European Skeletal Dysplasia Network
www.esdn.org
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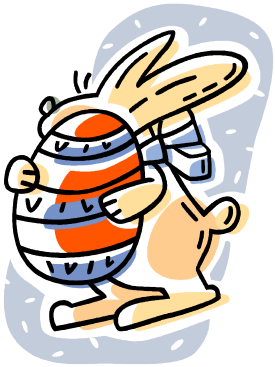
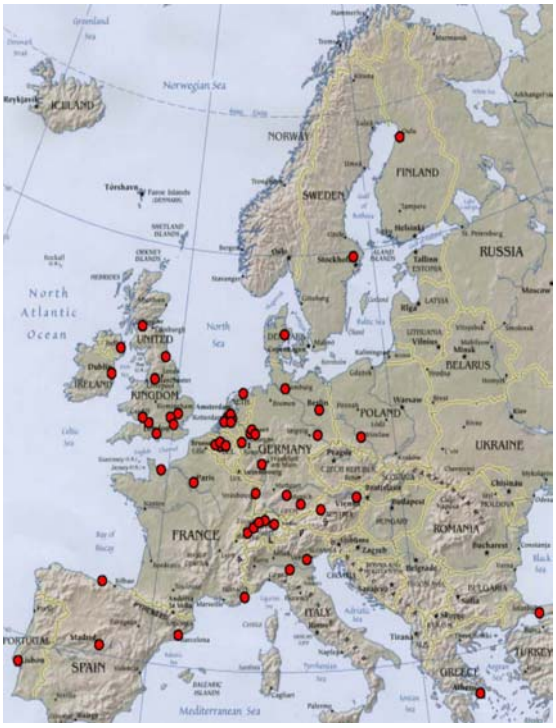
ESDN Newsletter



Milestones for 2002

- We have now established a pan-European referral and communication network that allows access to the ESDN diagnostic and research services from any country within the European Community. During 2002 ESDN received over 500 patient referrals, which came from 14 EU countries, 2 candidate states and 1 associated state. The geographic distribution of these referrals is indicated in the accompanying map.
- A molecular diagnostic service has been established, which includes the analysis of 21 genes in more than 28 disorders (All molecular genetics service profiles can be viewed on our website www.esdn.org).
- During 2002 over 290 diagnostic tests were performed under the auspices of ESDN and 150 mutations were found in patients and family members with skeletal dysplasias.



Editorial

Welcome to the Spring 2003 issue of the European Skeletal Dysplasia Network (ESDN) newsletter. We hope you enjoyed the December 2002 issue which introduced ESDN, and provided an outline of our proposed activities.

In this issue we have summarised ESDN activities for 2002.

Check out our "NEW LOOK" Website (www.esdn.org) for more information about the network.

Participants of the 3rd ESDN Management Meeting, January 2003 Lausanne, Switzerland

INSIDE THIS ISSUE:

an integrated research and diagnostic network of eight european centres of excellence

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Exciting new discoveries by members of ESDN

- In Gent, Geert Mortier and colleagues have delineated a new autosomal recessive skeletal dysplasia which they have named acrocapitofemoral dysplasia (ACFD) [Mortier G.R., *et al* J Med Genet 2003 40 (3):201-7]. Furthermore using homozygosity mapping they were able to link the phenotype to chromosome 2q35-q36 and subsequently identified *IHH* mutations in two unrelated families [Hellemans J., *et al* Am J Hum Genet 2003 72 (4):1040-6].
- In Paris, Valerie Cormier-Daire, Martine Le Merrer and colleagues were able to show that Dyggve-Melchior-Clausen Syndrome is caused by mutations in the novel gene Dymeclin located on 18q21.1. [El Ghouzzi V., *et al* Hum Mol Genet 2003 12 (3):357-64]. Independantly, Dan Cohn and colleagues also linked this gene to DMC [Am J Hum Genet 2003 Feb;72(2):419-28].
- In Newcastle, Judith Goodship, Michael Wright and colleagues have identified novel genes which when mutated lead to Ellis van Crevald and located them to 4p. [Ruiz-Perez V.L., *et al* Am J Hum Genet 2003 72 (3):728-32].

ESDN in Focus

Part II:

"New Faces to ESDN"

Dr. med. Andreas Zankl joined the ESDN as the Clinical Radiographic Review Facilitator (CRRF) in January this year. Andreas, who is based in Lausanne, is playing the important role of co-ordinating the review of cases referred to the ESDN. Andreas studied Medicine in Berlin and Lausanne and did his specialist training in Medical Genetics in Lausanne and Zurich. He joined the ESDN from the Institute of Medical Genetics at the University of Zurich after completing his specialist training.

Dr Linda Gibbs, is the new Post Doctoral Scientist who will join the ESDN group in Paris this April. She will be working with Jacky Bonaventure and colleagues on the development of an *in vivo* model for achondroplasia using a transgenic approach. Linda obtained her Ph.D at the University of London (Royal Veterinary College) and joins the Paris group after working as a research associate in the William Harvey Research Institute (Dept of Experimental pathology) in London.

Announcements

- Dr Geert Mortier is collecting familial and sporadic cases with **osteopoikilosis** for the genetic mapping of the condition. If you are interested in a collaboration on this project, please contact geert.mortier@rug.ac.be
- ESDN is actively collecting families with **Multiple Epiphyseal Dysplasia** in a collaborative effort to map the remaining disease genes. Please contact mike.briggs@man.ac.uk for further details.
- The Skeletal Dysplasia Center in Lausanne is collecting samples from individuals and families with **achondrogenesis type 1A**, **omodysplasia**, **costal and vertebral segmentation defects** (Jarcho-Levin and related phenotypes) and **Spondylo-epimetaphyseal dysplasia** with joint laxity and leptodactyly, in a collaborative research effort to identify responsible genes. Please contact asuperti@chuv.unil.ch for further details.
- ESDN is actively collecting individuals and families with **Spondylo-metaphyseal dysplasia**, Sutcliffe (corner-fracture) type, to refine diagnostic criteria and define clinical features, growth characteristics, diagnostic criteria and to identify causative gene(s). Please contact andreas.zankl@hospvd.ch or jacky.taylor@cmmc.nhs.uk for further details.
- The European Geleophysic Dysplasia Consortium is collecting sporadic and familial cases of **geleophysic dysplasia** and related conditions (**acromicric dysplasia**, **Moore-Federman**) in a collaborative effort to ascertain the causative gene(s) and refine clinical diagnostic criteria. Please contact g.mancini@erasmusmc.nl for further details.
- In the **NEXT ISSUE** find out about our **new interactive ESDN website** which will be online in a few months. Also **ESDN in Focus, Part III: "details on the Newcastle group".**