



Short Communication

## Ring14 International: Development of a National- Based Patient Association towards a “Global” Network Initiative to Fight a Chromosomal Disorder

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### Abstract

Chromosome 14 syndromes are rare genetic disorders. In some cases, one of the two chromosomes assumes the form of a ring resulting in “Ring14 syndrome,” characterized by early onset refractory epilepsy, motor and mental retardation, and a multitude of other physical challenges. In other chromosomal syndromes, the 14<sup>th</sup> pair loses or transfers genetic material (deletions or duplications) but often share a similar clinical picture. Ring14 Association, founded in Italy in 2002, has recently evolved into Ring14 International to facilitate the networking of different local chapters located worldwide with the mission to advance biomedical research towards best care and possible cure.

### Keywords

Ring chromosome 14; Chromosome 14 syndromes; Rare genetic disease; Patient advocacy group; Promotion of scientific activities

### Introduction

A group of families with children suffering from a rare genetic disease, called Ring14 Syndrome, founded Ring14 Italy as a non-profit organization in Reggio Emilia in May 2002. Ring14 Syndrome results from a circularization (ring) of the 14th chromosome, a genetic anomaly caused by the loss of chromosome distal ends and fusion of remaining portions [1]. Ring14 Italy choose to advocate for this syndrome and, indeed, all syndromes arising from anomalies on the 14th chromosome and has, for last ten years, successfully tried to improve the quality of life for all affected patients and their families.

Currently there are no effective therapies, there is no cure for these syndromes and families, and doctors have few tools at their disposal to establish a comprehensive description of symptoms and determine best treatments. Our organization has been the only reference point for hundreds of families around the world: we are present in five continents, 23 countries and we communicate in five languages. In addition to providing support to the families as they care for their children, Ring14 Italy has also promoted and funded

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biomedical research for rare diseases - a key role of patient advocacy organizations [2]. Since its founding, Ring14 Italy has consistently sustained and supported scientific projects. Indeed, Ring14 Italy has sponsored ambitious basic-science projects, such as the creation of Ring14 models (i.e. the first mouse model or stable iPSC cell lines) and the gene expression profiling of immortalized Ring14 cells, as well as funding phenotypic [3,4] and language development [5,6] studies.

### Ring14 International

About a decade later, these medical advances inspired the creation of Ring14 International, founded in 2014, through the merging of Ring14 Italy and Ring14 USA. The aim of this new organization is to promote the creation and coordinate the action of independent national chapters, which gather individual families from single countries. In particular, all research initiatives will be organized, funded and managed at the international level.

This new organizational structure will help assist families, divulge information better, and allow us to administer our resources more efficiently. This year, chapters representing other countries, such as France, United Kingdom, Spain and Netherlands have joined Ring14 International and many others are expected in future (Figure 1). Ring14 International, founded by families of people affected by the Ring14 Syndrome, aims to become the leading advocacy association representing any person affected by a 14<sup>th</sup> chromosomal rearrangement, even the most rare (and often nameless) syndromes.

### Scientific advisory board

In order to better promote research, Ring14 International has created an international Scientific Advisory Board (SAB) and appointed a scientific coordinator. The 1<sup>st</sup> SAB committee, which will be renewed on a 3-year basis, is chaired by Prof. Giovanni Neri (Università Cattolica in Rome, Italy) and composed by Profs. Pietro DeCamilli (Yale University, USA), Andrew DeWoody (Purdue University, USA), Stylianos Antonarakis (University of Geneva, Switzerland), and Phil Hieter (University of British Columbia, Canada). The SAB together with the scientific coordination office will be responsible for managing world scientists in order to develop

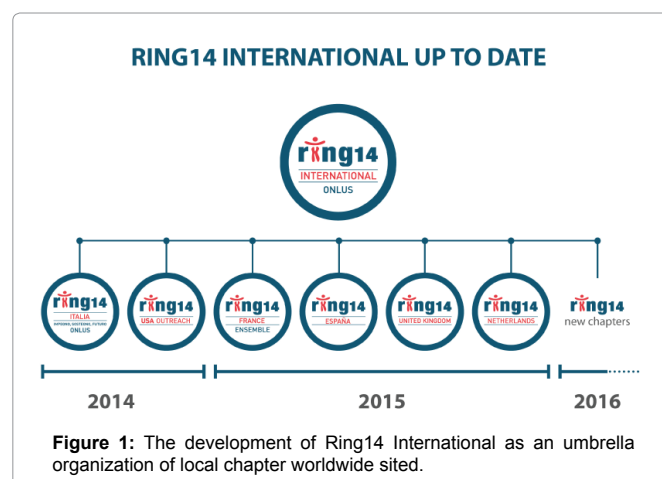


Figure 1: The development of Ring14 International as an umbrella organization of local chapter worldwide sited.

new treatments and cures for Ring14 and for defining new scientific strategies for future developments.

### Ring14 scientific activities

Ring14 sponsors basic and translational research through its strong commitment to projects aimed at advancing scientific knowledge on chromosome 14 disorders (Figure 2). Ring14 International sets up scientific activities dedicated to strategic planning of research initiatives and management of the project selection process. This work is carried out in compliance with best practice methods and employs dedicated professionals with specific scientific expertise in these areas. All research-funding decisions are therefore subject to peer-review by our international SAB.

### Biobanks

Biobanks are authorized centres that collect and store genetic material of patients and make it available to researchers around the world. In December 2009, Ring14 Italy signed an agreement with the Telethon Network of Genetic Biobanks (TNGB) for the collection of blood and skin samples of our patients and their relatives [7]. One of the Biobanks of the TNGB (the Galliera Genetic Biobank) now contains more than 300 biologic samples available to the international

scientific community.

### Database of clinical information

Ring14 also manages a database of clinical information collected from families. This database is extremely useful to help determine which symptoms might be related to these syndromes and to stimulate and develop translational research. The Ring14 database can be freely-accessed upon receiving a letter of intent/scientific project and is listed in several international research programs such as the FP7 RD-Connect project [8], which invited Ring14 International to join the “Core Implementation Group” [9].

### Workshops

The Association also organizes international workshops involving experts in the field of cytogenetics, molecular genetics, clinical applications and epilepsy with the aim of generating ideas and innovative approaches for the study of these diseases, and meetings/holiday camps for families and patients.

### Set-up kit

In order to facilitate the start-up of new national Ring14 Chapters, the International Association has developed and is offering a set-up



Figure 2: The timeline (2002-to date) of the most relevant milestones achieved by Ring14 Italy.

kit that includes:

- a) A copy of articles of association and internal regulations that each National Chapter will adapt to the relevant legislation of its home country,
- b) A national website linked to Ring 14 International website,
- c) Basic training on fund raising activities,
- d) Ring14 personalised trademark and literature,
- e) Ring14 pamphlets, posters, gadgets, as well as other forms of merchandising to promote and facilitate fund raising activities,
- f) A local meeting with families and doctors.

### Future perspectives

Future perspectives will focus on:

- a) Maintaining yearly calls for applications for international research grants,
- b) Organizing international workshops for researchers and meetings for families,
- c) Promoting biological samples and clinical data collections,
- d) Opening new Chapters all over the world.

### Final Remarks

The intent of this letter is to highlight how the advocacy work being done by a patient association can be expanded beyond just improving the visibility of an underdiagnosed orphan disorder. In fact, even a small group of patients, especially if internationally networked, can have a pivotal impact on stimulating excellent scientific activities, funding critical and innovative biomedical research, and encouraging clinicians to perform specific genetic tests to facilitate prompt diagnosis.

### Competing Interests

Ring14 International is a non-profit organization that raises

funds through fundraising activities. The authors does not have any financial or non-financial competing interests to disclose.

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### References

1. Zollino M, Ponzi E, Gobbi G, Neri G (2012) The ring 14 syndrome. *Eur J Med Genet* 55: 374-380.
2. Mavris M, Le Cam Y (2012) Involvement of patient organisations in research and development of orphan drugs for rare diseases in Europe. *Mol Syndromol* 3: 237-243.
3. Giovannini S, Marangio L, Fusco C, Scarano A, Frattini D et al. (2013) Epilepsy in ring 14 syndrome: a clinical and EEG study of 22 patients. *Epilepsia*. 54: 2204-2213.
4. Giovannini S, Frattini D, Scarano A, Fusco C, Bertani G et al. (2010) Partial epilepsy complicated by convulsive and nonconvulsive episodes of status epilepticus in a patient with ring chromosome 14 syndrome. *Epileptic Disord* 12: 222-227.
5. Zampini L, Zanchi P, D'Odorico L (2014) Developing with ring 14 syndrome: a survey in different countries. *Clin Linguist Phon* 28: 844-856.
6. Zampini L, D'Odorico L, Zanchi P, Zollino M, Neri G (2012) Linguistic and psychomotor development in children with chromosome 14 deletions. *Clin Linguist Phon* 26: 962-973.
7. Filocamo M, Baldo C, Goldwurm S, Renieri A, Angelini C et al. (2013) Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. *Orphanet J Rare Dis* 8: 129.
8. Thompson R, Johnston L, Taruscio D, Monaco L, Bérout C (2014) RD-Connect: an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. *J Gen Intern Med* 29: 780-787.
9. <http://rd-connect.eu/platform/registries/core-implementation-group>.

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