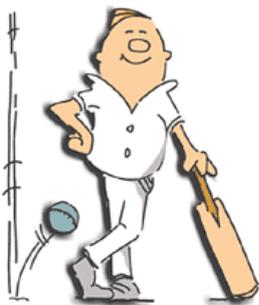




ESDN Newsletter



Introducing the ESDN-CMW

- An Electronic Communication, management and Web-portal system (ESDN-CMW) is being developed for ESDN. Users will be able to refer cases online to ESDN using this system. This tool will also allow us to manage ESDN to a high level of efficiency.
- To become an ESDN user please contact the ESDN Project Administrator, Jacky Taylor: info@esdn.org
- The site is currently undergoing testing but we hope to release the first online version in October.
- The following screen shots illustrate some of the features of the new system.

Editorial

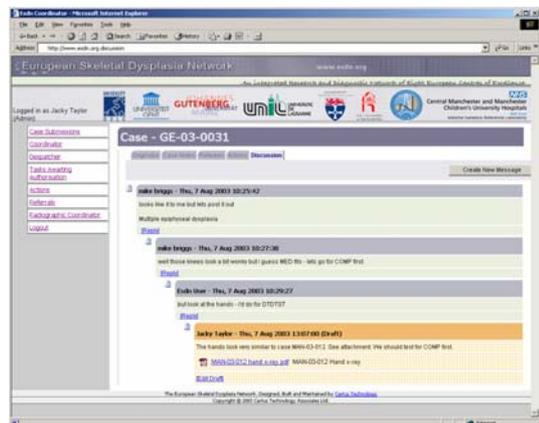
Welcome to the Summer 2003 issue of the European Skeletal Dysplasia Network (ESDN) newsletter.

The main focus of this issue will be to introduce our new interactive website which is being developed as we speak. If you would like to become a user please let us know: info@esdn.org. We are hoping it will be ready to use by everyone in October as it is currently undergoing testing.

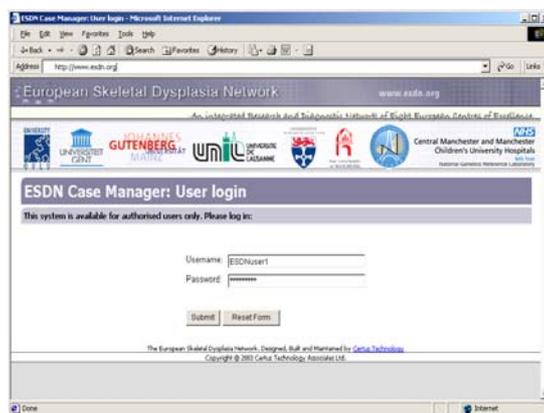
Further interest: We'll give you 3 guesses as to who is into cricket (see ESDN in focus part III for clues)



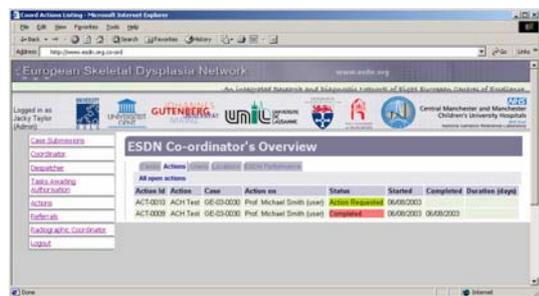
After you have submitted your case with all the appropriate information, it will be assigned a unique identifier and referred to ESDN.



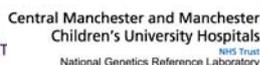
The case will be discussed by the ESDN Clinical Radiographic Management Group using an electronic discussion forum. Once a consensus has been agreed upon, the outcome will be communicated to the referring clinician who can then participate in the discussion.



Log onto the secure system with your personal user name & password.



The ESDN co-ordinator oversees the actions associated with each case, whether they be requests for further information, to obtain a DNA sample from the patient, routing of the sample to the appropriate laboratory for testing or communication of the final results.





ESDN in Focus

Part III: "Details on the Manchester Research group"

In Manchester the ESDN-Research component is performed in Mike Briggs's laboratory in the Wellcome Trust Centre for Cell Matrix Research at the University of Manchester (<http://www.wtccmr.man.ac.uk/>). They are using a multidisciplinary approach to study the pathobiology of pseudoachondroplasia and multiple epiphyseal dysplasia.

- Dr Faye Barker and Dr Gail Jackson are using genetic linkage and DNA sequencing to identify novel genes and mutations that can cause MED and related conditions including Perthes disease and osteoarthritis.
- Dr Sally Cotterill, Helen Attisha and Natalie Robinson are using *in vitro* approaches to study the effect of specific disease causing mutations on the structure and function of the relevant gene products.
- Dr Matt Leighton and Kasia Pirog are developing 'knock-in' *in vivo* models of PSACH and MED to understand the disease processes and identify other genetic factors which might influence disease severity.

Further information about their research can be found on the lab home page: <http://www.biomed2.man.ac.uk/briggsm/>



NEWSFLASH

- Last year ESDN received over 500 patient referrals, performed 290 diagnostic tests and identified mutations in 150 patients.
- In the first half of this year ESDN has received 307 patient referrals, performed 179 diagnostic tests and identified disease causing mutations in 124 patients.

Announcements

- Dr Valérie Cormier-Daire, Dr Frances Goodman and Dr Colin Johnson are collecting familial and sporadic cases of **asphyxiating thoracic dystrophy (Jeune syndrome)** in a collaborative effort to map and clone the causative genes, refine diagnostic criteria and identify possible prognostic features. If you are interested in collaborating on this project, please contact cormier@necker.fr, fgoodman@hgmp.mrc.ac.uk or c.a.johnson@bham.ac.uk.
- Dr Ed Wraith from the Royal Manchester Children's Hospital and Dr Charles Peters of Minneapolis University will be jointly hosting an Expert Meeting on the Orthopaedic Management of Mucopolysaccharide and Related Diseases, to be held in Manchester, UK, 2-4 Oct 2003. For further information please contact: Cheryl Pitt C.Pitt@mppsociety.co.uk.
- In the **NEXT ISSUE** find out more about our **new interactive ESDN website**. Also **ESDN in Focus, Part IV: "details on the Newcastle group"**.