



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

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## The DYSCERNE Network

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 management guidelines for selected dysmorphic syndromes.

## Why Develop Guidelines?

Delivering high quality genetic services requires healthcare professionals to develop evidence based guidelines which are subject to validation and quality. However, in clinical genetics, both the rarity of many conditions and lack of published, high grade evidence, means that very few guidelines are produced, and fewer still have been developed using a robust methodology.

## Which Conditions?

Four conditions were chosen. The decision was based on the results of a scoping exercise which identified where there was a lack of availability of clinical management recommendations and other information on a disease. The expertise and special interests of the DYSCERNE Partners, who are leading the guideline development groups was also taken into account.

The chosen conditions are:

- **Angelman Syndrome**
- **Kabuki Syndrome**
- **Noonan Syndrome**
- **Williams Syndrome**

The guidelines will include: criteria for diagnosis, protocols for review and screening, information on clinical management at different life stages, and when specialist referral is needed.

## Designing an Appropriate Methodology

DYSCERNE's guideline development process utilises a modified Scottish Intercollegiate Guidelines Network (SIGN) methodology. This internationally recognised and validated method involves the systematic review and grading of published evidence, and uses multidisciplinary groups of clinicians to achieve expert consensus.

The SIGN methodology assumes a rich evidence base and has been used to develop guidelines for a range of common diseases and interventions. However, for rare diseases, the evidence base is very small or in some cases, non-existent, so the process has been adapted to place more emphasis on expert opinion and consensus, whilst still maintaining systematic rigor and transparency. Figure 1 details the DYSCERNE approach to the guideline development process.

Developing guidelines is a very labour intensive process, and many clinical departments lack the resources and, especially for very rare conditions, the expertise to carry out this kind of work. The DYSCERNE organisation is ideally placed to take on this role, having developed in-house expertise in the development process, and also because of our extensive network of contacts in specialist centres throughout Europe who form our Network of Centres of Expertise.

## Guideline Development Process

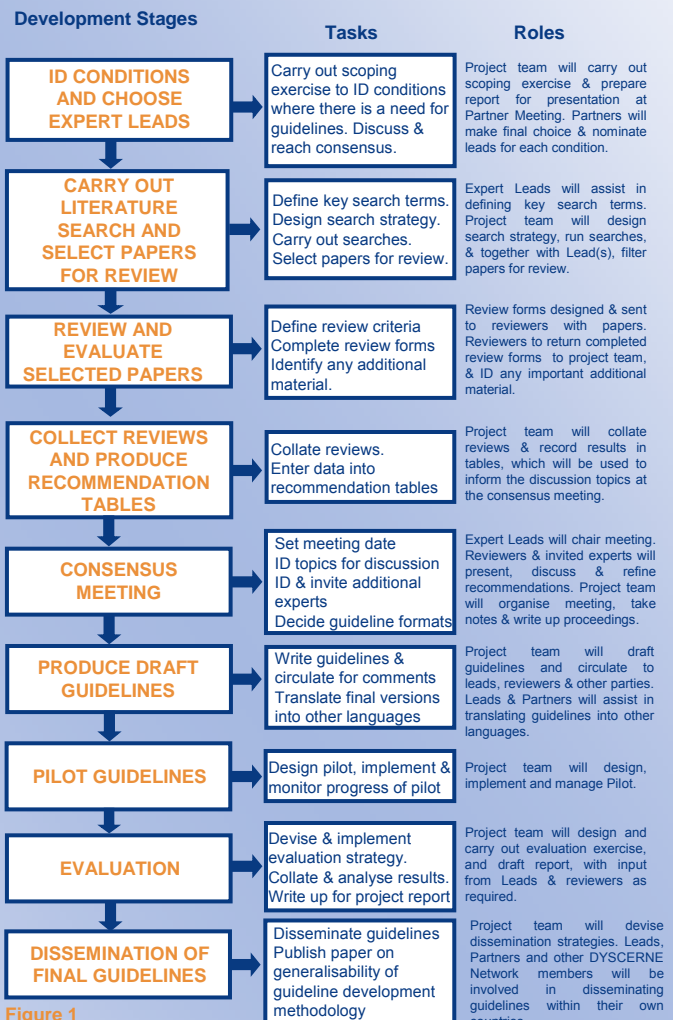


Figure 1

## Where Can I Find Out More?

Our programme of guideline development is ongoing and subject to continuous appraisal so modifications can be incorporated where necessary. The final development protocol will be available on the DYSCERNE website ([www.dyscerne.org](http://www.dyscerne.org)) and it is hoped that others will be encouraged to utilise this approach to develop much needed guidelines for rare diseases.

