

Editorial

A good thing comes to an end...

but leaves behind a defined network of dysmorphology experts, an online diagnostic tool and guidelines for four rare diseases



English scribe Geoffrey Chaucer long ago observed that all good things must end. But so soon? The [Dyscerne Network](#) launched in early June 2007 with three years of DG Sanco funding to improve the diagnostics, management, and dissemination of information for rare dysmorphic syndromes. The rationale behind the project was clear:

"Individually, most of the 2,500 recognised dysmorphic conditions are rare, but collectively they cause high morbidity, so it is important that patients are diagnosed correctly and promptly, and receive appropriate care. An experienced clinical dysmorphologist can recognize and diagnose conditions based on these features; however there are relatively few experts in clinical dysmorphology, and access to one of these specialists varies widely across the EU."

An initial goal thus identified centres of expertise for rare dysmorphologic disorders and created a network amongst those centres. Such a network could "capture complementary expertise, improve standards in diagnosis and management, facilitate patients' access to rare disease services, and act as a model for other networks of expertise." Over 80 dysmorphology experts from 31 European countries were invited to join the Network - either as members of an expert review panel or serving as contact points through which undiagnosed cases could be submitted. Other functions of the network include contributing to the definition of newly-identified dysmorphic conditions, and developing an evidence-base for the management of diseases. The network joined forces with existing organisations – specifically Orphanet and Eurocat – in order to disseminate best-practice guidelines.

The project also launched the Dysmorphology Diagnostic System (DDS), a Web-based software tool that allows clinicians to electronically submit cases for diagnostic purposes to expert dysmorphologists via an interface hosted at one of over 60 designated "nodes". Some 75 centres from 28 European countries were given access to the DDS software, which allows for the submission of undiagnosed dysmorphic cases for review via a standardised submission form, complemented by securely uploaded images and investigation results to members of a panel of 37 experts from 32 European Centres of Expertise. Within a remarkably brief 30 day period, submitting clinicians receive a report honed from a consensus of several expert opinions, complete with advice for further investigation - including molecular or cytogenetic testing, and suggestions for patient management. The traditional route for undiagnosed dysmorphology cases rests upon presentation at national or international meetings, taking place just a few times per year. A further advantage of the DDS is that it offers a wide pool of international expertise for each given case, for which experts can take the time to consult the literature and their own archives rather than trying to make an on-the-spot diagnosis that meeting consultations demand.

To date, the DDS has examined and suggested diagnoses for over 90 cases, involving a broad range of conditions – genetic, chromosomal, biochemical and environmentally caused. At least two or three new, previously-undescribed syndromes have been seen. The DDS allows experts to locate other similar cases and delineated specific phenotypes.

The final coup of the Dyscerne project is the completion of clinical practice guidelines for four rare conditions: [Angelman](#), [Kabuki](#), [Noonan](#), and [Williams](#) syndromes. These excellently executed documents discuss diagnosis and provide detailed evidence-based recommendations for each aspect of disease management and treatment, organised into sections depending on the different body systems, within which considerations for distinct age groups are presented. Key references and relevant links are also furnished. These publications are freely available from the Dyscerne website and are simultaneously disseminated by Orphanet. Dyscerne is looking for experts willing to translate these guidelines into other languages.

EU funding has now ended for the Dyscerne project. The website is being maintained on a good-will basis. Similarly, the network continues and cases are still being accepted because the experts want to keep diagnosing rare syndromes. The founders are brainstorming for ideas to keep Dyscerne alive – including local funding and engaging doctoral students at the University of Manchester. But Dyscerne is a European project – and most benefits those submitting undiagnosed cases. A diagnosis can lead to a specific, tailored treatment plan. In the maelstrom of the cross-border directive controversy, where patients may or may not be allowed to travel for expert care and diagnostics they lack at home – Dyscerne offers an elegant, cost-effective solution where neither the patient nor the expert are required to travel – just the data. Dyscerne still hopes to expand beyond Europe. Indeed, it donated its software to a practitioner in Sri Lanka who had no other means of diagnosing unknown syndromic disorders. Those interested in acquiring the DDS software (the licence cost per account is less than 100 euros), should [contact Dyscerne](#).

