

BASIC INFORMATION

Duration of the elective: 4 weeks /one month

Number of students: 2

Passing grade on PEDI 403; thorough knowledge of suggested readings of PEDI 403

Location(s): Outpatient clinics (Pediatrics room 24, Gynecology & Obstetrics room 7), consultations (NICU, Pediatrics, Oncology), Genetic Diseases Diagnostic Center (M-6th floor) and Zebrafish facility (KUTTAM; 4th floor)

CONTACT INFORMATION

Elective Director : Prof. Hülya Kayserili, MD, PhD

Elective Faculty : Asst. Prof. Umut Altunoğlu, MD

Other specific contacts: Şahin Avcı, MD (Medical genetics specialist); Gülleyla Kılıç, PhD (Chief of cytogenetics laboratory); Serpil Eraslan, PhD (Chief of molecular genetics laboratory); Esra Yücel, PhD (Chief of NGS & bioinformatics); Nihan Bilge Kamer, PhD (Chief of molecular cytogenetics laboratory); Elanur Yılmaz, PhD (zebrafish facility)

BRIEF DESCRIPTION OF STUDENT ACTIVITIES

Obligatory attendance to weekly activities of the Medical Genetics Department

Monday [15.30-17.00] Case presentations & discussion with laboratory findings

Thursday [15.30-17.00] Textbook review (chapters to be reviewed change each academic year; 30 minutes presentation + 15 minutes discussion), journal club (selected article related to the theoretical topic presented; 15 minutes presentation + 15 minutes discussion), case presentation (related to the topic or one with a strong message both for clinicians and laboratory geneticists; 15 minutes presentation + 15 minutes discussion)

Attendance to KUH meetings that medical Geneticists attend and/or contribute:

Monday [13.00-14.00] Neuromuscular case presentations, weekly

Tuesday [13.00-14.00 and 16.00-17.00] Endocrine, weekly [KUH&AH]

Wednesday [07.30-08.30] Morbidity & Mortality, twice monthly [KUH & AH]

Thursday [07.30-09.00] Hemato-oncology, weekly [KUH & AH]

Thursday [07.00-09.30] Multiple system oncology (thorax & respiratory, neurosurgery, gynecology, weekly [KUH & AH]

Friday [13.30-14.30] Gastroenterology, weekly [KUH & AH]

In the first two weeks, which is the observation period, attendance to clinical consultations and all genetic sessions is obligatory. During the last two weeks, two patients & families will be primarily consulted by the student. They will present the clinical data and decide on the follow-up and testing policies. They will be accompanied by a medical geneticist and their evaluation and input will be discussed twice, one at the date of the consultation and later at the weekly department meeting on Monday.

– The student will write clinical notes on two cases with confirmed laboratory diagnosis, including genetic counseling issues, which will be reviewed and handed to the patient or family by the student, under the watch of a medical geneticist).

– The student will prepare a theoretical chapter, journal club and case presentation, and present once at the weekly department meeting on Thursday.

– Attendance to research meetings, held two times a month is not obligatory but suggested. The meetings are on ongoing research projects, tasks related to writing projects or manuscripts.

LEARNING OBJECTIVES

At the end of the elective, a 5th grade student will have a broad understanding on various activities of the Medical Genetics Department. They will appreciate the role of clinical genetics in medicine, clinical evaluation leading to definite diagnosis, and genetic testing strategies. They will observe laboratory work, the interaction between clinicians and laboratory personnel target-driven diagnostic algorithms, , diagnostic reporting of the genetic results, and the impact of good communication skills between the clinicians and the patient / family with emphasis on genetic counseling and long term follow-up strategies.

Specific objectives are;

– be able to take detailed family history, construct a pedigree and evaluate pedigree data with its possible impact on inheritance pattern.

– be able to perform physical examination including dysmorphic evaluation, and additional measurements needed for detailed evaluation.

- be able to use genetic databases for genetic diagnosis and to list differential diagnoses, and plan for laboratory testing.

- be able to obtain informed consent both for genetic testing and photo / video release for publication purposes.

- be able to perform skin biopsy, and other samplings (buccal smear, saliva, hair etc.).

- be able to perform genetic material storage (banking).

- be able to write a clinical note covering family history, patient's personal history, physical examination findings, positive finding list, possible diagnosis and differential diagnoses and the testing algorithm for definite diagnosis.

- be able to write a clinical note for a patient / family with definite diagnosis covering all possible clinical features of the genetic disease, prognosis, follow-up strategies, inheritance pattern, family members to be tested for affected or carrier status, reproductive options, possible ongoing research related to possible treatment.

- be able to gain & update knowledge on the ideal genetic laboratory reporting formats and interpret its input for clinical purposes.

- be able to appreciate long term research objectives and how to pursue clinical research.

ASSESSMENT OR EVALUATION

- 1) Each student will be evaluated by their presentation on Thursday weekly meeting. This presentation covers the related topic of the text book chosen for the academic year (e.g. for 2020-21 academic year Genes by Lewin et al. was appointed), research article discussion (summed up in 10-12 slides) and case presentation.

- 2) Each student will write at least two detailed clinical notes, one will be a genetic consultation note for a patient with a definitive diagnosis, covering genetic counselling issues.

- 3) At weekly case discussion meetings and at weekly theoretical lecture time comprising journal club and case presentations, contribution of the student, and their attentiveness on the subject will be evaluated by the medical genetics core group (as of now; three MDs and five Postdocs). "KUSOM Clinical Performance Evaluation Form" is used for reporting.

Recommended readings/resources:

- 1) Emery's Elements of Medical Genetics, Peter Turnpenny
- 2) Congenital Malformations: Evidence-Based Evaluation and Management, Praveen Kumar
- 3) The Bedside Dysmorphologist, William Reardon
- 4) Management of Genetic Syndromes, Judith E. Allanson
- 5) Thompson & Thompson Genetics in Medicine, Robert L. Nussbaum
- 6) Pediyatrik Semiyoloji, Fatma Oğuz