Systematic scoping reviews (ScR) on rare disorders: a feasible first-step method for gaining overview of studies on rare congenital disorders?

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Background

The complexities of rare congenital disorders often require life-long, highly specialized and coordinated care involving multiple disciplines. Within each subtopic there is often very few papers, and studies frequently suffer from high risk of bias. Therefore systematic reviews that include an expedient grading of the evidence seems to be far ahead.

Objectives

To present key methodological issues and discuss our experiences with conducting two ScRs:

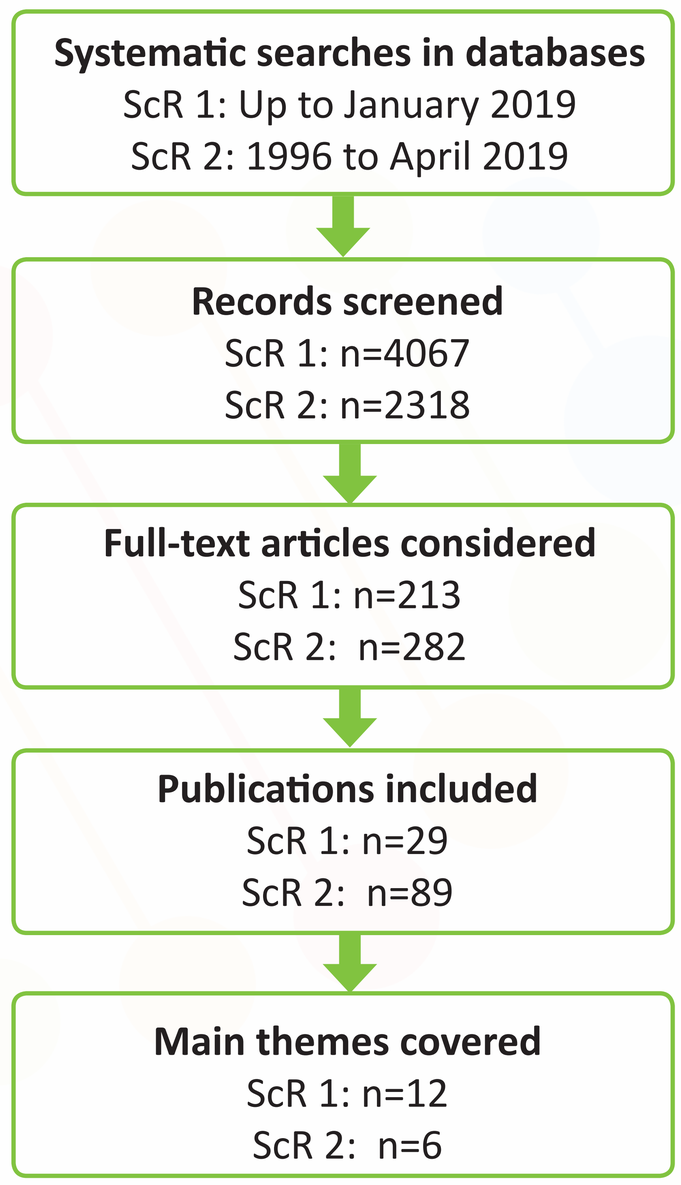
- Current knowledge of medical complications in adults with achondroplasia: A scoping review; ScR 1 (1)
- Pediatric patients with Marfan syndrome: A scoping review; ScR 2 (2)

Methods

The PRISMA-ScR guideline was followed (3)

Inclusion:

- Primary studies or systematic reviews
- Reporting separately on ≥ n=6 participants with a specified diagnosis
- Participants' age
- Mixed populations containing at least 80% of the population in interest, was accepted
- English and Scandinavian languages



Results

- Most were observational studies conducted in the USA (ScR 1) or Europe (ScR 2)
- Non-focused studies covering many issues, diciplines and mixed study populations
- Few studies within each subtopic
- Few participants; overlap of participants between studies suspected
- ScR 1 publications: Almost 50% were conducted > 20 years ago
- ScR 2 publications: Increasing numbers each year

Discussion

The ScR methodology was applied to very broad research questions to map clinical areas with known low frequency of publications. ScRs of complex rare disorder studies may include an overview of types of journals publishing and trends regarding subtopics, methodologies and measurements applied. Research gaps were easily identified. We specifically want to highlight the lack of high quality systematic reviews and that studies did not adhered to accepted reporting standards.

Conclusion

- The scoping review methodology is a feasible and excellent way of mapping and surveilling very broad research questions within fields with relatively low frequency of publications such as rare congenital disorders.
- ScRs within rare disorders is important to identify main concepts, theories, knowledge sources and to surveille the research knowledge and gaps to set future research agendas.

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2. Lidal IB, Bathen T, Johansen H, Velvin G. Pediatric patients with Marfan syndrome: a scoping review. (Under review in Acta Peadiatrica)
3. Tricco AC, Lillie E, Zarin W, O'Brien KK, Colquhoun H, Levac D, et al. PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation. Annals of internal medicine. 2018;169(7):467-73.

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Background: The complexities of rare congenital disorders often require life-long, highly specialized and coordinated care involving multiple disciplines – and many research questions arise. Within each subtopic, there is often very few papers, and studies frequently suffer from high risk of bias. Therefore, systematic reviews that include an expedient grading of the evidence seems to be far ahead. Recently, the PRISMA-ScR guidelines were published, describing the conduction of scoping reviews (ScRs) to map evidence on a topic and to identify main concepts, theories, sources, and knowledge gaps. Is this a feasible first-step method for gaining overview of studies on rare congenital disorders?

Objectives: To present results and discuss our experiences with conducting two ScRs.

Methods: We followed the PRISMA-ScR. Inclusion criteria for both reviews: a verified diagnosis; participants' age; systematic reviews, qualitative and quantitative primary studies with six or more diagnosed participants; English and Scandinavian languages. Excluded were studies on genetics, anatomy, pathophysiology, diagnostics (ScR-1) and treatment (ScR-1).

Both ScRs are presented with reference, aim, nationality, study design/methods, material, and the primary authors' conclusions. We also elucidated some methodological challenges, although we did not assess risks of bias or perform GRADE procedures.

Results: Included studies in ScRs covered nine and six topics respectively. Neurology and orthopedic issues were most frequent in ScR-1, while cardiovascular issues were most common in ScR-2. We found few studies on natural history, degree and extent of clinical manifestations in both adults with achondroplasia and in peadiatric Marfan patients, including few studies on psychosocial issues. Excluded papers were mainly due to study population's age (or not separate data between adults/ paediatric patients), design, or the study's aim was not within the scope of the review.

Conclusions: The scoping review methodology is an excellent way of mapping and surveilling research areas with relatively low frequency of publications, such as within the fields of rare congenital disorders. Identifying main concepts, theories, knowledge sources, and gaps to set future research agendas are important. Broad research questions are applicable and we thus avoid empty reviews or reviews with very few included studies. Scoping reviews may also serve as precursors for subsequent systematic reviews.

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