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## Project Outline

The DYSCERNE project aims to raise current standards for the diagnosis and management of rare dysmorphic conditions. A Network of Centres of Expertise in Dysmorphology has been established and a web-based electronic dysmorphology diagnostic system (DDS) developed, which enables clinicians to submit difficult to diagnose cases electronically for review by an Expert Panel. The DDS system will increase capacity and access to accurate diagnosis, improving definition and delineation of rare dysmorphic syndromes. Also, guidelines for the clinical management of selected dysmorphic conditions are being produced. These are being developed by expert consensus groups, and include criteria for diagnosis, protocols for review and screening, and information on management at different life stages. After piloting, the guidelines will be disseminated widely and their use evaluated, providing an evidence base for the development of future rare disease management strategies. The project will serve as a model for future EU Networks of Expertise for Rare Diseases, and Management. The project is coordinated by the University of Manchester, a centre with a strong record of leadership in Dysmorphology.

## Background

- Dysmorphology is the study of birth defects or malformations which form recognisable patterns of growth, development, behaviour and physical features.
- There are more than 2,500 of these rare and difficult to diagnose conditions.
- The rarity of these conditions means making a diagnosis can be problematic or delayed.
- Diagnosis is essential for management and for accurate information and counselling.

## Strategic Objectives

- Improve the diagnosis of dysmorphic conditions throughout the EU.
- Improve the definition and delineation of dysmorphic conditions.
- Develop a methodology for developing management guidelines for rare diseases.
- Link with and build on the experience of related European initiatives such as Orphanet, the European Skeletal Dysplasia Network and EUROCAT.

## Work Packages

### Project coordination (WP1)

- An infrastructure has been established to manage all administrative, technical, communication and financial aspects of the project.
- Protocols have been developed to facilitate effective communications between the project team, partners and collaborators. A combination of regular team meetings, network meetings, workshops and electronic communication has been utilised to achieve this.
- Systems and forms have been produced to help collect information and data to assist with project monitoring, reporting and evaluation.

### Dissemination (WP2)

- A project website ([www.dyscerne.org](http://www.dyscerne.org)) has been developed containing background information, news, contact details and links to other websites of interest.
- A presentation has been distributed to the Partners, which they use to introduce the project and raise the profile in the international clinical genetics community.
- Over 40 Posters and talks have been presented by DYSCERNE main partners at international conferences including EURODIS 2007, ESHG 2007 & 2008, and also at national and regional meetings.
- Formal links with Orphanet have been established, and the two organisations have agreed to work collaboratively wherever possible.

### Project Evaluation (WP3)

- The progress of individual work packages' is monitored against a detailed timeline.
- Collection of data on outcomes and indicators is ongoing and will be used inform the design of suitable tools to assist in the final project evaluation.

### DYSCERNE Network (WP4)

The DYSCERNE network comprises:

- 6 Main Partners and an Expert Diagnostic Panel, giving a total of 27 members from 20 centres of expertise for dysmorphology.
- Additionally there is an extended network of 78 centres acting as case submission nodes, in 30 different European countries.



Figure 1: The DYSCERNE Network

### DDS Development (WP5)

- System specifications and a detailed workflow diagram (fig 3) have been produced.
- Guidelines for the submission of cases have been developed.
- Patient and Clinician Information leaflets in the Partner languages (Dutch, English, French, Italian and Polish) are available. Further language versions are in development.



Figure 2: DDS Information Leaflets

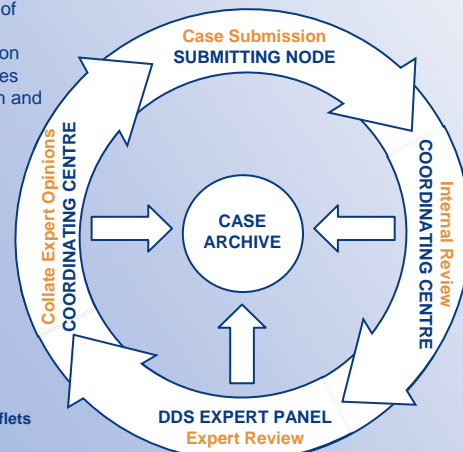


Figure 3: An overview of the DDS workflow

- Policies for data protection, privacy, and secure access to the DDS have been developed.
- The first of a proposed series of Educational resources (A guide to the examination of a fetus with congenital abnormality) has been developed and will be accessible to registered users on the website.

### Launch and Operation of the DDS (WP6)

- The initial DDS pilot began in December 2008 and involved 7 submitting nodes submitting a limited number of cases which were reviewed by the DYSCERNE Partners.
- After this successful test of the planned workflow and procedures, the full Expert Panel began reviewing cases in February 2009.
- The number of centres submitting cases will be increased in phases from March 2009, with all 78 submitting nodes having access to the DDS by June 2009.
- The case archive will be regularly reviewed to facilitate definition and delineation of new and existing condition.

## Summary

Having a correct diagnosis is the cornerstone of patient management, enabling clinicians to locate other patients with the same condition, share clinical expertise, and increase individual and collective knowledge about these rare conditions.

Through the development of management guidelines and educational resources, and the launch of the Network of Centres of Expertise and the DDS, the DYSCERNE project will raise current standards for the diagnosis, management and information dissemination of rare dysmorphic syndromes.

Where can I find out more? Our website is at [www.dyscerne.org](http://www.dyscerne.org), or you can contact: Pamela Griffiths, Project Manager, [pamela.griffiths@cmmc.nhs.uk](mailto:pamela.griffiths@cmmc.nhs.uk), Tel: +44 161 276 3209.