



SVEUČILIŠTE  
U SPLITU  
MEDICINSKI  
FAKULTET

## Clinical Genomics and NGS 30<sup>th</sup> 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.*

*Prijave na mail/mobitel:*

[teakastelan@gmail.com](mailto:teakastelan@gmail.com)

[maja.pivic@gmail.com](mailto:maja.pivic@gmail.com)

**+385 98 865 264;**

**+387 63 486 957**

**Podatci za uplatu:**

**IBAN: HR8523300031100071293**

**OIB: 02879747067**

**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*