

Convegno monotematico
Gruppo di Lavoro «Malattie Rare e Farmaci Orfani» SIF4RARE

INNOVAZIONE E MALATTIE RARE: DALLA RICERCA PRECLINICA AL PAZIENTE



23-24 Febbraio 2024
Bari, Camera di Commercio

Comitato Scientifico

Giuseppe Cirino (Presidente SIF)
Pier Luigi Canonico (Novara)
Annalisa Capuano (Napoli)
Emilio Clementi (Milano)
Annamaria De Luca (Bari)
Maurizio Taglialatela (Napoli)
Gianluca Trifirò (Verona)

Comitato organizzatore locale

*Unità di Farmacologia, Dipartimento di Farmacia – Scienze del Farmaco
Università Aldo Moro Bari*

**Annamaria De Luca, Domenico Tricarico, Sabata Pierno,
Antonella Liantonio, Paola Imbrici, Giulia Maria Camerino,
Michela De Bellis, Antonietta Mele, Ornella Cappellari,
Elena Conte, Paola Mantuano, Brigida Boccanegra**

Il convegno nasce con l'obiettivo principale di promuovere la ricerca traslazionale delle malattie rare, in particolare quelle di origine genetica non oncologiche. Le malattie rare rappresentano un importante "unmet medical need" ad elevato impatto socio-sanitario e sono numerosi gli sforzi mirati a sviluppare farmaci orfani, passando dal riposizionamento di farmaci all'identificazione di nuove molecole e terapie avanzate in grado di modificare significativamente il decorso delle patologie. Importanti traguardi sono stati raggiunti, ma numerose sono ancora le sfide aperte. È necessario identificare strategie per colmare le numerose incertezze su efficacia e sicurezza di nuovi agenti terapeutici mediante un confronto su approcci innovativi di ricerca farmacologica preclinica e clinica nonché di raccolta e analisi di dati in real-life per meglio affrontare le conseguenti problematiche di accesso e sostenibilità. In tal senso, e alla luce delle opportunità offerte dal nuovo Piano Nazionale delle Malattie Rare, è importante il dialogo da implementare, anche con il presente convegno, tra i diversi stakeholders, tra cui politici, decisori, associazioni dei pazienti, clinici e ricercatori.

Relatori e Moderatori su invito

Annemieke Aartsma-Rus, Professor of Translational Genetics, LUMC, The Netherlands
Giuseppina Annicchiarico, Coordinatrice CoReMaR_ ARESS Puglia
Maria Rosaria Bucci, Professore Farmacologia, Università Federico II Napoli
Pier Luigi Canonico, Professore Farmacologia, Università del Piemonte Orientale
Annalisa Capuano, Professore Farmacologia, Università della Campania Vanvitelli
Giuseppe Cirino, Presidente SIF, Professore Farmacologia, Università Federico II Napoli
Emilio Clementi, Professore Farmacologia, Università Statale Milano
Diana Conte, Professore Farmacologia, Università Aldo Moro Bari
Jean- Francois Desaphy, Professore Farmacologia, Università Aldo Moro Bari
Giuseppe d'Orsi, Direttore UOC Neurologia, IRCCS Casa Sollievo della Sofferenza (Fg)
Juanma Fernandez Costa, Senior Researcher, IBEC, BIST, Spain
Loretta Ferrera, Ricercatore Sanitario, Ospedale Gaslini, Genova
Fabrizio Gardoni, Professore Farmacologia, Università Statale Milano
Armando Genazzani, Professore Farmacologia, Università di Torino
Mattia Gentile, Direttore Laboratorio di Genetica Medica, Ospedale Di Venere, Bari
Marcello Gemmato, Sottosegretario alla Salute con delega alle Malattie Rare
Loreto Gesualdo, Presidente FISM, Professore di Nefrologia, Università Aldo Moro Bari
Maria Grazia Grilli, Professore Farmacologia, Università del Piemonte Orientale
Andrea Marcellusi, Presidente ISPOR Rome – Italy
Monica Montagnani, Professore Farmacologia, Università Aldo Moro Bari
Colin Nichols, Director of CIMED, Washington University St. Louis, USA
Enrico Piccinini, Coordinatore Focus Group Malattie Rare - Farmindustria
Patrizia Popoli, Direttore Centro Nazionale per la Ricerca e la Valutazione dei Farmaci, ISS - Roma
Annalisa Scopinaro, Presidente UNIAMO
Antonio Torsello, Professore Farmacologia, Università Milano Bicocca
Luigia Trabace, Professore Farmacologia, Università di Foggia
Gianluca Trifirò, Professore Farmacologia, Università di Verona
Federico Zara, Professore Genetica Medica, Ospedale Gaslini, Genova



12:00-13:15

Registration

13:15-14:00

WELCOME AND INTRODUCTION

- Giuseppe Cirino, *Presidente Società Italiana Farmacologia (SIF)*
- Annamaria De Luca, *Coordinatrice Gruppo di Lavoro SIF4RARE*
- Stefano Bronzini, *Magnifico Rettore Università degli Studi di Bari Aldo Moro*
- Marcello Gemmato, *Sottosegretario alla Salute con delega Malattie Rare*
- Michele Emiliano, *Presidente Regione Puglia*
- Filippo Anelli, *Presidente Ordine dei Medici Chirurghi e Odontoiatri (OMCeO) di Bari e Federazione Nazionale OMCeO*
- Luigi D'Ambrosio Lettieri, *Presidente Ordine Interprovinciale Farmacisti di Bari e Barletta-Andria-Trani*
- Francesco Leonetti, *Direttore Dipartimento di Farmacia – Scienze del Farmaco, Università degli Studi di Bari Aldo Moro*

14:00-15:30

ROUND TABLE

Therapy of rare diseases and innovation: science and politics at the service of patients

Chairs Pier Luigi Canonico, Annamaria De Luca

Panel Giuseppina Annicchiarico, Diana Conte, Marcello Gemmato, Armando Genazzani, Mattia Gentile, Andrea Marcellusi, Enrico Piccinini, Patrizia Popoli, Annalisa Scopinaro, Gianluca Trifirò

15:30-16:00

OPENING LECTURE

Chair Giuseppe Cirino

Antisense oligonucleotides in rare diseases: lessons learned from neuromuscular disorders to N-of-1 treatment in brain diseases

Annemieke Aartsma-Rus

16:00-16:15

Coffee break



SESSION 1

PATIENT-CENTERED DATA: FROM CLINICAL PRACTICE TO BASIC RESEARCH AND BACK
Chairs Annalisa Capuano, Emilio Clementi

16:20-16:40	<i>Invited talk</i> Autoimmune-based renal disorders: the link between basic research and clinic Loreto Gesualdo
16:40-16:55	<i>Focus on</i> Orphan drugs and advanced therapies: the value of patient-centered innovation and open challenges Patrizia Popoli
Oral Communications	
17:00-17:10	Epidemiological analyses through machine learning approaches to accelerate rare diseases diagnosis <u>Ingrasciotta Y</u> , Crisafulli S, Trifirò G (<i>Verona</i>)
17:10-17:20	Safety aspects of the gene therapy for SMA, Zolgensma®: a retrospective analysis of the European pharmacovigilance database Eudravigilance <u>Ruggiero R</u> , Balzano N, Mascolo A, di Napoli R, Capuano A (<i>Napoli</i>)
17:20-17:30	The therapeutic value of treatment for metachromatic leukodystrophy: analysis of health technology assessments of 3 European Countries <u>Gozzo L</u> , Romano GL, Longo L, Vitale DC, Drago F (<i>Catania</i>)
17:30-17:40	Exploiting routine laboratory test to identify patients with familial chylomicronemia syndrome (FCS) patients in a large Italian hospital <u>Pavanello C</u> , Pazzucconi F, Parolini M, Turri M, Mombelli GG, Castiglione S, Alberti A, De Maria R, Calabresi L (<i>Milano</i>)
17:40-17:50	Inborn errors of immunity: lessons learned from an ever evolving paradigm <u>Martire B</u> (<i>Barletta</i>)
17:50-18:00	The use of real world data for regulatory purposes in the rare diseases setting <u>Giannuzzi V</u> , Stoyanova-Beninska V, Hivert V (<i>Bari</i>)
18:00-18:20	Discussion
18:20-18:30	<i>Insight</i> The program of European Certified Pharmacologists (EuCP) Mariagrazia Grilli



POSTER VIEW AND DISCUSSION - *wine and cheese*

18:30-20:30

Poster topic 1

MUSCULAR DYSTROPHIES AND NEUROMUSCULAR DISORDERS

Chairs Maria Rosaria Bucci, Giulia Maria Camerino, Sabata Pierno

Poster topic 2

CHANNELOPATHIES

Chairs Paola Imbrici, Loretta Ferrera, Michela De Bellis

Poster topic 3

NEUROLOGIC, RENAL, METABOLIC AND OTHER RARE DISEASES

Chairs Antonella Liantonio, Antonietta Mele, Antonio Torsello

Poster topic 4

NEW MODELS AND METHODOLOGIES IN RARE DISEASES

Chairs Ornella Cappellari, Monica Montagnani, Domenico Tricarico



SESSION 2

ADVANCEMENT IN DIAGNOSIS AND PRECISION MEDICINE IN RARE DISEASES

Chairs Jean-Francois Desaphy, Armando Genazzani

8:30-8:50

Invited talk **Progresses in diagnostic-therapeutic pathways for precision medicine in rare epilepsies**
Federico Zara

8:50-9:05

Focus on **Glycogen storage disorders and drug repurposing: the example of Lafora disease**
Giuseppe d'Orsi

Oral Communications

9:10-9:20

Fluoxetine as a precision-medicine approach for epileptic encephalopathies caused by KCNT1 variants

Puzo D, Mosca I, Freri E, Ambrosino P, Belperio G, Granata T, Canafoglia L, Ragona F, Solazzi R, Filareto I, Castellotti B, Messina G, Gellera C, Trivisano M, De Dominicis A, Specchio N, DiFrancesco JC, Soldovieri MV, Taglialatela M (Napoli)

9:20-9:30

Innovative therapeutic strategies for Krabbe disease: nanoparticle-mediated enzyme replacement and autophagy induction

Del Grosso A, Carpi S, Gagliardi M, De Sarlo M, Scaccini L, Colagiorgio L, Alabed HBR, Pellegrino RM, Tonazzini I, Emiliani C & Cecchini M (Pisa)

9:30-9:40

Fenamates as ClC-1 chloride channels modulators for a potential repurposing in myotonia congenita

Saltarella I, Laghetti P, Campanale C, Ninni I, Altamura C, Desaphy JF (Bari)

9:40-9:50

ClC-39Na: a beacon of hope in the fight against Tubular Aggregate Myopathies
Pessolano E, Genazzani AA (Novara)

9:50-10:00

Reintroducing synthetic SIL1 protein to treat Marinesco-Sjogren Syndrome
Bellia E, Amodei L, Viele M, Potenza F, Ruggieri AG, Dufrusine B, Federici L, Salles M (Chieti-Pescara)

10:00-10:10

Heme oxygenase 1 a new possible target therapy against inflammation in squamous cell carcinoma in Epidermolysis Bullosa patients: a pre-clinical pilot study

Cicco G, Lospalluti L, De Marco A, Murciano M, Annicchiarico G (Bari)

10:10-10:30

Discussion

10:30-11:00

Coffee break



SESSION 3
INNOVATIVE PRECLINICAL PLATFORMS:
DRUGGABLE MECHANISMS AND VALIDATION OF NEW THERAPIES
Chairs Fabrizio Gardoni, Luigia Trabace

- 11:00-11:20** *Invited talk* **Translational research in rare diseases: old and new models**
Colin Nichols
- 11:20-11:40** *Focus on* **Patient-derived 3D skeletal muscle organoids: new platforms for studying personalized therapies in rare neuromuscular disorders**
Juanma Fernandez Costa

Oral Communications

- 11:45-11:55** **Exploiting pharmacological predictive fruit fly models for the identification of promising molecules in future Hereditary Spastic Paraplegia therapy**
Guarato G, Vantaggiato C, Dianin F, Rossato R, Gumeni S, Bassi MT, Orso G (*Padova*)
- 11:55-12:05** **Phosphodiesterases S-sulphydration contributes to human Malignant Hyperthermia**
Smimmo M, Vellecco V, Panza E, Bibli SI, Casillo GM, Villani R, Fleming I, Cirino G, Bucci M (*Napoli*)
- 12:05-12:15** **Is the D2-mdx mouse a better preclinical model for Duchenne muscular dystrophy? Insights from growth hormone secretagogues studies**
Boccanegra B, Mantuano P, Cappellari O, Tulimiero Li, Mele A, Cristiano E, Marinelli M, Conte E, Trisciuzzi D, Bresciani E, Torsello A, Denoyelle S, Nicolotti O, Liantonio A, De Luca A (*Bari*)
- 12:15-12:25** **A novel integrated pharmacological/antioxidant approach for Duchenne Muscular Dystrophy**
Perrotta C, Prata C, Zecchini S, Ottria R, Cervia D, Hrelia S, De Palma C, Clementi E (*Milano*)
- 12:25-12:35** **Drug repurposing strategy to identify novel activators of lysosomal Ca²⁺ channels via autophagy regulation in a preclinical model of amyotrophic lateral sclerosis**
Tedeschi V, Sisalli MJ, Ciancio R, Sapienza S, Castaldo A, Pannaccione A, Secondo A (*Napoli*)



12:35-12:45

Testing reliability of in vitro neuronal cultures in pharmacological treatment of a genetic form of migraine

Barbieri R, Misurale F, Alloisio S, Freilinger Tobias, Pusch M, Gavazzo P (*Genova*)

12:45-13:00

Discussion

13:00-13:30

Award ceremony for best oral communication and poster (under 38 SIF members)

Award presentation Giuseppe Cirino, Fabrizio Gardoni

Closing remarks

Giuseppe Cirino, Annamaria De Luca



Poster topic 1

MUSCULAR DYSTROPHIES AND NEUROMUSCULAR DISORDERS

Chairs Maria Rosaria Bucci, Giulia Maria Camerino, Sabata Pierno

1. Characterization of the effects of JMV2894, a synthetic growth hormone secretagogue, in a cellular model of Duchenne muscular dystrophy

Bresciani E, Rizzi L, Meanti R, Cappellari O, Mantuano P, Conte E, Sanarica F, Boccanegra B, Cerchiara AG, Liantonio A, Cantel S, Denoyelle S, Fehrentz J-A, Locatelli V, De Luca A, Torsello A

2. SRT2104, a new SIRT1 activator, is an effective metabolic enhancer that promotes muscle recovery in DMD

Giovarelli M, Zecchini S, Casati S, Clerici G, Mollica L, Cattaneo MG, Brunetti D, Banfi C, Perrotta C, De Palma C

3. SRT2104 effects on human muscle cell model of Duchenne muscular dystrophy, as a SIRT1 highly selective activator

De Santis C, Quarta R, Cristiano E, Cerchiara AG, Zecchini S, Barile SN, Lasorsa FM, Mouly V, De Palma C, Clementi E, De Luca A, Giovarelli M, Cappellari O

4. LKB1 as a novel diagnostic and therapeutic player in Duchenne muscular dystrophy: new insights from cellular and murine dystrophic models

Boccanegra B, Mantuano P, Conte E, Cerchiara AG, Tulimiero L, Quarta R, Forino M, Spadotto V, Cappellari O, Fossati G, Steinkühler C, De Luca A

5. Erucin, a natural hydrogen sulfide (H₂S) donor, improves DMD-induced SKM dysfunction

Smimmo M, Casale V, Bello I, Panza E, Bonomo M, Brancaleone V, Cirino G, Bucci M, Vellecco V

6. Dasatinib as a booster of mutation-specific molecular therapies in Duchenne muscular dystrophy: first assessment of safety in murine and human cell models

Cristiano E, Quarta R, De Santis C, Cerchiara AG, Cappellari O, Boccanegra B, Conte E, Mantuano P, De Luca A

7. Class I selective HDAC inhibitors as new potential treatment for DMD: in vivo and ex vivo readouts in D2-mdx mouse model

Tulimiero L, Boccanegra B, Licandro S, Decio A, Mantuano P, Cappellari O, De Luca A, Steinkühler C

8. Gut microbiota-endocannabinoid interplay in rare skeletal muscle myopathies: an intricate relationship that must be taken into consideration

Di Martino E, Pagano E, Panza E, Ercolano G, Silvestri C, Piscitelli F, di Marzo V, Iannotti FA

9. Targeting unfolded protein response reverts ER stress and ER Ca²⁺ homeostasis in cardiomyocytes expressing the pathogenic variant of Lamin A/C R321X

Pietrafesa G, De Zio R, Scorza SI, Armentano MF, Pepe M, Forleo C, Procino G, Gerbino A, Svelto M, Carmosino M

10. Potential application of Growth Hormone Secretagogues (GHS) for Amyotrophic Lateral Sclerosis (ALS) treatment: mechanisms of action and neuroprotective effects in human SH-SY5Y SOD1G93A cells

Meanti R, Rizzi L, Bresciani E, Licata M, Molteni L, Omeljaniuk RJ, Fehrentz J-A, Denoyelle S, Locatelli V, Torsello A

11. Effects of irisin treatment on the expression of genes associated with myogenesis, inflammation, mitochondrial metabolism, and neuroprotection in an Amyotrophic Lateral Sclerosis "in vitro" model

Carbone G, Canfora I, Conte E, Tarantino N, Camerino GM, Pierno S

12. Preclinical study showing a gender specific protective properties of conjugated linoleic acid (CLA) for amyotrophic lateral sclerosis

Bacchetti F, Bonifacino T, Torazza C, Balbi M, Ferramosca A, Tessitore S, Boccanegra B, Pierno S, Bonanno G, Bergamo P, Milanese M

13. In-vitro and in-vivo pre-clinical evidence unveiling the mGlu5 receptor as a promising pharmacological target for ALS clinical treatment

Milanese M, Bacchetti F, Bonifacino T, Torazza C, Provenzano F, Ravera S, Balbi M, Tessitore S, Ferrando S, Bonanno G

14. New therapeutic intervention for amyotrophic lateral sclerosis: analysis of the effects of acetazolamide on the biophysical properties of skeletal muscle in a transgenic mouse model

Canfora I, Tarantino N, Mantuano P, Cappellari O, Conte E, Camerino GM, Dobrowolny G, Musarò A, De Luca A, Pierno S

15. Gene therapy for the treatment of childhood SMA1: Onasemnogene abeparvovec. Observations of results of early Zolgensma administration thanks to newborn screening

Ferrante MP, Dell'Aera M, Console V, Tornabene A, Gagliardi D, Canzio E, Storelli S, Attolini E, Annicchiarico G

16. KIF5A, a protein involved in axonal transport, represents a new druggable target in a mouse model of spinal muscular atrophy

Valsecchi V, Kolici X, Baklou M, Laudati G, Brancaccio P, Pignataro G

Poster topic 2

CHANNELOPATHIES

Chairs Paola Imbrici, Loretta Ferrera, Michela De Bellis

17. Biallelic inheritance of two novel SCN1A variants results in loss of Nav1.1 channel function and developmental and epileptic encephalopathy

Dinai G, Conte E, Palumbo O, Benvenuto M, Coppola MA, Palumbo P, La Stella P, Boccanegra B, Di Muro E, Castori M, Carella M, Sciricchio V, de Tommaso M, Liantonio A, De Luca A, La Neve A, Imbrici P

18. SCN2A A1659V loss-of-function variant causes early infantile onset encephalopathy

Ferrera L, Ludovico A, Riva A, Morinelli L, Albini M, Bianchi A, Sterlini B, Lombardo G, Madia F, Lesca G, Falsaperla R, Corradi A, Zara F

19. Automated patch clamp for assessing the effects of mexiletine and its pyrrolidine derivative on Nav1.4 and Nav1.5: towards anti-myotonic drugs with improved safety profile

Cerchiara AG, Becker N, Fertig N, Cappellari O, Okeyo G, De Bellis M, Carocci A, Lentini G, Rolland J-F, Imbrici P, De Luca A

20. Combined in silico and in vitro approaches to repurpose drugs towards Kv1.1 and Kv1.2 potassium channels for epileptic encephalopathy and ataxia pharmacological treatment

Tondo AR, Trisciuzzi D, Siragusa L, D'Adamo MC, Liantonio A, De Luca A, Nicolotti O, Imbrici P

21. Structure-based identification and characterization of novel inhibitors of KNa1.1 potassium channels

Miceli F, Carotenuto L, Mosca I, Soldovieri MV, Ambrosino P, Carleo G, Iraci N, Ostacolo C, Campiglia P, Taglialatela M

22. De novo variants in KCNA3 cause developmental and epileptic encephalopathy

Belperio G, Soldovieri MV, Ambrosino P, Mosca I, Servettini I, Pietruni F, Syrbé S, Taglialatela M, Lemke JR

23. Potassium channels and TRPV1 modulators on SU-DIPG-36 and SU-DIPG-50 cells: in vitro effects on cell proliferation and channel currents characterization

Di Turi A †, Antonacci M †, Miciaccia M, Maqoud F, Perrone MG, Scilimati A, Tricarico D

24. Antiproliferative effects of tyrosine kinases (tk) inhibitors staurosporin/midostaurin on SU-DIPG cells and on cation currents: role of K⁺ channels and TRPV1 channels as drug targets

Antonacci M †, Di Turi A †, Miciaccia M, Maqoud F, Perrone MG, Scilimati A, Tricarico D

25. Kir6.1- and SUR2-dependent KATP overactivity caused intestinal tight junction protein alterations in the intestinal epithelium in murine models of Cantú syndrome

Maqoud F, Orlando A, Tricarico D, Nichols CG, Antonacci M, Russo F

26. New insights into the involvement of rare genetic variants in CLCN6 and CLCN7 associated with neurological diseases: a functional in vitro study

Coppola MA, Imbrici P, Liantonio A, Gavazzo P, Fong P, Pusch M

27. Preclinical evaluation of safinamide as an antimyotonic drug in myotonic ADR mouse model

Canfora I, Altamura C, Desaphy J-F, Vailati S, Caccia C, Padoani G, De Luca A, Pierro S

28. Functional and pharmacological characterization of sodium and chloride channel mutations in Italian families affected by non-dystrophic myotonias

Altamura C, Campanale C, Laghetti P, Ninni I, Imbrici P, Saltarella I, Desaphy J-F

Poster topic 3

NEUROLOGIC, RENAL, METABOLIC AND OTHER RARE DISEASES

Chairs Antonella Liantonio, Antonietta Mele, Antonio Torsello

29. Fingerprinting cardiolipin in leukocytes by MALDI-TOF mass spectrometry as a screening tool for Barth Syndrome

Lobasso S

30. The Keap1/Nrf2/ARE pathway as a potential therapeutic target for the treatment of Huntington's disease

Pruccoli L, Sita G, Pagliarini B, Morroni F, Tarozzi A

31. A clinical case of corpus callosum agenesis: a neuroscience multidisciplinary evaluation

Galletta D, de Bartolomeis A, Zeppetella Del Sesto FS, Marzullo A, Flace P

32. A clinical case report of cerebellar vermis hypoplasia related to deletion 15q21.3-22.31

Flace P, de Bartolomeis A, Gelato M, Zeppetella SF, Marzullo A, Galletta D

33. Calretinin in the human brain: a light microscopy immunohistochemical study

Flace P, Galletta D, Marzullo A

34. Genetic variants and inborn errors of immunity in bone marrow failure: novel potential drug targets for precision medicine

Desantis V, Andriano A, Marasco C, Pappagallo F, Di Marzo L, Ingravallo G, Di Paola R, Tabares P, Beilhac A, Vacca A, Solimando AG, Montagnani M

35. Pharmacological block of prokineticin system and microglia inhibition counteract pain in a murine model of Fabry-Anderson disease

Galimberti G, Franchi S, Amodeo G, Magni G, Riboldi B, Ceruti S, Sacerdote P

36. Exploring the link between GALC and GPR65: implications for Krabbe disease

Carpi S, Ferrero G, Del Grosso A, De Sarlo M, Colagiorgio L, Scaccini L, Battini R, Santorelli FM, Cutrupi S, Tonazzini I, Cecchini M

37. Phosphodiesterase 5 inhibitors as a new treatment for maternally inherited Leigh syndrome

Pedrotti G, Zink A, Santanatoglia C, Henke M-T, Di Donfrancesco A, Brunetti D, Decimo I, Adamo A, De Sanctis F, Tiranti V, Schuelke M, Prigione A, Bottani E

38. The β 3-AR agonist BRL37344 ameliorates the main symptoms of X-linked nephrogenic diabetes insipidus in the mouse model of the disease

Milano S, Saponara I, Gerbino A, Carmosino M, Svelto M, Procino G

39. Dual targeting of the G protein-coupled receptors CaSR and V2R for treating autosomal dominant polycystic kidney disease (ADPKD)

Di Mise A, Venneri M, Ferrulli A, Centrone M, Ranieri M, Caroppo R, Tamma G, Pellegrini L, Torres VE, Valenti G

40. What is hidden in patients with unknown nephropathy? The genetic screening could represent the missing link in the diagnosis and management in kidney transplantation

Mitrotti A, Giliberti M, di Bari I, Franzin R, Conserva F, Stea ED, Rossini M, Fiorentino M, Castellano G, Pontrelli P, Gesualdo L

41. Characterization of lipid and lipoprotein profile in Alagille syndrome

Ossoli A, Cananzi M, Turri M, Pavanello C, Belotti L, Gomaraschi M, Vidal E, Calabresi L

42. Riboflavin transporter deficiency type 2 (RTD2, OMIM #614707): a focus on the endoplasmic reticulum responses

Tolomeo M, Console L, Nisco A, Magliocca V, Persichini T, Compagnucci C, Barbaro R, Colella M, Bertini E, Massey K, Indiveri C, Barile M

43. Off-label use of liraglutide counteracts the immune dysregulation associated with Wolfram syndrome

Panfili E, Gargaro M, Orabona C, Mondanelli G, Fallarino F, Pallotta MT

44. Anti-yo mediated paraneoplastic cerebellar degeneration: a case report

Place P, Galletta D, Stucci LS, De Caro M, Mesto C, Pascazio L, Livrea P, Marzullo A, Liaci G

45. CD90-TGF β 1 co-expression promotes chemoresistance in intrahepatic cholangiocarcinoma

Pizzuto E, Mancarella S, Gigante I, Serino G, Dituri F, Giannelli G

Poster topic 4

NEW MODELS AND METHODOLOGIES IN RARE DISEASES

Chairs Ornella Cappellari, Monica Montagnani, Domenico Tricarico

46. Human cellular preclinical models to understand congenital myopathies: from pathogenic mechanism to new therapeutical targets in TAM and SEPN1-RM diseases

Conte E, Imbrici P, Dinoi G, Maggi L, De Luca A, Liantonio A

47. In vitro 3D-model of mitochondrial myopathy human skeletal muscle

Di Leo V, Tejedera A, Fernández-Garibay X, Ramón-Azcón J, Gorman GS, Russell OM, Vincent AE, Fernández-Costa J

48. Optogenetic skeletal muscle-on-chip as a drug screening and disease modelling platform for rare neuromuscular disorders.

Quarta R, Han M, De Santis C, Arduino I, Denora N, Fiermonte G, Gaio N, Rossini N, De Bellis M, De Luca A, Cappellari O

49. Characterization of cellular differentiation in an in vitro model of neuromuscular junction using a co-culture system: a preliminary study of a drug testing platform.

Canfora V, Carbone G, Pierno S, Camerino GM

50. Urine-derived stem cells and derived skeletal muscle cells as a functional model to study calcium homeostasis perturbation in neuromuscular diseases.

Talmon M, Lecchi G, Fresu LG

51. Patient-specific neuronal stem cells as an in vitro model for screening drug safety and efficacy for Aicardi-Goutières pediatric patients

Pugnetti L, Braidotti S, Ferraro RM, Irshad M, Franca R, Marinozzi V, Tommasini A, Lucafò M, Decorti G, Giliani S, Stocco G

52. Impaired bioenergetic profile in neuron progenitor cells from iPSCs of patients affected by AGC1 deficiency

Barile SN, Magnifico MC, Palmieri L, Distelmaier F, Viggiano L, Pignataro A, Petralla S, Hentschel J, Porcelli V, Poeta E, Pisano I, Fiermonte G, Anderson SA, Monti B, Lasorsa FM

53. A novel renal collecting duct model to study secondary nephrogenic diabetes insipidus associated to cystinosis

Ferrulli A, Gijsbers R, Di Mise A, Cairoli S, Van den heuvel LP, Levchenko E, Valenti G

54. A molecular docking-based virtual screening study to tackle the cure for Lafora disease

Trisciuzzi D, Imbriani P, Gambacorta N, Dinoi G, Conte E, Mantuano P, Palumbo O, Nicolotti O, Carella M, De Luca A, d'Orsi G, Altomare CD, Liantonio A

55. Computational screening for Lafora disease: repurposing of approved drugs as a novel treatment strategy

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57. Target therapy for high grade neuroblastoma treatment: integration of regulatory and scientific tools is needed

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