



European Cytogeneticists Association



Dear colleagues,

With this e-mail we would like to inform you about an initiative to explore the feasibility of forming a European Reference Network on Rare **Chromosome** Disorders.

What is a rare chromosome disorder?

This is a disorder that is (predominantly) caused by a chromosomal imbalance and that occurs in less than 1 in 2000 newborns.

What is a European Reference Network (ERN) for a rare disorder?

An ERN is a network connecting centres of expertise of highly specialized healthcare, for the purpose of improving access to diagnosis, treatment and the provision of high-quality healthcare for patients with conditions requiring a particular concentration of resources or expertise no matter where they are in Europe. ERNs will promote expertise and support health care providers in order to bring local, regional and national provision of health care closer to the patients.

Who can be part of an ERN?

To participate in an ERN you should be recognized by your own government as centre of expertise (i.e. fulfil the EUCERD criteria).

How can an ERN apply for assessment by the Board of Member States?

The 1st call will be in February 2016 and details of the assessment procedure can be found at the links at the bottom of this e-mail. An ERN should consist of at least 10 centres of expertise out of 8 member states. It is important not to wait for this call, but start already now if we think an ERN for rare chromosome disorders is feasible. After the call, there will be only 3 months to finalize the application.

DEKON Association Management Company (AMC)

provides association management services to and on behalf of
E.C.A. (European Cytogeneticists Association)

Adress : Gayrettepe Mah. Esentepe Yıldız Posta Cad. Akın Sitesi
1. Blok No:6 Kat:1 D:3 34394 Şişli - İstanbul - TURKEY
Phone : +90 212 347 63 00
Fax : +90 212 347 63 63
E-mail : esra.guler@dekon.com.tr
Visit www.dekon.com.tr for more information!

E.C.A. (European Cytogeneticists Association)

Prof. Konstantin Miller
E.C.A. General Secretary
Institute of Human Genetics Hannover Medical School
30623 HANNOVER / GERMANY
Phone: +49 511 532 3572
Fax: +49 511 532 6555
E-mail: miller.konstantin@mh-hannover.de



European Cytogeneticists Association



What are the main tasks/requirements of an ERN?

- Provide multidisciplinary and highly specialized patient care (including guidelines, standards of care)
- Collaborate with patient organizations and develop PROMS (patient reported outcome measures).
- Patient registries/databases
- Education and training
- eHealth
- research (but the emphasis is on good clinical care and patient empowerment)

Why should we apply for an ERN for rare chromosome disorders?

Formalizing a network will improve the quality of patient care, allow cross-border care and facilitate research. The official recognition as an ERN is a quality mark that may be beneficial for funding.

Moreover, we think that many prerequisites for an ERN are already (almost) in place. The only thing we still need to do is to connect the officially recognized centres of expertise and formalize a governance structure.

For example,

- Most of us already collaborate with patient organizations (Unique, Wolf-Hirschhorn, Phelan-McDermid, VCFS, etc.). Especially Unique is an important and very well organized partner.
- We already have ECARUCA and DECIPHER, but more importantly, in the light of patient participation and eHealth, we are currently validating a multi-lingual, social media-based patient/parent interactive website that has been developed for our chromosome 6 project, and which can easily be adapted to all chromosome disorders.
- We already have an annual European course on clinical cytogenetics that is attended by clinical geneticists and clinical laboratory geneticists and has a very high standard. This can easily be complemented by exchange internships in the centres of expertise.

I am interested, what should I do?

More information can be found via the links at the bottom of this e-mail. We are working on a dedicated web page on the ECA website and will let you know as soon as this is ready.

If you are a centre of expertise for one or more rare chromosome disorders, please let us know and we will list you on our web page.

DEKON Association Management Company (AMC)

provides association management services to and on behalf of
E.C.A. (European Cytogeneticists Association)

Adress : Gayrettepe Mah. Esentepe Yildiz Posta Cad. Akın Sitesi
1. Blok No:6 Kat:1 D:3 34394 Şişli - İstanbul - TURKEY
Phone : +90 212 347 63 00
Fax : +90 212 347 63 63
E-mail : esra.guler@dekon.com.tr
Visit www.dekon.com.tr for more information!

E.C.A. (European Cytogeneticists Association)

Prof. Konstantin Miller
E.C.A. General Secretary
Institute of Human Genetics Hannover Medical School
30623 HANNOVER / GERMANY
Phone: +49 511 532 3572
Fax: +49 511 532 6555
E-mail: miller.konstantin@mh-hannover.de



European Cytogeneticists Association



If you are interested in joining or coordinating the ERN, please send an e-mail to c.m.a.van.ravenswaaij@umcg.nl.

With kind regards

Nicole de Leeuw, clinical laboratory geneticist

Conny van Ravenswaaij, clinical geneticist

ECA Permanent Working Group on Clinical and Molecular approaches to cytogenetic syndromes

Links

http://ec.europa.eu/health/ern/policy/index_en.htm

http://ec.europa.eu/health/ern/docs/faq_establishing_ern_en.pdf

http://ec.europa.eu/health/ern/implementation/index_en.htm

DEKON Association Management Company (AMC)

provides association management services to and on behalf of
E.C.A. (European Cytogeneticists Association)

Adress : Gayrettepe Mah. Esentepe Yıldız Posta Cad. Akın Sitesi
1. Blok No:6 Kat:1 D:3 34394 Şişli - İstanbul - TURKEY
Phone : +90 212 347 63 00
Fax : +90 212 347 63 63
E-mail : esra.guler@dekon.com.tr
Visit www.dekon.com.tr for more information!

E.C.A. (European Cytogeneticists Association)

Prof. Konstantin Miller
E.C.A. General Secretary
Institute of Human Genetics Hannover Medical School
30623 HANNOVER / GERMANY
Phone: +49 511 532 3572
Fax: +49 511 532 6555
E-mail: miller.konstantin@mh-hannover.de