In collaboration with

Fondazione CARIPARO

Associazione Veneta per la Lotta contro la Talassemia

ALT - Associazione Lotta alla Talassemia di Ferrara

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Theranostics in β-Thalassemia

Ferrara, October 20
Room C3, Palazzo Manfredini, Via Monsignore Bovelli 59 - 44121 - Ferrara – Italy
Entrance: Via Ludovico Muratori, 9

Organized by: Department of Life Sciences and Biotechnology, Ferrara University, Italy

Program

Speakers

Sjaak Philipsen (EMC, ERASMUS UNIVERSITAIR MEDISCH CENTRUM ROTTERDAM, The Netherlands)
Roberto Gambari (UNIFE, UNIVERSITY OF FERRARA, Italy)
Laura Breda(CU, CORNELL UNIVERSITY CORPORATION, NY,USA)
Carsten Lederer (CING, THE CYPRUS FOUNDATION FOR MUSCULAR DYSTROPHY RESEARCH, Cyprus)
Marios Phylactides (CING, THE CYPRUS FOUNDATION FOR MUSCULAR DYSTROPHY RESEARCH, Cyprus)
Alessia Finotti (UNIFE, UNIVERSITY OF FERRARA, Italy)
Effrossyni Boutou (LAIKO GENERAL HOSPITAL, ATHENS, Greece)
Eleni Katsantoni (BRFAA, BIOMEDICAL RESEARCH FOUNDATION ACADEMY OF ATHENS, Greece)

THALAMOSS (THALAssaemia MOdular Stratification System for personalized therapy of beta-thalassaemia)

The specific aims of the THALAMOSS Project are based on the development of novel methods for associating variation in genomic data with phenotypic variation. THALAMOSS combines cutting-edge computing technology with optimized algorithms to mine the unique datasets provided through the proposed project for biologically and medically relevant patterns that can be reliably associated with specific patient groups, treatment response and disease progression. In addition to stratification of patient samples for their molecular properties, the proposed project is analysing responsiveness to advanced therapeutic approaches to classify patient samples, analysing this responsiveness both as a consequence of molecular properties and as a determinant of the success of novel therapies. At present, the most promising novel approaches to β-thalassaemia treatment are the application of chemical inducers of endogenous foetal haemoglobin (HbF) and transduction of haematopoietic precursor cells with lentiviral vectors expressing exogenous β-globin. In this vein, the proposed project has standardized a high-throughput *in vitro* differentiation protocol of patient-derived erythroid precursor cells and to be used to test established inducers of HbF and established β-globin-expressing lentiviral vectors for their therapeutic efficacy and cytotoxicity in a large number of representative cultures from β-thalassaemia patients, in order to identify sample characteristics compatible with palliative chemical or curative gene therapy (GT) intervention. In addition to basic vectors over-expressing β-globin, enhanced vectors (additionally down-regulating disease modifiers and aberrant β-globin mRNA species by an established shRNA co-expression strategy) are tested in patient samples, in order to assess their potential utility as tools for personalised medicine. Results of these large-scale analyses will have wide-ranging implications for chemical and lentiviral treatments, including the establishment of markers for potentially successful chemical HbF induction and of minimum efficiency requirements for basic and enhanced GT vectors. Taken together, the expected outcome of the THALAMOSS project will be a landmark shift in our approach to the treatment of β-thalassaemia, based on detailed genotype-phenotype correlations, novel markers and a set of standardised analysis procedures for the stratification of patients for optimised disease management and personalised therapy.

Program

8.30-9.00	Registration
9.00-9.05	**Roberto Gambari** (UNIFE, Italy): Introductory remarks
9.05-9.40	**Sjaak Philipsen** (EMC, The Netherlands): Genetic and epigenetic regulation of hemoglobin switching
9.40-10.10	**Laura Breda** (CU/CHOP, USA): Use of forced chromatin looping to reactivate HbF in beta-globinopathies: achievements and challenges
10.10-10.40	**Carsten Lederer** (CING, Cyprus): Genome editing and personalized gene therapy for hemoglobinopathies
10.40-11.00	**Alessia Finotti** (UNIFE, Italy): Modulation of BCL11A gene expression in erythroid cells
11.00-11.15	*Coffee break*
11.15-11.45	**Eleni Katsantoni** (BRFAA, Greece): Transcriptomics and proteomics for beta-thalassemia
11.45-12.15	**Marios Phylactides** (CING, CYPRUS): Investigation of genetic factors affecting drug response and clinical phenotype in Cypriot β-thalassaemia patients
12.15-12.40	**Roberto Gambari** (UNIFE, Italy): Theranostics and drug repurposing: from the laboratory to the clinic
12.40-13.10	**Effrossyni Boutou** (LGHA, Greece): Molecular basis of ongoing clinical trials for beta thalassaemia
13.10-13.30	**Carsten Lederer** (CING, Cyprus): ITHANET: an information and database portal for hemoglobinopathies