



# An electronic Dysmorphology Diagnostic System (DDS)

S Gardner<sup>1</sup>, P Griffiths<sup>1</sup>, K Strong<sup>1</sup>, R Day<sup>1</sup>, D Donnai<sup>1</sup>, B Kerr<sup>1</sup>, K Metcalfe<sup>1</sup>, H Brunner<sup>2</sup>, B Dallapiccola<sup>3</sup>, K Devriendt<sup>4</sup>, M Krajewska-Walasek<sup>5</sup>, N Philip<sup>6</sup>, J Clayton-Smith<sup>1</sup>  
<sup>1</sup>University of Manchester, UK <sup>2</sup>UMC St Radboud, Netherlands <sup>3</sup>RCCS – CCS, Italy <sup>4</sup>KU Leuven, Belgium, <sup>5</sup>IP-CZD, Poland <sup>6</sup>Hopitaux de Marseille, France

## Background

- The rarity of dysmorphic conditions means that even in Centres of Expertise, experience may be limited, resulting in delayed or uncertain diagnoses.
- One of the main aims of the DYSCERNE project ([www.dyscerne.org](http://www.dyscerne.org)) is to develop an electronic Dysmorphology Diagnostic System (DDS) which will link over 50 European Centres of Expertise in dysmorphology to form a powerful diagnostic tool and educational resource for rare dysmorphic conditions.

**DYSCERNE** is an EU funded project. The main partners are:

Jill Clayton-Smith, Bronwyn Kerr,  
 Dian Donnai & Kay Metcalfe  
**Manchester, UK (Lead Partner)**



Han Brunner  
 & Ineke Van der Burgt  
**Nijmegen, NETHERLANDS**



Bruno Dallapiccola  
**San Giovanni Rotondo, ITALY**



Koenraad Devriendt  
**Leuven, BELGIUM**



Malgorzata Krajewska-Walasek  
 & Krystyna Chrzanoswska  
**Warsaw, POLAND**



Nicole Philip  
**Marseille, FRANCE**



● = DYSCERNE Partner & Expert Panel Member  
 ● = Expert Panel Members

## 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.

## What will be the benefits of using DDS?

### For clinicians:

- Access to a range of expert opinions and management advice.
- An online teaching resource will improve diagnostic skills.
- Regular reviews of the DDS Archive will improve definition and delineation of new and existing conditions.

### For patients, accurate diagnosis will:

- Alleviate uncertainty and anxiety, and provide reassurance to parents and clinicians that care and advice is appropriate.
- Facilitate access to new treatments and therapies as they become available.
- Facilitate accurate genetic counselling and discussion of reproductive options.
- Help patients access appropriate support groups or other families and individuals diagnosed with the same condition.

## Summary

- The DDS will facilitate timely and equitable access for clinicians from all EU countries to expert opinions.
- It will increase capacity and accuracy of diagnoses and decrease time from presentation to diagnosis.
- This will improve definition and classification of rare dysmorphic conditions and promote further clinical research into these complex disorders.
- A pilot of the DDS will begin in June 2008, with the full launch of the system anticipated in October 2008.

## Where can I find out more?

Our website is at [www.dyscerne.org](http://www.dyscerne.org), or you can contact:

- Sara Gardner, DDS Network Coordinator, [sara.gardner@cmmc.nhs.uk](mailto:sara.gardner@cmmc.nhs.uk), Tel: +44 161 276 4248

## How will cases be submitted to the DDS?

