



J Clayton-Smith¹, R Day¹, S Gardner¹, P Griffiths¹, K Strong¹, D Donnai¹, B Kerr¹, K Metcalfe¹, H Brunner², B Dallapiccola³, K Devriendt⁴, M Krajewska-Walasek⁵, N Philip⁶,
¹University of Manchester, UK ²UMC St Radboud, Netherlands ³IRCCS – CCS, Italy ⁴KU Leuven, Belgium, ⁵IP-CZD, Poland ⁶Hopitaux de Marseille, France

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 will be established and a web-based electronic dysmorphology diagnostic system (DDS) will be developed, enabling 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 will be produced. Developed by expert consensus groups, these will 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 dysmorphic conditions means that even in EU designated Centres of Expertise, experience may be limited and diagnosis might be delayed.
- Diagnosis is essential for management and for accurate information and counselling.

Strategic Objectives

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

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 that to assist with project monitoring, reporting and evaluation.
- 1st year interim reports on have been completed and submitted to PHEA.

Progress on Work Packages

DDS Development (WP5)

- The elements of the system have been specified and a detailed workflow diagram produced.
- Guidelines for the submission of cases have been developed.
- Patient Information & Consent forms and Clinician Information leaflets have been produced and translated into the Partner languages (Dutch, English, French, Italian and Polish).

Dissemination (WP2)

- A project website (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 has raised the profile of the project in the international clinical genetics community, and widened network participation.
- 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.
- A detailed report on dissemination activities has been included in the year 1 interim report.

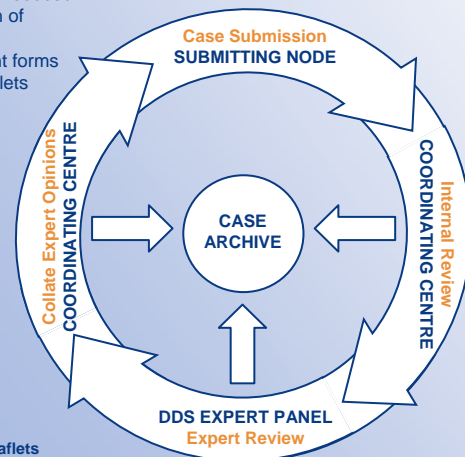


Figure 2: DDS Information Leaflets

- A data protection and privacy policy has been developed.
- System development meetings between the system designers and the Lead Partner have taken place to define and refine system design and functionality.
- A policy for secure access, similar to that used by the European Skeletal Dysplasia Network (ESDN) has been developed and will ensure only authorised users can access the system.
- The first of a proposed series of Educational tools (A guide to the Examination of the Dysmorphic Fetus) has been developed and is accessible to registered users on the website.

Figure 3: An overview of the DDS workflow

Project Evaluation (WP3)

- Monitoring of individual work packages' progress against a detailed timeline of all aspects of the project is ongoing.
- 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 six Main Partners and an Expert Diagnostic Panel, giving a total of 26 members from 20 centres of expertise for dysmorphology.
- There is also an extended network of centres with varying levels of experience in dysmorphology who will act as case submission nodes. There are currently 78 centres in this extended network which includes at least one centre from every EU Country.



Figure 1: The DYSCERNE Network

Guideline Development (WP7)

- The DYSCERNE Partners have identified four rare conditions where there is a need for clinical management guidelines.
 - Angelman Syndrome
 - Kabuki Syndrome
 - Noonan Syndrome
 - Williams Syndrome
- For each condition an Expert Review group will hold consensus meetings and draw up draft guidelines.
- Guidelines will include; criteria for diagnosis, protocols for review and screening, and information on management at different life-stages.
- The guidelines will be piloted in participating centres and their impact and their usage audited and evaluated.

Launch and Operation of the DDS (WP6)

- The pilot of the DDS began in the Summer of 2008 with the full launch of the system anticipated in late Autumn 2008.

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. The DYSCERNE project will raise current standards for the diagnosis, management and information dissemination of rare dysmorphic syndromes. This Network aims to achieve these objectives by:

- Forming a European Network of Centres of Expertise for Dysmorphology.
- Designing & implementing an electronic case submission system for difficult to diagnose cases.
- Developing guidelines for specific syndromes, and a valid guideline development methodology for rare diseases.
- Creating tools to facilitate the education of non-dysmorphologists.

Where can I find out more?

Our website is at www.dyscerne.org, or you can contact: Pamela Griffiths, Project Manager, pamela.griffiths@cmmc.nhs.uk, Tel: +44 161 276 3209.



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Leader Organisation: University of Manchester, UK
 Contact Person: Pamela Griffiths, Project Manager
 Web site: www.dyscerne.org