



Klinički bolnički centar
KBC
Split



HRVATSKO DRUŠTVO
ZA HUMANU GENETIKU



**Tečaj trajne edukacije liječnika I. kategorije s međunarodnim sudjelovanjem
Kombinirani (Hybrid) tečaj s "Course in Medical Genetics" u organizaciji
EuroMediterranean University Center of Ronzano, Bologna, Italia. Medicinski fakultet u
Splitu i Klinika za dječje bolesti KBC Split**

Pod pokroviteljstvom HDHG (Hrvatsko društvo za humanu genetiku) i ESHG (European Society of Human Genetics)

Voditeljica: Prof. dr. sc. Vida Čulić, pedijatar genetičar

Mjesto održavanja: Medicinski fakultet Sveučilišta u Splitu, Šoltanska 2, Split **Vrijeme održavanja: 11. svibanj – 15. svibanj 2014.**

Tečaj je namijenjen liječnicima obiteljske medicine, pedijatrima, ginekolozima, internistima, specijalizantima i specijalistima raznih grana. Tečaj je I kategorije s međunarodnim sudjelovanjem, koji će tako biti bodovan od **liječničke komore**.

Struktura Tečaja:

- **jutarnji program** je on-line prenos iz Bolonje, Ronzano, Italija iz EuroMediterranean University Center of Ronzano (27th Course in Medical Genetics) u kojem sudjeluju najznačajniji genetičari iz određenih područja (dizmorfologija, spinalne mišićne atrofija, starenja, epigenetika, X- vezanih bolesti, mentalne deficijencije)

- **popodnevni program** nastavljaju naši i inozemni predavači s praktičnim radionicama u ambulanti za genetičko infomiranje, Laboratoriju za humanu genetiku i Klinici za dječje bolesti, KBC Split, Spinčićeva 1. Tijekom tečaja biti će podijeljeni tekstovi predavanja iz Bolonje.

Cijena kotizacije je 200 kn za liječnike (zbog potvrdnice Hrvatske liječničke komore), uplatiti na **račun** : Medicinski fakultet U Splitu, Broj žiro računa 2330003-1100071293, OIB 02879747067, Matični broj 01315366, poziv na broj 02703

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27th Course in Medical Genetics May 11-15, 2012 EuroMediterranean University Center of Bologna (Italy)

Course description: Medical Genetics is postgraduate level course addressed to both researchers and clinicians seeking an up-to-date overview of the field of the medical genetics today. It provides an overall view of the clinical developments which are taking place as major applications of modern genetics different medical specialities. The topics covered in the present edition are: Introduction to medical Genetics and Genome Analysis, Cytogenetics and Clinical Genetics, New Approaches in medical Genetics, Complex Genetic Disorders, Therapy, Technology, Epigenetics and Ethical Issues. At the end of the course, on May 24th afternoon, a Public Awareness of genetics event in honor of an Alumni reunion of the European School of genetic Medicine.

Directors: H. Brunner (Nijmegen), G.Romeo (Bologna), B Wirth (Bonn)

Speakers H. Brunner (Nijmegen), M.Capecci (Salt Lake City), E Colonna (Cambridge), D.Donnai (Manchester), J Gecz (Adelaide), B. Horsthemke (Essen), S. Lyonnet (Paris), N. Katsanis (Durham), A. Read (Manchester), G. Romeo (Bologna), D. Valle (Baltimore), J Veltman (A. Nijmegen), Jermeesh (Leuven), B Wirth (Bonn), B. Wollnick (Bonn).

ZAJEDNIČKI PROGRAM:

Nedjelja, Sundy, 11 May Online Medical Genetics, from Ronzano, Italy (Šoltanska 1)

Morning Session: Introduction to Human Genome Analysis

9.00-9.15 Registration to the course
9.15-9.30 Introduction to the course G. Romeo
9.30-10.15 Medical Genetics Today D. Donnai
10.15.-11.00 Genotypes & phenotypes H. Brunner

11.00.-11.30 Coffe Break

11.30.-12.15 NGS J. Veltman

12.15-13.00 How to deal with next generation sequencing output, C. Gilissen

13.00.-14.00 Lunch Break

Afternoon Session: Split (Šoltanska 2)

14:30 – 16.30 h Seminar i **Vježbe: V. Čulić: Obiteljsko stablo**

Ponedjeljak, Monday, 12 May Online Medical Genetics, from Ronzano, Italy

Morning Session: Approaches to Clinical and Molecular Genetics

9.00-9.50 Linkage and association in a conceptual and historic perspective, A.Read

9.50.-10.40 Arrays and CNVs, J. Vermeesch

10.40.-11.10 Coffe Break

11.10-12.00 Dysmorphology. D. Donnai

12.00-12.50 Molecular syndromology in the NGS-era: which phenotype which family witch strategy? B. Wollnick

13.10.-14.00 Lunch Break

Afternoon Session: Split (Spinčićeva1)

14:30 – 16:30 h M. Meštrović (Split), Branka Polić (Split) : Klinika za dječje bolesti, JILD obilazak

Utorak, Tuesday, 13 May Online Medical Genetics, from Ronzano, Italy

Morning Session: From monogenic to complex genetic disorders

9.00-9.50 Marfan syndrome, related diseases and therapy, B.Loeys

9.50.-10.40 Aging Phenotypes B. Wollnik

10.40.-11.10 Coffe Break

11.10.-12.00 Oligogenic inheritance N. Katsanis

12.00.-12.50 Complex genetics, C. Wijmenga

13.00.-14.00 Lunch Break

Afternoon Session: Split (Spinčićeva1)

14:30–16:30h V. Čulić (Split): **Vježbe:** Ambulanta za genetičku informaciju

– **Primjeri u praksi**

Srijeda, Wednesday, 14 May Online Medical Genetics, from Ronzano, Italy

Morning Session: Therapy and Gene Regulation (Šoltanska2)

9.00-9.50 Inherited cancer and prospects for therapy, J. Burn

9.50-10.40 SMA: From gene and modifiers to therapy, B. Wirth

10.40.-11.10 Coffe Break

11.10-12.00 Epigenetics and disease, K. Temple

12.00-12.50 Long Distance Regulation, E. Klopocki

13.10.-14.00 Lunch Break

Afternoon Session: Split (Spinčićeva 1)

14:30 – 16:30 h **Vježbe:** Tea Kaštelan, Ivana Gunjača (Split): Laboratorij za humanu genetiku (citogenetika)-postnatalna, prenatalna, tumorska

Četvrtak, Thursday, 15 May Online Medical Genetics, from Ronzano, Italy

Morning Session: Mitochondria and evolution

9.00 – 9.50	Mitochondrial inheritance and disease, Rugarli E.
9.50-10.40	Mitochondria and cancer, G. Gasparre
10.40-11.00	Coffee Break
11.00 – 11.50	Population genetics in the genome era: The Sardinia project , F. Cucca
11.50-12.40	Best Poster Presentations by students
12.40	Wrapping up of the course
12.40-14.00	Lunch

Afternoon Session: Split (Spinčićeva 1)

14:30 – 15:30 h **A. Schinzel (Zurich):** t.b.t

16:00- 17:00: Ispit

Završetak tečaja



European Genetics Foundation