European Skeletal Dysplasia Network
www.esdn.org
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ESDN Newsletter

Special points of interest:

- Main contact for ESDN - Jacky Taylor
- Please register with the ESDN
- ESDN partners
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Editorial

Welcome to the first European Skeletal Dysplasia Network (ESDN) newsletter. In this issue I will introduce the ESDN, what we do currently and what we plan to do in the future.

I would be grateful if you and your colleagues would register with the ESDN if you are interested in receiving regular information about the network.

To register please email me your contact details at the address above.

Season's greetings,
Jacky Taylor.

Main contact for ESDN:

Dr Jacky Taylor
ESDN Project Administrator
c/o National Genetics Reference Laboratory (Manchester)
Regional Genetics Services
St. Mary's Hospital
Hathersage Road
Manchester M13 0JH UK
jacky.taylor@cmmc.nhs.uk (Email)
+44 (0) 161 276 6741 (Tel)
+44 (0) 161 276 6606 (Fax)
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What is the ESDN?

Currently there are 8 ESDN partners in 6 European countries (**Table 1**). The ESDN provides an integrated **research** and **diagnostic** network for skeletal dysplasias. The objectives of ESDN are:

➡ To understand what cellular, molecular and genetic factors cause bone dysplasias.

➡ To develop effective approaches for the diagnosis and treatment of bone dysplasias.

Ultimately this integrated and multidisciplinary approach will promote the correct diagnosis and targeted treatment for many skeletal dysplasias.

The ESDN is funded by the European Commission 5th Framework Programme.

European Commission

Participating Centres

Dr Michael Briggs ESDN Co-ordinator	Wellcome Trust for Cell Matrix Research School of Biological Sciences, University of Manchester 2.205 Stopford Building, Oxford Road, Manchester M13 9PT **UK** www.biomed.man.ac.uk/
Dr Rob Elles ESDN Co-ordinator **Dr Jacky Taylor** ESDN Project Administrator	National Genetics Reference Laboratory (Manchester), Regional Genetics Services, St. Mary's Hospital, Hathersage Road, Manchester M13 0JH **UK**
Dr. Michael Wright, Dr Judith Goodship ESDN partners	Institute of Human Genetics, International Centre for Life, Central Parkway, Newcastle-upon-Tyne NE1 3BZ **UK** www.ncl.ac.uk/ihg
Dr. Geert Mortier, Dr Anne De Paepe ESDN partners	Department of Medical Genetics, University Hospital of Ghent 185 De Pintelaan, B-9000 Ghent **Belgium**
Dr Jacky Bonaventure ESDN partner	Unité INSERM 393, Hopital Necker-Enfants Malades 149 rue de Sèvres, 75743 Paris, Cedex 15 **France**
Dr Andrea Superti-Furga ESDN partner **Dr Andreas Zankl** Clinical Radiographic Review Facilitator	Division of Molecular Pediatrics, Centre Hospitalier Universitaire Vaudois Rue du Bugnon, CH-1011 Lausanne **Switzerland** www.homepage.mac.com/asuperti
Dr Bernhard Zabel ESDN partner	Children's Hospital, University of Mainz, LangenBeckstr. 1 D-55101 Mainz **Germany**
Dr Leena Ala-Kokko ESDN partner	Department of Medical Biochemistry, Biocentre, University of Oulu, PO Box 5000, Aapistie 7, 90014 Oulu **Finland**

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Molecular Diagnosis of Skeletal Dysplasias

ESDN diagnostics: provides molecular diagnosis for more than 28 skeletal dysplasias through an integrated network approach, whose aims are to:

- Provide a robust European referral pathway with efficient routing of samples to appropriate centres.
- Provide effective quality assessment systems.
- Provide diagnostic service "best practice" guidelines.
- Provide efficient dissemination of results and clinically relevant information.

How to refer a patient to the ESDN:

For an efficient and effective diagnostic service it is vital that a full clinical description and radiographs of the patient are provided.

Submission details can be obtained from our website (www.esdn.org) or by contacting:

Dr Jacky Taylor:

jacky.taylor@cmmc.nhs.uk (Email)

+44 (0) 161 276 6741 (Tel)

+44 (0) 161 276 6606 (Fax)

Services offered by the ESDN:

Centre	Disease specialities	Genes screened	Molecular diagnosis	Research
Manchester	Pseudoachondroplasia, Multiple Epiphyseal Dysplasia (AD)	*COMP, COL9A1, COL9A2, COL9A3* and *MATN3*	yes	yes
Newcastle	Ellis-van Creveld syndrome	*EVC*	no	yes
Ghent	Osteogenesis Imperfecta Types I-IV	*COL1A1* and *COL1A2*	yes NB a charge will be made for this service	yes
	Achondrogenesis II, Hypochondrogenesis, Kniest dysplasia, Spondyloepiphyseal dysplasia, Spondyloepimetaphyseal dysplasia, Stickler dysplasia	*COL2A1*	yes	yes
Oulu	Stickler dysplasia, Otospondylomegaepiphyseal dysplasia, Marshall Syndrome, Weissenbacher-Zweymuller Syndrome	*COL11A1 and COL11A2*	yes	yes
Paris	Thanatophoric dysplasia -Types I and II, Achondroplasia, Hypochondroplasia Other FGFR3 disorders, SADDAN dysplasia	*FGFR3*	yes	yes
Lausanne	Diastrophic dysplasia, Achondrogenesis 1B, Atelosteogenesis type II, Multiple Epiphyseal Dysplasia (R) Other DTD variant disorders	*DTDST (SLC26A2)*	yes	yes
	Spondylocostal dysostosis	*Dll3*	yes	yes
	Cartilage-hair hypoplasia	*RMRP*	yes	yes
	Progressive pseudorheumatoid dysplasia	*WISP-3*	yes	yes
Mainz	Metaphyseal chondrodysplasias Schmid type Other SMD variants	*COL10A1*	yes	yes
	Dyschondrosteosis Léri-Weill Langer mesomelic dysplasia	*SHOX*	yes	yes
	Acromesomelic dysplasias - Grebe type Hunter-Thompson type	*CDMP1*	yes	yes
	Osteoonychodysplasia (Nail-Patella syndrome)	*LMX1B*	yes	yes
	Cleidocranial dysplasia	*OSF2/CBFA1*	yes	yes

Table 2. These services are currently being provided free of charge to genetic centres (or equivalent) within EU Member States or States associated with the EC 5th Framework Programme (with the exception of OI).

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Jacky Taylor

Participants of the 2nd ESDN Management Meeting, June 2002, Ghent, Belgium

Research on Skeletal Dysplasias

ESDN research: will use an integrated approach for identifying the molecular genetic basis and cell-matrix pathology of skeletal dysplasias. This will be achieved through 6 complimentary programmes:

- Identifying novel genes involved in human skeletal dysplasias by EST screening.
- Identification and characterisation of susceptibility and modifier genes.
- Genetic linkage studies and positional candidate cloning.
- Investigation of candidate genes.
- Analysis of protein function and dysfunction through structural & functional studies.
- A proteomics approach for candidate gene identification and investigating the molecular cell pathology of skeletal dysplasias.

Announcements

- Dr Geert Mortier is collecting familial and sporadic cases with osteopoikilosis for mapping the condition. If you are interested in a collaboration on this project, please contact geert.mortier@rug.ac.be
- Members of the Department of Genetics Paris, France, have now identified the gene for Dyggve-Melchior-Clausen syndrome (OMIM 223800). This new gene encodes a new protein called dymeclin with an unknown function (El Ghouzzi et al. Hum Mol Genet, 2003 in press).

ESDN in Focus

Part I:

"The Clinical and Radiographic Management Group" (ESDN-CRMG)

The CRMG comprises some of the world's leading experts in the clinical and radiographic diagnosis of skeletal dysplasias: -
Geert Mortier (Ghent); Jurgen Spranger and Bernhard Zabel (Mainz); Michael Wright (Newcastle); Andrea Superti-Furga (Lausanne); Martine Le Merrer and Valerie Cormier-Daire (Paris)
ESDN-CRMG can provide expert diagnostic services and will act as a forum for the discussion of individual cases in which the clinical diagnosis is inconsistent with the results obtained by molecular testing. To ensure the efficient running of this group a Clinical Radiographic Review Facilitator is located in Lausanne.