



SVEUČILIŠTE
U SPLITU
MEDICINSKI
FAKULTET

Clinical Genomics and NGS 30th course jointly organised by ESGM, ESHG and CEUB

Medicinski fakultet Sveučilišta u Splitu i KBC Split
Organiziraju online tečaj iz Bertinora (Italija):

Od 2.5. do 5.5. 2017. maksimalni broj polaznika je 40, Hrvatska liječnička komora ocijenila tečaj I kategorije. Skup je kategoriziran kao Međunarodni tečaj prve kategorije te je aktivnim sudionicima dodijeljeno 30 bodova, a pasivnim sudionicima dodijeljeno je 20 bodova.

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Podatci za uplatu:

IBAN: HR8523300031100071293

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Iznos: 500,00 KN

Matični broj: 01315366

Poziv na broj: 02703

Kotizacija se može platiti i na dan tečaja u iznosu od 600,00KN

Tuesday MAY 2.

Morning lectures: Medical Genetics concept and principals

08:30 – 09:00	Participants Registration
09:00 – 09:15	Introduction to the course - Giovanni Romeo
09:15 – 10:00	Genomic Medicine - Dian Donnai
10:00 – 10:45	Phenotype to genotype - Han Brunner
10:45 – 11:00	<i>Pause</i>
11:00 – 11:45	Cytogenetics and arrays - Eva Klopocki
11:45 – 12:30	Complex disorders and classical gene identification - Andrew Read
12:30 – 13:15	Discussion of the morning lectures
13:30 – 14:30	<i>Pause</i>
14:30 – 15:00	Basics of next generation sequencing technology - Alexander Hoischen
15:00 – 15:30	Basics of NGS bioinformatics - Christian Gilissen
15:30 – 15:45	Coffee break
15:45 – 16:15	NGS in the clinic - Anita Rauch
16:15 – 16:45	Future NGS technologies - Jonathan O'Halloran
16:45 – 17:00	Discussion of the afternoon lectures

Wednesday MAY 3.

Morning Lectures: Therapy and prenatal diagnostics in the NGS era

- 09:00 – 09:45 Therapy and cancer - John Burn
09:45 – 10:30 SMA: From gene and modifier to therapy - Brunhilde Wirth
10:30 – 11:00 *Pause*
11:00 – 11:45 Non-invasive prenatal testing – Janneke Weiss
11:45 – 12:30 Mitochondrial pathologies - Caterina Garone
12:30 – 13:15 Discussion of the morning lectures
13:30 – 14:30 *Pause*

Afternoon Lectures: Complex mechanisms of disease

- 14:30 – 15:00 Discovering structural variants in cancer using NGS data - Tobias Rausch
15:00 – 15:30 Epigenetics, imprinting, clinical - Karen Temple
15:30 – 16:00 *Pause*
16:00 – 16:45 Non-coding mutations/long-range effects - Eva Klopocki
16:45 – 17:30 Oligogenic diseases - Nicholas Katsanis
17:30 – 18:00 Discussion of the afternoon lectures

THURSDAY MAY 4.

Morning Lectures: Novel NGS applications

- 09:00 – 09:45 Molecular inversion probes and Saturation Genome editing – A. Hoischen
09:45 – 10:30 Long-read sequencing - Evan E Eichler
10:30 – 11:00 *Pause*
11:00 – 11:45 GWAS with NGS - Carlo Sidore
11:45 – 12:30 Data integration - Lude Franke
12:30 – 13:15 Discussion of the morning lectures
13:30 – 14:30 *Pause*
14:30 – 14:45 Croatian scientist presentation
14:45 – 15:00 Discussion
15:00 – 15:15 Visiting lab at. University of Split School of Medicine
20:00 *Diner*

FRIDAY MAY 5.

Morning Lectures: Large scale NGS

- 09:00 – 09:45 Visiting lab for Human Genetics at UHC Split, Paediatrics Clinic
09:45 – 10:30 Genomics England - Augusto Rendon
10:30 – 11:00 *Pause*
11:00 – 11:45 Phenotype and NGS integration / HPO benefits - David Fitzpatrick
11:45 – 12:30 Single cell sequencing and applications to PGD - Thierry Voet
12:30 – 13:15 Discussion of the morning lectures
13:30 – 14:30 *Pause*