

# ESDN Newsletter

## Update on the ESDN-CMW



### Editorial

Welcome to the Spring 2004 issue of the European Skeletal Dysplasia Network (ESDN) newsletter.

In this issue:

- news on the ESDN Case Manager - the new efficient way of referring cases to the ESDN. If you would like to become a user please let us know, email: [info@esdn.org](mailto:info@esdn.org) stating your full mailing address with telephone and fax numbers.

- welcome Christine Hall, a new member of the **ESDN Clinical and Radiographic Management Group**.

- find out about the "Swiss ESDN node" in Part V of "ESDN in Focus". (If you are confused about the Swiss node please email: [asupert@chuv.unil.ch](mailto:asupert@chuv.unil.ch))

The ESDN electronic Communication, Management and Web-portal system (ESDN-CMW), also known as (AKA) the ESDN Case Manager, is a secure interactive website which allows us to manage ESDN diagnostics to a high level of efficiency whilst at all times protecting the confidentiality of all patient referrals and ensuring that adequate consent procedures are in place.



Full official use of the ESDN Case Manager, to registered users, began in February 2004.



We currently have 72 verified, registered users.



We are now up to case number ESDN-00079, and all is going well.



Registered users can use the ESDN Case Manager to submit known and unknown skeletal dysplasia cases directly to the ESDN.



Relevant information will automatically be referred to a panel of skeletal dysplasia experts (the **ESDN-CRMG**).



The Clinical-Radiological Review Co-ordinator (email: [Andreas.Zankl@hospvd.ch](mailto:Andreas.Zankl@hospvd.ch)) ensures the efficient running of the **ESDN-CRMG**, through the ESDN Case Manager.



For further information about the ESDN Case Manager and to become an user, please contact the ESDN Administrator: Jacky Taylor, email: [info@esdn.org](mailto:info@esdn.org)

## ESDN-CRMG

**Welcome Christine Hall, a new member of 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: -

Andrea Superti-Furga (Lausanne); Geert Mortier (Ghent); Jurgen Spranger and Bernhard Zabel (Mainz); Michael Wright (Newcastle); Martine Le Merrer and Valerie Cormier-Daire (Paris). We are now pleased to welcome Christine Hall (London) on board.

ESDN-CRMG can provide expert clinical diagnostic services and will act as a forum for the discussion of individual cases through the ESDN Case Manager.

The Clinical-Radiological Review Co-ordinator ([Andreas.Zankl@hospvd.ch](mailto:Andreas.Zankl@hospvd.ch)) ensures the efficient running of the ESDN-CRMG, through the ESDN Case Manager.

# ESDN in Focus

## Part V: "Details on the Lausanne group, AKA the Swiss ESDN node "

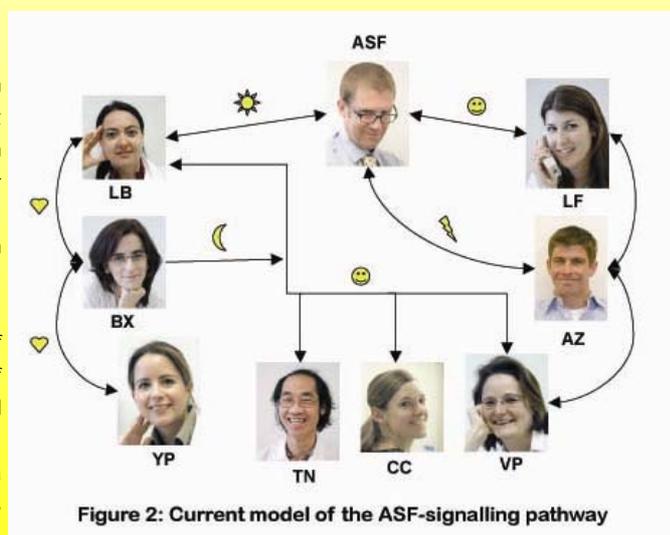
The Swiss node of the ESDN is based at the Division of Molecular Pediatrics at the University Hospital of Lausanne ([www.pediatrics.ch](http://www.pediatrics.ch)), on the shores of Lake Geneva. The Division is involved in clinical care and diagnosis of pediatric patients with metabolic, skeletal, and other genetic diseases. Within ESDN, the Lausanne team has a triple role: it staffs the Clinical-Radiological Review Co-ordinator, who ensures that all necessary clinical and radiological information is available to the ESDN experts and oversees the clinical-radiological review process. It also provides a clinical-radiological expert and laboratory services. Diagnostics focuses on 4 skeletal dysplasias: the Diastrophic Dysplasia spectrum, Cartilage Hair Hypoplasia, Spondylocostal Dysostosis and Progressive Pseudorheumatoid Chondrodysplasia.

**Figure 1** (left to right): Dr Belinda Xavier (post-doc), Carole Chiesa (technician), Dr Luisa Bonafé (head of the diagnostic and research laboratory), Prof. Andrea Superti-Furga (head of the division and one of the clinical-radiological experts of ESDN), Lucia Fortunato (secretary), Valerie Pittet (technician), Dr Yannick Perrin (pediatrician), Dr Andreas Zankl (Clinical-Radiological Review Co-ordinator), Thanh Nguyen (technician).



The interactions within the Swiss ESDN node are very complex and still await full clarification. **Figure 2**, below, summarizes our current understanding of the ASF-signalling cascade.

- **ASF**: highly reactive transmembrane receptor molecule involved in bone metabolism. Activating ligands seem to be quite heterogeneous but many pertain to the JPEG family. Attempts to knock-out ASF have been unsuccessful. Activities transmitted to cytoplasm via LF, LB and AZ, or redirected towards other cells by EMAIL signalling.
- **LB**: usually first transducer in the ASF-activated pathways. Acts as an effector with feed-back mechanisms to stabilize the system. Highly involved in the stress response.
- **AZ**: The physically most robust member of the pathway. A modifier of the ASF-signalling pathway, particularly for cybernetic output. Has bursts of activities that are typically silenced by ASF. Also known for ASF-unrelated submarine activity.
- **BX**: A recently purified member of the ASF-signalling pathway with a peculiar nocturnal expression pattern. Relatively resistant to ASF or LB activation.
- **CC/VP/TN**: Effective multifunctional proteins with high DNA sequencing affinity. Preferentially activated by prenatal events, highly involved in the LB-ESDN crosstalk.



**Figure 2: Current model of the ASF-signalling pathway**

# Announcements

- Do you want to use the new secure electronic ESDN Case Manager to refer your cases to the ESDN? If so Email: [info@esdn.org](mailto:info@esdn.org) with your full mailing address, telephone and fax numbers
- Do you have any announcements for the next ESDN newsletter? If so Email: [info@esdn.org](mailto:info@esdn.org)