

# DYSCERNE's Dysmorphology Diagnostic System (DDS): results from the first year and current progress

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DYSCERNE's online dysmorphology diagnostic system (DDS) was launched in late December 2008. After a short pilot, involving 7 centres (Istanbul, Leuven, Manchester, Marseille, Nijmegen, San Giovanni Rotondo & Warsaw,) the DDS was launched across Europe in May 2009 and by the end of its first year (December 2009) a total of 76 centres in 27 countries were able to submit their difficult to diagnose cases for review by the DDS Expert Panel.

During the DDS's first year a total of 67 cases were submitted by 21 centres. Of these 63 (94%) were accepted for review by the DDS Expert Panel. The average number of expert reviews per case was 7.8, with over half the cases (33/63) reviewed by 6 – 12 experts.

Summary reports of the expert opinions received for each case were prepared an average of 5 weeks after submission to the DDS, depending on the timing & number of reviews, with over 80% completed within 6 weeks. The summary reports included suggested diagnoses and investigations:

- 98% had diagnoses suggested with an average of 2.8 diagnoses per case. Overall a consensus clinical diagnosis was reached in 15% of cases.
- 92% cases had further investigations recommended, with molecular or cytogenetic tests suggested for 49 cases (78%).
- 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.

Cases which the expert panel reached a consensus clinical diagnosis for included Acro-Cardio-Facial syndrome, Kabuki syndrome, Cerebro-Oculo-Facio-Skeletal syndrome, Macrocephaly-Cutis-Marmorata-Telangectasia Congenita and Mhyre Syndrome.

For one case, the suggested DDS diagnosis of Coffin-Lowry syndrome was confirmed at a molecular level by the detection of a RSK2 mutation.

There are also 2 pairs of siblings on the system who have what is thought to be a new recessively inherited multiple abnormalities mental retardation syndrome. Features include short stature, microcephaly and developmental delay, malocclusion with prominent upper incisors / maxilla, scoliosis, tapering fingers and Dandy Walker malformation. Another pair of siblings on the system has raised the possibility of a separate new recessive condition. We are hoping all these cases will be published in the near future as case reports.

These results from the first year of the DDS, coupled with positive feedback from case submitters and expert panel members, indicate that the DDS is an effective diagnostic system which provides a valued service to clinicians with difficult to diagnose dysmorphic cases.

Work is currently ongoing to evaluate formally the impact of the DYSCERNE Network and the DDS on patient management.

## Current Progress

Since the end of 2009 the membership of the DDS network has increased to 86 centres in 33 countries throughout

Europe and now also includes a small number of centres in North America, Asia, Australasia and Africa.

The DDS Expert Panel of reviewers has also increased to a total of 37 allowing for more reviews per case and on average the DDS has received 6-8 case submissions per month in 2010.

We are pleased that so many colleagues are using the system and now have a total of 84 accepted cases. Over the next year we look forward to a continued increase in the use of the DDS among the existing DYSCERNE Network. The DDS is undoubtedly a success and a valuable tool for practicing dysmorphologists.

If you would like more information on the DYSCERNE Network and the on-line diagnostic service, visit our website at [www.dyscerne.org](http://www.dyscerne.org) or contact Sara Gardner, DDS Coordinator ([sara.gardner@cmft.nhs.uk](mailto:sara.gardner@cmft.nhs.uk)).