# Rare Disease Publications: How Accessible Are They?

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## Objective

- To investigate accessibility and metrics of peer-reviewed journals publishing paediatric rare disease articles.

## Background

- The publication of rare disease research is essential to increase awareness, further clinical research and inform trial designs in these rare conditions and related common diseases.
- Publishing in this field is, however, associated with a wide array of challenges, such as small sample sizes, lack of available peer-reviewers, and a small readership.
- One key dilemma faced by rare disease researchers is the selection of a target journal, as many rare diseases have a wide range of symptoms that could fall into a range of therapy areas.
- In addition, given the importance of patient engagement in the field of rare diseases, authors must consider open access publishing and an accompanying lay summary, which may be associated with increased costs.
- As 75% of rare diseases affect children,[1] the analyses reported here focused on published paediatric rare disease research articles and the metrics of their respective journals.

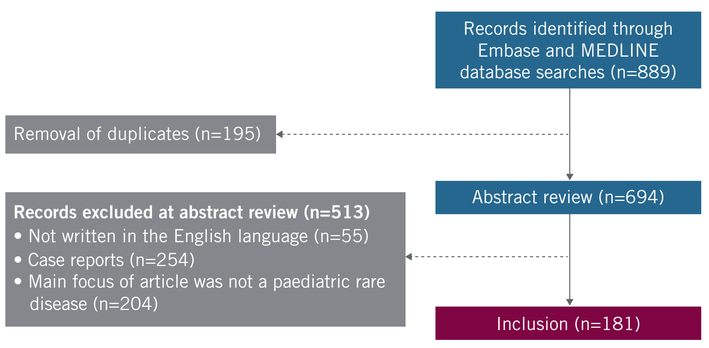
## Methods

- A pragmatic literature review was conducted in September 2017 to identify paediatric rare disease articles published in 2017.
- Embase and MEDLINE databases were simultaneously searched via the OvidSP platform, using paediatric and rare disease search terms.
- Abstracts were screened by a single reviewer to include English language articles with a main focus on a paediatric rare disease; case studies were excluded.
- The journals in which the eligible articles were published were classified according to the journal focus as reported on the journal website. Three groups were identified: general medical journals, journals with a rare disease focus and those focused on a common disease.
- Additional information and metrics were then extracted from the journal websites, including the most recently reported impact factor, whether the journal gave the option to publish open access and/or publish lay summaries, and the date the journal was established.

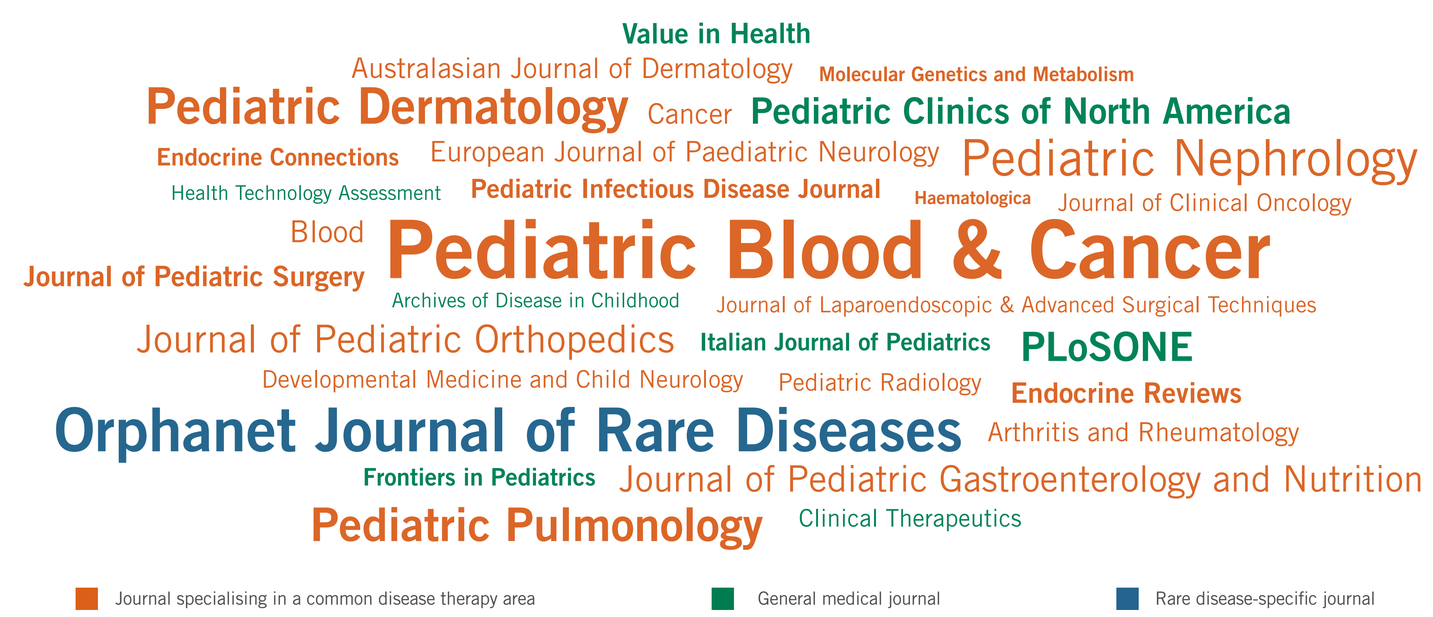
## Results

- A total of 889 abstracts were identified in the database searches, 181 of which met the eligibility criteria **(Figure 1)**.
- The 181 included abstracts were published in a total of 132 journals.
- Most articles (72%; 130/181) were published in journals specialising in a common disease therapy area.
- Only 4% (7/181) of articles were published in rare disease-specific journals.
  - Of these, 71% (5/7) of articles were published in the *Orphanet Journal of Rare Diseases*.
- 24% (44/181) of articles were published in general medical journals that did not specify a therapy area.
  - Of these, 56.8% (25/44) of articles were published in journals that exclusively publish paediatric research.

### Figure 1 | PRISMA diagram

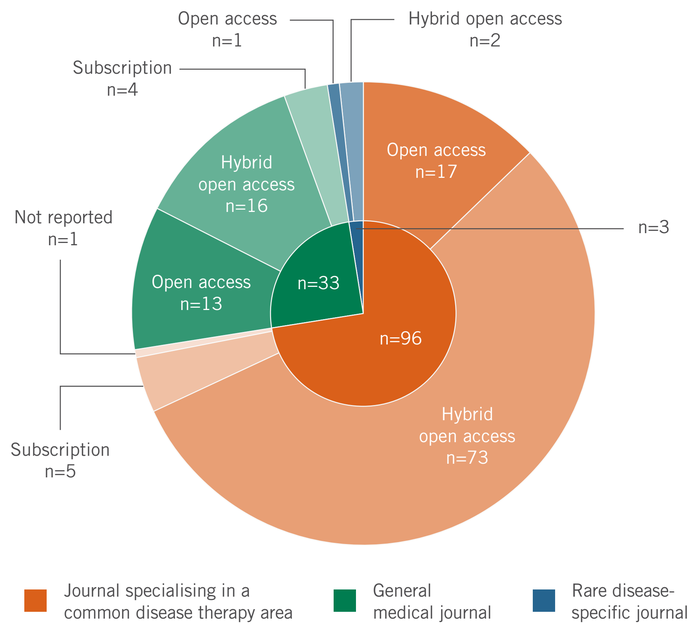



### Figure 2 | Journal word cloud




Journals with ≥2 included abstracts were included in the word cloud. Text size corresponds to the number of included abstracts found that were published by that journal.

### Figure 3 | Types of journal and their open access opportunities




Open access journals were defined as journals that publish exclusively open access. Hybrid open access model journals were defined as those that offer both open access and non-open access publication options. Subscription journals were defined as those that did not provide an open access option.

- *Pediatric Blood & Cancer* published the largest number of articles in this study (6 articles; impact factor: 2.634), followed by the *Orphanet Journal of Rare Diseases* (5 articles; impact factor: 3.478), *Pediatric Dermatology* (5 articles; impact factor: 0.99) and *Pediatric Nephrology* (5 articles; impact factor: 2.516) **(Figure 2)**.
- The majority of the journals identified had a common disease focus (73% [96/132]), while 25% (33/132) were general medical journals and only 2% (3/132) were rare disease-specific **(Figure 3)**.
- 23% (31/132) of journals were exclusively open access and 69% (91/132) used hybrid open access models, where the journal offered both open access and non-open access publication options.
  - A higher proportion of general medical journals (39% [13/33]) were open access, as compared to rare disease-specific journals (33% [1/3]) and those with a common disease focus (18% [17/96]) **(Figure 3)**.
  - Elsevier published the highest proportion of both open access journals and hybrid open access model journals (16.1% [5/31] and 26.4% [24/91], respectively).

### Table 1 | Most recently reported impact factor stratified by journal type

| Journal type | n | Median impact factor | Interquartile range |
|---|---|---|---|
| Journal specialising in a common disease therapy area | 96 | 2.416 | 2.820 |
| General medical journal | 33 | 1.921 | 1.927 |
| Rare disease-specific journal | 3 | 3.478 | 1.785 |

### Table 2 | Date of establishment stratified by journal type

| Journal type | n | Median year of establishment | Interquartile range |
|---|---|---|---|
| Journal specialising in a common disease therapy area | 96 | 1985 | 27 |
| General medical journal | 32* | 1977 | 42 |
| Rare disease-specific journal | 3 | 2006 | 9 |

*One journal did not report date of establishment.

- The median impact factor for the identified rare disease-specific journals was higher than in the general medical journals and journals specialising in a common diseases **(Table 1)**.
- Journals focusing on rare diseases were established much later than other journals, with a median date of establishment of 2006, compared with 1977 for general medical journals and 1985 for journals specialising in common diseases **(Table 2)**.
- 4.5% (6/132) of journals required an accompanying lay summary.

## Conclusions

- The majority of paediatric rare disease articles are published in journals specialising in a common disease area.
- A relatively small number of rare disease articles are published in rare disease-specific journals, of which the *Orphanet Journal of Rare Diseases* appears to be the most popular choice.
- Future analyses of patient engagement via alternative sources, e.g. social media and blogs, are recommended in order to further assess the accessibility of rare disease research.

## Abstract

### Objective

- Patient engagement in rare disease research is vital to ensure that relevant clinical questions and patient-centred health outcomes are addressed in future studies. It could therefore be expected that more attention is given to the accessibility of target journals when submitting rare disease articles compared to those describing common conditions. As 75% of rare diseases affect children,[1] we aimed to investigate the accessibility of paediatric rare disease publications.

### Research Design and Methods

- A pragmatic literature review was conducted in September 2017 using EMBASE and Ovid MEDLINE® to identify paediatric rare disease articles published in the English language in 2017. Abstracts were screened by a single reviewer.

### Results

- A total of 889 abstracts were identified, 181 of which met the eligibility criteria. Of these, 4% (7/181) were published in rare disease-specific journals (71% [5/7] of articles published in these journals were published in the *Orphanet Journal of Rare Diseases*), 72% (130/181) in journals specialising in a common disease therapy area (excluding rare diseases and paediatrics), and 24% (44/181) in general medical journals which did not specify therapy area. 54.5% (24/44) of the journals in the final category exclusively published paediatric manuscripts. 24% (43/181) of articles were published in open access journals whilst 1% (2/181) were published in journals requiring lay summaries.

### Conclusions

- Improved accessibility to orphan disease literature is essential for the rare disease community, as only a small proportion of rare disease articles are published in open access journals, with fewer still providing accompanying lay summaries. Further analyses into individual metrics of these journals and their correlation with visibility are recommended.

## References

**1.** Great Ormond Street Hospital. Scale of Rare Diseases. Available at: www.gosh.org/what-we-do/research/zayed-centre-research-rare-disease-children/rare-diseases/scale-rare-diseases [Last accessed 22.12.17].

## Author Contributions

Substantial contributions to study conception/design, or acquisition/analysis/interpretation of data: ET, MH, AG; Drafting of the publication, or revising it critically for important intellectual content: ET, MH, AG; Final approval of the publication: ET, MH, AG.

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