RARE DISEASE DAY IN DIMED

RESEARCH PROJECT

Università degli Studi di Padova - Dipartimento di Medicina - DIMED
Progetto DIMAR

Padova

Friday 28th - Saturday 29th February 2020

Palazzo Bo' - Aula Magna - Archivio Antico

Aula Nievo

Via VIII Febbraio, 2 - Padova

Organizers

Roberto Vettor and Fabrizio Fabris

Scientific Commitee

Cristina Degan, Gabriella Milan,
Carlo Agostini, Pietro Maffei,
Sandra Casonato, Monica Facco,
Matteo Fassan, Caterina Mian,
Lorenzo Calò, Luca Iaccarino

Rare is many worldwide

Rare is strong every day

Rare is proud everywhere

RAREDISEASEDAY.ORG

Friday 28th February 2020
Palazzo Bo' – Via VIII Febbraio, 2 - Padova
Aula Magna – Archivio Antico

Language: Italian

8.30	Registration
	PLENARY SESSION I Aula Magna
9.00	Authorities' Welcome
9.20	Introduction to the Rare Disease Day in DIMED. *Roberto Vettor and Fabrizio Fabris*
9.30	Lecture. PATIENT'S RIGHTS IN CROSS-BORDER HEALTHCARE. *Paolo Giovanni Casali,* *Istituto Nazionale dei Tumori, Università di Milano*
	Coffee break 10.10 -10.40
	PARALLEL SESSION I Aula Magna
	Rare Diseases in Rheumatology and Autoimmunity Chairs: *Andrea Doria and Diego Cecchin*
10.40	Safety of oral anticoagulation withdrawal in patients with systemic lupus erythematosus and secondary antiphos pholipid antibody syndrome who had become seronegative. *Margherita Zen, Reumatologia*
10.50	A low-penetrance NLRP3 variant in a NOD2 carrier possibly worsens the phenotype of Blau syndrome. *Paola Galozzi, Reumatologia*

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11.00	Predictors of interstitial lung disease onset and progression in patients with systemic sclerosis and anti-topoisomerase I positivity. *Francesco Benvenuti, Reumatologia*
11.10	Persistent low-grade vascular inflammation in remitted large vessel vasculitis patients is associated with an increased risk of relapse. A longitudinal study using fully integrated 18F-FDG PET/MR. *Roberto Padoan, Reumatologia*
11.20	Genetic profile of anti-mag peripheral neuropathy helps to identify new therapeutic approach. *Andrea Visentin, Ematologia e Immunologia Clinica*
11.30	DISCUSSION Chairs: *Emilio Quaia and Paolo Sfriso*
11.50	Presentation and outcomes between 2000-2010 and 2011-2018 decades in granulomatosis with polyangiitis and microscopic polyangiitis: a 20 years followup monocentric study. *Mara Felicetti, Reumatologia*
12.00	Osteoporosis in Systemic Autoinflammatory Diseases: a case-control study. *Sara Bindoli, Reumatologia*
12.10	Severe preeclampsia related to antiphospholipid syndrome: an European descriptive study of 40 women. *Maddalena Larosa, Reumatologia*

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12.20 Prevalence and outcome of interstitial lung disease in patients with idiopathic inflammatory myopathies.
Mariele Gatto, Reumatologia

12.30 Are MR radiomic features the new radiological biomarkers for anti-Jo1 Syndrome?
Anna Sara Fraia, Radiologia

12.40 DISCUSSION

PARALLEL SESSION I Archivio Antico

Multidisciplinary approach to Rare Diseases (I)

Chairs: *Angelo Paolo Dei Tos and Claudio Ronco*

10.40 Hypertension and cardiovascular-renal remodelling: the human model of Bartter's and Gitelman's syndromes.
Matteo Rigato, Nefrologia

10.50 A new skin directed therapy for mycosis fungoides: near-infrared photoactivable immunotherapy.
Irene Russo, Dermatologia

11.00 Intimal Sarcoma: a Clinicopathologic and Molecular Analysis of 25 Cases.
Marta Sbaraglia, Anatomia Patologica

11.10 Whole Exome Sequencing in genetically unresolved cases of Dent disease.
Monica Ceol, Nefrologia

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11.20	CT Texture Analysis of Adrenal Glands of Primary Aldosteronism Patients. *Filippo Crimì, Radiologia*
11.30	DISCUSSION Chairs: *Carla Scaroni and Mario Plebani*
11.50	Fecal Eosinophil Cationic Protein as potential marker of disease activity in patients with eosinophilic esophagitis. *Andrea Padoan, Medicina di Laboratorio*
12.00	9-cis retinoic acid decreases POMC expression and cell viability in experimental model of Ectopic Cushing Syndrome. *Daniela Regazzo, Endocrinologia*
12.10	Role of transient elastography to detect Fontan-associated liver disease in rare congenital univentricular heart corrected by Fontan circuit. *Chiara Zanon, Clinica Medica 5ª*
12.20	Spinal and bulbar muscular atrophy: genetic aspects and clinical manifestations. *Maria Santa Rocca, Servizio Patologia Riproduzione Umana*
12.30	The European Registry of Fibromuscular dysplasia. *Livia Lenzini, Clinica dell'Ipertensione*
12.40	DISCUSSION

LUNCH 13.00-14.00

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PLENARY SESSION Aula Magna

Chairs: *Carlo Agostini, Pietro Maffei, Roberto Vettor*

14.00 SPAZIO DI INCONTRO:
Associazioni dei Pazienti, amministratori, politici, medici e ricercatori del territorio

Chairs: *Roberto Padrini and Andrea Alimonti.*

15.30 Lecture. ORPHAN DRUG AND RARE DISEASE'S TREATMENT.
Andrea Cignarella, Dipartimento di Medicina
Università degli Studi di Padova

16.00 OPEN QUESTIONS

PARALLEL SESSION II Aula Magna

Rare Diseases in Hemato-Oncology and Coagulation

Chairs: *Maria Luigia Randi and Livio Trentin*

16.10 Identification of a stat3/mir-146b/fasl axis in the development of neutropenia in T-large granular lymphocyte leukemia.
Antonella Teramo, Ematologia e Immunologia Clinica

16.20 Clinical prognostic factors for the management of patients with acquired Thrombotic Thrombocytopenic purpura (aTTP).
Irene Bertozzi, Clinica Medica 1ª

16.30 The bleeding risk in families with congenital factor V deficiency.
Chiara Simion, Malattie Trombotiche ed Emorragiche

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16.40 Study of a large cohort of patients with idiopathic and congenital erythrocytosis: focus on PHD2 mutations
Elisabetta Cosi, Clinica Medica 1ª

16.50 Combined contraceptives and the risk of venous thromboembolism in antithrombin, protein C and protein S defcient women. A prospective cohort study.
Daniela Tormene, Malattie Trombotiche ed Emorragiche

17.00 DISCUSSION

Chairs: *Paolo Simioni and Francesco Piazza*

17.20 Perioperative and long term management of severe congenital factor XIII deficiency in cardiac surgery: a case report.
Elena Campello, Malattie Trombotiche ed Emorragiche

17.30 Stat3 mutations impact on overall survival in large granular lymphocyte leukemia: a single center experience of 205 patients.
Gregorio Barilà, Ematologia e Immunologia Clinica

17.40 Are the two different computing programs MyPKfit and Wapps-Hemo comparable in PK-assessment for a haemophilia A population treated with octocog-alfa?
Samantha Pasca, Malattie Trombotiche ed Emorragiche

17.50 CK2a is overexpressed in classical Hodgkin lymphoma and regulates critical signaling pathways as well as PD-L1 expression.
Edoardo Ruggeri, Ematologia e Immunologia Clinica

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18.00 B cells from poor prognosis chronic lymphocytic leukemia express activated focal adhesion kinase (FAK) that is correlated to cytoskeletal-connected molecules.
Federica Frezzato, Ematologia e Immunologia Clinica

18.10 DISCUSSION

PARALLEL SESSION II Archivio Antico

Endocrine-Metabolic and Hepatic Rare Diseases
Chairs: *Angelo Avogaro and Luca Busetto*

16.10 A novel Melanocortin-4 Receptor mutation MC4R-F313S caused an impairment in α-MSH-induced cAMP signalling, resulting in severe obesity.
Elisabetta Trevellin, Clinica Medica 3ª

16.20 METABASE project: a multi-dimensional resource for assessing outcomes in adults with inherited metabolic diseases.
Nicola Vitturi, Malattie del Metabolismo

16.30 Adipose tissue characterization in Multiple Simmetric Lipomatosis.
Chiara Compagnin, Clinica Medica 3ª

16.40 Lipohypertrophy in acromegaly: exploring the mechanisms of growth hormone antagonist action on adipose tissue.
Francesca Favaretto, Clinica Medica 3ª

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16.50	Influence of 20 yr GH replacement on bone mineral density in adult hypopituitary patients *Alice Spinazzé, Clinica Medica 3ª*
17.00	DISCUSSION Chairs: *Patrizia Pontisso and Ambrogio Fassina*
17.20	Regulatory function and prognostic significance of SerpinB3 in Cholangiocarcinoma. *Andrea Martini, Clinica Medica 5ª*
17.30	Role of cellular senescence in the natural history of primary sclerosing cholangitis. *Samantha Sarcognato, Anatomia Patologica*
17.40	Response to treatment with terlipressin and albumin improves post liver transplant outcomes in patients with hepatorenal syndrome *Salvatore Piano, Clinica Medica 5ª*
17.50	Effects of miR-21 silencing in MZ-CRC-1 and TT Medullary Thyroid Cancer cell lines. *Loris Bertazza, Endocrinologia*
18.00	PD1 and PDL1 expression in medullary thyroid carcinoma. *Francesca Galuppini, Anatomia Patologica*
18.10	DISCUSSION

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APERICENA 18.30

19.30 CONCERTO

DIMAR for Patients

Asclepio Ensemble (University Hospital Orchestra)

SALA DEI GIGANTI, PALAZZO LIVIANO

Saturday 29th February 2020
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8.30	Registration
8.50	**PhD 34 - 35 Course in Rare Diseases: Introduction** Chairs: *Paolo Angeli and Gian Paolo Rossi*
9.00	Lecture. LIVER AND PANCREATIC ORGANOIDS: FROM BASIC RESEARCH TO THERAPEUTIC APPLICATIONS. *Luigi Aloia, Wellcome Trust-Cancer Research UK Gurdon Institute University of Cambridge, UK*
9.30	OPEN QUESTIONS
9.45	Rare Diabetes and Obesity: Hepatic stiffness evaluation with Shear Wave Elastography in Alström Syndrome. *Silvia Bettini, 2nd year, PhD in Clinical and Experimental Sciences*
10.00	Molecular characterization of the skin sodium compartment in aldosteronomas causing human hyperaldosteronism, a rare form of human hypertension. *Francesca Torresan, 2nd year, PhD in Arterial Hypertension and Vascular Biology*
10.15	Congenital and hereditary erythrocytosis: study of functional mechanisms of iron overload. *Andrea Benetti, 1st year, PhD in Clinical and Experimental Sciences*

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Language: English

10.30 Mechanisms of damage and vascular remodeling in patients with primary immunodeficiencies.
Alessandro Bressan, 1st year, PhD in Arterial Hypertension and Vascular Biology

10.45 OPEN QUESTIONS

Coffee break 11.00 - 11.30

Specific Projects in Rare Diseases 2020_2022

Chairs: *Carlo Agostini and Roberto Vettor*

11.30 Primary Immune Deficiencies (PIDs).
Francesco Cinetto, Medicina Interna e Centro Malattie Rare Immunologiche, Treviso

11.45 Interstitial Lung Disease in Idiopathic Inflammatory Myopathies: predictors of disease progression and performance of candidate serum biomarkers.
Elisabetta Zanatta, Reumatologia

12.00 The impact of the immune system in the risk-stratification of myelodysplastic syndromes: a deep characterization of T and NK cell compartments.
Cristina Vincenzetto, Ematologia e Immunologia Clinica

12.15 Medullary thyroid carcinoma (MTC).
Simona Censi, Endocrinologia

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12.30 Exploring the role of Alström Syndrome Protein in the Leptin/Melanocortin Pathway.
Pietro Maffei, Clinica Medica 3ª

12.45 OPEN QUESTIONS

LUNCH 13.00 - 14.10

Language: Italian

SESSION III

Multidisciplinary approach to Rare Diseases (II)

Chairs: *Gianpietro Semenzato and Fabrizio Fabris*

14.10 Lecture. RARE MYELODYSPLASTIC SYNDROMES
Valeria Santini, NDS Unit, AOU Careggi, Università di Firenze

14.50 OPEN QUESTIONS

15.00 A novel RUNX1 variant is related to thrombocytopenia in a sporadic form of myelodysplastic syndrome.
Fabrizio Vianello, Ematologia e Immunologia Clinica, Clinica Medica 1ª

15.10 Phenotypic and molecular characterization of T and NK cells in hypocellular myelodysplastic syndromes.
Giulia Calabretto, Ematologia e Immunologia Clinica

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15.20 Neurocognitive assessment in Alström Syndrome
Francesca Dassie, Clinica Medica 3ª

15.30 Obstructive Sleep Apnea in Acromegaly and the Effect of Treatment: A Systematic Review and Meta-Analysis.
Matteo Parolin, Clinica Medica 3ª

15.40 Muscle involvement in a familiar form of tubular aggregates myopathy.
Amalia Lupi, Istituto di Radiologia

15.50 18F-FDG PET/MRI with DWI: one-stop shop for pediatric sarcomas.
Giovanna Orsatti, Istituto di Radiologia

16.00 DISCUSSION

SESSION IV

Endocrine and Renal Rare Diseases

Chairs: *Carlo Foresta and Lorenzo Calò*

16.20 A novel thyroid hormone receptor beta mutation (G357R) in a family with resistance to thyroid hormone beta.
Jacopo Manso, Endocrinologia

16.30 Digenic inheritance: two rare cases of Gitelman syndrome.
Franca Anglani, Nefrologia

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16.40	Radiomics: a new tool to diagnose adrenal carcinoma from adenoma. *Filippo Ceccato, Endocrinologia*
16.50	The glomerular damage in three cases of Dent disease: the role of nephrin downregulation in podocytes. *Lisa Gianesello, Nefrologia*
17.00	Cardiovascular risk and metabolic profile in adult patients with congenital adrenal hyperplasia *Mattia Barbot, Endocrinologia*
17.10	Involvement of Oxidative Stress in the pathophysiology of Fabry disease. Is there anything to do? *Giovanni Bertoldi, Nefrologia*
17.20	Prevention and Emergency management of adrenal crisis: a multidisciplinary program of education for patients and healthcare workers. *Chiara Sabbadin, Endocrinologia*
17.30	DISCUSSION
17.50	ECM Test
18.15	Concluding Remarks and DIMAR Awards. *Roberto Vettor and Fabrizio Fabris*

NOTES

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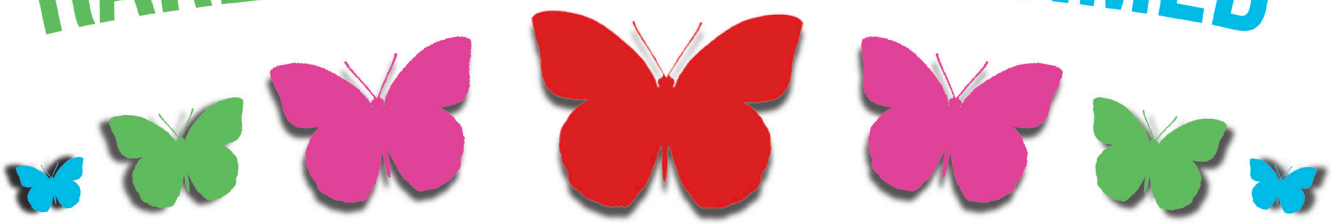
RARE DISEASE DAY IN DIMED



RESEARCH PROJECT

Modalità d'iscrizione:
il corso è gratuito con iscrizione obbligatoria attraverso il seguente link
https://forms.gle/5Y8CiPG9XUFz7SyU8

è riservato a n. 150 partecipanti per le seguenti figure professionali:
Medico Chirurgo (tutte le discipline), Biologo, Chimico, Farmacista, Tecnico Sanitario di Laboratorio Biomedico, Tecnico sanitario di radiologia medica, Psicologo, Infermiere, Infermiere pediatrico, Dietista, Fisioterapista.

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Con il patrocinio di:

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