

Eurochromnet Newsletter

For anyone affected by a rare chromosome disorder and for those who work with them

We were alone... Now we are a lot 1st International Congress Ring 14

By Paola Martinelli

'We were alone Now we are a lot' . This was the title of the presentation of a mother at the first International Congress of the Association Ring 14 held in Reggio Emilia on October 13-14th 2006 and it reflected the real climate of sharing and friendship that we had felt during those days.

The Congress, the first ever organized in the world on these rare syndromes, has been a very important moment: the Association presented the work of the Ring 14 Association and the research they fund to the medical and scientific community

It was aimed at the physicians and all those practitioners involved in assisting and rehabilitating these children, as well as at families as the principal protagonists of the projects. Just to indicate the international character of the meeting, 23

families participated in this important event from Italy, United States, Brazil, Australia, France and Great Britain. It was greatly satisfying to see the gratitude in the eyes of all these parents who have found

appreciated?" "Will it be worth the effort?". We can now say: Yes, without doubt! It has been a great pleasure to see our beautiful children all play together with



Participants of the first International Congress Ring 14

in us the principal motor of the search for knowledge into the illnesses of their children and some of them have indicated their availability to open branches in their countries.

It was really hard to organize the Congress and during the preparation we had a lot of doubts: "Will families participate?", "Will the chosen subjects be

so many worthy persons in the playroom, to have the opportunity of getting to know the other families, to exchange mutual experiences and perceiving expressions of gratitude in their eyes because FOR THE FIRST TIME EVER so many doctors, researchers and scientists were together for discussing ONLY about the diseases of our children.

Continued on page 2

**Eurochromnet:
European
Networks for
Rare
Chromosome
Disorders on
the Internet**

**Eurochromnet
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In this Issue:

<i>We were alone... Now we are a lot 1st Ring 14 Congress</i>	1
<i>Workshop Rare Disease Patient Re- gistries</i>	2
<i>Photo Contest</i>	2
<i>At last... The Ring 14 Conference</i>	3
<i>Partners, meetings, subscription, editorial</i>	4

Rare Disease Patient Registries: an Essential Tool in the Development of Therapies

By Caroline van Heesewijk
Board member European
Chromosome 11q Network



www.11q.org

Eurordis5th Workshop “Rare Disease Patient Registries: an Essential Tool in the Development of Therapies?”

Date: – November 20th, 2006

During the conference in Berlin it became evident for Eurordis that they had to organise a workshop about patient registries. This workshop was organised in Paris and it was successful. We had some presentations regarding classification of rare diseases in different countries with different examples.

Subjects were:

- Discovering registries
- Role of registries as major tools for medicinal product development in the pre and post-marketing phases
- Legal and ethical issues related to registries in Europe
- Rare Disease registries: real-life experiences
- Registries for Rare Diseases: legitimacy of all interested parties in the creation, management access and ownership of patient registries. Respective responsibilities, best practice, etc...

Between presentations we had discussion time. It was really interesting because we had two points of view: (a) The medical side with pharmaceuticals and researchers and (b) Patient's organisations.

Summary:

1. Working together is a matter of good coordination and cooperation. This should not be limited to just scientific models. Next Question to be answered: How can we find a global

approach?

2. Patient registries are very costly.
Next Question to be answered: How to fund?
3. Patient registries should not only be done for drug development.
Next Question to be answered:
 - What information should be captured, and
 - How can we make the classification of patients?
4. Information should be kept confidential and secure to non-authorised people.
Next Question to be answered: What is the best practice to achieve information confidentiality e.g. Consent Forms, Confidentiality Charters

Conclusion:

Eurordis will take the lead in this project with the help of all of its members. Because of the magnitude of scale and complexity and different interest groups, this will be a long-term and large project.

Eurordis' 2007 Photo Contest: On your marks - Point - Shoot!

“Following the success of its 2006 contest, Eurordis is launching its second photo contest in December.

This time, the contest is open to everyone having an interest in rare diseases in Europe, whether members of Eurordis or not. The contest will be open from 1 December 2006 till 31 March 2007.

Winners will be announced at the Eurordis Annual Membership Meeting Paris 2007. Prizes will be awarded to the best three pictures. Terms and conditions as well as entry forms and prizes are posted on Eurordis' website”.

(Announcement by Eurordis)

more info: www.eurordis.org

Ring 14

Continued from page 1

Generally all of us, parents of "special rare children", are used to receive only vague answers or, often, not any answer; now it seems some answers are coming even if, obviously, a lot of questions are still pending, but we shall try, step by step, with patience but DETERMINATION to achieve answers.

Some families have agreed to try to establish a seat or a branch of an Association like "Ring14" in their countries. This is the best gift for our children, Do not lose your feeling for it! you will surely achieve the target and we shall support you in any way!

We go on, and our children will join us. We know that for now this rare illness cannot be prevented. But we started "to besiege" it three years ago, and we are nearer to achieving our objectives.

We know that we are not alone, and the new contacts reaching us from every part of Europe and worldwide inspire us to dare more and more, to plan new interventions and to believe in what we are doing. We believe for our children in a certain level of rehabilitation, in a better quality of the life, in the possibility of more specific treatment of the symptoms and we hope that our Association will become a sounding box in that aim.

Everybody interested in receiving the material related to the congress (actions, recording) can send an Email

to our E-mail addresses:
info@ring14.com or
azzali.stefania@asmn.re.it

The congress material is also available on our web site
www.ring14.com

For any further information please contact : Stefania Azzali, President Association Ring 14
 Cell. 3408681962; e-mail:
info@ring14.com

At Last.... The Ring 14 Conference

Finally the expected day had arrived!

After so many months of hard work for the organizers of the first Ring 14 conference, we were here, all excited and deeply moved like



Alessandro



children on their first day of school, anxiously waiting for the arrival of the participants, but primarily waiting for the families with their splendid children: 23 families... from Italy - Great Britain - France - Brazil - U.S.A. - Mozambique - Australia.

We had been full of anxiety and doubts in organizing the playroom in the best way possible for our children afraid not to be capable to manage our children and instead..... it was so simple! Thanks to the help of many volunteers, everything went very well and our children too showed to be able to deal with this situation and new experience.

The Thursday afternoon was devoted to a first meeting of the families and our medical staff in informal way, to make each other acquaintance and to exchange opinions and experiences. On Friday morning the fantastic experience of the conference started; the intervention of any presenter enriched our mind satisfying our hunger to know. Also all of us parents, we did not spare ourselves in asking them a lot of

questions.

On Saturday morning all the parents with their children had the possibility of meeting our Scientific Committee for a medical and genetic visit allowing at the same time the doctors to examine in this unique

occasion many cases of ring and deletion of chromosome 14. For all of us parents it was a great pleasure to be together, to talk about ourselves, of our experiences and on our faces it showed the new hope of a better future for our children.

At the end of the conference the common feeling was that we were one big family! A

family that for distance reasons will not have the possibility of a regular meeting but that will always be in our heart of hearts for all what we learnt, for all people we had the pleasure to get to know, for all what we have received and also given.



Daniela,
Alessandro's mum, Ring14

Partners in Eurochromnet

- Angelman eV, Germany
- Association GEN - Poland
- Association Ring 14
- Chromosome Help-Station
- Cri du Chat Switzerland
- ECARUCA
- European Chromosome 11q Network
- Eurordis
- IDIC 15 Europe
- Leona eV - Germany
- Ring Chromosome 20 Foundation
- Unique
- UniqueDanmark
- Valentin Apac - France
- VSOP - The Netherlands
- Zeldzaam/FVO - The Netherlands

Links to their websites can be found at:
www.chromosomehelpstation.com

**Deadline articles,
meetings and
suggestions for
Eurochromnet
Newsletter 2:
7 February 2007**

Meetings

- 3-4 March 2007: 22q11.2 Deletion Syndrome: Practical Implications for Behaviour and Learning, is to be held in Sacramento California. Location: UC Davis MIND Institute, Sacramento, CA, USA. More information is available online at www.elwyngenetics.org.
- 9-11 March 2007, Leona active members working meeting, near Fulda, Germany. More info: wwleona-ev.de
- 10 or 17 March 2007 meeting 'Zeldzaam', Dutch network for rare chromosome disorders. Theme: communication with your child. More info: www.zeldzame-syndromen.nl
- 24 March 2007: 7th meeting for Klinefelter, Turner, Triple X and Double Y, France. More info: www.valentin-apac.org
- 22-25 March 2007: 5th International Conference European Chromosome 11q Network, Pforzheim-Hohenwart, Germany. More info: www.11q.org
- 4-5 May 2007, Eurordis Xth anniversary and annual membership meeting, Paris, France, Institut Pasteur. More info: www.eurordis.org
- 17-20th May 2007: 6th International IPWSO Conference (Prader Willi International) in Cologne, Germany. More info: www.ipwso.org
- 1-3 June 2007: Eurochromnet meeting, Copenhagen-Oslo.
- 21-24 June 2007: IPWSO Conference (Prader Willi), Cluj-Napoca, Romania. More info: www.apwromania.ro
- 28-30 September 2007: Leona F=family meeting, Heinrich-Lübke-Haus, Möhnesee-Günne, Germany. More info: www.leona-ev.de
- Autumn 2007: Family Day Valentin APAC, France. More info: www.valentin-apac.org
- 27-28 November 2007: 4th European Conference on Rare Diseases (ECRD)

Subscription Eurochromnet Newsletter

This Newsletter is sent to whoever is interested in rare chromosome disorders and networks that support affected people.

Subscription costs for 2007 are € 15 per year (persons) or € 30 per year (organisations, networks, institutes). These can be paid in this way:

- Inhabitants of the European Union: please pay to the Chromosome Help-Station, Else Mauhsstraat 7, 6708 NJ Wageningen, The Netherlands. Postbank nr 9311090. Bank account: ING foreign operations, PO box 1800, 1000 BV Amsterdam, The Netherlands. IBANnr: NL36PSTB009311090. BIC (Swift) code: PSTBNL21.
- Inhabitants of the Netherlands: same address, Postbank nr 9311090.
- Payment via creditcard, Visa or Mastercard/Eurocard. More information: www.chromosomehelpstation.com

Editorial

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