

Program of the second course
**Basics in human genetic diagnostics –
A course for CLGs* in education**

*Clinical Laboratory Geneticists

25th - 29th September 2017



Taking place at: Kostis Palamas Building, Akadimias 48 & Sina, 106 79, Athens, Greece

Main organizers: Thomas Liehr and Anja Weise (Jena, Germany),
Myrto Poulou (Athens, Greece)
Isabel Marques Carreiras (Coimbra, Portugal)

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Main sponsor - *European Society of Human Genetics*

ADS BIOTEC (Glasgow, UK)

Carl Zeiss (Jena, Germany)

Face2Gene

MetaSystems (Altlussheim, Germany)

MRC-Holland (Amsterdam, Netherlands)

ZytoVision (Bremerhaven, Germany)



Detailed program

DAY 1 – 25.09.2017

Basics of Human Genetics and Genetics Counselling		
09.15-09.30	1-1: Opening of the course	Aspasia Tsezou
09.30-10.00	1-2: Short introduction in human genetics	Giannis Georgiou
10.00-10.30	1-3: Basics of genetics	Jan Traeger
10.30-11.00	1-4: Genomic analysis: from karyotype to next generation sequencing	Aspasia Tsezou
11.00-11.15	Coffee break	
11.15- 12.45	1-5: Mendelian and non-Mendelian inheritance, including basics of Hardy-Weinberg-law	Anja Weise
12.45-13.45	Lunch break	
13.45-14.15	1-6: Syndromology and genetic syndromes	K. Varvagiannis
14.15-14.45	1-7: Basics of genetic counselling	Loukia Katsichti
14.45-15.15	1-8: Risk calculations in genetic counselling	Loukia Katsichti
15.15-15.30	Coffee break	
15.30-16.00	1-9: Next Generation Phenotyping with Face2Gene in the clinic, laboratory and for research	Nicole Fleischer (FDNA)
16.00-17.15	1-10: Workshop: How to do own risk-calculations	K. Varvagiannis Loukia Katsixti
17.15-17.30	break	
17.30-18.00	Written test	

DAY 2 – 26.09.2017

Genetic Syndromes, Teratology and Cytogenetics		
09.15-9.30	Correct answers of test from day before	Anja Weise
09.30-10.10	2-1: 'Frequent' chromosomal syndromes and databases	Anja Weise
10.10-10.30	2-2: Teratology - basics	Isabel M..Carreira
10.30-11.00	2-3: ADS Biotec: A One-Stop Shop for Automation in a Cytogenetic laboratory	Joachim Fischer (ADS Biotec Ltd)
11.00-11.15	Coffee break	
11.15-12.45	2-4: Classical and banding cytogenetics – techniques in pre- and postnatal diagnostics	Anja Weise
12.45-13.45	Lunch break	
13.45-14.30	2-5: Tumor cytogenetics	Isabel M. Carreira
14.30-15.45	2-6: Reporting in prenatal, postnatal and tumor cytogenetics incl. guidelines	Anja Weise
15.45-16.00	Coffee break	
16.00-16.40	2-7: Human chromosomes 1-22, X and Y	Anja Weise
16.40-18.00	2-8: Workshop: identify human chromosomes on a metaphase picture, cut and glue a karyogram	Anja Weise
18.00-18.10	break	
18.10-18.40	Written test	
20.30	Common dinner	

DAY 3 – 27.09.2017

Molecular Cytogenetics and Molecular Genetics I		
09.15-9.30	Correct answers of test from day before	Anja Weise
09.30-09.50	3-1: Molecular cytogenetics - basics	Anja Weise
09.50-10.20	3-2: Interphase-FISH and Multicolour-FISH	Anja Weise
10.20-10.30	3-3: FISH-applications in diagnostics	Anja Weise
10.30-10.45	Coffee break	
10.45-11.10	3-4: CGH and array-CGH - introduction	Maria Tzetzis
11.10-12.00	3-5: Array-CGH – principles and applications,	Andreas Polten (Agilent)
12.00-12.20	3-6: Epigenetics – uniparental disomy and imprinting	Isabel M. Carreira
12.20-12.40	3-7: Gene therapy – any relevance for diagnostics?	Isabel M. Carreira
12.40-13.45	Lunch break	
13.45-14.45	3-8: Workshop: Reporting incl. databases and confirmation of aCGH-results - aCGH results interpretation	Lina Florentin
14.45-15.30	3-9: Overview of Molecular Genetics	Jan Traeger
15.30-16.15	3-10: Workshop: Biological databases and Gene-specific databases	Eirini Fylaktou
16.15-16.30	Coffee break	
16.30-17.20	3-11: PCR techniques and applications	Christos Kroupis
17.20-17.30	Break	
17.30-18.00	Written test	

DAY 4 – 28.09.2017

Molecular Genetics II		
09.15-9.30	Correct answers of test from day before	Isabel M. Carreira
09.30-10.15	4-1: NIPT	Isabel M. Carreira
10.15-10.40	4-2: MLPA, related techniques and applications	Nikoleta Selenti
10.40-11.15	4-3: Examples from diagnostics and research for PCR/MLPA approaches - reporting and databases	Nikoleta Selenti Myrto Poulou
11.15-11.30	Coffee break	
11.30-12.15	4-4: MLPA applications (MLPA and MRC Holland)	Jan Smout (MRC Holland)
12.15-13.15	Lunch break	
13.15-14.00	4-5: Preimplantation genetic diagnosis and preimplantation genetic screening	Georgia Kakourou
14.00-15.15	4-6: Designing a PGD case	Thalia Mamas
15.15-15.30	Coffee break	
15.30-16.50	4-7: Technical basis of Sanger sequencing	Eirini Fylaktou
16.50-17.20	4-8: Technical basis of NGS	Nikolaos Marinakis
17.20-17.40	break	
17.40-18.10	Written test	

DAY 5 – 29.09.2017

Molecular Genetics III and future directions of the field		
09.15-9.30	Correct answers of test from day before	Isabel M. Carreira
09.30-10.20	5-1: NGS principles and applications	Jeroen Adema (Illumina)
10.20-11.10	5-2: Cancer genetics diagnostics and case studies	Florentia Fostira
11.10-11.30	Coffee break	
11.30-13.00	5-3: Reporting and databases for sequencing results	Florentia Fostira Nikolaos Marinakis
13.00-14.00	Lunch break	
14.00-15.00	5-4: FISH as a routine diagnostic tool in pathology labs	Sven Hauke (Zytovision)
15.00-15.15	Coffee break	
15.15-15.55	5-5: Inborn errors of metabolism: from symptoms to diagnosis	Evridiki Drogari
15.55-16.35	5-6: Neurogenetics diagnostics and case studies	Georgia Karadima
16.35-17.05	5-7: The future of diagnostics with Proteomics	Makis Zoidakis
17.05-17.20	Break	
17.20-18.00	Written test	
18.00-18.15	Concluding session	
18.15-18.30	Break (e.g. to go to hotels)	
19.15-21.00	Guided Tour of Acropolis museum	
21.00	Common dinner – Acropolis restaurant	
	Results of all test and certificates will be sent out by Dr. Thomas Liehr asap	

