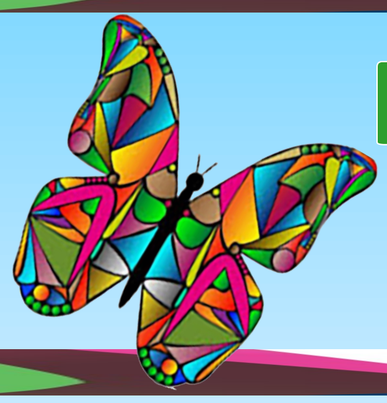
PROGETTO DIMAR
DIMED-MALATTIE RARE

Dipartimento di Medicina DIMED

Università degli Studi di Padova



Rare Disease Day in DIMED

Research Projects

Thursday 28th February 2019

Orto Botanico, Padova

Scientific Committee: F. Fabris and R. Vettor

A. Dei Tos, L. Iaccarino, P. Maffei, G. Milan, F. Piazza, C. Scaroni

8.30 Registrazione dei partecipanti

9.00 Saluto delle Autorità

9.30 DIMAR (DImed MAlattie Rare): Development of a scientific, educational and management platform for Rare Diseases 2018-2022
C. Agostini (Comitato DIMED-DIMAR)

10.00 *Lecture*. EUROWABB: an EU rare disease registry
T. Barrett, University of Birmingham, UK

Coffee break 10.30 -10.50

I Session: Focus on Endocrine and Metabolic Rare Diseases

Chairs: A. Avogaro and C. Foresta

10.50 Alterations of circulating stem cells in Fabry disease
G.P. Fadini, Malattie del Metabolismo
11.00 The Alström Syndrome a rare ciliopathy: patient networks, clinical aspects and molecular mechanisms
F. Favaretto, Clinica Medica 3
11.10 Familial and sporadic hyperaldosteronism: from chimeric gene to channelopathy
L. Lenzini, Clinica dell'Ipertensione
11.20 Primary Ciliary Dyskinesia: genetic aspects and clinical manifestations
M. Santa Rocca, Servizio Patologia Riproduzione Umana
11.30 Discussion

II Session: Focus on Rare Tumours

Chairs: F. Fallo and C. Scaroni

11.50 EURACAN: A European Reference Network Consortium on Rare Solid Cancers of Adults
A. Dei Tos, Dipartimento di Patologia, Treviso
12.00 Thyroid medullary carcinoma: old and new prognostic markers
C. Mian, Endocrinologia
12.10 Von Hippel-Lindau, a paradigm of multidisciplinary
A.M. Ferrara, Istituto Oncologico Veneto
12.20 Discussion

12.40 *Lecture*. European Networks and Rare Disease grants
A.M. Colao, Università Federico II, Napoli

Lunch and Poster Slide Session 13.10-14.30

Chairs: F. Fabris and R. Vettor

XXXIV PhD Course in Rare Diseases

Chairs: G. Rossi and P. Angeli

14.30 Rare Diabetes and Obesity: NAFLD and Alström Syndrome
S. Bettini, PhD in Clinical and Experimental Sciences
14.40 Molecular characterization of adosteronomas in human hyperaldosteronism
D. Schiavone, PhD in Arterial Hypertension and Vascular Biology
14.50 Discussion

III Session: Focus on Liver and Kidney Rare Diseases

Chairs: E. Quaia and L. Calò

15.00 Pathology and Pathogenesis of Idiopathic Non-Cirrhotic Portal Hypertension: current knowledge and future research
S. Sarcognato, Anatomia Patologica
15.10 MR cholangiography in primary sclerosing cholangitis: current role and future perspectives
R. Motta, Istituto di Radiologia
15.20 The Serpin B3 polymorphism (SCCA-PD) is associated with the severity portal hypertension and complications onset in patients with advanced liver disease
A. Martini, Clinica Medica 5
15.30 Oxidative stress and the altered reaction to it in Fabry Disease: a possible target for cardiovascular-renal remodeling
G. Bertoldi, Nefrologia
15.40 Polycistic Kidney Disease: epidemiology, genetics and potential treatments
F. Gastaldon, Nefrologia Vicenza
15.50 Discussion

Coffee break 16.10 -16.30

IV Session: Focus on Immunological, Rheumatological and Hematological Rare Diseases

Chairs: G. Semenzato and A. Doria

16.30 Sarcoidosis: a paradigmatic granulomatous disease
F. Cinetto, Internal Medicine and Immunological Rare Disease Center, Treviso
16.40 Advanced diagnostic techniques in autoimmune blistering disorders
I. Russo, Dermatologia
16.50 The role of PTX3/antiPTX3 antibody arrangement in SLE and ANCA associated vasculitis
M. Gatto, Reumatologia
17.00 Glycosylated ferritin in Adult-onset Still disease and acquired hemophagocytic lymphohistiocytosis
M. Marinova, Medicina di Laboratorio
17.10 Lack of von Willebrand factor multimer organization associated with the c.2269_2270del mutation which acts through a non-canonical splicing site: a new pathogenic mechanism of von Willebrand disease
V. Daidone, Clinica Medica 1
17.20 Rare inherited bleeding disorders: novel therapeutic strategies
C. Bulato, Malattie Tombotiche ed Emorragiche
17.30 Novel insights on rare γδ T-cell lymphoproliferative disorders
A. Teramo, Ematologia e Immunologia Clinica
17.40 Discussion

18.00 Living with an immunological Rare Disease
A. Gressani, IPOPI, International Patient Organization for Primary Immunodeficiency

18.15 Concluding Remarks and DIMED Rare Disease Awards.
F. Fabris and R. Vettor

18.30 ECM Questionnaire

AZIENDA OSPEDALIERA UNIVERSITA' PADOVA

QR CODE FOR REGISTRATION

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Modalità d'iscrizione: il corso è gratuito con iscrizione obbligatoria attraverso la seguente link https://goo.gl/forms/F3Cq6liynlkXZnRN2 e riservato a n. 170 partecipanti per le seguenti figure professionali: Medico Chirurgo (tutte le discipline), Biologo, Chimico, Farmacista, Tecnico Sanitario di Laboratorio Biomedico, Psicologo.

ECM: Ai fini dell'attestazione dei crediti ECM è necessaria la presenza effettiva degli iscritti al 90% dell'intera durata dell'attività formativa, con verifica tramite firma di frequenza in entrata e in uscita, la compilazione e restituzione della scheda di valutazione/gradimento, della scheda anagrafica e del questionario ECM di apprendimento alla fine del corso.