## 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:

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<tr>
<th>First name</th>
<th>Last name</th>
<th>Email</th>
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<tbody>
<tr>
<td>Hui</td>
<td>Zhang, MD</td>
<td><a href="mailto:h.zhang@yale.edu">h.zhang@yale.edu</a></td>
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 Staff contact (M-F, 8:30am to 5:00pm):

<table>
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<tr>
<th>First name</th>
<th>Last name</th>
<th>Title or role</th>
<th>Email</th>
<th>Phone</th>
<th>Location/Other</th>
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<tbody>
<tr>
<td>Irene</td>
<td>White</td>
<td>Sr. Administrative Assistant, Genetics Consultation Service</td>
<td><a href="mailto:irene.white@yale.edu">irene.white@yale.edu</a></td>
<td>203-785-2660 (option # 1)</td>
<td>WWW 313 – check in 1st day at 8:30am with Irene White.</td>
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 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 (YAH) 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 YAH 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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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.