

Ring14 International: a worldwide network to fight a chromosomal disorder



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Who we are

Chromosome 14 syndromes are rare genetic disorders. In some cases, one of the two chromosomes assumes the form of a ring resulting in the "Ring14 syndrome" (omim #616606), characterized by early onset refractory epilepsy, motor and mental retardation, autism and a multitude of other physical challenges. In other 14th chromosome syndromes, the chromatid pair loses or transfers genetic material (deletions or duplications) sharing usually 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.



Ring14 International is a 2nd-level non-profit worldwide organization having an umbrella structure, functioning as a coordination centre to promote clinical and scientific activities and to provide the best standard cares to affected children and their families.

Ring14 International consists of the two founders (Italy & USA) and other chapters connected from new countries.

Ring14 International is a reference point:
 - we are present in **26 countries** & communicate 5 languages
 - to date, we represent **242 families** (50.8% with Ring14)



The management
Board of Directors - The BoD is the governing body, which defines and annually reviews general policies for the Network and its members.

Scientific Coordination office

Scientific Advisory Board - The SAB is responsible for providing scientific advice and evaluating new projects to be selected through peer-reviewed annual calls.

Current projects & activities

Field	Objective	Partnership	Results	
Supporting research projects	<i>In vivo/In vitro</i> models	These projects are focusing on making of Ring14 models: - In mouse, with rearrangements homologous to the human chr14; - By CRISP/CAS9 genome editing, to introduce loxP sites on the chr14 ends in wild-type iPSCs; - iPSC derived neurons in culture.	- Univ. Strasburg (FR) - Case Western Univ. (USA) - Harvard Univ. (USA)	- Investment: 314K Euro - 11 research centers expressed interest to work on Ring14 (6 got funded) - 10 Scientific articles acknowledging R14 published (+ 1 paper directly authored by Ring14 International) - 4 new manuscripts to be submitted in 2016
	Language skills	To describe the characteristics of communicative development of children affected by chr14 abnormalities.	Univ. Milan-Bicocca (IT)	
	Genotype-phenotype correlation	To correlate the quantity (and type) of lost genetic material (deletions) or rearranged material (translocations or ring structures) with the clinical symptoms.	- Univ. Pavia (IT) - Univ. La Cattolica (Rome, IT)	
	Gene expression profiling	To determine the molecular aetiology of seizures in R14 and verify if ring chromosomes have an altered expression profiling.	Univ. Pennsylvania (USA)	
Biobanking	We signed an agreement with the TNGB for taking custody of Ring14 bio-samples and make them available to the international scientific community.	- Galliera hospital (Genoa, IT) - Telethon Network of Genetic Biobanks (TNGB, IT)	To date, 669 samples (mainly blood & fibroblasts) of patients and relatives have been already collected	
Scientific meetings	We organize international workshops for researchers & clinicians and meetings among families.	/	5 planned meetings with tot. 617 delegates (18,5% scientists/clinicians)	
Clinical database	We collect and keep clinical records, which are anonymized and digitalized in a database.	RD-Connect (a FP7 EU-funded project)	The database is free-accessible online	

Next steps

To publish **Standard Guidelines** for the management of children affected by Ring14 syndrome

To solicit new scientists on chr14 diseases research

To open chapters in new countries by 2016

To plan a new International meeting for both scientists & families

To partner in a large-scale scientific project

➔ A dedicated task force is working on the SGs: expected release on **early 2017**

➔ New Calls issued on **May 2016**:
 • Autistic traits (€ 5.000, entirely sponsored by R14 Italy)
 • Exploratory projects (€ 50.000)

➔ **Two countries** expressed interest to join R14 International

➔ Next meeting is scheduled on **Summer 2017**

➔ An application to a major funding agency on **early 2017**

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