



DYSCERNE: Presenting a Methodology for Developing Clinical Management Guidelines for Rare Diseases



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The DYSCERNE Project

The DYSCERNE Network of Centres of Expertise for Dysmorphology aims to raise standards in the diagnosis, management, and information dissemination for rare dysmorphic conditions. The Network's principal activities are:

- Developing and managing a European Network of Centres of Expertise in Dysmorphology.
- Providing an on-line dysmorphology diagnostic system (DDS).
- Developing clinical management guidelines for selected dysmorphic syndromes.

Why Develop Clinical Management Guidelines?

Due to the rarity of many syndromes and a lack of published, high grade evidence, relatively few guidelines are produced for dysmorphic conditions, and fewer still have been developed using a robust, validated methodology.

Which Conditions?

Four conditions were chosen, on the basis of a scoping exercise which identified the dysmorphic conditions for which there was a lack of existing information available on optimum clinical management. The expertise and special interests of the DYSCERNE Partners, who led the guideline development groups, was also taken into account.

The conditions selected were:

- **Angelman syndrome**
- **Kabuki syndrome**
- **Noonan syndrome**
- **Williams syndrome**

The guidelines include: criteria for diagnosis, information on recommended screening, reviews, and clinical management (including specialist referral) at different life stages, growth charts where applicable, and pointers to further sources of information and support for healthcare professionals and families.

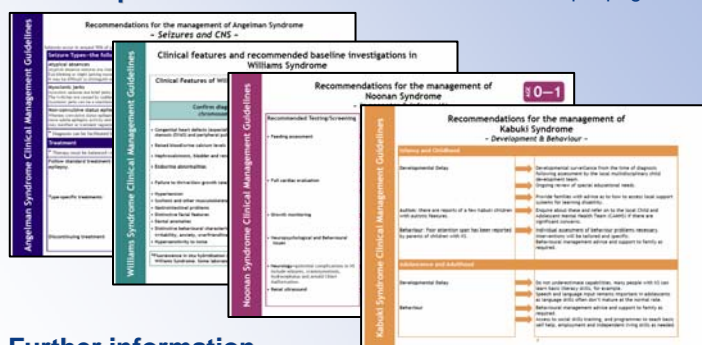
Methodology

DYSCERNE's guideline development methodology was adapted from the Scottish Intercollegiate Guidelines Network (SIGN) approach, which utilises the systematic review and grading of published evidence, in addition to consultation with multidisciplinary groups of clinicians, to achieve expert consensus.

This approach has been adapted to place more emphasis on expert opinion & consensus, whilst maintaining systematic rigor, (Fig. 1), as the evidence base for rare diseases is relatively limited.

The results of this iterative process have been well received– initial feedback on the guidelines has been very positive. Additionally, taking part in the process has been beneficial to guideline development group members – reviewing the literature and attending consensus meetings has been educational and informative.

The completed DYSCERNE Guidelines – example pages



Further information

The four sets of clinical management guidelines are available from the DYSCERNE website (www.dyscerne.org).

Any comments on the guidelines would be welcomed. The guidelines will be subject to regular review and any comments or suggestions made will be considered at review. Please email Kate Strong, Guidelines Developer, with any enquiries or comments: kate.strong@cmft.nhs.uk

Guideline Development Process

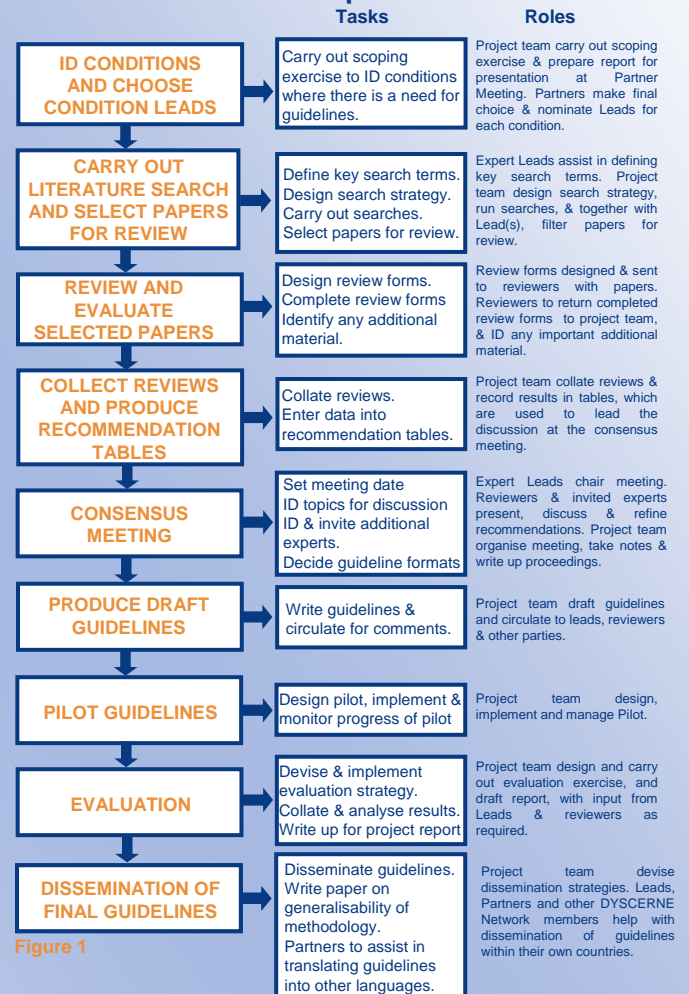


Figure 1

Conclusions

The methodology developed and utilised by DYSCERNE has produced robust, clinically approved guidelines for the selected rare conditions, which will improve the clinical management of patients with these conditions.

DYSCERNE has developed in-house expertise in the development process, and due to our contacts in Europe (members of the Network of Centres of Expertise for Dysmorphology), is ideally placed to take on this guideline development role for further conditions in the future.

