Last announcement of the fifth course

**Basics in human genetic diagnostics –**
*A course for CLGs* in education

*Clinical Laboratory Geneticists*

**Dates:** 05th - 09th September 2022; Monday, 9.00 hrs to Friday 18:30 hrs

**Location:** Hotel Mercure, Av. 25 de Abril 22, 3080-086 Figueira da Foz, Portugal

**Main organizers:** Isabel M. Carreira, Joana B. Melo, Susana Ferreira (Coimbra, Portugal)

**Co-Advisor:** Thomas Liehr (Jena, Germany)

**Registration:** Last call until 30 June 2022

**Costs:** EUR 500


**Travel and stay:** to be organized by the participants themselves

**Preliminary program**

- Each day starts at 9.00 hrs and finishes around 18:30 hrs;
- Two coffee and one lunch breaks are scheduled each day;
- Every day will contain a practical part;
- At the end of each day a written exam will be taken;
- A certificate will be issued at the end of the course (in case of successful participation).

**Topics:**

Introduction in human genetics; Basics of genetics; Genomic analysis: from karyotype to next generation sequencing (NGS); Mendelian and non-Mendelian inheritance, including basics of Hardy-Weinberg-law; Syndromology and genetic syndromes; Basics of genetic counselling; Risk calculations in genetic counselling; Frequent chromosomal syndromes and databases; Teratology – basics; A One-Stop Shop for Automation in a Cytogenetic laboratory; Classical and banding cytogenetics – techniques in pre- and postnatal diagnostics; Human chromosomes 1-22, X and Y; Identification of human chromosomes on a metaphase picture, plus working package to cut and glue a karyogram; Molecular cytogenetics – basics; FISH-applications in diagnostics; Tumour cytogenetics; Repetitive Elements; FISH as a routine diagnostic tool in clinic laboratory genetics; Epigenetics – uniparental disomy and imprinting; Reporting in prenatal, postnatal and tumour cytogenetics including guidelines; CGH and array-CGH – principles and applications; Overview of Molecular Genetics; fundamentals and applications in MLPA, MS-MLPA, digital MLPA. Fragile-X, PCR techniques and applications: Examples of clinical cases; Technical basis and principle of Sanger sequencing and NGS; Reporting and databases for sequencing results; NGS applications.; Biological databases and Gene-specific databases; UCSC Genome Browser as an important tool in variant interpretation in the clinical context; Cancer genetic diagnosis and discussion of clinical cases; Gene therapy and its relevance for diagnostics Optical mapping – a new approach to chromosomes; Discussion of challenging clinical cases in: Neurogenetics; Inborn errors of metabolism.; Prenatal Diagnosis: Invasive and non-invasive
Return this form to: Susana Ferreira cimago@fmed.uc.pt

Personal Data: (PLEASE FILL WITH COMPUTER OR CLEAR HANDWRITING!!)

Name, first name, title: ____________________________________________________________________________

Institutional Address: ____________________________________________________________________________

Telephone / FAX: _________________________________________________________________________________

e-mail: _________________________________________________________________________________________

I herewith register for the fifth course:

Basics in human genetic diagnostics – A course for CLGs* in education
*Clinical Laboratory Geneticists

Costs: EUR 500

Travel and stay: to be organized by the participants themselves

☐ I herewith declare that I will transfer the course fee within 3 weeks of receiving the corresponding payment details by Susana Ferreira.

☐ It is ok for me that my name and contact is printed in the final program and used openly in mails to all participants and sponsors. Please consider that if you do not tick the box it may cause problems in contacting you!

☐ I am ESHG member: Trainee/Regular/Non-Member
(please check and underline if something is true)

______________________________________________________________________________________________ (place, date, signature, stamp)