

# A European Network of Centres of Expertise for Dysmorphology



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## Background

Dysmorphology is the study of birth defects or malformations which form recognisable patterns of growth, development, behaviour and physical features.

Over 2,500 rare and difficult to diagnose conditions presenting with patterns of birth defects have been identified. The rarity of these dysmorphic conditions means that even in EU designated Centres of Expertise, experience may be limited and a diagnosis may not be made. Correct diagnosis is the cornerstone of patient management, enabling clinicians to locate other patients with the same condition, share clinical experience, and increase individual and collective experience about these rare conditions. For patients and their families, the importance of having a diagnosis cannot be emphasised. It can help them come to terms with the condition, reassure them that they are receiving appropriate care, and may facilitate making contact with other affected individuals and families for support and advice.

## Introducing Dyscerne

The DYSCERNE Network aims to raise current standards for the diagnosis, management and information dissemination of rare dysmorphic syndromes. The project is funded by DG Sanco, Rare Diseases Programme and comprises six designated centres of expertise for dysmorphology. The project's lead partner, the University of Manchester, will be the coordinating and managing centre for the Network. DYSCERNE will link with other European Rare Disease Networks including Orphanet.

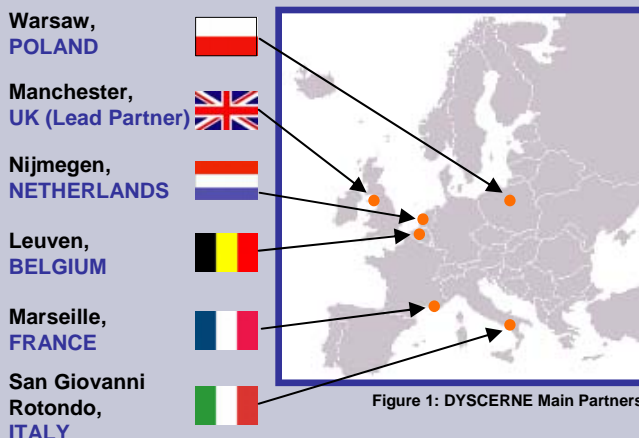


Figure 1: DYSCERNE Main Partners

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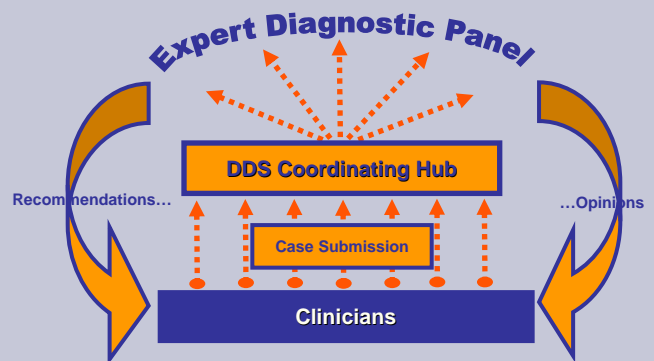
## Network Aims

- ★ Form a network of centres of expertise for dysmorphology
- ★ Establish a web-based, electronic Dysmorphology Diagnostic System (DDS)
- ★ Develop management guidelines for selected dysmorphic syndromes

## DDS Overview

The Dysmorphology Diagnostic System (DDS) will enable clinicians throughout the EU to submit cases electronically for diagnosis via a 'node'. Initially there will be 50 designated nodes: at least one in every EU member state. An on-line training package, guidelines for submission and on-line proformas will ensure that submissions are made in a standard format using preferred descriptive terms. The system will allow clinicians to upload photographic images and results of investigations including imaging studies to a secure, searchable archive. After 7 days, recommendations and opinions from the expert diagnostic panel will be collated and sent back to the referring clinician.

Figure 2: Electronic Dysmorphology Diagnostic System



## Guideline Development

The Network will carry out a scoping exercise to identify four dysmorphic conditions where guidelines for clinical management are needed. An expert review group for each condition, will be created and consensus meetings held to draw up draft guidelines. The 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.