Rare Disease Publications: How Accessible Are They?  
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Objectives
- 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 design 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, 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.
- Articles 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 website, 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 999 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.
- 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.

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 classified as having common conditions. As 75% of rare diseases affect children, we aimed to investigate the accessibility of available 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 999 abstracts were identified, 181 of which met the eligibility criteria. 4% (5/181) 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) (5 journals specialising in a common disease therapy area (including rare diseases and paediatrics), and 24% (44/181) (2 general medical journals which did not specify therapy area. 54.5% (43/78) 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 hybrid open access journals.

Conclusions
- Improved accessibility in the rare disease community 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.

Table 1: Most recently reported impact factor stratified by journal type

<table>
<thead>
<tr>
<th>Journal type</th>
<th>Median impact factor</th>
<th>Interquartile range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Open access in a common disease therapy area</td>
<td>3.83</td>
<td>2.79-5.85</td>
</tr>
<tr>
<td>General medical journal</td>
<td>3.02</td>
<td>1.92-5.15</td>
</tr>
<tr>
<td>Rare disease-specific journal</td>
<td>2.7</td>
<td>1.68-3.74</td>
</tr>
</tbody>
</table>

Table 2: Date of establishment stratified by journal type

<table>
<thead>
<tr>
<th>Journal type</th>
<th>Date of establishment (n=132)</th>
<th>Interquartile range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Open access in a common disease therapy area</td>
<td>2000</td>
<td>1996-2005</td>
</tr>
<tr>
<td>Rare disease-specific journal</td>
<td>2006</td>
<td>2005-2006</td>
</tr>
</tbody>
</table>

Table 3: Types of journal and their open access opportunities

- Pediatr 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).
- 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 2).
- 23% (31/132) of journals were exclusively open access and 69% (93/132) used hybrid open access models, where the journal offered both open access and non-open access publication options. Subsequent journals were defined as those that did not provide open access options.

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 disease (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).

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.

References
1. Great Ormond Street Hospital. Scale of Rare Diseases. Available at: www.gosh.org.uk/wat- we/do/research/medical-centre/research-focussing-on-rare-diseases/rare-disease-scale/

Author Contributions
Substantial contributions to study conception and design, acquisition of data, or analysis and interpretation of data, and drafting the article or revising it critically for important intellectual content: E. Thurtle, M. Haughton, A. Griffiths.

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Figure 1: PRISMA diagram

Figure 2: Journal word cloud

Figure 3: Types of journal and their open access opportunities


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