

FIRST EUROCHROMNET MEETING Luxemburg, 20 June 2005, Hotel Ibis Aeroport

10.00 – 17.00 h

Participants:

Chromosome Ring 20 Foundation: Pierra Roberts
Ecaruca: Ilse Feenstra
European Chromosome 11q Network: Arne Eiwen, Caroline van Heesewijk
Frambu: Lisen Mohr, Bente Hunn
Leona eV: Harriet Hoste, Sabine Lehnert, Thorsten Randt
UniqueDanmark: Jette Ziegler
Unique: Beverly Searle, Prisca Middlemiss
VSOP: Pauline Evers (morning)
Chromosome Help-Station: Annet van Betuw (report)

1. Welcome

Beverly Searle chaired the meeting and welcomed everybody who was present.

This was the first formal meeting of Eurochromnet. Eurochromnet is based on the publication European Rare Chromosomes United, by Chromosome Help-Station and VSOP. On CHS' website some pages on Eurochromnet are published.

Valentin Apac was not able to come because of financial reasons. Gen could not come either for financial reasons and their own conference that was organised in the beginning of June. Stefania Azzali (Ring 14) could not come after all for personal reasons. It is important to find money for the next conference, in order to make it possible that everybody can come. We see that there is a big difference between culture and care supplied in different countries. We must share experiences and hold hands.

2. Notes meeting 15 October 2003

No comments.

3. Presentation of support groups and other interested visitors

– also update of actions support groups

Everybody told in headlines his/her personal involvement with rare chromosome disorders. Next to that followed an update of actions of the support groups:

Unique/Beverly Searle: more than 4500 member families in 64 countries (25 European countries). More and more people from eastern Europe (eg Russia). There are 2 full-time and 3 part-time people employed. Beverly is development director, Prisca is Unique's information officer. Unique is now publishing family-friendly leaflets on chromosome disorders as well as on behaviour, siblings, legislation, special education, etc. The Little Yellow Book has been translated into Italian, Finnish, Russian and Norwegian. In addition Unique is participating in the EC funded projects Eurogentest and SAFE (see agenda point 5). At this moment Unique is changing its legal status, to be sure that the personal liability of the trustess' is limited.

11q/Arne Eiwen: Although the European Chromosome 11q Network is still a small network (75 members in 12 countries) it is growing, as are the activities. Therefore a new board structure is needed. Active members have learned to delegate; now people have been made responsible for sub-themes. The board will meet once a year. In October 2003 the fifth International Conference was organised. It was a success. Information on that meeting can be found on the website and in the report which will be published soon.

Leona eV/Sabine Lehnert: For two years Leona has had five instead of three board members. It is hard to mark out tasks, but it is functioning well now. Once a year there is a meeting for active members, chaired by a professional from outside. This resulted in much more effective and productive meetings. There are 600 contact families, 300 of them are member families. Maybe it is time for Leona to go

professional? It is important to raise awareness. Leona wants to make presentations which can be used by people in the field to promote Leona at hospitals etc.

Frambu/Lisen Mohr: Rare chromosome disorders is a group within Frambu. The aim of Frambu is to learn to live with the rare disease, you cannot cure. Families stay with Frambu for one or two weeks, learning how to cope with the disease, also information is given on possibilities for help, schooling, work, housing, etc. Frambu is located close to Oslo. Families come to Frambu, where 20 families can be accommodated. There is also a lot of info via the web (www.frambu.no). Professionals can also take part in courses. Frambu is entirely state funded. Agrenska in Sweden works the same way. People can ask for help and Frambu also travels throughout the country to give support.

Unique/Danmark/Jette Ziegler: The network continued the activities and is growing. There are now 70 members. Unique is supported by the government.

Ring 20/Pierra Roberts: The Foundation is rather new. It is based in London and New York and had 25 members in 1,5 year.

Chromosome Help-Station/Annet van Betuw: Annet started the 11q network in 1997; she will step down as president in 2007 at the 6th conference. She started her own advisory Chromosome Help-Station in 2002. One of the activities is organising this meeting and publishing the Eurochromnet Newsletter since January 2005.

VSOP/Pauline Evers was present as advisor for Eurochromnet. She was to visit another meeting in the afternoon and tell there about this meeting. *Ilse Feenstra* was to give an update on *Ecaruca* at Agenda Item 6.

During this update a short discussion on liability started. **Conclusion:** never give medical advice, that is the responsibility of the doctor. And put a disclaimer on your website.

4. Update actions of Eurochromnet

- What happened since October 2003, by Annet van Betuw

The presentation in PowerPoint will be put on the website. The conclusions and proposals for next steps are discussed on Agenda Item 8.

One of the goals of Eurochromnet is exchange information. Annet thinks that it is important to have a multilingual website for Eurochromnet. Now it is a part of the site of the Chromosome Help-Station. Moreover: this multilingual website must be set up in 6 languages. There was an attempt to have money for this site within the Safe project (see item 5), but it was rejected. Now there are plans to start a foundation in the Netherlands for rare chromosome disorders or rare diseases with major goal fundraising. The first project will be the multilingual website.

5. Meet projects SAFE and Eurogentest

- by Beverly Searle

Also these presentations will be put on the website.

SAFE = Special Advances in Fetal and Neonatal Evaluation Network. Prof Maj Hulten in Warwick is the project leader. Key mission: implement routine non invasive diagnostic testing for chromosome disorders, foetal and neo-natal. There are 55 partners in 18 countries. Unique is involved from patient side, others are all researchers.

Unique is especially involved in work packages 5, risk assessment, 6, socio-economics and 7, ethical aspects, as well as the education and spreading of excellence activities. Research for non invasive techniques is far away from actual use.

The goal of Eurogentest is harmonisation and validation of testing and standardisation of tests throughout Europe. There are over 1000 diagnostic laboratories in Europe; there is a limited network and no Europe-wide accreditation system. Moreover there is sometimes poor counselling.

6. Ecaruca

- An online database for rare chromosome aberrations, by Ilse Feenstra

Ecaruca is a EU wide database assembling data on as many people with rare chromosome aberrations as possible: cytogenetic, molecular and clinical. That can be entered by physicians but also by patients themselves. By collecting data at a bigger international level insight in the course of disease can be enlarged. By publication of the combined results there will be improved information and patient care.

Ilse gives a short explanation of the database. Ecaruca contains no info on balanced translocations. Parents can enter data of the child; not only for the original diagnosis but also follow-up data throughout the lifetime of the patients. In that way the natural course of the disorder can be followed. On Beverly's question, Ilse answers that each case has an identifying number; identical twins will have that too. Until now Ecaruca has not come across identical twins yet. Currently there are 3.800 cases in the database from Prof. Schinzel in Germany; and also 200 new cases. Most of the new cases are from the Netherlands, some UK. Ilse checks all the data and if necessary she corrects after consultation with the physicians. Arne wonders how she can find out if her geneticist has entered information. Prisca asks if there is family involvement on an advisory board or a regular newsletter. Ilse answers no, but that it is a good idea. Until now the focus was on the database and getting it started and running. Annet suggests that Ecaruca can use the Eurochromnet Newsletter. Then Ilse explains that the proposal for the continuation of the present project with the EU was rejected. The people present at this meeting are shocked. The reason given for the rejection was there were insufficient number of collaborators on the project. Ilse says that this is untrue. Unfortunately there is no possibility to protest against the decision. The rejection was received shortly before the Eurochromnet meeting. Therefore Ilse cannot inform us yet if and how other funding will be found. For the moment there is no problem, because the project has enough money to continue.

In the discussion, the participants in the Eurochromnet meeting exchange experiences in communication with geneticists (many families no longer visit a geneticist); not having access to records or even records that are destroyed; tests with different results like Caroline's son who turns out to have a disorder on 11p and not 11q; and medical students and not enough geneticists who are not aware of chromosome aberrations.

7. Unique's Information Project

- Development of medically-verified, family friendly information flyers and booklets about a wide range of rare chromosome disorders, by Prisca Middlemiss.

This presentation will be added to the Eurochromnet web

The goal of this project is to produce chromosome disorder-specific information for families and give them an outlook when they are told their children has a chromosome disorder. The information in the leaflets is taken from the medical literature, from Unique's members' database (which details the longitudinal histories of 5500+ individuals, the vast majority of cases never having been published in the medical literature) as well as more in-depth surveys of individual member families. Areas to be covered in each leaflet are: learning, education, lifestyle, achievements, care needs, therapies, work, independence, collaborative organisations and medical literature. All texts are verified before publication.

8. Discussion and agreement on next steps

- A. *Goals*: The goals of Eurochromnet as published in the posters were approved again.
- B. *Structure*: The discussion on the structure showed that there is no need for a leader; the structure with a small group and an 'all' group is ok. Problems within both groups are time and English. We always have to remember this.
- C. *List of talents*: Pierra suggested that the small group identifies tasks, breakdown and find out how people can help. Beverly suggested an inventory list of talents.
- D. *Ecaruca*: How can Eurochromnet help Ecaruca? Get parents and patients aware and submit data. The problem is that many patients are no longer seeing a clinical geneticist. Further there is a need to translate data for patients. Would it be useful that Ecaruca has leaflets from Unique? Ilse answered yes, moreover she can translate leaflets into Dutch. Ecaruca's goal is to add 1000 more cases before the end of this part of the project.
- E. *Next conference*: It was agreed that in the next Newsletter the conference from May-June 2007 will be announced, so that people can start raising money to attend this meeting. Jette and Arne suggest to make money for meetings. Annet needs money for the website.
- F. *Next conference and general*: Eurochromnet will continue as a virtual network. We will have an update the end of May/beginning June 2007 on the boat from Copenhagen to Oslo. In Oslo

we will be visiting Frambu. We will try to have a meeting every two years. It is important that it must be organised in a region where one of the members lives and can organise it.

9. Involvement of researchers

– Discussion on participation of researchers and/or institutes in Eurochromnet
Eurochromnet is a virtual network for support groups. Researchers, institutes and others should not be members. Articles for the Newsletter must not promote institutions. Eurochromnet should maintain control. Annet (the Chromosome Help-Station) edits the Newsletter and publishes. She is then in contact with Pauline and Beverly. If there are problems they will discuss it in the 'small' group.

10. Funding issues

This is discussed at other items. See 8 E.

11. Other business

- *Eurochromnet Newsletter*: It is no problem for Annet to produce 10 issues a year. Prisca wondered if she would mind featuring organisations outside of Europe. Annet as the other participants do not mind, as long as the articles are short.

- *Not very rare disorders*: Eurochromnet accepts the definition of the EU on rare: $1 < 2000$ patients. This means that Klinefelter, Turner and other chromosome disorders are not rare. On the other hand: the Eurochromnet Newsletter can be used as a vehicle to give information on not very rare disorders, because everybody is in need of information.

12. Close

Beverly thanked everybody for attending the meeting. We will meet again in May/June 2007 on the boat from Copenhagen to Oslo.

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