



EU Project Follow-up

Following a successful pilot test, DYSCERNE's Dismorphology Diagnostic System launches in 26 European countries



The [21 November 2007](#) issue of *OrphaNews Europe* reported the debut of a new European network created to improve the diagnosis, management and information dissemination for rare dysmorphic syndromes. [DYSCERNE](#), the European Network of Centres of Expertise for Dismorphology, kicked off in October 2007. In May of this year, the project successfully launched its electronic Dismorphology Diagnostic System ([DDS](#)). All of the 76 proposed submitting centres have been invited to apply for DDS accounts and at the end of August 2009, [67 centres](#) from 26 European countries have access to the on-line dysmorphology diagnostic system DDS.

The aim of the DDS is to provide rapid and equitable access for clinicians to expert dysmorphologists' opinions throughout Europe. The DDS software links [27 expert dysmorphologists](#) from 20 European Centres of Expertise, forming a powerful web-based diagnostic resource. It allows clinicians to submit difficult to diagnose dysmorphic cases for review by this expert panel. A diagnostic report including suggestions for further investigation and clinical management of the case is prepared from the consensus of opinions received and sent to the submitting clinician.

Prior to the full launch of the DDS, a four-month pilot test of the system was conducted, which involved seven centres (Istanbul, Leuven, Manchester, Marseille, Nijmegen, San Giovanni Rotondo, and Warsaw,) all of which were invited to submit cases for review by the DDS Expert Panel. During the pilot phase, 20 cases were reviewed by an average of five expert panel members with seven cases receiving between 6-10 reviews. All the cases could be described as complex phenotypes with combinations of dysmorphic features, varying congenital abnormalities affecting different body systems and a range of neurocognitive disabilities. Diagnoses (an average of 2.5 per case) and further investigations (an average of 3 per case) were suggested for all the cases, with molecular or cytogenetic tests being suggested for 19 of the 20 cases. Summary reports of the expert opinions on each case were prepared an average of six weeks after case acceptance onto the system. The traditional route for these difficult to diagnose dysmorphic cases would be presentation at national and international dysmorphology meetings - a process which can take many months and still not result in a diagnosis. Feedback on one of the pilot cases has already confirmed a suggested DDS diagnosis at the molecular level (Coffin Lowry Syndrome with a RSK2 mutation). Another case on the system is a possible new recessive condition that will be published in the near future as a case report.

These promising early results, coupled with positive feedback from the pilot participants, indicate that the DDS provides a rapid and effective diagnostic service. Since the full launch of the DDS in May 2009, case submissions have averaged over five per month with summary reports of the expert opinions on the majority of cases being produced within 30 days of case acceptance. The DYSCERNE Coordinating Centre, based at Manchester University, UK, is anticipating that case submissions will steadily increase over the next few months as the centres with DDS accounts familiarise themselves with the system and incorporate it into their clinical practice. For more information on the DYSCERNE Network and the DDS visit the DYSCERNE [website](#) or contact [Pam Griffiths](#), Project Manager.

