European Skeletal Dysplasia Network www.esdn.org

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ESDN Newsletter

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Don't forget to use your ***ESDN Case Manager*** account. If you would like to become a user or find out more- please let us know, email: info@esdn.org

A message from the ESDN Co-ordinator - Mike Briggs

I would like to take this opportunity to thank you all for your continued support and interest in the European Skeletal Dysplasia Network and to wish you a successful 2006.

As many of you are aware the current EC funding period for ESDN finished in December 2005, however, I am very happy to tell you that thanks to the continued commitment by all the partners of ESDN the network will continue to provide a clinical and molecular diagnostic service for skeletal dysplasias.

- Jacky Taylor (Jacky.Taylor@CMMC.nhs.uk) will continue as ESDN project administrator and will be based in the European Projects Office in the Nowgen Centre in Manchester.
- The clinical and radiographic evaluation of cases will continue through the ESDN Case Manager and the co-ordination of this facility will be performed jointly by Andreas Zankl (andreas.zankl@gmail.com) and Sheila Unger (sheila.unger@uniklinik-freiburg.de). Please note that Sheila will be the first point of contact should you need to send hard copies of x-rays with your referrals.
- All contact details will soon be available on the ESDN web site (www.ESDN.org), which is currently undergoing a revamp.
- Please be aware that as referring clinicians you should continue to supply high quality clinical x-ray data to the ESDN Clinical and Radiographic Review Group using the ESDN Case Manager system. This will allow us to review your cases as quickly as possible and make a recommendation. Please also be aware that some of the ESDN laboratories are now charging for the molecular analysis. This will continue until we can secure future funding.

Once again many thanks for your continued support of ESDN
Mike Briggs

Results of the ESDN user feed back questionnaire

Thank you to those of you who completed the ESDN user feed back questionnaire. The information you provided was very useful and important to us. We will be presenting the information to the European Commission and using it to further develop the existing network and the ESDN Case Manager.

- An interesting outcome of the questionnaire was that over 90% of you said that the confirmation of a definite final diagnosis was in itself an important benefit to the patient.
- In addition, 90% said a confirmed diagnosis was important to provide correct genetic counselling of the patient.
- 71% of you also indicated that a confirmed diagnosis is helpful in the efficient management of the patients' condition.
- 44% highlighted that a confirmed diagnosis was interesting at an academic level.

Further results:

- The majority of you (58%) see 1-10 skeletal dysplasia patients per year whilst a further 35% see 10-50. Only 5% see 50-100 with 2% seeing over 100. This confirms a common impression that the majority of practicising physicians see a relatively small number of patients with skeletal dysplasias in their day to day work.
- 85% of you have referred 1-10 skeletal dysplasia patients to the ESDN. This suggests that most people are using the ESDN for an expert opinion on a small number of cases rather than as a method for obtaining a diagnosis in routine cases. Together these points prove to us the need for a system like ESDN.
- The majority of respondents (85%) said they would be prepared to pay for molecular genetic testing services for skeletal dysplasias but only when carried out in a quality assured laboratory.
- 45% of respondents said they would be prepared to pay a maximum of €600 for molecular genetic testing, 17% said they would pay €800.
- 70% of you said the ESDN Case Manager was easy to use which is reassuring but you will be glad to hear that we hope to make further improvements to the ESDN Case Manager. These should make it even easier to use, and will take into account as many of your very useful comments as we can.

All your additional comments and support were gratefully received. Please continue to send in any comments or questions if you have them (email: info@esdn.org).

ESDN in Focus

Part VIII: "Details on the Oulu group"

In Oulu the ESDN diagnostic and research work is carried out in Leena Ala-Kokko's laboratory located at the Department of Medical Biochemistry and Molecular Biology, University of Oulu.
As part of the ESDN the Oulu group perform a molecular diagnostic service for the type XI collagenopathies (Stickler and Marshall Syndrome, OSMED).
Research in the field of skeletal dysplasias has concentrated on phenotype-genotype correlation on Stickler/ Marshall syndrome and MED in collaboration with the Gent and Manchester groups, respectively. Since high myopia is a common feature in syndromic disorders such as Stickler, we have extended our research on the molecular genetics of high myopia. One of the goals is to study the role of the small leucine rich repeat proteins (SLRPs) expressed in the eye and the exon 2A of COL2A1 in high myopia.
Our other research interests include the genetics of common musculoskeletal disorders: osteoarthritis, intervertebral disc disease and osteoporosis. Further information about our research can be found on: http://www.biocenter.oulu.fi/projects/alakokko.html

Fig 1. The Oulu group. Including Leena Ala-Kokko, Minna Mannikko, Marja Majava and colleagues.

Fig 2. A view of the University of Oulu. Oulu is located on the shore of the Gulf of Bothnia in Finland.

Announcements

The UK Melorheostosis Association
Go to www.melo.eu.com and find out about the UK Melorheostosis Association. This association is a not-for-profit organisation dedicated to finding the cause, treatments and cure for melorheostosis and associated rare bone diseases.

ESDN Case Manager - Please contact us by email at info@esdn.org if you would like to become a user, stating your position, full mailing address with telephone and fax numbers.

Orphanet (www.orpha.net) is an online resource, which aims to contribute to the improvement of diagnosis, care and treatment of patients with rare diseases. Information on ~4000 conditions is listed on Orphanet from a number of disciplines including: clinical and molecular genetics, cytogenetics, biochemistry, haematology, immunology or any other discipline involved in the research, diagnosis and treatment of rare diseases. For each condition, Orphanet lists relevant testing laboratories, specialist clinics, research projects, clinical trials and support groups. Orphanet is free to use, for more information check out the website: www.orpha.net or turn to page 6 of the November 2005 (Issue 31) BSHG newsletter. For anyone in the UK wishing to have information listed on the site please contact **Emma Gillaspy** (Emma.Gillaspy@CMMC.nhs.uk).