



## **Dysmorphology Diagnostic System (DDS)**

### **Information for Clinicians**

**DYSCERNE: A Network of Centres of Expertise for Dysmorphology**

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European Commission Public Health Executive Agency (DG Sanco) Project: 2006122

- A European Network of Centres of Expertise for Dysmorphology

# Introducing DYSCERNE

## A European Network of Centres of Expertise for Dysmorphology

DYSCERNE is a European project which aims to improve the diagnosis, management and availability of information on rare dysmorphic syndromes. The project is funded by the European Commission Public Health Executive Agency and comprises six main partners from existing Centres of Expertise for dysmorphology who are:

- Jill Clayton-Smith - University of Manchester (UK)
- Han Brunner – Universitair Medisch Centrum Sint Radboud (Netherlands)
- Bruno Dallapiccola - Ospedale Casa Sollievo Della Sofferenza (Italy)
- Koenraad Devriendt - Katholieke Universiteit, Leuven (Belgium)
- Malgorzata Krajewska-Walasek - Instytut Pomnik-Centrum Zdrowia Dziecka, Warsaw (Poland)
- Nicole Philip, Assistance Publique – Hôpitaux de Marseille (France)

The project's lead partner, the University of Manchester, will be the coordinating centre for the Network.

The main aims of the DYSCERNE project are:

- To identify Centres of Expertise for dysmorphology and create a formal network of these centres.
- To develop a web based electronic Dysmorphology Diagnostic System (DDS)
- To develop diagnostic and clinical management guidelines for selected dysmorphic syndromes.

# **What is the purpose of the Dysmorphology Diagnostic System (DDS)?**

The electronic Dysmorphology Diagnostic System (DDS) will link over 50 European Centres of Expertise in dysmorphology, creating a powerful diagnostic service and educational resource.

A core group of experts (the DDS Expert Panel) will collaborate to review your patient's medical history, and will provide recommendations and opinions on possible diagnoses, and suggest further investigations and management strategies.

## **Who is DDS for?**

Clinicians with patients who have dysmorphic features and/ or malformations where the diagnosis is unclear or confirmation of a rare diagnosis is required.

These cases would usually have been presented at local, national or international meetings without a diagnosis being confirmed.

## **What are the benefits of using DDS?**

### **For Clinicians**

The DDS offers unique access to a range of expert opinions and management advice.

An on-line teaching resource available only to DDS registered users will improve diagnostic skills in the evaluation of rare dysmorphic syndromes.

Regular review of the submitted cases in the DDS Archive will lead to improved definition and delineation of new and existing conditions.

## **For Patients**

Receiving an accurate diagnosis helps to alleviate uncertainty and anxiety, and reassures patients that they are receiving appropriate care and advice.

Accurate and early diagnosis may help with access to new treatments and therapies as they become available.

Accurate diagnoses facilitate genetic counselling and discussion of reproductive options.

Patients can be put in touch with appropriate support groups or other families and individuals diagnosed with the same condition.

## **Does a detailed medical history need to be submitted?**

For the DDS Expert Panel to give an accurate opinion they need full details of your patient's medical history, examination findings, results of any investigations or specialist opinions and clinical photographs.

Please submit as much background information as possible. As a minimum we need to know the patient's sex, age, ethnicity, maternal & paternal age and number of siblings (and if they are also affected) as well as current growth parameters (serial measurements would also be useful).

We will also ask questions about the patient's birth and antenatal history, early & general development and any investigations carried out. If you do not have this information the Expert Panel can still look at your patient's case but any background information you can give us will help the DDS Expert Panel to make more informed decisions about possible diagnoses and management.

Good quality photographs are important. Images should be in focus and relevant to the case. As the appearance of some syndromes can change as the patient ages, photographs of the subject at different ages may be useful. Photographs should be uploaded to the DDS site during the case submission process.

To facilitate storage and retrieval of patient data, we are asking case submitters to describe their patients' symptoms using London Dysmorphology Database ([www.lmdatabases.com](http://www.lmdatabases.com) and [www.dyscerne.org](http://www.dyscerne.org)) terminology.

## **What happens after a medical history has been submitted to DDS?**

The DDS coordinating centre will check the submission to make sure it is suitable for the DDS. Your patient's anonymised medical history and photographs will then be reviewed by the DDS Expert Panel.

The Experts will send their recommendations and opinions on possible diagnoses to the coordinating centre.

The coordinating centre will collate the expert opinions into a summary report to the case submitter.

## **What about storing the information?**

If the patient agrees, their medical history and photographs will be stored in the DDS Archive. Anonymised information from the Archive will be used to help measure the effectiveness of the system and improve the DDS for future users. It will also be used to define and classify new and rare dysmorphic conditions and increase understanding and knowledge.

If patients choose not to allow their anonymised medical history and clinical photographs to be stored in the DDS Archive, their decision will not affect the ability of the Expert Panel to offer an opinion on their case. Patients are free to change their decision at any time and can write to us at the address on the front of this leaflet.

## **How do I submit a patient's case to DDS?**

Please ask your patient to read the DDS Patient Information & Consent Form. If they are happy for their medical history and photographs to be sent to the DDS please ask them to complete and sign two copies of the Consent Form. One copy should be filed with the patient's medical records, the other is for the patient to keep. Cases without the appropriate consent will not be accepted.

Initially, only a limited number of centres (nodes) will be licensed to submit cases to DDS. In each node one person will be coordinating submissions so you will need to refer the case to that person.

If you are not located within a submitting centre, you will need discuss your case with your nearest submitting centre who will submit it on your behalf if you download and complete the case submission form. There is a list of submitting centres on the DDS website.

During submission, partially completed cases can be saved and the submission process completed later. At the end of the submission process there is the facility to print a copy of the submitted data for inclusion in the patient's medical notes.

Applications for a licence to become a submitting centre can be sent to the DYSCERNE coordinating office at the address on the front of this leaflet.

## **Policy for publication of data held on DDS Archive**

We may ask for your consent to mention instructive cases at national and international dysmorphology meetings as part of a DYSCERNE presentation. We will not use your patient's information in a presentation or publication without your prior consent, and if your case is used you will be appropriately acknowledged. Submitting clinicians will be encouraged to publish case reports in appropriate journals.

Any publication which refers to diagnoses, recommendations or management plans made by the DDS Expert Panel or to data held in the DDS Archive should specifically acknowledge the DYSCERNE Project (**DYSCERNE: A European Network of Centres of Expertise for Dysmorphology, funded by the European Commission Public Health Executive Agency (DG Sanco), Project No. 2006122**)

## **Where can I find out more?**

You can contact us at the address or telephone number on the front of this leaflet. There is more information about DYSCERNE and how to submit cases to the DDS on our website at **[www.dyscerne.org](http://www.dyscerne.org)**

**Thank you for reading this information leaflet**

## DYSCERNE Partners:



Assistance Publique  
Hôpitaux de Marseille

## Supporting Organisations:



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