

Elective Description:

Primary Objectives:

Skills in diagnosis of genetic disorders:

Congenital malformations and mental retardation

Recognition and assessment of multiple congenital anomalies, with or without mental retardation

Effects of major teratogens and mechanisms of teratogenesis

Clinical diagnosis based on physical exam, history, and literature review

Use of cytogenetic & molecular genetic tests to diagnosis congenital malformations and mental retardation

Use of literature to consider single gene disorders

Predictive screening for genetic disease

Principles of newborn screening; principles of follow up for abnormal tests

Principles of carrier screening for single gene disorders based on ethnic background

Principles of screening for disease pre-disposition

Issues in advising patients and parents about the risks and benefits of screening

Recognition of single gene disorders causing specific phenotypes

Skeletal dysplasias

Distinctive dysmorphology or physical findings (e.g. NF, facial dysmorphic syndromes)

Major organ abnormalities (renal, cardiac, gi, pulmonary, cns)

Use of the physical examination, history, imaging studies, literature and molecular testing for diagnosis

Disorders of intermediary metabolism

Recognition of cardinal signs and symptoms of inborn errors

Principles of acute management of metabolic emergencies

Formulation of a differential diagnosis

Use of the laboratory, history, and physical exam in diagnosis of inborn errors

Principles of long term management of inborn errors

Genes and disease: mechanisms of inheritance

Genes and cancer

Unusual mechanisms of gene action (trinucleotide repeats, imprinting)

Common disorders, e.g. diabetes

Skills in management of genetic disease

Treatment of genetic disease

Environmental management

Replacement of missing protein or gene product

Gene therapy

Use of genetic information

How novel scientific discoveries can be evaluated in a clinical context

How to obtain a useful genetic history

Principles of communicating genetic information to parents and families

Cultural and social issues in genetic information

Problems in confidentiality

Delivery of bad news

Use of genetic consultants

Faculty Contact:

First name	Last name	Email
Hui	Zhang, MD	h.zhang@yale.edu

Staff contact (M-F, 8:30am to 5:00pm):

First name	Last name	Title or role	Email	Phone	Location/Other
Irene	White	Sr. Administrative Assistant, Genetics Consultation Service	irene.white@yale.edu	203-785-2660 (option # 1)	WWW 313 – check in 1 st day at 8:30am with Irene White.

IMPORTANT INFORMATION!

Dr. Hui Zhang asks that every resident scheduled for the Genetics elective **go to WWW 313 or call Irene White at 203-785-2660 (option # 1) two (2) weeks PRIOR to your start date.** Your 1st day you'll report to Irene White in WWW 313 at 8:30 a.m. We do not want you to call the fellow through the YNHH Page Operator.

All Yale Affiliated Hospitals Program (YAHP) Residents ARE REQUIRED to **check in at the YNHH Housestaff Office** (Enter 330 Cedar Street, 2nd Floor, Tompkins 209) **the morning of their 1st day** of this elective rotation. The YNHH Housestaff Office will provide YAHP residents information about obtaining a YNHH temporary ID and parking. You may call for the YNHH Housestaff Office at 203-688-2259 for office hours.

Will Affiliated Hospitals' residents be required to take EPIC Training in order to participate in this elective?

Yes. Residents from Yale Affiliated Hospitals: **At least 1 month PRIOR** to starting your Genetics Elective Rotation at YNHH you will need to **make arrangements for EPIC computer Training.** Your home program coordinator will provide you with the paper work required to initiate the EPIC Computer Training. For additional information, please contact Tracy Crosby at tracy.crosby@yale.edu, the contact for Yale Affiliated Hospital Residents, for additional assistance regarding your elective, and/or EPIC Computer Training.

Maximum number of residents accepted at a given time:

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2

PGY Levels accepted from Yale Internal Medicine Residency Programs:

PGY1, PGY2, PGY3, PGY4

PGY Levels Accepted from Yale Affiliated Hospitals Internal Medicine Programs:

Yale Affiliated Hospitals are: Bridgeport Hospital, Danbury Hospital, Greenwich Hospital, Griffin Hospital, Norwalk Hospital, St Mary's Hospital and Waterbury Hospital.

PGY2, PGY3. YAHP Interns may not be scheduled prior to March 1st of their intern year for electives at YNHH or VA.

Which Blocks will you be CLOSED for scheduling?

We do not want any medicine residents scheduled for the Genetics Elective during Blocks 7B and 8A.