



An electronic Dysmorphology Diagnostic System (DDS)

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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) 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.

The DYSCERNE Network

- 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.



How will cases be submitted to the DDS?



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 that the care and advice they are receiving 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.
- The system will improve the definition and classification of rare dysmorphic conditions and will promote further clinical research into these complex disorders.
- A pilot of the DDS is due to begin in late Summer 2008, with the full launch of the system anticipated in the late Autumn.

Where can I find out more?

Our website is at www.dyscerne.org, or you can contact:
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