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DYSCERNE: a European Network of Centres of Expertise for Dysmorphology

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There are over 2,500 identifiable dysmorphic conditions, which individually are rare but together form a significant proportion of referrals to a genetic service. The rarity of these diseases means that even in Centres of Expertise established in many EU countries, experience can be limited, resulting in delay or uncertainty of diagnosis.

To improve diagnosis of dysmorphic syndromes across the EU, a formal European Network for Dysmorphology was created within the EU-funded DYSCERNE project (2007-2010) coordinated by the UK (University of Manchester). It links a total of 85 centres, including 32 centres of expertise with the remaining centres acting as case submission nodes for a web-based electronic Dysmorphology Diagnostic System (DDS). Making a correct diagnosis is essential for patient management and for providing accurate information and counselling. The DDS allows rapid access for clinicians from across Europe to expert opinions increasing accuracy of diagnosis. It will also facilitate definition and classification of rare dysmorphic disorders and promote further research. Linked to DDS, educational tools in a modern, on-line format were created aimed to guide and educate clinicians throughout Europe on key aspects of clinical dysmorphology (http://www.dyscerne.org). One of the principal activities of the DYSCERNE network was developing best practice management guidelines which use a robust methodology. Management protocols for four selected conditions: Angelman, Noonan, Kabuki and Williams Syndromes will be available on the DYSCERNE website soon.

There is a need to continue activities initiated by DYSCERNE, in particular to sustain European networking, which helps to make a correct diagnosis and continue development of management guidelines for further rare diseases.

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