

Clinical Practice Guidelines for Rare Diseases: the Orphanet Database

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Background

Clinical practice guidelines (CPGs) for rare diseases (RDs):

- are scarce,
- may be difficult to identify from Internet search,
- may vary greatly in quality depending on the source and methodology.

Aim

- Facilitate access to freely available, reliable and specific recommendations for the management of RDs,
- Contribute to improve diagnosis, treatment and patient care.

Method

Flow chart for CPG dissemination

Orphanet inclusion criteria

- Any language
- Developed by expert group
- Provides recommendations for clinical practice
- Specific for a RD/group of RDs
- Cover all aspects of management
- Cover all concerned population ages
- < 5-years-old, preferably <3-y-old

Identification

Evaluation

AGREE-II* criteria

- Scope and purpose
- Stakeholder involvement
- Editorial independence
- Rigour of development
- Clarity of presentation
- Applicability

Free access to full text

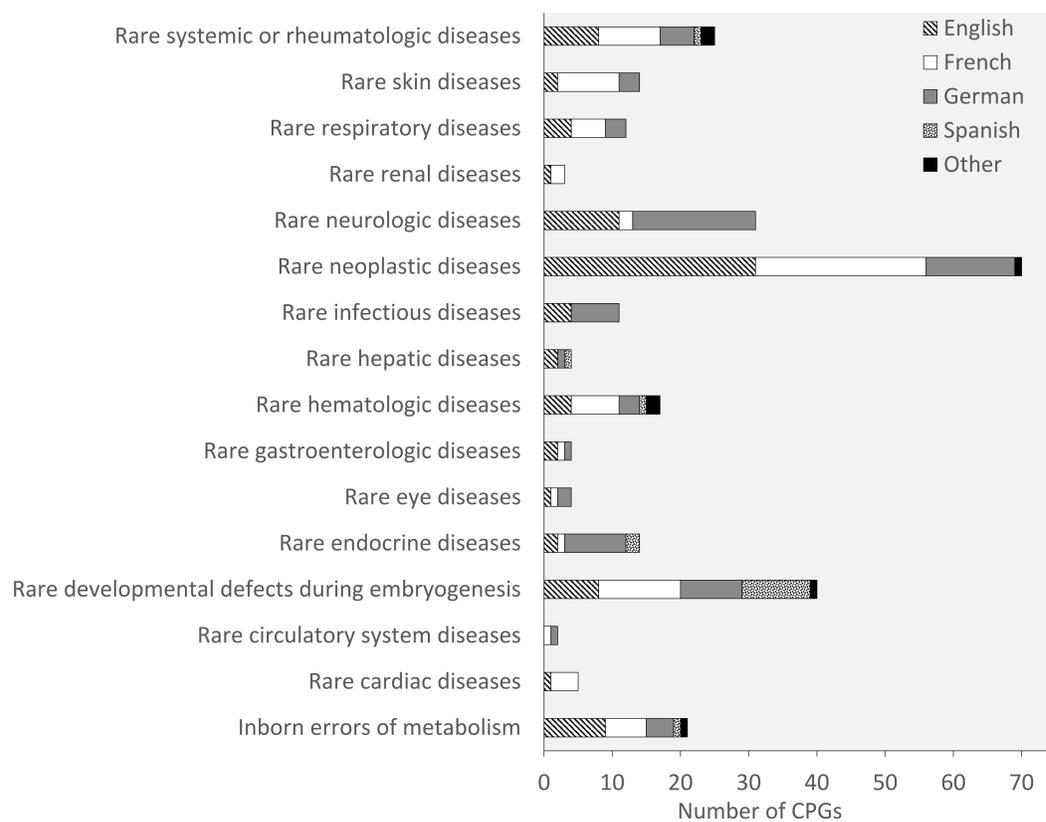
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Dissemination

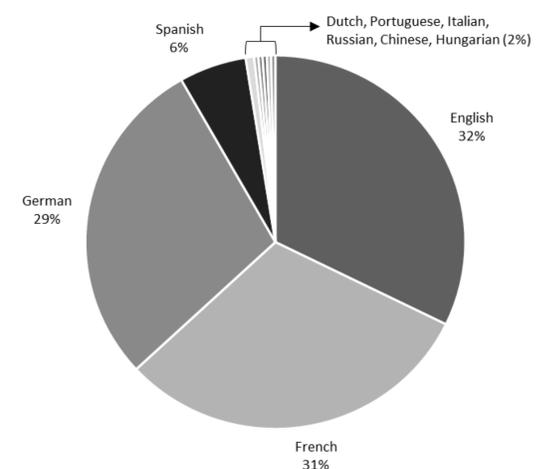
(*Appraisal of Guidelines for Research & Evaluation II, www.agreetrust.org)

Results

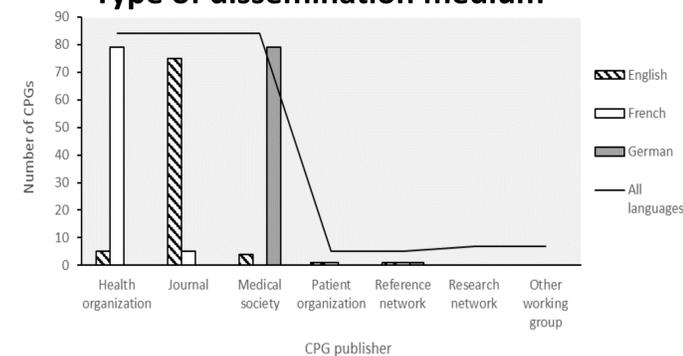
> 300 CPGs, distributed over 16 medical specialties



Languages



Type of dissemination medium



Conclusions

- More than 300 CPGs disseminated since January 2012; continuously growing collection; available at www.orpha.net
- The CPG database is included in the larger Orphanet project that establishes a link between diseases, good-quality textual information on RDs and the appropriate services for patients, researchers and healthcare professionals, and thus, it serves a range of stakeholders and purposes.

