During the course of the Medical Genetics residency training program, each trainee must acquire an understanding of basic genetic principles and familiarity with all of the major sub disciplines of the field. This will include (1) detailed knowledge regarding the basic science of genetics in general and human genetics in particular, (2) knowledge of genetic diseases in humans as well as related diseases which might be genetic or have a genetic component, (3) familiarity with the tests used in evaluation of genetic disorders, and (4) the ability to apply these to the evaluation of patients suspected to have a genetic disease. In addition, trainees should be able to obtain relevant family and medical histories, perform a physical examination, develop a differential diagnosis, select and interpret appropriate laboratory tests, discuss the prognosis of most common genetic disorders, provide clinical management of these disorders, interact with other health-care professionals in the provision of services for patients with genetically influenced disorders and provide accurate medical and genetic counseling to patients and families.

This training will require extensive experience in clinical evaluation of patients, information gathering relevant to the specialty of medical genetics, genetic counseling, and preparing lectures and seminars to many different groups. In addition, trainees must learn how to conduct clinical or laboratory-based research, and write manuscripts and research grants. A broad spectrum of clinical cases, coursework, conferences, and readings will cover general medical genetics, dysmorphology and teratology, prenatal diagnosis and screening, metabolism, cancer genetics, neurogenetics, and metabolic, molecular and cytogenetic laboratory technology.

Duration and Structure of the Overall Program

The joint Medical Genetics residency program sponsored by UC/NU is a 2 year program, which includes 18 months of significant exposure to clinical genetics, as mandated by the Residency Review Committee for Medical Genetics (RRC). The final 6 months may be devoted to either research or clinical electives, including electives in other programs offered at UC or NU. Residents will also attend a continuity genetics clinic once per week during the 6 months of research. The clinical genetics component of the program will consist of rotations on the inpatient consultation service at two different hospitals, in a variety of general and subspecialty genetics outpatient clinics at both institutions, and in each of the appropriate genetic testing laboratories. While the program will include enough flexibility to accommodate individual interests, a typical residency will be weighted toward general clinical genetics and research (Table 1). For example, residents interested in a career in Prenatal Genetics will be allowed to increase their time on this rotation to a maximum of 3 months, and continue to attend prenatal genetics clinic as their continuity clinic during their research months. The research/elective component will usually be taken as a block in the second year. All residents must keep a detailed patient log that includes all the information required by the ABMG in their application for certification.

Table 1. Structure of the standard UC/NU Medical Genetics residency program

Year 1	7 months	Clinical genetics (UC)
	2 months	Prenatal genetics (UC or NU)
	0.5 month	Biochemical laboratory genetics (NU)
	1.5 months	Clinical cytogenetics and laboratory molecular genetics (UC)
	1 month	Cancer cytogenetics and clinical cancer genetics (UC)
Year 2	6 months	Clinical genetics (NU)
	6 months	Research experience or clinical electives (UC or NU)

UC, The University of Chicago; NU, Northwestern University and Children's Memorial Hospital

Each trainee will be also expected to attend the Core Lecture Series in Clinical Genetics once per week when offered, complete one graduate level course in Human Genetics, and attend at least 80% of the weekly or monthly clinical and research conferences sponsored by the Department of Human Genetics. We have developed two coursework options for trainees to learn basic and human genetics, as described in the Basic Medical Education section of this application. Each trainee will also be expected to complete a basic science or clinical research project under the supervision of a faculty member that would be suitable for publication, and attend at least one national genetics meeting per year, such as the ASHG or ACMG annual meetings.

We will encourage all residents interested in an academic career to complete at least one year of additional training completely devoted to research. During this time, we will encourage the trainee to attend Clinical Genetics Conferences and Genetics Grand Rounds, and to hold a weekly half-day general genetics clinic. Funding for the third and subsequent research years will be through individual or training grants if available, or other funding sources arranged prior to the year. Individuals wishing to sit for the American Board of Medical Genetics certification exam in a clinical laboratory specialty in addition to the Clinical Genetics exam must take an additional one or two years of training to do so, for which they must be accepted into the program in advance.

During the course of the Clinical Rotations the resident will be expected to investigate the current literature and texts regarding specific genetic conditions when preparing for patient contact in clinic and/or the consult service. This investigation should specifically focus on the current standard of practice for the evaluation, diagnosis and management of these conditions. The resident will be expected to present one of these cases a month at case conference reviewing the standard of practice. In addition, for these case presentations, the resident will perform a cost analysis of the evaluation and management plan to gain a better understanding of the cost benefit analysis in patient care. This activity is designed to facilitate training in the core competencies **Practice Based Learning and Improvement** and **Systems-Based Practice**.

Clinical Genetics Rotations

Rotations in general clinical genetics, prenatal genetics, and several subspecialty clinical genetics areas will be available at both UC and NU, with the subspecialties

differing between the two institutions, thus offering a wide exposure to trainees. Trainees with an interest in prenatal genetics will be able to take up to 3 months on this rotation, plus any time they choose from their elective months. The required laboratory rotations will be located where teaching is most readily available. The complementary nature of lab strengths was one of the most compelling considerations in establishing a joint program.

Residents will usually spend 3 months on this rotation during the first year, and 3 months during the second year. However, this may be reduced by 1-2 months to accommodate trainees interested in prenatal genetics.

Goals: During these rotations, trainees will learn issues related to **Patient Care** specifically to obtain family and medical genetic histories, perform physical exams with specific emphasis on dysmorphology, appropriate use of and interpretation of specific genetic tests, diagnose genetic conditions, and manage the medical complications of these disorders. They will increase their **Medical Knowledge** through obtaining familiarity with the resources for these disorders, including current text books, journals, and scientific literature. They will learn **Practice-Based Learning and Improvement** by using information technology such as computer databases, internet services, and family support group web sites. They will develop **Interpersonal and Communication Skills** necessary for genetic counseling and provide genetic counseling regarding genetic diagnosis to patients and their families. They will be expected to communicate with other health care professionals by direct contact and through preparation of consultation notes regarding the medical and psychosocial implications of genetic diagnosis and the tests used to arrive at these diagnoses. They will be expected to develop **Professionalism** skills by timely medical reports, communication with relevant health care professional and demonstrating a commitment to ethical principles, confidentiality, compassion, sensitivity and integrity in all patient care activities. This will be accomplished by participation in general and specialty genetics clinics, and the inpatient consult service. In order to learn issues related to **Systems Based Practice**, trainees will gain an understanding of the evaluation, diagnosis, management and natural history of specific genetic disorders, and interact with multi-specialty teams through participation in specific subspecialty genetic clinics. In addition, the resident will be expected to play an active role in patient care and develop an understanding of the different health care provider systems with regards to pursuing pre-approval of genetic tests and services.

Rotation 1. Clinical Genetics at The University of Chicago

Residents will usually spend 3 months on this rotation during the first year, and 3 months during the second year. However, this may be reduced by 1-2 months to accommodate trainees interested in prenatal genetics.

Objectives:

1. Participate in the evaluation of a wide spectrum of general genetic and specialty referrals
2. Develop skills in obtaining and analyzing pedigrees
3. Use physical exam findings and literature review to make diagnoses of specific genetic disorders
4. Gain an appropriate understanding of laboratory testing and interpretation of results to assist in establishing a diagnosis
5. Perform genetic counseling regarding specific conditions
6. Develop teaching skills by participating in resident education and teaching conferences

The Inpatient Consult Service (UC)
The resident will participate in the consult service while on clinical genetics blocks at The University of Chicago. The resident will be primarily responsible for all requests for genetic consultation from all services including the Departments of Pediatrics and Medicine. The resident will carry the consult beeper 24 hours a day and requests for consults will either come through the genetics offices or to the resident directly. Few consults are requested on weekends, and the resident will be expected to carry the consult beeper for two weekends of each month. The resident will be responsible for answering questions and seeing consults in a reasonable time. All duties will be under the supervision of the attending physician.

Requirements:

1. Obtain relevant information from the service requesting the consult, identifying pertinent medical history and the nature of the question being asked of the genetics service.
2. Review the history, and obtain and review records and laboratory testing.
3. Interview the patient/family to assess their understanding of the genetic questions and medical issues involved, obtain further history and construct a detailed family history.
4. Perform a thorough physical exam with attention to the dysmorphology exam where appropriate.
5. Write a detailed consultation note reviewing each of the above points and create a detailed assessment of the patient including any pertinent literature. Formulation and communication of the assessment and plan to the requesting team should be done only after review with the attending physician. The attending physician will review the history, examine all patients, review exam findings with the resident and participate in formulation of the plan.
6. Communicate the findings and plan to the parents, initially under the supervision of the attending physician, and to the house-staff.
7. Follow-up on results of tests and studies, gather additional medical information needed and communicate any additional, pertinent information to the attending physician on the daily rounds.
8. Participate in education of the medical residents in regards to the consults requested.

9. Present patients with interesting exam or laboratory findings at the weekly Clinical Genetics and Laboratory Case Conference and briefly review medical literature and scientific advances specific to the case presented.

Staff: Darrel J. Waggoner, MD and William B. Dobyns, MD
GC: Rebecca Brown, MS, CGC

Outpatient Genetics Clinics (UC)

Outpatient Genetics Clinics are held at least 3 days per week, and include both general and specialty clinics. While on the Clinical Genetics rotation at UC, residents will be expected to attend all of these clinics, with some preparation ahead of time expected. General genetics clinics are held on Tuesday and Friday mornings from 8 AM to 12 PM at the DCAM outpatient center at The University of Chicago. All activities will be done under the direct supervision of the attending physician.

Requirements for Genetics Clinics:

1. Residents are expected to see a minimum of 2 patients at each clinic.
2. Review medical records, testing and pertinent history prior to the scheduled patient visit.
3. Develop a preliminary differential diagnosis based on information obtained, which will direct subsequent reading and literature review.
4. Interview patients and families to obtain any additional history needed, obtain family history and perform a physical exam.
5. Review patients with the attending physician.
6. Develop a differential diagnosis and management plan, and participate in family counseling.
7. Dictate a note to the referring physician reviewing the history, exam findings, investigations ordered or recommended, and the diagnosis when known.
8. Communicate results of all tests or additional information obtained to the attending physician, referring physician, and patient or family.

Staff: Darrel J. Waggoner, MD and William B. Dobyns, MD
GC: Rebecca Brown, MS, CGC ; Melissa Dempsey, MS, CGC; Rachelle Lorenz, MS, CGC

Specialty Genetics Clinics

Several specialty genetics clinics are conducted on a weekly or monthly basis. Attending these clinics will help trainees develop a knowledge base regarding the evaluation and treatment of specific genetic disorders. This, in turn, will help them understand the natural history, treatment options and genetic recurrence risks associated with these specific genetic conditions, become comfortable participating in multispecialty clinics, and gain exposure to patients with a broad variety of health concerns in all medical practices including both adults and children. The general requirements are the same as for general

genetics clinics, but in addition the residents will be expected to attend any conferences specific for the specialized clinic and participate in the presentation of any patients at these conferences.

Craniofacial Clinic

Craniofacial clinics are held on the first and third Thursday of each month from 9 AM to 12 PM. In addition to expectations for all Genetics Clinics, residents will be expected to read directed materials as determined by the attending physician, and attend the post-clinic conference held directly after each clinic.

Staff: Darrel J. Waggoner, MD

Neurogenetics Clinic

Neurogenetics clinics are held once or twice a month (depending on number of patients) on Thursday afternoons from 1 PM to 5 PM. Residents will read directed materials as determined by the attending physician, develop an understanding of neurogenetics and gain experience in working with the Neurosurgery team in joint patient evaluations, and become familiar with developmental brain abnormalities, neurodegenerative diseases, and related conditions.

Staff: Darrel J. Waggoner, MD, and William B. Dobyns, MD

Metabolic Clinic

One clinic per month is designated as a Metabolic Clinic. However, patients with inborn errors of metabolism are seen during General Genetics Clinics as well, providing that the dietician is available to attend, which is usually the case. Residents will attend clinic and evaluate patients with known or suspected metabolic disorders. During this clinic, they will learn to interpret biochemical laboratory tests, analyze growth patterns and specialized diets, and become familiar with the state screening program. They will also answer questions from residents and community physicians regarding follow-up of abnormal results.

Staff: Darrel J. Waggoner, MD

Cancer Risk Clinic

Trainees will be rotate through the large Cancer Risk Clinic at UC during their Cancer Cytogenetics and Molecular Genetics Rotation (Rotation 6).

Rotation 2: Clinical Genetics at Northwestern University

Residents will usually spend 4 months on this rotation during the first year, and 3 months during the second year. However, this may be reduced by 1-2 months to accommodate

trainees interested in prenatal genetics. The goals and objectives for this rotation are the same as listed for Clinical Genetics at UC (Rotation 1). The outpatient experience will include both general and specialty clinics. Most of the specialty genetics clinics differ from those available at UC, and several of them are multi-specialty clinics attended by one or more pediatric specialists in other areas. In all of these clinics, residents will learn the skills needed for medical management of these disorders, in addition to diagnosis.

Genetics Inpatient and Consult Service (CMH)

The resident will be assigned to the inpatient Genetics Service while on Clinical Genetics rotation at Children's Memorial Hospital. The resident will be primarily responsible for all requests for genetic consultation, and should be available by beeper 24 hours a day. This service does admit patients directly, usually patients with inborn errors of metabolism. Few consults are requested on weekends, and the resident will be expected to carry the consult beeper for two weekends of each month. The resident will be responsible for answering questions and seeing consults in a reasonable time. All responsibilities will be under the supervision of the attending physician on call.

Staff: Barbara Burton, MD, Brad Angle, MD, and Joel Charrow, MD
GC: Katherine Kim, MS, CGC; Karen Niedermeyer, MS, CGC; Andrea Paras, MS, GCC; and Danielle Hartung, MS

General Genetics Clinic

General Genetics Clinic is held on Tuesday and Wednesday afternoons and Friday morning, with additional satellite Genetics Clinics held at other times twice per month. The volume of patients is large, and includes some patients who would also be eligible to attend a specialty clinic, such as patients with metabolic diseases.

Staff: Barbara Burton, MD; Brad Angle, MD, and Joel Charrow, MD
GC: Karen Niedermeyer, MS, CGC; Andrea Paras, MS, GCC, Dinele Pond, MS

Jewish Genetics Diseases Clinic

This clinic has developed based on the expertise of Dr. Charrow in this area, particularly in the management of Gaucher disease. It is held on Tuesday afternoons twice per month, although the number of patients is large so they are also seen at other times. In addition, most patients with Gaucher disease come into the Day Hospital every other week for intravenous infusion of enzyme replacement therapy.

Staff: Joel Charrow, MD
GC: Laura A. Niewadomski, MS

Neurofibromatosis Clinic

This clinic specializes in caring for patients with neurofibromatosis, most of whom have

NF type 1. Both of the medical staff have extensive clinical and research experience with this disease. The residents will learn the natural history of NF1, which will allow them to take a directed history and order appropriate lab studies searching for known complications of this protean disease. It is held weekly on Thursday morning. This is a multi-specialty clinic, which is also attended by Pediatric Ophthalmology.

Staff: Joel Charrow, MD; Robert Listernick, MD (pediatrician)
GC: Katherine Kim, MS, CGC; Dinele Pond, MS

Marfan Syndrome Clinic

This multi-specialty clinic focuses on evaluation of patients with Marfan syndrome, and is also attended by a pediatric cardiologist. This clinic also serves patients with other connective tissue dysplasias such as Ehlers-Danlos syndrome and contractural arachnodactyly. It is held on once per month on Friday morning

Staff: Barbara Burton, MD; Rae-Ellen Kavey, MD (Pediatric Cardiology), Marla Mendelson, MD (adult Cardiology)
GC: Karen Niedermeyer, MS, CGC

Neurogenetics Clinic

Neurogenetics clinic is held on Monday afternoons once per month in conjuction with Pediatric Neurology. Patients seen in this clinic have a wide variety of neurologic diseases, such as tuberous sclerosis, complex neurodegenerative diseases and malformations.

Staff: Barbara Burton, MD; Charles Swisher, MD (Pediatric Neurology)
GC: Danielle Hartung, MS

Phenylketonuria Clinic

This busy clinic is held weekly on Wednesday afternoons with a dietician and social worker present. In this clinic, residents will learn basic aspects of dietary management of inborn errors of metabolism, and detailed dietary management of PKU. In addition the standard medical history, residents will learn to assess dietary history, develop a dietary plan in conjunction with nutrition, interpret lab values relevant to PKU, assess psychosocial conditions which may interfere with dietary compliance, and assess the need for intervention.

Staff: Barbara Burton, MD
GC: Danielle Hartung, MS
Dietician: Rhoda Papanastassiou, R.D.
Social Worker: Hazel Vespa, MSW

Skeletal Dysplasia Clinic

This multi-specialty clinic is held once per month with pediatric orthopedics and radiology

also attending. All of the staff, specifically including the radiologist, have extensive experience with these disorders. The resident will learn to interpret bone X-rays to assist in making the correct clinical diagnosis, and learn the complex management skills required to care for individuals with these disorders.

Staff: Joel Charrow, MD; John Sarwark, MD (Pediatric Orthopedics), Andrew Poznanski, MD (Radiology)
GC: Katherine Kim, MS, CGC

Rotation 3: Prenatal Genetics at The University of Chicago

Residents will usually spend two months on this rotation or Rotation 4 during the first year of the program. However, trainees interested in prenatal genetics will have the option of spending up to 4 months on prenatal genetics rotations during their 18 months of required clinical genetics experience, and may spend additional elective time on these rotations as well.

Prenatal genetics clinics are held at the DCAM outpatient center at The University of Chicago on three half days per week. Residents are expected to attend all of these clinics. All activities will be done under the direct supervision of the attending physician and genetic counselor.

Objectives:

1. Recognize genetic and non-genetic disorders that present during fetal life.
2. Develop an understanding of prenatal testing and screening procedures.
3. Become familiar with genetic counseling prior to pregnancy, during pregnancies complicated by risk factors such as maternal age or teratogen exposure, during pregnancies complicated by known fetal anomalies, and following pregnancy loss.

Requirements for Prenatal Clinic:

1. Attend clinic on Monday afternoon, and Tuesday and Thursday mornings while on Prenatal Genetics rotation. Residents are expected to see 2 patients at each clinic.
2. Participate in preconception counseling, pregnancy loss counseling, and counseling for teratogen exposures.
3. Participate in evaluation of and counseling for prenatal genetic disorders.
4. Review medical records, testing and pertinent history prior to the scheduled patient visit, interview patients and families to obtain family history and any relevant medical history, and review patients with the attending physician.
5. Develop a differential diagnosis and management plan.
6. Dictate a note to the referring physician reviewing the history, risk factors, tests ordered or recommended, and the diagnosis when known.

7. Communicate results of all tests or additional information obtained to the attending physician, referring physician, and patient.

Staff: Marion Verp, MD
GC: Elyse Muller, MS

GOALS, OBJECTIVES AND STRUCTURE OF LABORATORY GENETICS ROTATIONS

Residents will spend 3 months during the first year rotating through the laboratory genetics specialties. Some of these will be combined with related clinical genetics experience. For each lab, trainees should become familiar with the indications for testing, methods used, standard terminology and any special nomenclature systems, content of reports, and time required to complete testing. They are specifically not expected to become proficient in performing these tests on their own during these rotations.

Rotation 5: Clinical Cytogenetics and Molecular Genetics Laboratories

This rotation will expose residents to the standard cytogenetic and molecular genetic techniques used for clinical diagnosis of most genetic disorders, especially congenital disorders. The goals and objectives of this rotation are as follows: During this rotation, trainees are expected to gain an understanding of **Patient Care and Medical Knowledge** in regards to genetic laboratory testing. They will learn gain an appreciation for the techniques, capabilities, limitations and applications of genetic testing performed in the clinical cytogenetics and clinical molecular genetics laboratories. This rotation will provide understanding of the latest testing procedures and the scientific basis of their usefulness in clinical diagnostics. The resident will be expected to develop **Practice Based Learning and Improvement** skills by gaining an appreciation of the use of electronic databases and resources used in developing and interpreting laboratory testing. In addition, the goals of **Professionalism and Interpersonal Skills and Communication** will be addressed as the resident will learn to recognize and appreciate the working relationship that exists between the clinical geneticist and the laboratories and gain experience in communication genetic test results to other. Finally, the resident is expected to develop **Systems-Based Practice** skills by developing an understanding of the cost-benefit ratio with regards to genetic tests for specific diseases as well as in screening programs.

The Clinical Cytogenetics and Molecular Genetics Laboratories at The University of Chicago are located in the same area, share the cell culture and DNA isolation facility, and hold joint conferences. This will therefore be scheduled as a single 1.5-month rotation. Trainees will focus on clinical cytogenetics including both amniocytes for prenatal diagnosis and lymphocytes or fibroblasts for congenital disorders during the first 3 weeks, and on clinical molecular genetics during the last 3 weeks.

Requirements during the entire rotation:

1. Attend weekly Clinical Case Conference - Tuesday @ 2:30 pm MARP 300
2. Present a 20-30 minute seminar on a topic related to clinical cytogenetics or molecular genetics on the last Tuesday of rotation at the 2:30 pm case conference; the topic must be pre-approved by the cytogenetics or molecular laboratory director at least two weeks in advance.
3. Be prepared to discuss abnormal laboratory results (cytogenetic and molecular) at the Tuesday Clinical Case Conference (i.e., research the abnormality using appropriate databases and literature, and have photocopies of pertinent articles available for laboratory directors, clinicians and counselors).
4. Attend weekly Cytogenetics Laboratory Technologists Meeting.
5. Attend Wednesday bi-weekly journal club or cancer pedigree review - MARP300.

Rotation 5A: Clinical Cytogenetics

The first half of the rotation will focus on clinical cytogenetics. Trainees will learn to establish cultures of several different types of tissue, recognize each of the human chromosomes under the microscope, arrange karyotypes, recognize abnormalities especially common numerical and structural abnormalities, and describe many different types of abnormalities using correct nomenclature. This part of the rotation will have several different sections, based on the procedures used.

Objectives:

1. Participate in cytogenetic laboratory activities, and become familiar with procedures used in routine and specialized cytogenetics including fluorescence *in situ* hybridization (FISH).
2. Learn how to interpret results of tests performed in the lab.
3. Learn what tests are available on a clinical and research basis.

Requirements:

1. Blood section. Trainees will set up, harvest, and analyze their own blood sample.
2. Prenatal section. Prepare and analyze amniotic fluid and chorionic villi samples under the direction of one of the cytogenetic technologists.
3. FISH (molecular cytogenetics) section. Prepare and analyze patient samples by FISH.
4. Tissue section. Prepare and analyze chromosomes from tissue samples such as skin biopsies and products of conception.

Staff: Stuart Schwartz, PhD

Rotation 5B: Molecular genetics

The last half of this rotation will focus on clinical molecular genetics. Trainees will learn to isolate DNA, perform several relevant laboratory procedures such as PCR, Southern blot, sequencing and genotyping, analyze and report results.

Objectives:

1. Participate in molecular genetics laboratory activities and become familiar with procedures used such as southern blotting, PCR, sequencing, and genotyping.
2. Learn how to interpret results of tests performed in the lab.
3. Learn what tests are available on a clinical and research basis.

Requirements:

1. Perform DNA extraction.
2. Perform several routine PCR-based tests, such as tests for uniparental disomy, and methylation testing for Prader-Willi and Angelman syndromes.
3. Perform specialized testing such as genotyping, DNA sequencing, and direct mutation analysis.

Staff: Soma Das, PhD; and Carole Ober, PhD

Rotation 6: Cancer Genetics and Cytogenetics

This rotation will expose residents to the specialized genetics knowledge needed for the clinical and laboratory evaluation of patients with cancer, both hereditary and sporadic. The clinical and laboratory components will be closely integrated throughout the rotation to allow maximum exposure.

During the first 3 weeks, trainees will be assigned primarily to the Cancer Cytogenetics Laboratory, but will also attend the Cancer Genetics Clinic once per week and several Cancer Genetics conferences. During the fourth week, residents will be assigned full time to the Cancer Genetics program, and will attend two Cancer Risk Clinics staffed by Genetic Counselors, the Cancer Genetics Clinic, and several related conferences.

Rotation 6A: Cancer Cytogenetics

The Cancer Cytogenetics Laboratory at The University of Chicago is located in a different area from Clinical Cytogenetics, and analyzes both hematologic and solid tumor samples. The lab coordinates many didactic sessions with the Cancer Genetics Clinic. During this part of the rotation, residents will gain an understanding of the techniques, capabilities, limitations and applications of cytogenetic testing performed for the diagnosis and subclassification of human tumors. They will already have rotated through the regular Cytogenetics Laboratory, and so will be familiar with most of the techniques used. They will learn the procedures involved in the specialized techniques of fluorescence in situ hybridization (FISH) and spectral karyotyping of cancer cells, and learn what tests are available on a clinical and research basis.

Requirements:

1. Leukemia and Lymphoma. Trainees will participate in culture initiation and harvest, slide preparation of bone marrow, peripheral blood, lymph node, and tumor mass specimens, and analyze metaphase cells from tumor specimens.
2. Solid Tumor section. Trainees will participate in initiating cultures, and analyze metaphase cells from tumor specimens.
3. FISH and SKY (Molecular Cytogenetics) section. Trainees will participate in hybridization of probes to detect recurring chromosomal abnormalities and analysis of hybridized slides.
4. Attend twice weekly Clinical Cancer Cytogenetics Case Review Conference – Tuesday and Friday at 10:30 am, Room I303.
5. Attend bi-weekly case review conference with cytogeneticists, medical oncologists and hematopathologists– Thursday at 10:00 am, Room E215.
6. Attend monthly Hematopathology Conference with pediatric and medical oncologists, molecular geneticists, cytogeneticists, and hematopathologists – Thursday at 9:00 am, Room TW040.
7. Attend weekly Pediatric Tumor Board - Wednesday at 3:30 pm, Room C400.
8. Attend weekly research conference on cytogenetics, molecular cytogenetics, and molecular analysis of human tumors- Wednesday at 12:30 pm, Room E215.
9. Present a 20-30 minute seminar on topic related to cancer cytogenetics during the last week of rotation at Clinical Cancer Cytogenetics Case Review Conference. The topic must be pre-approved by laboratory director at least 1 week in advance.
10. Be prepared to discuss abnormal laboratory results at the Tuesday and Friday Cancer Cytogenetics Case Review Conference, and Thursday bi-weekly case review conference (i.e., research the abnormality using appropriate databases, literature, and photocopy pertinent articles for laboratory directors and clinicians, and pathologists).

Staff: Michelle Le Beau, PhD; and Stuart Schwartz, PhD

Rotation 6B: Clinical Cancer Genetics

The Cancer Genetics Service evaluates patients with a variety of solid tumors that may be familial. Residents will attend the Cancer Genetics Clinic and relevant clinical conferences each week during the entire rotation. In clinic, residents will conduct a thorough physical examination on each patient, learn to obtain family histories relevant to hereditary forms of cancer, recognize the familial forms of cancer, and provide genetic counseling fulfilling the core competency **Patient Care.** They will have additional exposure during the last week when they will attend all clinics and perform inpatient consults with the Cancer Program genetic counselors and attending physician.

This exposure of **Medical Knowledge** will allow them to learn and identify the genes for which genetic testing is now standard of care and incorporate such testing into patient

care. They will also learn the process of genetic testing for colon, breast and ovarian cancer, and become familiar with the psychosocial, legal, ethical and reimbursement issues related to cancer genetics fulfilling the competency **Systems Based Practice**. This rotation will allow them to gain an understanding of medical management and treatment options for individuals at increased risk for cancer, and learn to effectively incorporate this into genetics practice fulfilling the competency **Practice-Based Learning and Improvement**. This will also highlight the relationship between cancer genetics and the development of cancer. They will attain skill, experience in diagnosis, counseling and management of hereditary cancer syndromes in adults, and obtain familiarity with the journals, books, computer databases, and other resources concerning those conditions.

Requirements for Cancer Risk Clinic and Genetic Counseling Service:

1. Attend specialized cancer clinic held each Friday morning.
2. Attend risk assessment and genetic counseling consultations on Monday and Wednesday mornings during the fourth week of the rotation.
3. Perform inpatient consults on cancer patients during the fourth week.
4. Interact with the physician requesting the consult to precisely define what question is being asked of the cancer genetic service.
5. Present and discuss one or two patients each week at the weekly Cancer Risk Clinic Program meeting.
6. Read selected materials and conduct literature searches on the disorders seen in clinic in order to accumulate a base of reference material.

Staff: Olufunmilayo Olopade, MD (oncologist); and Marion Verp, MD
GC: Shelly Cummings, MS; Melody White, MS, CGC

Rotation 7: Biochemical Genetics Laboratory and Metabolic Clinic (NU)

The Biochemical Genetics Laboratory at Children's Memorial Medical Center, affiliated with Northwestern University, is one of only two such laboratories located in the greater Chicago area and provides diagnostic testing to a large population. During this rotation, trainees will gain experience with the diagnosis, laboratory interpretation, management, and genetic counseling of patients with various metabolic abnormalities (**Patient Care and Medical Knowledge**). They will be expected to attend Metabolic Clinic during this rotation, and during clinical genetics rotations in both years.

During this two week block and during all subsequent Metabolic Clinics, residents will participate in the evaluation and management of biochemical genetic disorders in both outpatients and inpatients, including some adults. They will gain experience with the state screening program (**Systems Based Practice)** and learn to manage patients with inborn errors of metabolism, including phenylketonuria, galactosemia, biotinidase deficiencies and Gaucher disease, work closely with metabolic dieticians to develop an understanding of the dietary management of patients with metabolic disorders, and learn to interpret laboratory testing including the evaluation of amino acid and organic acid analysis, and become familiar with various enzymatic analysis.

Requirements:

1. Observe biochemical laboratory testing
2. Participate in the weekly laboratory meeting interpreting and reviewing test results for amino acid and organic acid analyses
3. Participate in metabolic clinics, including PKU clinic at Children's Memorial Medical Center and in the clinical and laboratory evaluation of new patients at both Children's Memorial and The University of Chicago (and metabolic patients in clinics on T pm and F am (about half of all patients in clinic on those days). Day hospital for Gaucher, occasional inpatient with disorder.
4. PKU meeting Friday pm
5. Meet with dietician for introduction x 1-2 hours during this two week block.
6. Review the family, medical and dietary histories of patients with known and suspected biochemical disorders
7. Participate in medical and dietary interventions for metabolic disorders
8. Communicate results of testing and dietary interventions to families and physicians

Staff: Joel Charrow, MD, Brad Angel, M.D. and Barbara Burton, MD
Dieticians: Rhoda Papanastassiou, R.D.

SCHEDULE OF CONFERENCES AND OTHER DIDACTIC SESSIONS

During all rotations at UC, trainees will be expected to attend the Clinical and Laboratory Genetics Case Conference held on Tuesday afternoon, the Clinical Genetics Specialties Conference or Journal Club held on Wednesday afternoons, and the Human Genetics Seminar series on selected Thursday afternoons. During rotations at NU, trainees will be expected to attend the Thursday afternoon Clinical Case Review from 1:30 to 3:00 pm, when all patients from the previous week are briefly reviewed, and the Genetics Lab conference from 3:00 to 4:00 pm when all abnormal lab findings from the previous week are reviewed. This time is also used to review patients expected during the coming week. The PKU team at CMH meets Friday afternoon for about 1 hour. While circumstances always dictate that some conferences will be missed, we will require 80% attendance by each trainee.

SUPERVISION AND PROGRESSING RESPONSIBILITIES OF RESIDENTS

A faculty member will supervise the residents at all times during their training program. During outpatient clinics, they will be expected to review charts ahead of clinic, and conduct the initial evaluation of at least two patients or families per half-day clinic. Each patient will be reviewed with and examined by the attending physician. The resident will be responsible for the dictations and follow-up. All inpatient consults will be handled the same way, with the initial evaluation conducted by the resident and then reviewed with the attending physician. Rare consults during the evenings and weekends will be reviewed with attending physicians by phone, and then may be staffed the following day or in the outpatient clinic within the next week when the patients are not acutely ill.

However, most evening and weekend consults to Medical Genetics are urgent, often with known or suspected inborn errors of metabolism, and will require immediate evaluation by the attending physician. Similarly, the laboratory directors will review all laboratory studies interpreted by the residents.

As residents gain experience, they will be encouraged to conduct more and more of the patient and family counseling themselves, initially with the attending physician present and later on their own. However, current federal billing guidelines require the presence and significant involvement by the attending physician both in clinic and on the hospital wards. This has the potential of diminishing resident responsibility, but no alternatives currently exist. We will mitigate this factor for inpatient consults by encouraging them to pre-round on all patients.

As residents become more senior and their skills in counseling improve the residents will be allowed to provide genetic counseling to families without the direct supervision of a counselor or attending physician. The session will be reviewed before and afterward to assist in preparation and to assess the adequacy of the counseling sessions.

TEACHING AND RESEARCH

Residents will have the opportunity to participate in teaching activities in multiple ways. For hospital consultations they will be responsible for communicating the findings of the team and recommendations to the house staff officers and medical students directly involved with the patient in question. As part of this communication the resident will be able to incorporate teaching basic genetic principles, medical information regarding specific genetic diseases and medical management issues. As the genetics resident progresses through the program they may share a larger responsibility for teaching any medical students or other residents rotating on the genetics service. In addition, the second year genetics residents will participate in the teaching of the first year medical school genetics class by leading small group sessions.

Residents will have the opportunity in the second year to spend up to six months on a research project. The research projects may include both clinical and basic sciences research experiences. Residents interested in pursuing further basic sciences research training may spend additional years in a mentored research environment depending on the availability of funding by the individual faculty mentoring the research training.