International Conference
Conferenza internazionale

Rare Diseases and Orphan Drugs
Malattie rare e farmaci orfani

November 5-8 2007
5-8 novembre 2007

Organised by / *Organizzato da*
National Institute of Health
Istituto Superiore di Sanità

Aula Pocchiari
Istituto Superiore di Sanità
Viale Regina Elena, 299 – Rome

International Conference on Rare Diseases and Orphan Drugs / *Conferenza internazionale su malattie rare e farmaci orfani*

November 5-6, 2007
5-6 Novembre, 2007

Day 1: November 5th - 5 Novembre

8.30 Registration / *Registrazione*
9.30 Welcome speech / *Saluto di benvenuto*
E. Garaci
President of Istituto Superiore di Sanità / *Presidente dell'Istituto Superiore di Sanità*

Session I / I *Sessione* (9.45 – 13.00)

Round Table

Rare diseases in Italy: actions to tackle rare diseases and orphan drugs
Malattie rare in Italia: azioni per affrontare le malattie rare ed i farmaci orfani
Chairperson / *Moderatore*: **E. Garaci**

9.45 Priority actions on rare diseases and orphan drugs
Priorità nel settore delle malattie rare e dei farmaci orfani
Minister of Health / *Ministro della Salute*
Sen. Livia Turco
Patient's needs: report of the Consulta working groups
I bisogni dei pazienti: presentazione dei gruppi di lavoro della Consulta Nazionale delle Associazioni dei Pazienti di malattie rare
Representatives of the Italian "Consulta" of Patients' Associations / *Rappresentanti della Consulta Nazionale delle Associazioni dei Pazienti di malattie rare*

Participants / *Partecipanti:*
Representatives of the Ministry of Health / *Rappresentanti del Ministero della Salute*
F. Palumbo, G. Zotta
Italian Medicinal Agency / *Agenzia Italiana del Farmaco*
N. Martini
Interregional Co-ordination / *Coordinamento Interregionale*
M. Romeri
National Institute of Health / *Istituto Superiore di Sanità*
D. Taruscio

12.15 Discussion / *Discussione*

13.00 Lunch / *Pausa pranzo*

Session II / *II Sessione* (14.00 –17.30)

A focus on the European initiatives and perspectives
Un focus sulle iniziative e gli orientamenti europei
Chairperson / *Moderatore*: **G. Tarsitani**

14.00 EU Commission priorities and actions on rare diseases
Le priorità e le nuove attività della Commissione Europea sulle malattie rare
K. Freese

15.00 National plans for rare diseases: an upcoming priority for EU Member States?
Piani Nazionali per le Malattie Rare: la prossima priorità per i Paesi Membri?

15.00 Bulgaria **(R. Stefanov)**
15.20 France **(C. Nourissier)**
15.40 Portugal **(L. Nunes)**

16.00 Recommendations for rare diseases national plans development
Raccomandazioni per la definizione di piani nazionali per le malattie rare
D. Taruscio

16.20 The revision of the International Classification of Diseases: an opportunity for rare diseases coding and classification
Il processo di revisione della classificazione internazionale delle malattie: un'opportunità per migliorare la codifica e la classificazione delle malattie rare
S. Aymé

16.40 Discussion / *Discussione*

17.30 Conclusions / *Conclusioni*

Day 2: Novembre 6th - 6 Novembre

Session I / I Sessione (9.00 – 13.30)

Promoting international collaboration for rare diseases and orphan drugs research
Promuovere la collaborazione internazionale per la ricerca sulle malattie rare ed i farmaci orfani
Chairpersons / *Moderatori:* **G. D'Agnolo, D.Taruscio**

9.00 Research priorities on rare diseases in the FP 7
FP7: le priorità della ricerca sulle malattie rare
C. Berens

9.20 Building on success: the example of E-Rare
Un esempio di collaborazione internazionale: il progetto E-Rare
I. Beitia Ortiz de Zàrate

9.40 ERA-Net plus and rare diseases
Le malattie rare nel contesto dello schema "ERA-Net plus"
B. Wetterauer

10.00 The point of view of industries representatives
Il punto di vista dei rappresentanti dell'industria
Interventi preordinati

10.30 Discussion / *Discussione*

11.00 Coffee break / *Pausa caffè*

11.15 Examples from Member States

11.15 Independent Research in Italy
Il bando per la ricerca indipendente promosso dall'Agenzia Italiana del Farmaco
G. Traversa

11.30 The ISS-NIH call for proposals in Italy
Il bando per la ricerca promosso nell'ambito della collaborazione ISS-NIH
D. Taruscio, S. Groft

11.45 Research initiatives in Spain
Le iniziative per la ricerca sulle malattie rare in Spagna
M. Posada de la Paz

12.00 Research National Initiatives on rare diseases in Germany
Le iniziative per la ricerca sulle malattie rare in Germania
R. Schuster

12.15 Research National Initiatives on rare diseases in The Netherlands
Le iniziative per la ricerca sulle malattie rare in Olanda
S. van Weely

12.30 Discussion / *Discussione*

13.30 Lunch / *Pausa pranzo*

Session II / *II Sessione* (14.30 – 18.30)

Orphan drugs: emerging issues and initiatives
Farmaci orfani: problematiche emergenti ed iniziative
Chairpersons / *Moderatori:* **C. Tomino, S. Vella**

14.30 Eurordis survey on orphan drugs availability in Europe
Indagine di Eurordis sulla disponibilità di farmaci orfani in Europa
F. Bignami

14.50 How many orphan drugs in the coming years?
Quanti farmaci orfani nei prossimi anni?
F. Bignami, A. Trama

15.10 How many orphan drugs in Italy?
Quanti farmaci orfani sono disponibili in Italia?
P. Folino Gallo, C. Tomino

15.30 The Italian Orphan Drugs Register
Il Registro Italiano dei Farmaci Orfani
D. Pierannunzio, A. Trama

15.50 Coffee break / *pausa caffè*

16.00 Rare diseases treatment costs challenges in Italy: some examples
I costi del trattamento della malattie rare: alcuni esempi

16.00 Regione Lombardia
G. Baraldo

16.15 Regione Piemonte
D. Roccatello, S. Baldovino

16.30 Regione Sicilia
L. Borsellino

16.45 Regione Toscana
L. Giorni

17.00 Regione Veneto
P. Facchin

17:15 I.R.C.C. S. "Burlo Garofolo"
R. Paparazzo

17.30 Discussion / *Discussione*

18.30 Conclusions / *Conclusioni*

Workshop

"Presentation of research projects funded in the frame of the bilateral Italy (ISS) – USA (NIH – Office for rare diseases) agreement on joint research and development of public health actions on rare diseases"

"Presentazione dei progetti finanziati nell'ambito dell'accordo bilaterale Italia (ISS) – USA (NIH – Office for rare diseases) su ricerca e sviluppo di azioni di sanità pubblica sulle malattie rare"

November 7-8, 2007
7-8 Novembre, 2007

Day 3: November 7th - 7 Novembre

9.00 Welcome and objectives of the workshop
Saluto di benvenuto e obiettivi del workshop
E. Garaci, S. Groft
Director of Office for Rare Diseases, National Institute of Health
President of Istituto Superiore di Sanità
Presidente dell'Istituto Superiore di Sanità

Session I / I Sessione (9.30 – 13.00)

Aspects of pathogenesis / *Patogenesi*
Chairpersons / *Moderatori*:
F. Belardelli, M. Pocchiari

9.30 β-dystrobrevin interaction with iBRAF: a new role for dystrobrevin in neuronal differentiation
B. Artegiani, C. Labbaye, P. Torreri, M.T. Quaranta, C. Ramoni, M. Ceccarini, T.C. Petrucci, P. Macioce

9.50 Physical and functional interaction between the Rett Syndrome-associated factor Mecp2 and the Pro-Apoptotic factor HIPK2
G. Bracaglia, B. Conca, S. Giglio, F. Moretti, C. Kilstrup-Nielsen, N. Landsberger, S. Soddu

10.10 A novel pharmacological approach and identification of peripheral cellular biomarkers in Niemann-Pick C disease patients
C. Frank, S. Rufini, D. Merlo, G. Biagini, G. D'Arcangelo

10.30 Pharmacological and genetic regulation of TSC2-/- cell phenotype. A novel insight for TSC and LAM
A. Gorio

10.50 Coffee break and parallel poster sessions / *Pausa caffè e sessioni poster parallele*

11.20 Study of the genetic susceptibility and environmental factor involvement in the etiopathogenesis of autism
F.R. Guerini, M. Chiappedi, A. Ghezzo, E. Maggioni, P. Spelta, S. Manca

11.40 Does NMMHCIIA (MYH9) play a role as a transcriptional regulator?
C. Ferrai, V.M. Diaz, A. Bachi, F. Blasi, M.P. Crippa

12.00 Molecular genetics of infantile Pompe Disease in Italy
S. Dominissini, M. G. Pittis, A. Dardis, A.L. Montalvo, M. Donnarumma, M. Stroppiano, G. Ciana, M. Di Rocco, B. Bembi, M. Filocamo

12.20 Thrombin generation in severe haemophiliacs with different clinical phenotype
E. Santagostino, M.E. Mancuso, A. Tripodi, V. Chantarangkul, M. Clerici, S. M. Siboni, P.M. Mannucci

12.40 Molecular approaches for the diagnosis of genetic lymphedema
A. Caprini, F. Orsenigo, M. François, P. Koopman, E. Dejana

13.00 Lunch / Pausa pranzo

13:30 Parallel poster sessions / *Sessioni poster parallele*

Session II / II Sessione (14.30 – 18.00)

Pathogenesis and diagnosis / *Patogenesi e diagnosi*
Chairpersons / *Moderatori*:
T. C. Petrucci, G. Zambruno

14.30 Clinical and genetic evaluation of a large sample of patients with cerebellar ataxia syndromes
S. Bonato, M.G. D'Angelo, C. Megliani, A.C. Turconi, R. Borgatti, C. Zucca, L. Villa, A. Tonelli, G. Airoldi, C. Crimella, P.L. Baron, G. Silvestri, M.T. Bassi, N. Bresolin

14.50 Metachromic leukodystrophy – gene sequencing provides further evidences of genotype-phenotype correlation
A. Biffi, M. Cesani, F. Fumagalli, U. del Carro, C. Baldoli, S. Gerevini, G. Comi, M.G. Roncarolo, M. Filocamo, S. Regis, M. Sessa

15.10 Autosomal recessive spastic paraplegia with thinning of corpus callosum and periventricular white matter changes: clinical, molecular and neuroimaging studies
P. S. Denora, A. Tessa, G. Silvestri, E. Bestini, F. Zara, F. Garaci, F. M. Santorelli

15.30 Development of new diagnostic approaches for transmissible spongiform encephalopathies
F. Cardone, P. Parchi, G. Zanusso, S. Monaco, F. Tagliavini, M. Pocchiari

15.50 Coffee break and parallel poster sessions / *Pausa caffè e sessioni poster parallele*

16.20 Role and prevalence of GATA4, NKX2.5, FOG2 and novel candidate genes' mutations in specific subsets of congenital heart defects
A. Sarkozy, F. Lepri, G. Esposito, R. Ferese, F. Consoli, B. Marino, M. Tartaglia, M.C. Digilio, B. Dallapiccola

16.40 Activating mutations in SOS1 cause a distinctive form of Noonan syndrome
M. Tartaglia, L.A. Pennacchio, V. Fodale, F. Lepri, M.L. Dentici, A. Sarkozy, S. Martinelli, C.a Neri, C. Carta, F. Pantaleoni, V. Petrangeli, V. Cordeddu, M. Magliozzi, C. Zhao, K. K. Yadav, B. Pandit, K. Oishi, W. Schackwitz, A. Ustaszewska,

J. Martin, J. Bristow, A. Selicorni, O. Gabrielli, F. Faravelli, M.C. Digilio, G. Zampino, B. Dallapiccola, D. Bar-Sagi, B. D. Gelb

17.00 Tackling rare diseases yet lacking diagnosis and/or prognosis: a pilot project integrating data collection and experimental studies
D. Taruscio, A. Antoccia, G. Azzalin, R. Devito, A. Di Masi, C. La Rocca, S. Lorenzetti, G. Macino, A. Magrelli, A. Mantovani, F. Maranghi, C. Tanzarella, F. Tosto, M. Salvatore, S. Tait

17.20 The TP53 NOINS-PRO haplotype is not associated with breast and ovarian cancer in hereditary cases negative for mutations in BRCA1 and BRCA2
P. Peterlongo, G. De Vecchi, P. Verderio, S. Pizzamiglio, S. Manoukian, L. Bernard, V. Pensotti, S. Volorio, F. Ravagnani, M. Pierotti, P. Radice

17.40 Discussion / *Discussione*

18.00 Conclusions and general remarks / *Conclusioni*

Day 4: November 8th - 8 Novembre

Session I / I Sessione (9.00 – 13.30)

Diagnosis / *Diagnosi*
Chiarpersons / *Moderatori*:
S. Groft, L. Larizza

9.00 Genomic diagnosis and classification of rare disorders with mental retardation using high throughput technologies
B. Dallapiccola, L. Bernardini, A. Novelli

9.20 Is the "Chromosomal Phenotype" a clue for the diagnosis of de novo cryptic chromosomal rearrangements in people with mental retardation?
C. Romano, F. Calì, S. Reitano, D. Greco, P. Failla, V. Chiavetta, P. Schinocca, O. Galesi, D. Di Benedetto, L. Castiglia, R. Ciccone, M. Fichera, O. Zuffardi

9.40 Genotype-phenotype correlations in the CMT neuropathies: definition of a clinical and genetic diagnostic flow-chart
S. Coviello, A. Colombo, S. Benedetti, I. Spiga, P. Dacci, R. Fazio, M. Ferrari, G. Comi, S. P revitali, A. Bolino, A. Quattrini

10.00 Genotype/phenotype correlation in CDLS: Italian experience
A. Selicorni, S. Russo, C. Gervasini, D. Milani, F. Menni, M. Cerutti, P. Castronovo, M.Masciadri, A. Musio, L. Larizza

10.20 Alteration of striatal synaptic activity in a mouse model of DYT1 dystonia
G. Sciamanna, P. Bonsi, G. Martella, D. Cuomo, P. Platania, A. Tassone, P. Popoli, G. Bernardi, A. Pisani

10.40 Coffee break and parallel poster sessions / *Pausa caffè e sessioni poster parallele*

11.10 Genotype/phenotype analysis of neurodegenerative and aging-prone syndromas caused by mutations in the DNA damage response/repair pathway
D. Delia, P. Pichierri, M. Bignami, L. Chessa

11.30 CS-B patients with unusual clinical features
T. Nardo, R. Oneda, D. Orioli, E. Botta, H. Fawcett, A.R. Lehmann, M. Stefanini

11.50 Clinical, genetic and morphological investigation of HRPT2-related familial hyperparathyroidism
G. Masi, L. Barzon, M. Iacobone, G. Viel, A. Porzionato, V. Macchi, R. De Caro, G. Favia, G. Palù

12.10 Salivary gland tumors: comparative genomic hybridization in paraffine-embedded samples
G. Floridia, F. Censi, M.P. Foschini, V. Falbo, D. Taruscio

12.30 Classification of parathyroid tumours for optimal detection of HRPT2 mutations
V. Guarnieri, A. Scillitani, C. Battista, M. Bisceglia, R. Viti, I. Chiodini, M. Iacobellis, N. Malavolta, S. Minisola, L.A. Muscarella, L. D'Agruma, M. Carella, D.E.C. Cole, A. Spada

12.50 Lunch / *Pausa pranzo*

13:30 Parallel poster sessions / *Sessioni poster parallele*

Session II / II sessione (14.30 – 18.00)

Treatment and clinical management / *Trattamento e gestione clinica*
Chairpersons */ Moderatori:*
G. Russo, S. Vella

14.30 Adipose tissue-derived stem cells for the treatment of muscular dystrophy
I. Gatto, A. Gentile, S. Straino, A. Mangoni, M.C. Capogrossi, G. Di Rocco

14.50 Testing in vitro and in vivo treatments for inclusion body myositis
S. Saredi, C. Di Blasi, P. Bernasconi, L. Morandi, R. Mantegazza, M. Mora, C. Sancricca, E. Ricci, P.A. Tonali, M. Mirabella

15.10 Autoimmune pemphigus: quality of life, alternative therapeutic approaches and dynamics of autoreactive B cells
G. Cianchini, S. Tabolli, G. Di Zenzo, D. Abeni, G. Zambruno, A. Lanzavecchia, B. Didona

15.30 Stimulation of erythropoiesis and fetal hemoglobin reactivation induced by stem cell factor in human β-thalassemia
M. Gabbianelli, O. Morsilli, A. Massa, L. Pasquini, P. Cianciulli, U. Testa, C. Peschle

15.50 Coffee break and parallel poster sessions / *Pausa caffè e sessioni poster parallele*

16.20 Mesenchymal stem cells for the treatment of tibial congenital pseudarthrosis associated with type I neurofibromatosis
D. Granchi, V. DeVescovi, E. Leonardi, S.R. Baglio, O. Donzelli, M. Magnani, N. Baldini

16.40 Tumor angiogenesis and inflammation as a therapeutic target of Retinoblastoma and other rare ocular tumors
A. Albini, R. Venè, G. Fassina, M. Nicolò, R. Cammarota, M. Barberis, D.M. Noonan, G. Arena, F. Tosetti

17.00 Novel experimental approaches for investigation on new therapies against rare human bone tumors
A. De Milito, F. Lozupone, R. Canese, M. Marino, K. Scotlandi, F. Podo, S. Fais

17.20 Innovative Burkitt's Lymphoma therapy
G. Cutrona, Lidia C. Boffa, M.R. Mariani, S. Matis, M. Ferrarini

17.40 Therapy-oriented large scale genomic and gene expression analysis in thymomas, mesotheliomas and lung carcinoids
E. Belloni, F. Toffalorio, G.Pelosi, G. Veronesi, L. Spaggiari, T. De Pas, G. Pelicci, F. De Braud

18.00 Conclusions and general remarks / *Conclusioni*

Poster Session / Sessioni Poster
Novembre 7th – 8th / 7 - 8 Novembre

Improving diagnostic skills for inherited thrombocytopenias: identification of "novel" forms and characterization of "classical" forms to develop a DNA microchip
C.L. Balduini, C. Ambaglio, F. Faletra, A. Savoia

Gastroesophageal reflux disease in patients with systemic sclerosis: any relationship between reflux and pulmonary involvement?
R. Barbera, C. Gambaro, L. Belloli, B. Marasini, A. Malesci

Systematic diagnosis of rare erythroenzymopathies: generation of guidelines and study of the genotype/phenotype correlation
W. Barcellini, P. Bianchi, E. Fermo, G. Valentini, A. Zanella

Preliminary results on knowledge about reproductive risks in families with a child affected by Cornelia De Lange syndrome
M.F. Bedeschi, V. Bianchi, F. Lalatta, D. Milani, F. Menni, M. Cerutti, A. Selicorni

Trauma and risk of amyotrophic lateral sclerosis: a population-based case-control study
Ettore Beghi, Andrea Millul, Adriano Chiò, Giancarlo Logroscino

Preclinical studies aimed to develop target genes-based therapies for the treatment of amyotrophic lateral sclerosis
C. Bendotti, M. Peviani, T. Borsello, R. Piva

Dysbindin, the product of the DTNBP1 gene associated to schizophrenia susceptibility and mutated in HPS7 syndrome, interacts with dystrobrevin: possible involvement in intracellular trafficking and signalling
S. Benvegnù, C. Bucalossi, P. Torreri, G. Macchia, P. Macioce, T. C. Petrucci, M. Ceccarini

Type 1 Neurofibromatosis: advanced diagnostics
D. Bianchessi, F. Orzan, M.R. Balestrini, S. Guzzetti, D. Riva, F. Nataci, F. Lalatta, G. Finocchiaro

Natural history of CMT1A including qol within 2 year
P. Caliandro, C. Pazzaglia, D. Pareyson, I. Aprile, T. Cavallaro, A. Quattrone, N. Rizzuto, G. Vita, P. Tonali, A. Schenone, L.Padua, Italian CMT QoL Study Group

Variables influencing quality of life and disability in CMT patients: italian multicenter study
P. Caliandro, C. Pazzaglia, D. Pareyson, I. Aprile, T. Cavallaro, A. Quattrone, N. Rizzuto, G. Vita, P. Tonali, A. Schenone, L. Padua, Italian CMT QoL Study Group

Efficient molecular diagnostics and guidelines for rare genodermatoses affecting epithelial adhesion
D. Castiglia, M. Castori, G. Floriddia, M. D'Alessio, M. El Hachem, M. Colombi, D. Taruscio, P. Salerno, G. Zambruno

Development of an epidemiological and molecular integrated approach for the prevention of congenital hypothyroidism: a model for other rare diseases
R. Cerone, M. De Felice, R. Di Lauro, E. Medda, L. Persani, M. Tartaglia, D. Taruscio, M. Tonacchera, A. Olivieri

Cytoreductive surgery and hyperthermic intraperitoneal chemotherapy in the treatment of diffuse malignant peritoneal mesothelioma
M. Deraco, S. Kusamura, D. Baratti, A.D. Cabras, N. Zaffaroni

Characterization of a novel splicing mutation causing glycogen storage disease type II
S. Dominissini, A.L. Montalvo, E. Buratti, M.G. Pittis, B. Bembi, A. Dardis

The Italian external quality assessment in genetic tests: development of a web-based system
V. Falbo, G. Floridia, M. Salvatore, F. Tosto, F. Censi, D. Taruscio

Rare diseases: infant botulism
L. Fenicia, F. Anniballi, D. De Medici, E. Delibato, D. Lonati, C.Locatelli

Reliability and efficacy of the current diagnostic approach in narcolepsy and search for new genetic markers
R. Ferri, P. Bosco, O. Bruni, L. Ferini-Strambi, G. Plazzi

Inhibition of PDGFR phosphorylation: a pathogenetic treatment of systemic sclerosis
A. Gabrielli, G. Pomponio, P. Fraticelli, M. Luchetti, S. Svegliati, G. Moroncini, R. Giacomelli, P. Cipriani, A. Marrelli, V. Liakouli, E.Pingiotti, V. Dolo, D. Millimaggi, S. D'Ascenzo, I. Giusti, S. Guiducci, M. Matucci-Cerinic, S. Generini, G. Ferraccioli, B. Tolusso, M.a De Sanctis, W. Malorni, A.M. Giammarioli, E. Straface, M. Pierdominici, A. Maselli, L. Somma, S. Vettori, G. Abignano, G. Valentini, P. Rovere-Querini, A.A. Manfredi, M.G. Sabbadini

Surveillance of rare cancers in Europe
G. Gatta, R. Capocaccia, L. Ciccolallo, S. Sowe

Neural tube defects and folic acid: an integrated, evidence-based approach to primary prevention in the italian context
M. Grandolfo, F. Baldi, S. Brescianini, A. Mantovani, D. Taruscio

New genetic syndromes with aortic tortuosity and dissection
M. Grasso, N. Marziliano, E. Disabella, M. Diegoli, E. Porcu, A. Pilotto, M. Tagliani, M. Concardi, M. Agozzino, F. Inzani, M. Pasotti, A. Serio, F. Gambarin, S. Mannarino, A. Brega, E. Arbustini

Clinical and diagnostic course for patients affected by Prader Willi syndrome
T. Greggi, L. Sangiorgi, E. Pedrini, P. Parisini

A family-based linkage analysis of HLA and 5-HTTLPR gene polymorphisms in Sardinian children with autism spectrum disorder
F.R. Guerini, S. Manca, S. Sotgiu, S. Tremolada, M. Zanzottera, C. Agliardi, L. Zanetta, M. Saresella, R. Mancuso, A. De Silvestri, M.L. Fois, G. Arru, P. Ferrante

Is there a role of oxidative DNA damage in Cockayne Syndrome?
T. Lemma, P. Degan, A. Calcagnile, L. Narciso, M. Stefanini, M. D'Errico, E. Dogliotti

Proposal for an integrated approach to rare diseases: a study between basic laboratory models and clinical epidemiology in Amyotrophic Lateral Sclerosis (ALS)
A. Loizzo, D. Taruscio, L. Nisticò, M. Vichi, E. Beghi, A. Chiò, G. Campana, C. Campanella, S. Conti, L. Costa, R. Cotichini, S. D'Alfonso, M. Leone, G.Logroscino, S. Loizzo, A. Fortuna, M. Masocco, L. Mazzini, N. Nasuelli, C. Petrini, Paolo Salerno, S. Spampinato, V. Toccaceli, N. Vanacore

Compound heterozygosity for mutations in LMNA in a patient with a myopathic and lipodystrophic mandibuloacral dysplasia type A phenotype
F. Lombardi, F. Gullotta, M. Columbaro, A. Filareto, M. D'Adamo, A. Vielle, V. Guglielmi, A.M.Nardone, V. Azzolini, E. Grosso, G. Lattanzi, M.R. D'Apice, S. Masala, N.M. Maraldi, P. Sbraccia, G. Novelli

Biochemical and cellular real-time biomarkers of diagnostic and prognostic value in the management of Kawasaki's and Henoch-Schonlein Purpura diseases
W. Malorni, M. Viora, M. Minetti, D. del Principe

Anti-tumor activity of the new dual PI3K/MTOR inhibitor, NVP-BEZ235, in sarcomas
M.C. Manara, S. Nosari, D. Zambelli, G. Nicoletti, L. Landuzzi, M. Sauveur-Michel, C. García-Echeverría, P. Picci, K. Scotlandi

The Bladder Extrophy-Epispadias complex and exogenous risk factors: the blade project
A. Mantovani, C. La Rocca, M. Luconi, E.M. Faustman, S. Lorenzetti, M. Maggi, S. Tait

Callosal agenesis: a brain malformation with poligenic 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
S. Marelli, R. Grasso, C. Bonaglia, R. Giorda, M.T. Bassi, R. Borgatti

Preliminary data of a tissue microarray (TMA) - based multicenter study of thymic epithelial cell tumors with clinical implications
M. Marino, L. Lauriola, R. Martucci, G. Chichierchia, G. Palmieri, F. Facciolo, P. Granone, L. Ruco, E. Rendina, S. Ascani, I. Bravi, F. Puma, G. Merola, R. Perrone Donnorso, M. Piantelli, A. Evoli

Diverse mechanisms underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes
S. Martinelli, P. Torreri, M. Tinti, L. Stella, G. Bocchinfuso, E. Flex, A. Grottesi, A. Palleschi, G. Cesareni, B.D. Gelb, L. Castagnoli, T.C. Petrucci, M. Tartaglia

Usefulness of MLPA in the molecular diagnosis of lissencephalies and neuronal migration disorders: high diagnostic yield in p>a lissencephaly
D. Mei, E. Parrini, S. Gana, C. Marini, R. Guerrini

From proteomic to structural biology: a comprehensive approach to amyloid diseases
G. Merlini, L. Obici, G. Palladini, F. Lavatelli, M. Nuvolone, S. Giorgetti, S. Raimondi, M. Stoppini, V. Bellotti

Cytokine-based immunotherapy in preclinical models of cutaneous and ocular melanoma
C. Mosci, B. Carnemolla, L. Borsi, A. Rubartelli, S. Ferrini

Genetic abnormalities of complement regulatory molecules in hemolytic uremic syndrome N. 526D/9
C. Mossali, G. Pianetti, F. Castelletti, J. Caprioli, E. Bresin, G. Monteferrante, G. Remuzzi, M. Noris

Characterization of the molecular and cellular mechanisms underlying the liver pathogenesis in Hemophagocytic Syndromes (HS)
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N. Zaffaroni, M.G. Daidone, R. Villa, D. Baratti, S. Kusamura, M. Deraco

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