ERDERA

European Rare Diseases
Research Alliance

Establish a solid Network: A perspective from a Charity

Eleonora Passeri

May 2nd 2025

Athens, Greece

Co-funded by the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.
Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them.

Disclaimer

Ministero della Salute

Coordinator of the Comitato Nazionale Malattie Rare (CoNaMR)

R.S.P.
RARE SPECIAL POWERS

Founder and President of the Charity Association Rare Special Powers (RSP)

My Personal Journey

From Academia and Bench Research…

McLean Hospital
HARVARD MEDICAL SCHOOL AFFILIATE

Georgetown University

JOHNS HOPKINS
MEDICINE

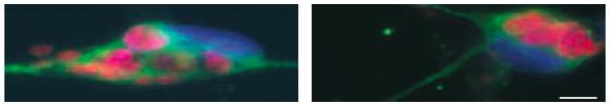
EBERHARD KARLS
UNIVERSITÄT
TÜBINGEN

UNIVERSITÀ
CATTOLICA
del Sacro Cuore

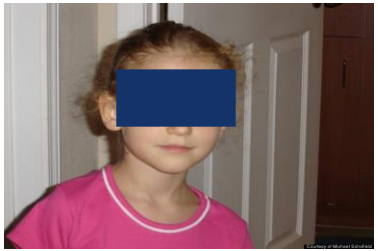
Neuroscientist – SfN 2014, Washington DC

The “turning points”

Cells (Induced Neurons, iN cells)



Children with
Childhood Onset Schizophrenia



The Johns Hopkins Schizophrenia Center hosts:

Mind the Gap

aspiring to bring clinicians and scientists together

2013-2014 Schedule

September 25th 2013, 4-5pm:
Professor Paul McHugh, M.D.
Meyer 8 Library

October 30th 2013, 4-5pm:
Psychiatric Epidemiology
Hongxing Wang, M.D.,Ph.D. & Paul Nestadt, M.D.
Meyer 8 Library

December 18th 2013, 4-5pm:
Comorbidity with Physical Disorders
Mari Kondo, Ph.D. & Matthew Peters, M.D.
Meyer 8 Library

January 29th 2014, 4-5pm:
Scientific Medication
Mehdi Omidvari, M.D. & Elizabeth Wise, M.D.
Meyer 8 Library

March 26th 2014, 4-5pm:
Pregnancy and Mental Illness
Lindsay Hayes, Ph.D. & Zina Meriden, M.D.
Meyer 8 Library

April 30th 2014, 4-5pm:
Stigma in Psychiatry
Nao Gamo, Ph.D. & Sarah Ramsay, M.D.
Meyer 8 Library

May 28th 2014, 4-5pm:
Mental State Examination in Transculturally Psychiatry
Teppei Tanaka, M.D. & Helen Bellete, M.D.
Meyer 8 Library

The Johns Hopkins Schizophrenia Center hosts:

Mind the Gap

aspiring to bring clinicians and scientists together

2014-2015 Schedule

November 11, 2014, 1-2pm:
Structuring a Department of Psychiatry for Clinical Practice and Research
Professor Paul McHugh, M.D.
Meyer 8 Library

December 16, 2014, 1-2pm:
Dimensional Perspective: Cognitive decline, mental retardation, and its effects on life.
Zachary Cordner, M.D./Ph.D. student & Elizabeth Prince, D.O.
Phipps 240

January 13, 2014, 1-2pm:
Life Story Perspective: The microbiome, diet, and mental disorders.
Geetha Kannan, Ph.D. & Traci Speed, M.D., Ph.D.
Harvey / Nelson 712

February 10, 2014, 1-2pm:
Behavioral Perspective: Marijuana and Psychosis.
Ross McFarland, Ph.D. student & Paul Nestadt, M.D.
Phipps 360

March 24, 2014, 1-2pm:
Disease Perspective: Sex Differences in Schizophrenia.
Joelle Lavoie, Ph.D. & Jacob Taylor, M.D., M.H.S.
Phipps 340

April 7, 2014, 1-2pm:
Neurodevelopment: A Fifth Perspective vs. RDoc Framework
Paul R. McHugh, MD, J. Raymond DePaulo Jr. MD, James C.O. Harris, MD, Solomon H. Snyder, MD, Solange Brown, MD, PhD. Chair: Akira Sawa, M.D., Ph.D.
Phipps 240

See the persons behind the “patients”

Patients are *not* numbers

Rare Disease (RD) in numbers

- 7.000 – 8.000 RDs
- 80% are genetic and 20% the etiology is not yet well defined (e.g. Infections, environmental toxic factors, etc.)
- 50-70% of patients are children (onset during childhood)
- 50% are undiagnosed
- Strong association between RDs and neurodevelopment disorders
- 95% do NOT have a cure

Health & Rights

FIFTY-FOURTH WORLD HEALTH ASSEMBLY **WHA54.21**

Agenda item 13.9 **22 May 2001**

International classification of functioning, disability and health

The Fifty-fourth World Health Assembly,

1. ENDORSES the second edition of the International Classification of Impairments, Disabilities and Handicaps (ICIDH), with the title International Classification of Functioning, Disability and Health, henceforth referred to in short as ICF;

2. URGES Member States to use the ICF in their research, surveillance and reporting as appropriate, taking into account specific situations in Member States and, in particular, in view of possible future revisions;

3. REQUESTS the Director-General to provide support to Member States, at their request, in making use of ICF.

Ninth plenary meeting, 22 May 2001
A54/VR/9

http://www.un.org/esa/socdev/enable/rights/convtexte.htm
6 December 2006

United Nations Convention on the Rights of Persons with Disabilities

Preamble

The States Parties to the present Convention,

(a) *Recalling* the principles proclaimed in the Charter of the United Nations which recognize the inherent dignity and worth and the equal and inalienable rights of all members of the human family as the foundation of freedom, justice and peace in the world,

(b) *Recognizing* that the United Nations, in the Universal Declaration of Human Rights and in the International Covenants on Human Rights, has proclaimed and agreed that everyone is entitled to all the rights and freedoms set forth therein, without distinction of any kind,

(c) *Reaffirming* the universality, indivisibility, interdependence and interrelatedness of all human rights and fundamental freedoms and the need for persons with disabilities to be guaranteed their full enjoyment without discrimination,

(d) *Recalling* the International Covenant on Economic, Social and Cultural Rights, the International Covenant on Civil and Political Rights, the International Convention on the Elimination of All Forms of Racial Discrimination, the Convention on the Elimination of All Forms of Discrimination against Women, the Convention against Torture and Other Cruel, Inhuman or Degrading Treatment or Punishment, the Convention on the Rights of the Child, and the International Convention on the Protection of the Rights of All Migrant Workers and Members of Their Families,

Reality…

Health economics

AIMS

- Inform
- Educate
- Raise awareness

- On Rare Diseases and Neurodevelopmental Disorders
- Relevant stakeholders

R.S.P.
RARE SPECIAL POWERS

RUNTS (DD n. 6680, 20/06/2023)

CF 94183560542

Literally “build” THE Network

The RSP strategy…

National and European Affiliations

Consolidate RSP in such scenarios.

Nationally – Italy

RSP is affiliated to…

In Europe

RSP is a member of..

Agreements with Healthcare professional associations

Nurses

18 Different healthcare professionals, such as technical radiologist, dieticians, speech therapists, etc.

Pharmacists

Physioterapists

Agreements with other RDs patient associations

Establish a Network with other RDs stakeholders

I

II

III

Municipalities

Direct collaborations with about 20 Umbrian Municipalities for the Light a Monument project carried out toghether with Anffas per Loro patient association for 2 Awareness days, the:

- Rare Disese Day – Feb. 28th/29th
- Autism- April 2nd

ERDERA

European Rare Diseases
Research Alliance

Scientific Committee

To align RSP actions with the scientific community and build up a social participation in scientific contents where the voice of persons can be heard and included.

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Spread RSP voice!

Communication strategy

How, who, when.

Il Rubino – Local magazine in Assisi – Every month

Avis Assisi magazine (blood donor association) – Every #3 months

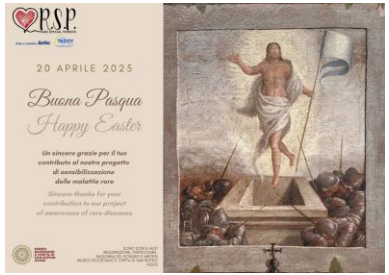
Umbria Network web-radio - Every month

Social media: Facebook, Instagram, YouTube, Whatsapp, LinkedIn

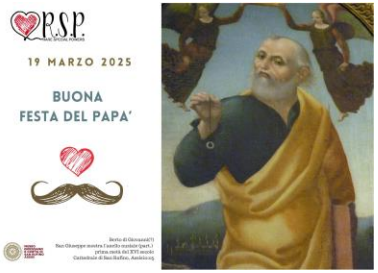
Science & Art

To disseminate scientific contents reaching out to new and unaware audiences.

Rare Art project in collaboration with the Museo Diocesano di San Rufino in Assisi (PG) - Italy



MUSEO DIOCESANO
E CRIPTA DI SAN RUFINO
ASSISI



RSP Supports... project carried out with more than 10 Italian artists

Other initiatives and actions

Organize and participation to events, fund raise initiatives, and much more…

Upcoming…

Klaipeda University

EUCONEXUS

SOCIAL INNOVATIONS FOR SUSTAINABLE REGIONAL DEVELOPMENT

RSP as a facilitator actor

Build Connections, establish Networks, consolidate Relationships

Solve the unsolved: Communication issues

- Different languages among the scientific community
- Different language between scientists/researchers and general society
- Lay language to connect with patients (e.g., red flags)

Tools and challenges

Opportunities vs limits. Pros & Cons.

Social media? Website? Newsletter? Events? Brochure/Flyers? Oral/poster presentation?

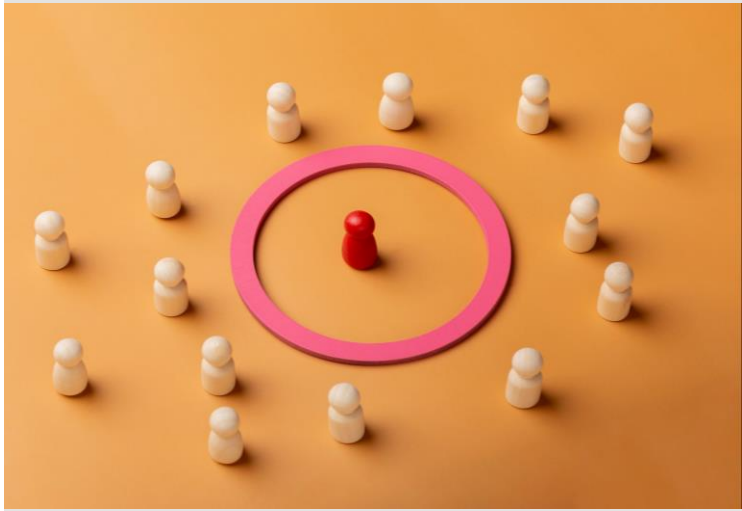
Language barrier (especially in Europe)

Scientific terminology

Time you have

Engange with stakeholders

- Scientific Community (MDs? PhD?)
- Patients/patient orgs
- Policy makers
- Press media
- Lay audience



Are you talking about YOUR project or are you the “voice” of someone else (e.g., organization, institution, company)?

We are in this together

Photo credit: Erica Baciocchi

Patient Advocacy

Alone you can go faster, but together you can go further.

It is all about…

Empathy

Building social connections

The change starts today, it starts with each of us taking actions

Photo credit: Alessia Torregrossa

Some suggestions…

- Reduce the fragmentations
- Joint calls
- Share good practices
- Collaborate
- Engage
- Learn from the past, especially from our mistakes
- Share knowledge, experience, expertise, skills, *HOPE and DREAMS*
- Lead by example
- Empowerment and support (e.g. patients, women)

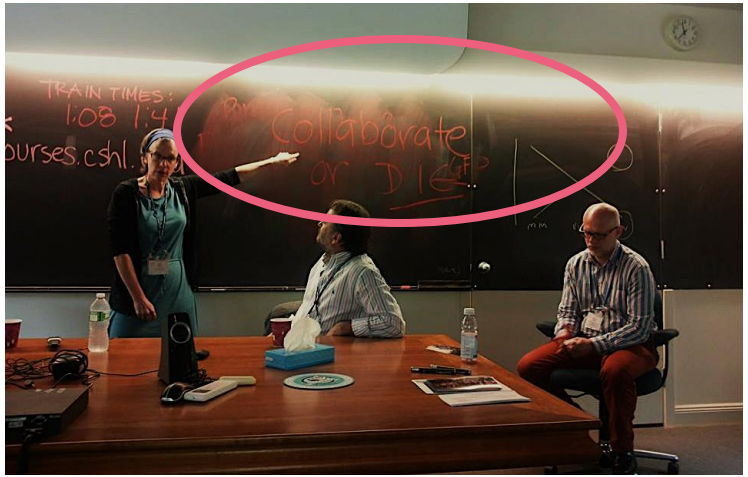
Virtual circle

- Awareness
- Information
- Knowledge
- Learning
- Share
- Network

Butterfly wing effect

Photo credit: Alessia Torregrossa

Take home messages…



Think outside the box…

…There is no box at all.

European Rare Diseases Research Alliance

Thank you!

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rarespecialpowers@pec.it

drpas84ele@gmail.com

RSP website is under construction! Stay tuned!

RARE SPECIAL POWERS

WhatsApp channel

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