

International Congress

RARE DISEASES AND ORPHAN DRUGS

February 22nd – 25th, 2010

Istituto Superiore di Sanità

**Aula Pocchiari
Viale Regina Elena, 299
00161 – Roma
Italy**

February 22nd, 2010

8.30 Registration

9.30 Welcome address

Enrico Garaci

President of the Istituto Superiore di Sanità

Introduction

Ferruccio Fazio (to be confirmed)

Minister of Health

Session I

Research on rare diseases and orphan drugs: from Europe to national actions

Chairpersons:

9.45 European Council Recommendation on rare diseases

Antoni Montserrat

10.15 European actions on research for rare diseases

Catherine Barends

10.45 Coffee break

11.15 Research and orphan drugs

Guido Rasi

11.30 Research and rare diseases in Italy

Massimo Casciello

11.45 Research on rare diseases at the Istituto Superiore di Sanità: bridging molecular studies through to EUROPLAN

Domenica Taruscio

12.00 How to promote research into Rare Diseases - EURORDIS' position

Terkel Andersen

12.15 The voice of Italian patient Associations

UNIAMO (**Renza Barbon Galluppi**)

La Consulta Nazionale delle Malattie Rare (**Flavio Bertoglio**)

La Consulta nazionale delle malattie neuromuscolari (**Mario Melazzini**)

13.00 Discussion

13.30 Lunch

Session II

ERA-NET for rare diseases: E-Rare

Chairperson:

14.30 The E-Rare Network

Sophie Kutouzov

14.50 European Network on Rett Syndrome (EuroRETT)

Laurent Villard

- 15.10 European and Mediterranean network on spastic paraplegias (EUROSPA)
Alex Brice, Giovanni Stevanin
- 15.30 Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6 (SCA1, SC2, SCA3, SCA6) (RISCA)
Thomas Klockgether
- 15.50 International Kindler Syndrome Network (Kindlernet)
Leena Bruckner-Tuderman
- 16.10 Benign Familial Neonatal Seizures (BFNS) as disease model for human idiopathic epilepsies: expansion of the genotype-phenotype correlations and insights into novel disease mechanisms (EUROBFNS)
Maria Roberta Cilio, Maurizio Tagliatela
- 16.30 PodoNet: Consortium for Clinical, Genetic and Experimental Research into Hereditary Diseases of the Podocyte (PodoNet)
Franz Schaefer
- 16.50 New Genes and Therapeutic Approaches to Osteopetrosis (OSTEOPETR)
Alberto Giovanni Ugazio, Anna Maria Teti
- 17.10 International Hirschsprung Disease Consortium (HSCR)
Stanislas Lyonnet, Isabella Ceccherini
- 17.30 Understanding the WHIM syndrome and search for new therapies: molecular analysis of CXCR4 functions in leukocyte trafficking and activation (WHIMPath)
Antonella Viola
17. 50 Discussion
- 18.15 Conclusions

23rd February 2010

ASPECTS OF PATHOGENESIS

Session I

Chairpersons:

- 9.00 Cornelia de Lange syndrome: clinical and molecular data of a large Italian cohort.
Angelo Selicorni, Cristina Gervasini, Silvia Russo, Donatella Milani, Anna Cereda, Alice Passarini, Paola Castronovo, Maura Masciadri, J Azzolini, Emanuele Basile, Roberto Borgatti, Francesca Bedeschi, Vera Bianchi, Faustina Lalatta, Larizza Lidia
- 9.15 Dysregulated RAS signaling in Noonan syndrome and related disorders: disease gene discovery and functional studies
Viviana Cordeddu, Simone Martinelli, Claudio Carta, Valentina Fodale, Elisabetta Flex, Francesca Pantaleoni, Anna Sarkozy, Francesca Lepri, Giuseppe Zampino, Maria C. Digilio, Laura Mazzanti, Giovanni B. Ferrero, Cesare Rossi, Lorenzo Stella, Len A. Pennacchio, Bruce D. Gelb, Bruno Dallapiccola, Marco Tartaglia
- 9.30 Characterization of the molecular and cellular mechanisms underlying the liver pathogenesis in hemophagocytic
Antonio Sica, Angela Santoni, Luigi D. Notarangelo, Raffaele Badolato, Silvano Sozzani, Cristina Capuano, Angela Santoni, Ricciarda Galandrini
- 9.45 Molecular targets of PITX2 in the pathogenesis of Axenfeld Rieger Syndrome.
Paola Briata, Roberto Gherzi
- 10.00 Genotype/phenotype analysis of neurodegenerative and aging-prone syndromes caused by mutations in the DNA damage response/repair pathway
Domenico Delia, Luciana Chessa, Pietro Pichierri, Margherita Bignami
- 10.15 Coffee break

Session II

Chairpersons:

- 10.30 Role of the dystrophin-associated glycoprotein complex in limb-girdle and congenital muscular dystrophies: from molecular pathophysiology to potential therapy (7DR1)
Pompeo Macioce, Enzo Ricci, Andrea Brancaccio, Marina Ceccarini, Tamara C. Petrucci
- 10.45 Molecular and functional characterization of the newly identified interaction between the Rett syndrome-associated factor MeCP2 and the pro-apoptotic factor HIPK2
Silvia Soddu, Giorgia Bracaglia, Barbara Conca, Anna Bergo, Laura Rusconi, Zhaolan Zhou, Michael E. Greenberg, Nicoletta Landsberger, Charlotte Kilstrup-Nielsen
- 11.00 X-linked or autosomal rare mental retardation syndromes: phenotypic analysis in transgenic mouse models.
Giovanni Laviola, Laura Ricceri, Bianca De Filippis, Carla Perrone-Capano, Maria Giuseppina Miano
- 11.15 Study of the genetic susceptibility and environmental factor involvement in the etiopathogenesis of autism
Franca R. Guerini, Elisabetta Bolognesi, Marina Saresella, Roberta Mancuso, Sonia Usai, Salvatorica Manca, Mario Clerici
- 11.30 A novel pharmacological approach and identification of peripheral cellular biomarkers in Niemann-Pick type C disease patients
Claudio Frank, Daniele Grossi, Giovanna De Chiara, Mauro Racaniello, Giuseppe Biagini, Virginia Tancredi, Stefano Rufini, Daniela Merlo, Giovanna D'Arcangelo
- 11.45 Risk factors investigation in the pathogenesis of the bladder extrophy-epispadias complex
Sabrina Tait, Cinzia La Rocca, Alberto Mantovani

- 12.00 Cellular and animal models for the MYH9-related diseases
Massimo P. Crippa, Roberto Ravazzolo, Patrizia Noris
- 12.15 Improving diagnostic skills for inherited thrombocytopenias
Anna Savoia, Daniela De Rocco, Mariateresa Di Stazio, Federica Melazzini, Alessandro Pecci, Patrizia Noris, Carlo L. Balduini
- 12.30 Hypertrophic cardiomyopathy associated to Fabry's disease: cardiomyocytes and cardiac stem cell functional analysis
Maurizio Pesce, Antonia Germani, Roberto Gradini, Matteo Russo, Andrea Frustaci

12.45 Lunch

Session III

Chairpersons:

- 13.45 Tumor angiogenesis and inflammation as therapeutic targets of retinoblastoma
Adriana Albini, Francesca Tosetti
- 14.00 Transcriptional study of p63alpha mutants found in ectodermal dysplasia syndromes
Gerry Melino, Elena Candi, Anna Maria Lena, Alessandro Terrinoni
- 14.15 Tackling Rare Diseases Yet Lacking Diagnosis And/Or Prognosis. A Pilot Project Integrating Data Collection And Experimental Studies: the Hepatoblastoma experience
Marco Salvatore, Armando Magrelli, Mara Viganotti, Fabrizio Tosto, Gianluca Azzalin, Antonio Antocchia, Alessandra Di Masi, Rita Devito, Stefano Lorenzetti, Francesca Maranghi, Giuseppe Macino, Alberto Mantovani, Caterina Tanzarella, Domenica Taruscio
- 14.30 Genetic, molecular and functional characterization of Cockayne syndrome, a rare transcription/repair defective hereditary disease
Guido Frosina, Eugenia Dogliotti, Elena Botta, Angelo Calcagnile, Paolo Degan, Mariarosaria D'Errico, Mara Foresta, Tiziana Lemma, Laura Narciso, Tiziana Nardo, Roberta Oneda, Donata Orioli, Monica Ropolo, Miria Stefanini
- 14.45 Genetic heterogeneity of hereditary predisposition to breast cancer: characterization of molecular markers at both somatic and at germline level.
Manuela Gariboldi, Paolo Peterlongo, Monica Barile, Loris Bernard, Irene Catucci, Elisabetta Crippa, Loris De Cecco, Giovanna De Vecchi, Stefano Fortuzzi, Laura Galastri, Lara Lusa, Siranoush Manoukian, Edoardo Marchesi, Bernard Peissel, Valeria Pensotti, Sara Pizzamiglio, Fernando Ravagnani, James. F. Reid, Laura Tizzoni, Paolo Verderio, Sara Volorio, Carlo M. Croce, Marco A. Pierotti, Paolo Radice
- 15.00 Trauma and risk of amyotrophic lateral sclerosis
Ettore Beghi, Elisabetta Pupillo, Giancarlo Logroscino, and Adriano Chiò for the EURALS Consortium
- 15.15 A genome wide non-synonymous SNP scan of amyotrophic lateral sclerosis (ALS)
Vincenzo Silani, Isabella Fogh, Antonia Ratti, Nicola Ticozzi, Cinzia Gellera, Ferdinando Squittirei
- 15.30 Coffee break
- 15.45 Preclinical studies aimed to develop target genes-based therapies for the treatment of amyotrophic lateral sclerosis.
Caterina Bendotti, Marco Peviani, Dario Lidonnici, Massimo Tortarolo, Tiziana Borsello, Roberto Piva
- 16.00 Dynamic NAD(P)H post-synaptic autofluorescence signals for the assessment of mitochondrial function in a neurodegenerative disease: monitoring the primary motor cortex of G93A mice, an amyotrophic lateral sclerosis model
Stefano Loizzo, Massimo Pieri, Alberto Ferri, Maria Teresa Carri, Cristina Zona, Andrea Fortuna, Stefano Vella
- 16.15 Evaluation of genetic and environmental factors in a cohort of twins with amyotrophic lateral sclerosis
Lorenza Nisticò, Maurizio Leone, the Italian Study Group on ALS affected Twins

- 16.30 Gastroesophageal reflux in systemic sclerosis (SSC): relationship with pulmonary involvement
Camilla Gambaro, Laura Belloli, Roberta Barbera, Nicoletta Carlo Stella, Paoletta Preatoni, Bianca Marasini, Alberto Malesci
- 16.45 The Italian External Quality Assessment (EQA) in genetic tests: the VI EQA scheme
Vincenzo Falbo, Giovanna Florida, Marco Salvatore, Manuela Marra, Fabrizio Tosto, Federica Censi, Domenica Taruscio
- 17.00 Rare cancers in Italy
Gemma Gatta, Annalisa Trama, Stefano Ferretti, Lisa Licitra, Paolo Casali, Paolo Angelo Dei Tos, Riccardo Capocaccia, Roberta De Angelis, Sandra Mallone, Mariano Santaquilani, Andrea Tavilla, the RITA working group

DRAFT

24th February 2010

DIAGNOSIS

Session I

Chairpersons:

- 8.30 Usefulness of MLPA in the molecular diagnosis of lissencephaly and neuronal migration disorders
Davide Mei, Elena Parrini, Carla Marini and Renzo Guerrini
- 8.45 Genomic diagnosis and classification of rare disorders with mental retardation using high throughput technologies
Laura Bernardini, Rita Mingarelli, Bruno Dallapiccola
- 9.00 Usefulness of 244K array-CGH in the ascertainment of Copy Number Variations in Mental Retardation
Corrado Romano, Santina Reitano, Donatella Greco, Pinella Failla, Daniela Di Benedetto, Ornella Galesi, Lucia Castiglia, Mirella Vinci, Valeria Chiavetta, Giuseppa Ruggeri, Franco Calì, Marco Fichera
- 9.15 Metachromatic Leukodystrophy: new insights in the genotype-phenotype correlation and in the natural history of the disease
Maria Sessa, Mirella Filocamo, Alessandra Biffi, Stefano Regis, Martina Cesani, Francesca Fumagalli, Serena Grossi, Cristina Baldoli, Ubaldo Del Carro, Luigi Naldini, Giancarlo Comi
- 9.30 Genotype-phenotype correlations in the CMT neuropathies: definition of a clinical and genetic diagnostic flow-chart.
Alessandra Bolino, Sara Benedetti, Emanuela Di Pierri, Lara Piantoni, Federica Cerri, Marina Scarlato, Raffaella Fazio, Giancarlo Comi, Maurizio Ferrari, Stefano Previtali, Angelo Quattrini
- 9.45 Type 1 neurofibromatosis: an integrated program of clinical and molecular diagnostics
Donatella Bianchessi, Federica Natacci, Daria Riva, M. Eoli, Ettore Salsano, Veronica Saletti, Sara Guzzetti, Francesca Orzan, Franca Lalatta, Gaetano Finocchiaro
- 10.00 Callosal agenesis: a brain malformation with polygenic origin. Identification of candidate genes and loci through a multidisciplinary approach of clinical, cytogenetic and molecular studies of a large set of patient with corpus callosum anomalies
Susan Marelli, Rita Grasso, Clara Bonaglia, Marianna Rusconi, Roberto Giorda, Maria Teresa Bassi, Renato Borgatti
- 10.15 Autosomal recessive spastic paraplegia with thinning of corpus callosum and periventricular white matter changes. clinical, molecular, and neuroimaging studies
Filippo M. Santorelli, Paola S. Denora, Alessandra Tessa, Gabriella Silvestri, Federico Zara, Francesco G. Garaci, Giovanni Stevanin
- 10.30 Development of new diagnostic approaches for transmissible spongiform encephalopathies
Franco Cardone, Serena Principe, Piero Parchi, Gianluigi Zanusso, Salvatore Monaco, Fabrizio Tagliavini, Maurizio Pocchiari
- 10.45 Coffee break

Session II

Chairpersons:

- 11.00 Reliability and efficacy of the current diagnostic approach in narcolepsy and search for new genetic markers
Paolo Bosco, Giuseppe Plazzi, Camilla Bernardini, Fabrizio Michetti, Christian Franceschini, Raffaele Ferri
- 11.15 Evaluation and rehabilitation of swallowing dysfunction in patients with rare neurological disorders and movement disorders: first year of study
Fabrizio Stocchi, Davide Tufarelli, Laura Vacca, Eugenio Mercuri

- 11.30 Identification of genetic factors responsible for rare disorders with congenital heart defects
Anna Sarkozy, Alessandro De Luca, Francesca Lepri, Valentina Guida, Rosangela Ferese, Federica Consoli, Maria Lisa Dentici, Chiara Iannascoli, Maria Cristina Digilio, Bruno Marino, Marco Tartaglia, Bruno Dallapiccola
- 11.45 New Genetic Syndromes with aortic tortuosity and dissection
Maurizia Grasso, Fabiana Gambarin, Roberto Dore, Nicola Marziliano, Valentina Favalli, Alessandra Serio, Michele Pasotti, Eliana Disabella, Elena Antoniazzi, Mario Mosconi, Savina Mannarino, Attilio Odero, Duke Cameron, Luca Vricella, Harry C Dietz and Eloisa Arbustini
- 12.00 Systematic diagnosis of rare erythroenzymopathies: generation of guidelines and study of the genotype/phenotype correlation
Wilma Barcellini, Paola Bianchi, Elisa Fermo, Cristina Vercellati, Anna Paola Marcello, Giovanna Valentini, Alberto Zanella
- 12.15 Both familial and sporadic atypical hemolytic uremic syndrome are associated with genetic complement abnormalities and the genotype greatly impact outcome and response to treatment
Marina Noris, Jessica Caprioli, Elena Bresin, Chiara Mossali, Gaia Pianetti, Sara Gamba, Erica Daina, Chiara Fenili, Annalisa Sorosina, Rossella Piras, Giuseppe Remuzzi
- 12.30 Circulating microparticles and thrombin generation capacity in severe hemophiliacs with mild clinical phenotype: diagnostic and prognostic implications
Elena Santagostino, Maria Elisa Mancuso, Armando Tripodi, Veena Chantarangkul, Andrea Artoni, Gianluigi Pasta, Marigrazia Clerici, Lidia Padovan, Pier Mannuccio Mannucci
- 12.45 Establishment of a European Network of Rare Bleeding Disorders
Flora Peyvandi, Roberta Palla, Marzia Menegatti, Simona Maria Siboni, Majda Benedik-Dolnicar, Christoph Bidlingmaier, Tiraje Celkan, Paul Giangrande, Ruth Gilmore, Jorgen Ingerslev, Katheline Peerlink, Helen Pergantou, Florence Suzan, Paolo Lanzi
- 13.00 Lunch
- Session III**
Chairpersons:
- 14.00 Development of novel molecular approaches for the diagnosis and characterization of genetic lymphedema
Elisabetta Dejana, Fabrizio Orsenigo, Andrea Caprini, Francesco Bertolini
- 14.15 Investigation of genetic and epigenetic mechanisms underlying Beckwith-Wiedemann Syndrome (BWS) on a large cohort of Italian patients
Silvia Russo, Flavia Cerrato, Angela Sparago, Serena Ferraiuolo, Francesca Bedeschi, Faustina Lalatta, Donatella Milani, Valentina Giorgini, Angelo Selicorni, Luigi Fedele, Andrea Riccio, Lidia Larizza
- 14.30 Genetic and clinical aspects of rare lymphomas
Elisabetta Caprini, Francesca Sampogna, Marie Perez, Marcella Visentini, Cristina Cristofolletti, Diego Arcelli, Paolo Fadda, Mauro Helmer Citterich, Valeria Tocco, Armando Magrelli, Federica Censi, Paola Torreri, Marina Frontani, Enrico Scala, Giuseppe Alfonso Lombardo, Domenica Taruscio, Massimo Fiorilli, Giandomenico Russo
- 14.45 Biomarkers in thymic epithelial tumors (TET). An immunohistochemistry-based tissue micro array (TMA) multicenter study with preliminary genetic data on the epidermal growth factor receptor (EGFR) gene in TET
Mirella Marino, Rossano Lattanzio, Salvatore Conti, Libero Lauriola, Robert Martucci, Enzo Gallo, Amelia Evoli, Giorgio Palestro, Roberto Chiarle, Daniele Remotti, Roberto Pisa, Massimo Martelli, Stefano Ascani, Francesco Puma, Luigi Ruco, Erino Rendina, Mauro Truini, Gianni Tunesi, Antonella Barreca, Stefano Sioletic, Iaria Bravi, Francesco Facciolo, Sandro Carlini, Marcella Mottolese, Anna Di Benedetto, Giovannella Palmieri, Pierluigi Granone, Mauro Antimi, Maurizio Lalle, Anna Ceribelli, Massimo Rinaldi, Giuseppina Chichierchia, Gerardina Merola, Luigi Petillo, Raffaele Perrone Donnorso, Mauro Piantelli
- 15.00 Cytogenetic and molecular analysis of rare salivary gland tumors
Giovanna Floridia, Federica Censi, Stella Lanni, Manuela Marra, Vincenzo Falbo, Domenica Taruscio

- 15.15 Clinical and genetic features of parathyroid carcinomas
Sabrina Corbetta, Valentina Vaira, Silvano Bosari, Alfredo Scillitani, Vito Guarnieri, Anna Spada
- 15.30 Therapy-oriented large scale genomic and gene expression analysis in thymomas, mesotheliomas and lung carcinoids
Francesca Toffalorio, Elena Belloni, Caterina Fumagalli, Elvira Gerbino, Carla Micucci, Simone Paolo Minardi, Gabriele Bucci, Giuliana Pelicci, Giuseppe Pelosi, Lorenzo Spaggiari, Filippo de Braud, and Tommaso De Pas
- 15.45 A multidisciplinary approach for the investigation of Hyperparathyroidism-Jaw tumour syndrome
Luisa Barzon, Giulia Masi, Maurizio Iacobone, Giovanni Viel, Gennaro Favia, Andrea Porzionato, Veronica Macchi, Raffaele De Caro, Giorgio Palù

Session IV

Chairpersons:

- 16.00 Diagnostic and therapeutic target of systemic amyloidosis: validation of new diagnostic tools and development of new disease models
Giampaolo Merlini, Francesca Lavatelli, Laura Obici, Giovanni Palladini, Mario Nuvolone, Simona Donadei, Sofia Giorgetti, Palma Mangione, Sara Raimondi, Monica Stoppini, Vittorio Bellotti
- 16.15 Inherited epithelial adhesion diseases: development and validation of diagnostic tools and guidelines, and assessment of quality of life
Daniele Castiglia, Marina D'Alessio, Naomi De Luca, Cristina Di Pietro, Francesca Sampogna, Stefano Tabolli, Marina Colombi, May El-Hachem, Paolo Salerno, Domenica Taruscio, Giovanna Zambruno
- 16.30 Biochemical and cellular real-time biomarkers of diagnostic and prognostic value in the management of Kawasaki's disease
Donatella Pietraforte, Elisabetta Straface, Alessio Metere, Lucrezia Gambardella, Luciana Giordani, Maria Giovanna Quaranta, Elisabetta Cortis, Alberto Villani, Alessandra Marchesi, Giuseppe Palumbo, Domenico Del Principe, Marina Viora, Maurizio Minetti, Walter Malorni
- 16.45 Genetic analysis of arrhythmogenic inherited diseases
Elena Sommariva, Sara Benedetti, Francesca Zuffada, Chiara Di Resta, Chiara Redaelli, Simone Sala, Maurizio Ferrari, Carlo Pappone
- 17.00 Development of an epidemiological and molecular integrated approach for the prevention of congenital hypothyroidism
Roberto Cerone, Mario De Felice, Roberto Di Lauro, Emanuela Medda, Luca Persani, Domenica Taruscio, Massimo Tonacchera, Fabrizio Bianchi, Fabrizio Minichilli, Antonella Olivieri
- 17.15 Infant botulism
Lucia Fenicia, Fabrizio Anniballi, Dario De Medici, Elisabetta Delibato, Bruna Auricchio, Carlo Locatelli, Davide Lonati

25th February 2010

TREATMENT AND CLINICAL MANAGEMENT

Session I

Chairpersons:

- 8.30 Electrophysiological and pharmacological analysis of striatal neurons from mice expressing torsin A with the DYT1 dystonia mutation
Annalisa Tassone, Graziella Madeo, Rosa Luisa Potenza, Patrizia Popoli, Paola Platania, Giuseppe Sciamanna, Dario Cuomo, Giuseppina Martella, Paola Bonsi, Antonio Pisani
- 8.45 Testing in vitro and in vivo treatments for inclusion body myositis
Simona Saredi, Claudia Di Blasi, Pia Bernasconi, Lucia Morandi, Renato Mantegazza, Marina Mora, Cristina Sancricca, Enzo Ricci, Pietro Attilio Tonali, Massimiliano Mirabella
- 9.00 Enzyme Replacement Therapy in Late-Onset Phenotypes of Glycogenosis type 2
Bruno Bembi, Marco Confalonieri, Federica Edith Pisa, Giovanni Ciana, Agata Fiumara, Rossella Parini, Miriam Rigoldi, Arrigo Moglia, Alfredo Costa, Cesare Danesino, Maria Gabriela Pittis, Andrea Dardis, Sabrina Ravaglia
- 9.15 Adipose tissue-derived stem cells for the treatment of muscular dystrophy
Antonietta Gentile, Iliara Gatto, Gabriele Toietta, Maurizio C. Capogrossi, Giuliana Di Rocco
- 9.30 Evaluation of the pathogenetic role and pharmacology of isolated human TSC2 smooth muscle cells. Novel therapeutic perspectives
Alfredo Gorio, Stephana Carelli, Vera Grande, Silvia Ancona, Diego Brancaccio, Maria Paola Canevini, Anna Maria Di Giulio, Elena Lesma, Eleonora Isaia, Eloisa Chiaramonte, Raffaella Adami, Silvia Maria Sirchia, Silvano Bosari, Guido Coggi, Rosa Maria Alfano, Filippo Ghelma, Emanuele Montanari
- 9.45 Mesenchymal stem cells for the treatment of tibial congenital pseudarthrosis associated with type I neurofibromatosis.
Donatella Granchi, Valentina DeVescovi, Elisa Leonardi, Serena R. Baglio, Onofrio Donzelli, Marina Magnani, Armando Giunti, Nicola Baldini
- 10.00 New cell therapy approaches for infantile malignant osteopetrosis
Alfredo Cappariello, Anna Berardi, Barbara Peruzzi, Andrea Del Fattore, Alberto Ugazio, Gian Franco Bottazzo, Anna Teti, Barbara Tondelli, Harry Blair, Matteo Guerrini, Kenneth Patrene, Barbara Cassani, Paolo Vezzoni, Franco Lucchini
- 10.15 Characterization of new genetic defects in MAD
Giovanna Lattanzi, Cristina Capanni, Elisabetta Mattioli, Marta Columbaro, Rosalba Del Coco, Daria Camozzi, Elisa Schena, Maria Rosaria D'Apice, Anne Vielle, Francesca Lombardi, Francesca Gullotta, Diana Postorivo, Anna Maria Nardone, Monica D'Adamo, Paolo Sbraccia, Stefano Squarzone, Giuseppe Novelli, Nadir Mario Maraldi
- 10.30 Clinical and diagnostic course for patients affected by Prader Willi Syndrome
Tiziana Gregg, Konstantinos Martikos, Georgios Bakaloudis, Francesco Vommaro, Mario Di Silvestre, Giovanni Barbanti Brodano, Stefano Giacomini, Alfredo Cioni, Emanuela Pipitone, Luca Sangiorgi, Stefano Lari, Patrizio Parisini

10.45 Coffee break

Session II

Chairpersons:

- 11.00 Cytokine-based immunotherapy and tumor microenvironment in models of cutaneous and ocular melanoma
Anna Rubartelli, Laura Borsi, Barbara Canemolla, Carlo Mosci, Silvano Ferrini
- 11.15 Multidrug resistance dependence on cytoskeleton integrity in solid tumours
Daria Brambilla, Stefano Fais

- 11.30 Immunobiologic and clinical activity of DNA hypomethylating agents in human sarcomas
Luca Sigalotti, Giulia Parisi, Alessia Covre, Francesca Colizzi, Elisabetta Fratta, Hugues JM Nicolay, Sandra Coral, Vincenzo Canzonieri, Michele Maio
- 11.45 Innovative Burkitt's lymphoma therapy
Giovanna Cutrona, Lidia C Boffa, Maria Rita Mariani, Serena Matis, Manlio Ferrarini
- 12.00 Targeting the prognostic and metastasis-predicting surface proteoglycan NG2 for immunotherapeutic treatment of selected sarcomas
Roberto Perris, Sabrina Cattaruzza, Pier Andrea Nicolosi, Maria Teresa Mucignat, Katia Lacrima, Nicoletta Bertani, Laura Pazzaglia, Maria Serena Benassi, Lucia Sigalotti, Massimo Guidoboni, Michele Maio, William B Stallcup, Piero Picci
- 12.15 Innovative management of patients with diffuse malignant peritoneal mesothelioma
Marcello Deraco, Federica Perrone, Maria Rosaria Balestra, Dario Baratti, Antonello Domenico Cabras, Antonino Carbone, Maria Grazia Daidone, Genny Jocollè, Shigeki. Kusamura, Barbara Laterza, Marzia Pennati, Silvana Pilotti, Raffaella Villa, Nadia Zaffaroni
- 12.30 Phenotype correction of ADAMTS13 deficiency and protection from the development of thrombotic thrombocytopenic purpura through intravascular and skeletal muscle ADAMTS13 gene delivery in mice
Piera Trionfini, Susanna Tomasoni, Miriam Galbusera, Roberta Donadelli, Daniela Corna, Lorena Zentilin, David Motto, Mauro Giacca, Giuseppe Remuzzi, Ariela Benigni
- 12.45 New therapeutical approaches in the human beta-thalassemia treatment: in vitro and in vivo studies.
Ann Zeuner, Ornella Morsilli, Monica Bartucci, Luca Pasquini, Nadia Maria Sposi, Marta Baiocchi, Adriana Massa, Paolo Cianciulli, Ugo Testa, Cesare Peschle, Ruggero De Maria, Marco Gabbianelli
- 13.00 Effects of the inhibition of PDGFR phosphorylation on the activation of circulating platelets and neutrophils in patients with systemic sclerosis: results from a double-blind placebo-controlled clinical trial
Armando Gabrielli, Giovanni Pomponio, Paolo Fraticelli, Michele Luchetti, Silvia Svegliati, Gianluca Moroncini, Roberto Giacomelli, Paola Cipriani, Alessandra Marrelli, Vasiliki Liakouli, Elisa Pingiotti, Vincenza Dolo, Danilo Millimaggi, Sandra D'Ascenzo, Iaria Giusti, Serena Guiducci, Marco Matucci-Cerinic, Sergio Generini, Gianfranco Ferraccioli, Barbara Tolusso, Maria De Sanctis, Walter Malorni, Anna Maria Giammarioli, Elisabetta Straface, Marina Pierdominici, Angela Maselli, Laura Somma, Serena Vettori, Giuseppina Abignano, Gabriele Valentini, Patrizia Rovere-Querini, Stefano Franchini, Norma Mageri, Angelo Andrea Manfredi, Maria Grazia Sabbadini
- 13.15 Autoimmune pemphigus: dynamics of autoreactive B cells, an integrated database and diagnostic/therapeutic recommendations
Giuseppe Cianchini, Stefano Tabolli, Giovanni Di Zenzo, Damiano Abeni, Giovanna Zambruno, Antonio Lanzavecchia, Biagio Didona
- 13.30 Conclusion and general remarks
- 13.45 Lunch
- 14.30 ECM questionnaire

DRAFT

GENERAL INFORMATION

Scientific Director

Domenica Taruscio
Centro Nazionale Malattie Rare
Istituto Superiore di Sanità, Roma

Scientific Committee

Stephen Groft, Domenica Taruscio, Marina Ceccarini, Tamara Petrucci, Giandomenico Russo

Scientific Secretariat

Marco Salvatore, Paola Torreri

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Organising Secretariat

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Language

The official language is English.

ECM credits (for Italian participants only)

For Italian participants, ECM accreditation has been requested.

Certificate of attendance

A certificate of attendance will be provided upon request at the end of the Conference



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DOMANDA DI ISCRIZIONE A CONVEGNI/CONGRESSI ISS
A iscrizione gratuita

Istruzioni per la compilazione della domanda:

- 1) Scrivere in modo leggibile
- 2) Le domande illeggibili non potranno essere considerate
- 3) Indicare il titolo e la data della manifestazione al quale si richiede di partecipare
- 4) Compilare tutti i campi richiesti

TITOLO DELLA MANIFESTAZIONE: RARE DISEAS AND ORPHAN DRUGS

N° ID della manifestazione: 035D10 DATA DI SVOLGIMENTO: dal 22/02/10 al 25/02/10

DATI PERSONALI

Cognome:	Nome:	
Data di nascita (gg/mm/aa):	Luogo di nascita:	(Prov.)
Stato (se diverso da ITALIA)	Genere:	M <input type="checkbox"/> F <input type="checkbox"/>
Indirizzo: Via		
Città:	Provincia:	
CAP:	Regione:	
Telefono:	Fax:	
Posta elettronica:	Codice Fiscale:	

DATI PROFESSIONALI

Titolo di Studio:	
Profilo sanitario ECM:	Richiede attestato ECM se previsto: SI <input type="checkbox"/> No <input type="checkbox"/>
Ente di appartenenza:	
Indirizzo: Via	
Città:	Provincia:
CAP:	Regione:
Telefono:	Fax:

NB: questi recapiti verranno utilizzati dalla segreteria per le future comunicazioni



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DOMANDA DI ISCRIZIONE A CONVEGNI/CONGRESSI ISS
A iscrizione gratuita

POSIZIONE

Tipologia dell'Ente (Az. Sanitaria, Ministero etc.):		
Inquadramento nell'Ente:		
Di ruolo:	SI <input type="checkbox"/> No <input type="checkbox"/>	Se di ruolo, specificare quale:

MODALITA' DI ISCRIZIONE, SELEZIONE E AMMISSIONE:

La domanda, debitamente compilata, deve essere stampata, firmata e inviata al numero di fax 06 4990 4370 entro il 15 febbraio 2010.

Firma.....

Data.....

INFORMATIVA AI SENSI DEL D.LGS. 196/2003:

La informiamo che i dati raccolti verranno trattati e conservati secondo correttezza e utilizzati esclusivamente a fini amministrativi concernenti il rapporto di collaborazione con questo Istituto di cui alla presente richiesta. Non saranno oggetto di comunicazione né di diffusione. In ogni momento ha facoltà di esercitare i diritti previsti dall'articolo 13 della predetta legge ed in particolare chiedere la correzione, integrazione o cancellazione degli stessi. Si informa inoltre che il titolare del trattamento è l'Istituto Superiore di Sanità.

Firma.....

Data.....