax 309-655-2565

ROCKFORD AREA

Rockford Memorial Hospital
Department of Genetics
2400 N. Rockton St.
Rockford, IL 61101
Phone 815-971-5069
Fax 815-968-7830

SPRINGFIELD AREA

Southern Illinois University
School of Medicine
Department of Pediatrics
Genetic Diagnosis and Counseling Services
St. John's Pavilion
301 N. Eighth St., Third Floor, A169
Springfield, IL 62701
Phone 217-545-4839
Fax 217-545-5834

VANDALIA AREA

St. Louis University
Cardinal Glennon Children's Hospital
Department of Pediatrics
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Phone 314-577-5639
Fax 314-268-6411

For further information, please contact
Illinois Department of Public Health
Clinical Genetics Program
500 E. Monroe St., First Floor
Springfield, IL 62701
Phone 217-785-8101
Fax 217-557-5396

Note: No recommendations or endorsements are implied.

APPENDIX C.2. LOCAL PUBLIC HEALTH DEPARTMENTS
(LHD Genetic Grantees in BOLD)

Adams County Health Department 330 Vermont St. Quincy, IL 62301 217-222-8440	**Bond County Health Department** 503 S. Prairie St. Greenville, IL 62246 618-664-1442	Boone County Health Department 1204 Logan Ave. Belvidere, IL 61008-4031 815-544-2951
Brown County Health Department 111 W. Washington St. Mount Sterling, IL 62353 217-773-2714	Bureau County Health Department 526 Bureau Valley Parkway Princeton, IL 61356 815-872-5091	Calhoun County Health Department P. O. Box 158, 210 French St. Hardin, IL 62047 618-576-2428
Carroll County Health Department 822 S. Mill St. Mt. Carroll, IL 61053 815-244-8855	Cass County Health Department 331 S. Main St. Virginia, IL 62691 217-452-3057	**Champaign-Urbana Public Health District** 201 W. Kenyon Road Champaign, IL 61820-1488 217-531-4311
Chicago Department of Health DePaul University Center 333 S. State St., Second Floor Chicago, IL 60604 312-747-9872	Christian County Health Department 902 W. Springfield Road Taylorville, IL 62568 217-824-4113	Clark County Health Department 997 N. York St., P.O. Box 266 Martinsville, IL 62442 217-382-4207
Clay County Health Department 601 E. 12th St. Flora, IL 62839 618-662-4406	Clinton County Health Department 930A Fairfax St. Carlyle, IL 62231 618-594-8942	Coles County Health Department P.O. Box 1064, 825 18th St. Charleston, IL 61920-9391 217-348-0530
Cook County Department of Public Health 1010 Lake St., Suite 300 Oak Park, IL 60301 708-492-2010	**Crawford County Health Department** 202 N. Bline Blvd. Robinson, IL 62454 618-544-2723	Cumberland County Health Department P. O. Box 130 Toledo, IL 62468 217-849-3211
DeKalb County Health Department 2550 North Annie Glidden Road DeKalb, IL 60115 815-748-2438	DeWitt-Piatt Bi-County Health Department P. O. Box 518, 910 State Route 54 East Clinton, IL 61727	Douglas County Health Department 1250 East U.S. Route 36 Tuscola, IL 61953 217-253-4137

	217-935-3427	
DuPage County Health Department 111 N. County Farm Road Wheaton, IL 60187 630-682-7400	**East Side Health District** 638 N. 20th St. East St. Louis, IL 62205 618-271-8722 (Covering cities of Alorton, Brooklyn, Cahokia, Caseyville (southwest area), Centreville, E. St. Louis, Fairmont City, Lovejoy, Sauget and Washington Park..)	**Edgar County Health Department** 502 Shaw Avenue Paris, IL 61944 217-465-2212
Effingham County Health Department 901 W. Virginia St. P. O. Box 685 Effingham, IL 62401 217-342-9237	**Egyptian County Health Department** Rt 3, Box 90A 1412 U.S. Route 45 North Eldorado, IL 62930-9234 618-273-3326 (Covering: Galatin, Saline, White)	Evanston Health Department See Chicago Department of Public Health
Fayette County Health Department P.O. Box 340 509 W. Edwards St. Vandalia, IL 62471 618-283-1044	Ford-Iroquois Public Health Department 114 N. Third St. Watseka, IL 60970 815-432-2483	Franklin-Williamson Bi-County Health Department 120 Express Drive Marion, IL 62959-9808 618-993-8111
Fulton County Health Department 700 E. Oak St. Canton, IL 61520 309-647-1134	Greene County Health Department 310 Fifth St. Carrollton, IL 62016 217-942-1134	Grundy County Health Department 1320 Union St. Morris, IL 60450 815-941-3113
Hamilton County Health Department Courthouse, Room 5 McLeansboro, IL 62859 618-643-3522	Hancock County Health Department 671 Wabash St. Carthage, IL 62321 217-357-2171	**Henderson County Health Department** P.O. Box 220 Gladstone, IL 61437-0220 309-627-2812 (Covering: Warren County)
Henry County Health Department 4424 U.S. Route 34 Kewanee, IL 61443 309-852-0197	**Jackson County Health Department** 415 Health Department Road Murphysboro, IL 62966 618-684-3143	**Jasper County Health Department** 106 E. Edwards St. Newton, IL 62448 618-783-4436 (Covering: Richland County)

Jefferson County Health Department #1 Doctors Park Road, Ste. F Mount Vernon, IL 62864 618-244-7134	**Jersey County Health Department** 1307 State Route 109 Jerseyville, IL 62052 618-498-9565	Jo Daviess County Health Department 9483 U.S. Route 20 West P. O. Box 318 Galena, IL 61036 815-777-0263
Kane County Health Department 210 S. Sixth St. Geneva, IL 60134 630-208-3801	**Kankakee County Health Department** 2390 W. Station St. Bradley, IL 60901 815-937-3565	Kendall County Department of Health & Human Services 811 W. John St. Yorkville, IL 60560 630-553-9100
Knox County Health Department 1361 W. Fremont St. Galesburg, IL 61401 309-344-2224	Lake County Health Department 3010 Grand Ave. Waukegan, IL 60085 847-377-8328	**LaSalle County Health Department** 717 Etna Road Ottawa, IL 61350 815-433-3366
Lawrence County Health Department P.O. Box 516 Lawrenceville, IL 62439 618-943-3302	Lee County Health Department 309 S. Galena Ave., Suite 100 Dixon, IL 61021 815-284-3371	Livingston County Health Department Livingston County Health and Education Bldg P.O. Box 886, 310 E. Torance Ave. Pontiac, IL 61764 815-844-7174
Logan County Health Department 109 Third St., P. O. Box 508 Lincoln, IL 62656 217-735-2317	**Macon County Health Department** 1221 E. Condit St. Decatur, IL 62521-1405 217-423-6988	**Macoupin County Health Department** 805 N. Broad St. Carlinville, IL 62626 217-854-3223
Madison County Health Department 101 E. Edwardsville Road Wood River, IL 62095 618-692-8954	**Marion County Health Department** 600 E. Main St. Salem, IL 62881 618-548-3878	Marshall County Health Department 319 Sixth St., P.O. Box 156 Lacon, IL 61540 309-679-6000
Mason County Health Department U.S. Route 136 East, P.O. Box 557 Havana, IL 62644 309-543-2201	**McDonough Co. Health Department** 505 E. Jackson St. Macomb, IL 61455 309-837-9951	McHenry County Health Department 2200 N. Seminary Ave. Woodstock, IL 60098 815-334-4510

McLean County Health Department 200 W. Front St., Room 304 Bloomington, IL 61701 309-888-5450	Menard County Health Department 1120 N. Fifth St. Petersburg, IL 62675 217-632-3283	**Mercer County Health Department** 305 N.W. 7th St. Aledo, IL 61231 309-582-3759
Monroe County Health Department 901 Illinois Ave. Waterloo, IL 62298 618-939-3871	**Montgomery County Health Department** 11191 Illinois Route 185 Hillsboro, IL 62049-0128 217-532-2001	Morgan County Health Department 345 W. State St. Jacksonville, IL 62650 217-245-5111
Moultrie County Health Department 2 W. Adams St. Sullivan, IL 61951 217-728-4442 or 4114	Oak Park Health Department 6026 W. Roosevelt Road Oak Park, IL 60304 708-383-6400 Ext. 5482	Ogle County Health Department 907 W. Pines Road Oregon, IL 61061 815-732-3201, Ext. 247
Peoria City/County Health Department 2116 N. Sheridan Road Peoria, IL 61604 309-679-6609	**Perry County Health Department** 907 S. Main St., P. O. Box 49 Pinckneyville, IL 62274 618-357-5371	Piatt County Health Department 1020 S. Market Street Monticello, IL 61856 217-762-7911
Pike County Health Department 113 E. Jefferson St. Pittsfield, IL 62363-1420 217-285-4407	Putnam County Health Department 220 E. High St., Suite 102 Hennipin, IL 61327 815-925-7001	**Randolph County Health Department** 2515 State St. Chester, IL 62233 618-826-5007
Rock Island County Health Department 2112 25th Ave. Rock Island, IL 61201 309-793-1955	**St. Clair County Health Department** #19 Public Square, Suite 150 Belleville, IL 62220 618-233-7703 (Covering cities of Caseyville, Collinsville (southern tip), Dupo and rest of St. Clair County except as covered by East Side Health District)	**Sangamon County Department of Public Health** 2501 N. Dirksen Parkway Springfield, IL 62702 217-535-3100
Schuyler County Health Department 127 S. Liberty St. P. O. Box 320 Rushville, IL 62681 217-322-4373	Scott County Health Department 335 W. Cherry St. Winchester, IL 62694 217-742-8203	Shelby County Health Department R.R2 , Box 54 1810 W. S. Third St. Shelbyville, IL 62565 217-774-2355

Southern Seven Health Department 37 Rustic Campus Drive Ullin, IL 62992 618-634-2297 (Covering Alexander, Hardin, Johnson, Massac, Pope, Pulaski and Union counties)	**Stark County covered by Henry County**	Skokie Health Department 5127 Oakton St., P. O. Box 309 Skokie, IL 60077 847-933-8252
Stark County Health Department 4424 U. S. Highway 34 Kewanee, IL 61433 309-852-3115	Stephenson County Health Department 10 W. Linden St. Freeport, IL 61032 815-235-8370	Stickney Township Public Health District 5635 State Road Burbank, IL 60459 708-424-9200
Tazewell County Health Department 21306 Illinois Route 9 Tremont, IL 61568-9252 309-925-5511	Vermilion County Health Department 200 S. College St. Danville, IL 61832 217-431-2662	Wabash County Health Department 130 W. Seventh St. Mount Carmel, IL 62863 618-263-3873
Washington County Health Department 177 S. Washington St. Nashville, IL 62263 618-327-3644	Wayne County Health Department 405 N. Basin Road P.O. Box 645 Fairfield, IL 62837 618-842-5166	Whiteside County Health Department 1300 W. Second St. Rock Falls, IL 61270 815-772-7411 Ext. 16
Will County Health Department 501 Ella Ave. Joliet, IL 60433 815-727-8485	**Winnebago County Health Department** 401 Division St. Rockford, IL 61104 815-962-5092	Woodford County Health Department 109 S. Major St. Eureka, IL 61530 309-467-2371

APPENDIX C.3 - SUPPORT ORGANIZATIONS

Alliance of Genetic Support Groups provides a listing of national and local support organizations for specific disorders.

35 Wisconsin Circle, Suite 440
Chevy Chase, MD 20185
800-336-4363
Fax 301-654-0171

Division of Specialized Care for Children (DSCC) at the University of Illinois works to help children with disabilities by offering services that include reduced cost or no-cost diagnostic services or medical treatment.

General Information
800-322-3722
Fax 312-413-3445

Illinois Teratogen Information Service, a project funded by Illinois Department of Public Health, provides information about all types of exposures during pregnancy including any medication, chemical, infectious disease or environmental agent that might interfere with the normal development of a fetus and result in the loss of a pregnancy or in a birth defect or a pregnancy complication.

680 N. Lake Shore Drive, Suite 1230
Chicago, IL 60611
312-981-4360
Fax 312-981-4366

March of Dimes Birth Defects Foundation (MOD) is a non-profit organization dedicated to improving the health of babies by reducing birth defects and infant mortality and by providing written information about specific disorders.

Illinois Chapter
111 W. Jackson Blvd.
22nd Floor
Chicago, IL 60604
312-435-4007
Fax 312-435-0988

MUMS-Mothers United for Moral Support is a nationwide parent-to-parent network for families of children with any special needs.

150 Custer Court
Green Bay, WI 54301-1243
920-336-5333
Fax 920-339-0995

National Organization for Rare Disorders Inc. (NORD) is a federation of voluntary health organizations dedicated to helping people with rare orphan diseases.

P.O. Box 8923
New Fairfield, CT 06812
800-999-NORD
Fax 203-746-6481

	Genetic Internet Sites	
Subject	**Description**	**Address**
Genetic Center for Disease Control (CDC) site	CDC genetics site with many links	http://www.cdc.gov/genomics/default.htm
Genetics Home Reference Page	Online resource and starting point for genetic-counseling-related information.	http://ghr.nlm.nih.gov
March of Dimes	Educational information	http://www.modimes.org/
National Cancer Institute	Main online information center for patients and physicians for genetics information	http://cancernet.nci.nih.gov/p_genetics.html
Online Mendelian Inheritance in Man	Maintained by John Hopkins University School of Medicine, provides information on identified genetic syndromes	http://www3.ncbi/nlm.nih.gov/Omim/
National Organization for Rare Disorders	Information on rare orphan diseases on specific information on 6000 rare diseases	http://www.rarediseases.org/
National Human Genome Research Institute	Human Genome Project for National Institutes of Health	http://www.nhgri.nih.gov/
Genetic Alliance	Formerly The Alliance of Genetic Support Groups, Inc. - international coalition genetic support organizations	http://www.geneticalliance.org/
GeneTests	Funded by National Library of Medicine of the NIH and Maternal & Child Health Bureau of HRSA. Genetic resource and PowerPoint slideshow	http://www.genetests.org/
GeneClinics	Clinical information resource relating genetic testing to diagnosis, management and genetic counseling	http://www.geneclinics.org/
Kansas University Medical Center	For genetic professionals, updated regularly with clinical, research and educational resources	http://www.kumc.edu/gec/geneinfo.html

Family Village	Global community integrating information, resources and communication on specific diagnoses, adaptive equipment, etc.	http://www.familyvillage.wisc.edu/index.htmlx
National Coalition for Health Professional Education in Genetics	National effort to promote health professional education and access to information on human genetics American Medical Association (AMA), American Nursing Association (ANA) and National Human Genome Research Institute	http://www.nchpeg.org/
New York Online Access to Health (NOAH)	Full-text health information for an underserved population of health consumers supporting English and Spanish	http://www.noah-health.org/
Human Genome Project Information	Learn the basics about the human Genome Project	http://www.ornl.gov/hgmis
The National Heart, Lung, and Blood Institute (NHLBI)	Provides leadership for a national program in diseases of the heart, blood vessels, lungs, and blood; sleep disorders; and blood resources.	http://www.nhlbi.nih.gov/
American College of Medical Genetics	Organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals	http://www.acmg.net/
National Association for Down Syndrome	National organization providing information and links on Down Syndrome	http://www.nads.org/
Illinois Teratogen Information Service	Funded through support from IDPH	http://www.fetal-exposure.org/
Mothers United for Moral Support (MUMS)	National Parent-to-Parent Network. Also provides database of disorders/conditions	http://www.netnet.net/mums
National Organization on Fetal Alcohol Syndrome	Information on all aspects of fetal alcohol syndrome	http://www.nofas.org/
Autism Society of America	Diagnosis and further information on autism spectrum disorders	http://www.autism-society.org/site/PageServer?pagename=about.home
Ethnomed Homepage	Resource center for specific cultural beliefs with ability to trarnslate materials	http://www.ethnomed.org

	Genetics Basic Review Internet Sites	
1,2,3 Genomics	Learn basics and links to many sources	http://123genomics.com
DNA from the Beginning	Animated primer on the basics of DNA, genes, heredity – image gallery, video interview, problems and links	http://www.dnaftb.org/dnaftb/
Genetics@GlaxoSmithKline	Interactive information on genetic basics, DNA, mutations and genes	http://genetics.gsk.com/overview.htm
Tour of the Basics Genetic Science Learning Center – University of Utah	What is DNA? Gene? Protein? Chromosome? Heredity? Trait?	http://learn.genetics.utah.edu/units/basics/tour/
	Family Health History Chronic Diseases	
Cancer, Risks and Genetics	Cancer genetics overview	http://www.fhcrc.org/science/phs/cgn/under.htm
Fact Sheets on Chronic Diseases – Minnesota Health Department	Chronic Disease Genomics Project Family Health History Fact Sheets – breast, prostate, colorectal cancer, cardiovascular disease and diabetes	http://www.health.state.mn.us/divs/hpcd/genomics/resources/fs/breast.html
U.S. Surgeon General's My Family Health Portrait Tool	Fact sheets for diabetes, heart disease, hereditary breast and ovarian cancer, hereditary non-polyposis colorectal cancer syndrome and couple with history of inherited disorder	www.surgeongeneral.gov/familyhistory
Genetic Science Learning Center – University of Utah	Using family history to improve your health – facts and informational handouts for families on heart disease and stroke, asthma, diabetes, osteoporosis, cancer and blood pressure and cholestrol	http://learn.genetics.utah.edu/units/health/index.cfm
Your Disease Risk – Washington University	Reference site for families to determine disease risk for cancer, diabetes, heart disease, osteoporosis and stroke	http://www.yourdiseaserisk.siteman.wustl.edu
Does it Run in my Family	A guide for family health history supported by U.S. Department of Health and Human Services a collaborative project (16 pages)	http://www.geneticalliance.org/ksc_assets/pdfs/runinfamily.pdf

	Newborn Screening Internet Sites	
Subject	**Description**	**Address**
National Taskforce for Newborn Screening and Genetic Resource Center (NNSGRC)	Cooperative agreement between the Maternal and Child Health Bureau, Genetic Services Branch, Human Resources and Service Administation and University of Texas Health Science Center at San Antonio, Department of Pediatrics	http://genes-r-us.uthscsa.edu./
Illinois Department of Public Health Genetics/NBS Section	Illinois Department of Public Health Newborn Screening Web site	http://www.idph.state.il.us/HealthWellness/genetics.htm
Save Babies Through Screening	Non-profit organization that provides up-to-date information and resources on newborn screening and disorders as a parent support network with many links to other related sites	http://www.savebabies.org/
Fact Sheets for Use by Physicians and Parents	American College of Medical Genetics Action (ACT) Fact Sheets for Newborn Screening Disorder Algorithms	http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm
Amino Acid Disorders Maple syrup urine disease (MSUD)	MSUD Family Support Group, 24806 SR 110, Goshen, IN 46526, Phone: (219) 862-2992	http://www.msud-support.org/
Tyrosinemias Type I, II, III	Genetics Home Reference Page – definition and symptoms	http://ghr.nlm.nih.gov/condition=tyrosinemia
Homocystinuria	Genetics Home Reference Page – definition and symptoms	http://ghr.nim.nih.gov/condition=homocystinuria
Biotinidase Deficiency	Genetics Home Reference Page – definition and symptoms	http://ghr.nlm.nih.gov/condition=biottindasedeficiency
Congenital Adrenal Hyperplasia	MAGIC Foundation Informational Web site	http://www.magicfoundation.org/www/docs/100/congenital-adrenal-hyperplasia
Congenital Hypothyroidism (CH)	MAGIC foundation, general information (4 pages)	http://www.magicfoundation.org/www/docs/114.126/hypothyroidism-clinicalhypothyroidism.html

CH	Maternal Thyroid Deficiency during Pregnancy and Subsequent Neuropsychological Development of the Child, New England Journal of Medicine, 2 pages	http://content.nejm.org/cgi/content/short/341/8/549
Fatty Acid Oxidation Disorders	Fatty Oxidation Disorder Family Support Group 805 Montrose Drive, Greensboro, NC 24710 Phone 336-547-8682	http://www.fodsupport.org/
Galactosemia	Galactosemia Handbook – A Guide for Families by Texas Department of Health (16 pages)	http://www.dshs.state.tx.us/newborn/handbook.htm
Phenylketonuria (PKU)	PKU Organization of Illinois (3 pages and links)	http://www.pkuil.org
PKU	National PKU News	http://www.pkunews.org/
Sickle Cell Disease (SCD)		
SCD	New Hope for People with Sickle Cell Anemia	http://www.fda.gov/fdac/features/496_sick.html
SCD	American Academy of Pediatrics Policy Statement, 9 pages	http://www.aap.org/policy/re1011.html
SCD	Sickle Cell Disease Association of Illinois	http://www.sicklecelldisease-il.org/
SCD	Sickle Cell Information Center - Hemoglobins	http://www.scinfo.org/
Urea cycle disorders	National Urea Cycle Disorders Foundation, 4841 Hill Street, La Canada, CA 91011 Phone 800-38NUCDF	http://www.nucdf.org/
Newborn Hearing Screening	National Center for Hearing Assessment and Management - Utah State University	http://www.infanthearing.org/

	Sudden Infant Death Syndrome (SIDS) Internet Sites	
American SIDS Institute	Frequently Asked Questions and many links	http://www.sids.org/
Consumer Product Safety Commission (CPSC)	Information on bedding and suffocation, safe sleeping environments and safety recalls	http://www.cpsc.gov/
Back to Sleep	Campaign information	http://www.nichd.nih.gov/sids
Compassionate Friends	Support group, grief counseling and booklets	http://www.compassionatefriends.org/
CJ Foundation	With many links	http://cjsids.com/index.htm
National Institute of Child Health and Human Development (NICHD)	Policy and recommendations for children health issues and info on "Back to Sleep" with downloadable versions of campaign's brochures and basic fact sheets	http://www.nichd.nih.gov/
SIDS Alliance		http://sidsalliance.org/
SIDS International		http://www.sidsinternational.minerva.com.au/
SIDS Network	Web pages can be translated from English to Spanish, French, German, Italian and Portuguese using Alta Vista Translation	http://sids-network.org/

APPENDIX C. 5. RECOMMENDED GENETICS TEXTS

The following is a brief listing of several texts that geneticists and genetic counselors find to be useful references. These books often are referred to when diagnosing a genetic condition. However, they also can be helpful for the genetic coordinator in educating herself or himself about different conditions. If an on-site clinic is being established, the genetic coordinator may wish to ask the participating geneticist or genetic counselor which texts he or she desires to have available at the local health department.

Baraister, M., Winter, R.M. 1996. *Color Atlas of Congenital Malformation Syndromes*. Mosby-Wolfe: London.

Buyse, M.L. 1990. *Birth Defects Encyclopedia*. Blackwell Scientific Publications: Boston.

Jones, K.L. 2005. *Smith's Recognizable Patterns of Human Malformation*. W.B. Saunders Company: Philadelphia.

McKusick, V.A. 1998. *Mendelian Inheritance in Man*. Johns Hopkins University Press: Baltimore.

Rimoin, D., Connor, J.M., Pyeritz, R. 2006. *Emery and Rimoin's Principles and Practice of Medical Genetics*. Churchill Livingstone: New York.

Robinson, A. and Linden, M. 2002. *The Clinical Genetics Handbook*. Blackwell Scientific Publications: Boston.

Scriver, C.R., Beaudet, A.L., Sly, W.S., Valle, D. 2001. *The Metabolic and Molecular Bases of Inherited Disease*. McGraw-Hill, Inc.: New York.

Wynbrandt, James and Ludman, Mark 2008. *The Encyclopedia of Genetic Disorders and Birth Defects.* Facts on File: New York.

APPENDIX D. QUARTERLY REPORTING INSTRUCTIONS

GENERAL INSTRUCTIONS

In order to meet State Guidelines and to ensure uniform, adequate and timely reporting from all grant recipients, the following reporting forms have been implemented.

Timelines: Data from all Illinois Department of Public Health funded grantees for follow-up services related to genetics will be collected on a quarterly basis as follows:

Reporting Period	Quarter	Report Due Date
July 1-Sep 30	1	October 31
Oct 1 - Dec 31	2	January 31
Jan 1 - Mar 31	3	April 30
April 1- June 30	4	July 31

Data should be collected on four different forms:

1. Summary Client Data Form
2. Genetic Screening Tool Tracking Form
3. Educational Activities Form
4. Additional Information Form (optional)

Submit completed forms to:

Genetics Program
Division of Health Assessment and Screening
Illinois Department of Public Health
535 W. Jefferson, Second Floor
Springfield, Illinois 62761

What to Report

Data should be reported for any client requesting services related to genetics regardless of age and for any client being followed for an abnormal newborn screening test.

Instructions for Quarterly Reporting Forms (4)

If you are using Cornerstone for the Family Health History (FHH) Assessment, Cornerstone will be gathering statistics into report titled: Summary Client Data Form. However,in order for this to gather the correct information you must identiy these clients using the Program Code 15 with the appropriate diagnosis in PA39. The Genetic Screening Tool Tracking Form will be populated automatically after doing the FHH assessment in Cornerstone and the genetic coordinator has filled in the PA39 screen marking positive family heath histories, referred, appointment kept or refused. Please see Appendix F for further Cornerstone instructions. If you are not using Cornerstone for the FHH assessment and continue to use paper forms or assessing clients not in Cornerstone, those numbers and reports will have to be tabulated together and submitted on a quarterly basis.

1. Summary Client Data Form NOTE: Please complete this form by recording the **totals only** (numbers, no names) for each category. Totals in all categories should equal the total number of clients served, except for the "services provided category'' which will usually be greater than the number of clients seen for follow up genetic services, if multiple services were provided. This form is for educational services provided to anyone **with a genetic condition** that has a genetic diagnosis. **Do not report genetic screening tool numbers or those that were positive for a referral on this form. This form is to report only those that have been identified with a genetic condition and are receiving follow-up by your health department through WIC/FCM or other health department program.**

Agency Name: Indicate the name of your agency.
Reporting Period: List the month(s) and year included in this report.
County of Residence: List the number of clients seen by county of residence.
Sex: Indicate the total number of clients served by sex: Female, Male, Unknown

In each of the next two categories (Hispanic and Race) *please give a response under both categories*:

Hispanic: Indicate the totals of all clients regarding Hispanic ethnicity (Yes and No).
Race: Indicate the totals of all clients by race as indicated on form.
Source of Payment: Indicate totals under the appropriate category to indicate the method of payment for services.
Age: Indicate the total number of clients seen in each age category for this reporting period.
First Source of Referral: Indicate the totals for the initial source of referral using the following categories:

Health Department Programs with the following listings and definitions:
APORS (Adverse Pregnancy Outcome Reporting System)
WIC (Women, Infant and Children Program)
Family Case Management
Family Planning

Other LHD/IDPH - Other program at the local health department or IDPH referral
Genetic Center - Genetic Services at Medical Center
Physician, school, self-referral or other

Services Provided: Indicate the totals under each category to reflect the number of times that the following services were provided during the reporting period:

Phone call	Family Counseling	Other/Not Stated
Home Visit	Letter	Clinic Visit
Referred to Genetic Center	Educational Material Provided	

Diagnosis or Reason for Referral: Indicate the totals for each category. **NOTE**: *Each client should be listed under only one category - choose one primary diagnosis or suspected reason for referral for each client (see enclosed reason for referral/diagnosis explanation sheet*).

Newborn Screening (NBS) Disorders
-Amino Acid Disorders (not PKU)
-Biotinidase Deficiency
-Congenital adrenal hyperplasia
-Fatty Acid Disorders
-Galactosemia
-Hypothyroidism
-Organic Acid Disorders
-Metabolic/endocrine disorder
-Phenylketonuria
-Sickle Cell Disease
- Other hemoglobin disease
- Sickle Cell Trait
-SC Disease
-Other hemoglobin trait
- Urea Cycle Disorder
- Unsatisfactory NBS test
- Unsatisfactory NBS hearing test

Other Genetic
-Chromosomal syndrome
-Environmental teratogen anomaly
-Functional disorder (mental retardation,
- Growth delay, severe visual or hearing disorder, cerebral palsy (CP), psycho-social/behavioral disorder)
-Multiple congenital anomalies
-Neuromuscular disorders
-Reproductive risk
-Sudden Infant Death Syndrome (SIDS)
-Single gene disorder
-Single malformation
-Skeletal/Connective Tissue Disorder
-Blood Disorder (hemophilia, etc.)

Chronic Diseases
-Asthma, Arthritis, Diabetes (at this time we aren't referring patients for genetic counseling on these disorders so these will be left blank on the Summary Client Data Form.
-Cardiovascular Disease and Cancer could be filled out for a referral and follow up if they meet the guidelines for referral in the instructions of early family history and a pattern of inheritance or early death (please see revised guidelines)
-Other genetic disease (describe)

2. Genetic Screening Tool Tracking Form

Agency: Indicate the name of your agency.
Date of Services: List the month(s) and year included in this report.
Program Name: Indicate the name of the program utilizing the Genetic Screening Tool: (WIC/ Family Case Management, Family Planning, etc.).
Clients Screened: Indicate the number of clients screened in each program area.
#Clients with Positive Indicator: Indicate number of clients who had a positive indicator.
Information Required for Clients with Positive Indicator:
Please fill in the left hand side of the form for the clients that had a positive indicator. Total the number for each disorder and place that number by the corresponding disorder on the right side. Follow across the form by disorder to indicate the number referred to a geneticist or genetic counselor, the number keeping appointment, number refused/postponed, educational material provided. On this form, a client may have more than one positive indicator.
#Clients Referred to Genetics: Report the number of clients *actually referred* to a clinical genetics center or those that are being seen by a geneticist/genetic counselor.
#Clients Kept Appointment: Indicate the number of clients who kept their appointment with the clinical genetics center (This number may not be known during this quarter, report number as is it known.)
Clients Refused Referral/Postponed Referral: Indicate the number of clients who were referred to a clinical genetics center, but refused the referral or did not follow through with appropriate paperwork for referral.

3. Educational Activities Form

Agency: Indicate the name of your agency.
Reporting Period: List the month(s) and year included in this report.
Date: Record date of presentation.
Presentation Title: Include subject of presentation (this can include trainings on folic acid, newborn screening, genetics, SIDS).

Approximate Audience Size: Record audience size or indicate approximate size of radio, TV or health fair audience and place under appropriate columns indicating either consumer group or professional group by numbers of either or both in attendance.

4. **Additional Information Form (Optional)**

Agency: Indicate the name of your agency
Reporting Period: List the month(s) and year included in this report
Additional Comments: Describe any additional noteworthy activities that occurred during the reporting period. Any brochures developed should be approved prior to circulation. Please share your accomplishments and new ideas to educate your clients.

REASON FOR REFERRAL OR DIAGNOSIS CATEGORIES

Newborn Screening Disorders	
Disease Category	Specific Information
Biotinidase deficiency Congenital adrenal hyperplasia Cystic Fibrosis Galactosemia Hypothyroidism Phenylketonuria Sickle cell disease Other Hemoglobin Disease Sickle cell trait Other Hemoglobin Trait	Suspect on newborn screening (NBS) for, or diagnosed with the disorder identified. and hyperphenylalaninemia including SC and S/Beta Thalassemia including C and E trait
Amino acid disorders and sub- category of: Urea Cycle Disorders	Maple syrup urine disease (MSUD) Tyrosinemia -type I Homocystinuria (HCU)/hypermethioninemia (PKU is also an amino acid disorder but will continue to be listed separately) 5-Oxoprolinuria (Pyroglutamic aciduria) Citrullinemia aka argininosuccininate synthetase deficiency Argininosuccinic aciduria aka argininasuccinate lyase deficiency (ASAL) Argininemia aka arginase deficiency
Acylcarnitines: A. Fatty acid oxidation disorders	Carnitine palmitoyl transferase deficiency type II Carnitine/acylcarnitine translocase deficiency Glutaric aciduria type 2 Carnitine palmitoyltransferase 1A Long chain 3-hydroxy-CoA hydrogenase deficiency (LCHAD) Very long chain acyl-CoA dehydrogenase deficiency (VLCAD) Medium chain acyl-CoA dehydrogenase deficiency (MCAD) Short chain acyl-CoA dehydrogenase deficiency (SCAD) Medium/short chain L-3 hydroxyacyl-CoA dehydrogenase deficiency Trifunctional protein deficiency (TFPD) Isobutyryl-CoA dehydrogenase deficiency (IBCD) Glutaric Acidemia Type II aka multiple acyl-CoA dehydrogenase deficiency)

B. Organic acid disorders	2-methylbutyryl Co-A dehydrogenase deficiency 3-methylcrotonyl-CoA carboxylase deficiency 3-hydroxy-3-methylglutaric-CoA lyase def 3-methylglutaconic aciduria 3-Ketothiolase Deficiency aka mitochondrial Acetoacetyl-CoA Thiolase Deficiency Beta-ketothiolase deficiency Glutaric aciduria, type I Propionic acidemia Isovaleric acidemia Methylmalonic acidemia Malonic aciduria Multiple carboxylase deficiency
Lysosomal Storage Disorders	Fabry Disease Gaucher Disease Krabbé Disease Mucopolysaccharidoses Niemann-Pick Disease
Unsatisfactory NBS test	Any newborn screening test that must be repeated; including a test done before 24 hours of age, or a test where the specimen was improperly collected, or was inadequate for testing
Unsatisfactory Newborn Hearing Test	Test was unsatisfactory and needs to be repeated or referred to an audiologist for confirmation/testing.

Genetic Disorders	
Chromosomal syndrome	Any chromosomal abnormality, such as Down syndrome, trisomy 13 or 18, Turner syndrome, Klinefelter syndrome, or Fragile X syndrome
Environmental/teratogen induced anomality	Any disorder caused by prenatal exposure to an environmental agent that causes birth defects, such as exposure to alcohol, prescription drugs, infections, chemicals, radiation, etc.
Functional disorder	Mental retardation, developmental disability, growth retardation, failure to thrive, seizure disorders, sensory deprivation such as visual or hearing impairment, psychological, behaviorial or psychiatric disorder, cerebral palsy, etc.
Metabolic/Endocrine Disorder	Metabolic disorders other than those tested for on newborn screening, such as Tay Sachs disease, MPS (mucopolysaccharidoses), etc.
Multiple congenital anomalies	More than one malformation
Neuromuscular disorder	Muscular dystrophy, Huntington disease, etc.
Reproductive risk	Infertility, fetal loss, consanguinity (related parents), family history of a genetic concern, etc.
SIDS (Sudden Infant Death Syndrome)	Any sudden, unexpected death of an infant to be confirmed by autopsy
Single gene disorder	Any other disorder that is inherited and caused by a defect in a single gene
Single malformation	Any single malformation, such as a neural tube, limb, cardiovascular or renal/urogenital defect, cleft lip/palate, etc.

Skeletal/connective tissue disorder	Neurofibromatosis, tuberous sclerosis, dysplasia, Marfan syndrome, Ehlers-Danlos, or other disorders of the bones or connective tissue
Blood Clotting Disorders	Hemophilia, thrombophilias,i.e., Factor V Liden, protein C deficiency and protein S deficiency
Chronic Diseases	
Asthma	Not referred to genetic center, but can be counted on screening tool tracking form for statistic purposes
Diabetes	Not referred to genetic center, but can be counted on screening tool tracking form for statistic purposes
Cardiovascular Disease	May be referred to genetic center if criteria for high risk is involved; can be used on screening tool tracking form for positive indicator
Cancer	May be referred to genetic center if criteria for high risk is involved; can be used on screening tool tracking form for positive indicator
Arthritis	Not referred to genetic center, but can be counted on screening tool tracking form for Statistic purposes
Other genetic disease	Any inherited disorder that does not fit in one of the above categories – NBS/Genetics/Chronic Diseases

Illinois Department of Public Health
Summary Client Data Form - Genetics Follow-Up
Revised July 2010

Agency Name | Reporting Period

Use this form for follow-up services for clients diagnosed with genetic disorders or who have agreed to a referral to genetics or genetic counselor.

County of Residence (indicate # of residents from each county)

Adams		Henderson		Ogle	
Alexander		Henry		Peoria	
Bond		Iroquois		Perry	
Boone		Jackson		Piatt	
Brown		Jasper		Pike	
Bureau		Jefferson		Pope	
Calhoun		Jersey		Pulaski	
Carroll		Jo Daviess		Putnam	
Cass		Johnson		Randolph	
Champaign		Kane		Richland	
Christian		Kankakee		Rock Island	
Clark		Kendall		Saline	
Clay		Knox		Sangamon	
Clinton		Lake		Schuyler	
Coles		LaSalle		Scott	
Cook		Lawrence		Shelby	
Crawford		Lee		Stark	
Cumberland		Livingston		St. Clair	
DeKalb		Logan		Stephenson	
DeWitt		Macon		Tazewell	
Douglas		Macoupin		Union	
DuPage		Madison		Vermilion	
Edgar		Marion		Wabash	
Edwards		Marshall		Warren	
Effingham		Mason		Washington	
Fayette		Massac		Wayne	
Ford		McDonough		White	
Franklin		McHenry		Whiteside	
Fulton		McLean		Will	
Gallatin		Menard		Williamson	
Greene		Mercer		Winnebago	
Grundy		Monroe		Woodford	
Hamilton		Montgomery		Unknown	
Hancock		Morgan		Out of State	
Hardin		Moultrie		Total	0

Demographics

Sex		Age	
Female		<1mo	
Male		1-5 mo	
Unknown/Not Stated		>6 mos-1yr<	
Total	0	2-9 yrs	
Hispanic		10-19 yrs	
Yes		20-34 yr	
No		35-49 yrs	
Not Indicated		50+ yrs	
		Unknown/Not Stated	
Total	0		
Race		Total	0
White		**Source of Referral**	
Black		APORS	
Am Ind/AL Nat		WIC/PCW	
Asian/Pacific Isl.		Fam Ping	
Unknown/Not Stated		Other LHD Pgm	
Total	0	Genetic Ctr	
Source of Payment		Doctor	
Medicaid		School	
Nonmedicaid		Self	
Unknown/Other		Other	
Total	0	Total	0
Services Provided			
Phone Call		Fam Counseling	
Letter		Referred Genetics	
Home Visit		Other/Not Stated	
Clinic Visit		Educ Mat'l Provided	
Total			0

Diagnosis/Reason for Referral

NBS Disorders	
Amino Acid Disorders	
Biotinidase Deficiency	
Cystic Fibrosis	
Congenital Adrenal Hyperplasia	
Fatty Acid Oxidation Disorders	
Galactosemia	
Hypothyroidism	
Lysosomal Storage Disorders	
Organic Acid Disorders	
Phenylketonuria	
Sickle Cell Disease	
Other Hgb Disease	
Sickle Cell Test	
Other Hgb Trait	
Urea Cycle Disorders	
Unsat Newborn (NB) Screen	
Unsat NB Hearing Screen	

Genetic Disorders	
Chromosomal Syndrome	
Env/Teratogen Induced Anomaly	
Functional Disorder	
Metabolic/Endocrine Disorder	
Multiple Congenital Anomalies	
Neuromuscular disorder	
Reproductive Risk	
SIDS/Infant Death	
Single Gene Disorder	
Single Malformation	
Skel/Connective Tissue Disorder	
Blood Disorder (Hemophilia etc.)	

Chronic Diseases	
Cardiovascular Disease	
Cancer	
Breast Cancer	
Colon Cancer	
Total	0

rev. 07-10

Illinois Department of Public Health

Family Health History Positive Indicator Tracking Form

Revised July 2010

Agency:

Date of Services: (Month/Year)

Program Name	# Clients Screened	# Clients with Positive Indicator	Declined referral with high risk
WIC/FCM			
Family Planning			
APORS			
Other (List Program Area)			
Total	0	0	0

*Please fill in the **LEFT** hand side of the form for the # of clients screened and the # of clients that had a positive indicator. If high risk and declined referral indicate in column D. On the **RIGHT** hand side, indicate **all** the YES responses for positive family history for each client separated into each disease category.

Follow across the form by disorder to indicate the number provided with educational materials for disorder, those who agreed to a referral to geneticist or genetic counselor or had a diagnosed condition.

When you find out that they kept an appointment, please fill in that column even if not in this quarter. This column does not have to match any other numbers.

Note:** at this time these disorders are not referred to a geneticist or genetic counselor. Refer to **guidelines for other chronic disease family history and referrals.

rev.07-10

Information Required for Clients with Positive Indicator				
	Total Number			
Disorders Identified by Newborn Screening	Positive Indicators	Educ Material Provided	Referred to Geneticist/GC	Kept Appt
Amino Acid Disorder (not PKU)				
Biotinidase Deficiency				
Congenital Adrenal Hyperplasia				
Cystic Fibrosis				
Fatty Acid Oxidation Disorder				
Galactosemia				
Hypothyroidism				
Lysosomal Storage Disorders				
Organic Acid Disorder				
Phenylketonuria				
Sickle Cell Disease				
Other Hgb Disease				
Sickle Cell Trait				
Other Hgb Trait				
Urea Cycle Disorder				
NBS Hearing Disorder				
Other Genetic Disorders				
Chromosomal Syndrome				
Env/Teratogen Induced Anomaly				
Functional Disorder				
Metabolic/Endocrine Disorder				
Multiple Congenital Anomalies				
Neuromuscular Disorder				
Reproductive Risk				
SIDS				
Single Gene Disorder				
Single Malformation				
Skel/Connective Tissue Disorder				
Blood disorder (hemophilia, etc.)				
Chronic Diseases				
Asthma*				
Diabetes*				
Cardiovascular Disease				
Cancer				
Breast Cancer				
Colon Cancer				
Other (indicate type)				
Arthritis*				
Total	0	0	0	0

Illinois Department of Public Health
Genetic Services Educational Activities Form

Agency: _____

Reporting Period Dates: _____

Date	Presentations (Please List by Topic)	# Consumer Participants	# Professional Participants
Please total if you are filling out forms by hand.	**Total Participants:**	0	0

Illinois Department of Public Health
Additional Information Form
Genetic Services

Agency:

Reporting Period:

Additional Accomplishments:

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
Office of Health Promotion
REIMBURSEMENT CERTIFICATION / EXPENDITURE FORM

Fiscal Contact Person: _____
Telephone Number: _____
Email Address: _____
Date Submitted: _____

		In the box below , please enter reimbursement amounts submitted for your FY09 grant.					
		$0.00	July	$0.00	October	$0.00	January	$0.00	April
Agency Name:		$0.00	August	$0.00	November	$0.00	February	$0.00	May
FEIN #:		$0.00	September	$0.00	December	$0.00	March	$0.00	June
Contract #:								**$0.00**	YTD
Program Name:		**Billing Period:**							

Name / Vendor	Title / Purpose	Period / Date Incurred	Amount Claimed from IDPH	Componets (specify)
Personal Services				
Fringes				
Travel				
Contractual				
Supplies				
Equipment				
Grand Total			**$0.00**	

Certification: *This signed document hereby certifies the goods and/or services claimed are necessary expenditures for the program, appropriate purchasing procedures have been followed, payment has been made as indicated and a reimbursement has not previously been requested or received.*

Authorized Agency Official **Date**

May be submitted by e-mail to Paula.L.Mitchell@illinois.gov

State of Illinois
DEPARTMENT OF PUBLIC HEALTH
Office of Health Promotion

BUDGET ADJUSTMENT REQUEST

Grantee: _____
TIN: _____
Gant Agreement# _____
Program: _____

The following adjustments to the approved budget are requested in order to better attain the goals and objectives of the program.

NOTE Deadline below

LINE ITEM	CURRENT BUDGET	CHANGE REQUESTED	REVISED BUDGET
Personal Services, including Fringe Benefits			
Contractual Services			
Travel			
Supplies			
Equipment			
TOTAL			

Justification:

_____ _____

Grantee Signature **Date** **Program Administrator** **Date**

_____

Deputy Director **Date**

The last date to submit a budget adjustment request if May 15 in order to receive approval.

State of Illinois
DEPARTMENT OF PUBLIC HEALTH
Office of Health Promotion

Instructions for Completing the
BUDGET ADJUSTMENT REQUEST Form

Revision #: Fill in the revision number for this program within this grant.

Provider/Vendor: Fill in agency name as it appears in the Agreement.

TIN: Fill in the Taxpayer Identification Number as it appears in the Agreement.

Program: Fill in the program name as stated on the Agreement program attachment.

Contract #: Fill in the Department Agreement contract number which is located in the upper right hand corner of page one of the agreement.

Current Budget: Fill in the amounts by line item for EACH line of the current, approved budget for this program. The TOTAL must agree with the amount of the award as originally stated or as previously amended.

Change Requested: Fill in the amount of the requested adjustments for each appropriate line item (decreases are to be shown in parentheses). The total of the Change Requested column will be zero, unless the Budget Adjustment Request is submitted in support of an amendment which increases (or decreases) the award amount, in which case the total will be amount of the increase (or decrease).

Revised Budget: Fill in the adjusted amount for each line item. If there is no change to a line item, fill in the original amount for that line item. The total of the Revised Budget column must agree with the total amount of the award for the program as stated in the original Agreement or the most recent amendment for this program.

Justification: Provide a detailed description/justification for the revisions requested. This justification shall include the programmatic rationale for the change. All adjustments to the equipment line shall be itemized. Attach additional

sheets if needed.

Provider/Vendor:	Signed and dated by an authorized official of the Provider/Vendor.
Submit to:	Grants Manager: Fax: 217-782-1321

The request will be forwarded to the appropriate program for review. After program review, the request will be signed and dated by authorized program staff person indicating Departmental approval or explaining reason(s) for denial and be returned to the Office of Health Promotion. The Office of Health Promotion will enter the approved adjustments into the Department accounting system and return a copy of the Budget Adjustment Request form to the Provider/Vendor.

APPENDIX F. CORNERSTONE INSTRUCTIONS FOR THE FAMILY HEALTH HISTORY (FHH) QUESTIONNAIRE AND REPORTING

Cornerstone screens that apply to the Genetics Program.

ASO1 – FHHQ – Family Health History. This form provides questions which are asked of clients to determine if there are genetic problems within their family history to make clients aware of problems that may be prevented through education and the genetic services available for known problems identified through the questionnaire. Please use the comment section to give explanations of actual family members and their relationship to client for disorders or known problems to help determine relevance of a genetic referral.

HSPR0208 – Expanded Genetic Screening Tool. This is the actual FHH form (a four-page document) that can be printed out for the client's record.

PA39 screen - After the FHH assessment is completed, the genetic coordinator needs to review the assessment, determine if the client has a positive family history of genetic/chronic disease, and mark the individual conditions by checkmark along with checkmark if educational material was provided for the separate tabs of NBS, Genetics and Other Disorders, Chronic Diseases.Under the tab Disposition by Genetic Coordinator, fill in as much information as you can especially Source of Referral, if referral was made (to genetic counselor or geneticist or they are already being seen by those providers). Fill in the appointment kept whenever you get confirmation even if in a different quarter. Fill in the primary diagnosis (or reason for the referral). This will ensure that the client is counted under the built in reporting of Cornerstone for the quarterly reporting form –Expanded Genetic Screening Tool Tracking Form.

PA15 - Genetics Program Code – If the client has been diagnosed with a disorder, not just a positive indicator as having a family member with a history of disorder, then the client should be indicated as a genetic client and the PA15 screen will need to be accomplished. This will ensure that the client will be tracked on the Summary Client Data Form under report selection HSPRO0761, which will assist in the quarterly reporting. *It would be very beneficial and make reports more accurate if you go in and identify all your diagnosed genetic clients with the PA15 code and under PA39 screen indicate the primary diagnosis using only one condition under the appropriate tab.*

HSPR0760 – **Expanded Genetic Screening Tool Tracking Form**. For each FHH completed and PA39 screen filled out appropriately, this will populate this quarterly reporting form.

HSPR0761 – Summary Client Data Form. This form will be populated and can be used as part of the quarterly reporting and will automatically identify genetic clients (with the PA15 code and the PA39 diagnostic code filled in) who have been to the health department for services during the quarter. It also will help the staff know who has a genetic condition. The demographics will be populated automatically.

HSPR0762 – Family Health History. This report will provide the health department with a list of the client screened with the questionnaire by date values.

HSPR0763 – Genetic Services Educational Activities. This is a report that can be filled in indicating the number of educational activities by date, topic and number in attendance separated by consumers and health care professionals. In order to populate this report access it under participants, genetic

services.

HSPR0764 – Genetic Services. This report provides any additional information that you want to share and special counts by counties for clients who are offered information on genetics but do not participate through Cornerstone programming. Access this report same as for HSPR0763.

SVC 936 code – can be used by the health departments for clients served at their genetic clinics.

For clients who are screened and not entered into your Cornerstone system, you will still have to count them separately and add that to your quarterly reports.

Please call the Genetics Program at 217-785-8101 with any questions you may have.