Curriculum of Genetics in Pediatrics

13 lectures, Venue: Ist Department of Pediatrics, 2nd floor, Koós Aurél room, 16:15

02.21.	The outstanding role of genetics in pediatrics, Pathomechanism of the childhood disorders in function of the underlying genetic alteration.	Kálmán Tory
02.28.	Physical examination of the infant.	Kálmán Tory
03.06.	Way to diagnosis - Minor anomalies and chromosomal abnormalities recognizable by morphological features	Árpád Kovács
03.13.	I. Selection of the appropriate method of investigation: karyotyping, FISH, aCGH, QMPSF, NGS, Sanger sequencing, haplotype analysis	Irén Haltrich and Eszter Jávorszky
03.20.	Way to diagnosis – Symptoms suggestive of metabolic diseases. Diagnoses based on biochemical alterations.	Petra Zsidegh
03.27.	Evaluation of genetic results: distinction of pathogenic and benign variants.	Kálmán Tory
04.03..	Patient presentation, approach, diagnosis, risk calculation, counseling in numerical chromosomal abnormalities	Anett Fekete
04.10.	Patient presentation, approach, diagnosis, risk calculation, counseling in structural chromosomal abnormalities and balanced translocations	Éva Pinti
04.17.	Patient presentation, approach, diagnosis, risk calculation, counseling in autosomal recessive disorders	Kálmán Tory
04.24.	Patient presentation, approach, diagnosis, risk calculation, counseling in autosomal dominant and mitochondrial disorders	Anna Lengyel
05.08.	Patient presentation, approach, diagnosis, risk calculation, counseling in X-linked, incompletely penetrant and multifactorial disorders	Kálmán Tory
05.15.	Approach to a disorder with an unknown molecular basis – animal models and functional investigations of novel genes.	Tália Magdolna Keszthelyi
05.22.	Ethical issues, gene therapy, what the future will bring. Summary and consultation	Kálmán Tory