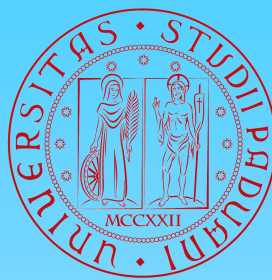




PROGETTO DIMAR
DIMED-MALATTIE RARE

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. Lenzi, *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 Polycystic 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 Trombotiche ed Emorragiche*

17.30 Novel insights on rare $\gamma\delta$ 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



QR CODE FOR REGISTRATION



Provider Ministeriale Nazionale id. 1884 - Dipartimento di Medicina - DIMED
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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.