



D6.10

Periodic electronic journal #1

Platform for sharing best practices for management of rare diseases

(RARE-Bestpractices)

Author(s)

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Beneficiary in Charge

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1. Rationale for the establishment of a Periodic Electronic Journal

1.1. Why is the journal needed?

Currently, there is a limited number of international scientific journals, which are focused on rare diseases issues. Moreover, rare diseases require a multidisciplinary, synergetic approach. Being published in a narrowly specialized journal, a rare diseases article loses a substantial part of its potential public. That is why it is important to put together in one single place all high quality research papers on rare diseases public health problems. That would further stimulate knowledge sharing and establishing partnerships, leading to an overall progress in rare diseases field. RARE-Bestpractices-affiliated journal would serve as a “meeting point” for all stakeholders, putting them together to collaborate and generate ideas.

1.2. What is the background of the journal in terms of the current and future development of the field?

The intended journal wouldn't be narrow-focused, because it has to serve all the rare diseases community. Several rare disease topics may be identified and covered by the journal. They include best practices guidelines, epidemiology and registries, health technology assessment, societal studies. This multiple combination would guarantee an immediate attention from numerous different fields that are professionally linked to rare diseases. It would provide a broad platform to establish and further develop the journal, ensuring a vast readership and impact.

1.3. How will the journal relate to relevant existing journals - and what will offer that these do not?

Rare diseases community needs a journal that is open in both aspects – publishing and accessing. Otherwise, some important outcomes could never become known. In 2013 several new rare diseases orientated journals were announced. It is a sign that the existing publishing resources are not sufficient and rare diseases stakeholders do need more accessible journals, where to present their work. So, the niche for a new synergetic open journal is still present.

1.4. How would the journal manage not only to find an audience, but also to generate sufficient high quality contributions?

Rare disease audience is growing as rare diseases and related issues are increasingly gaining attention from medical professionals and society as a whole. It is a multidisciplinary domain, which attracts researchers from different scientific fields. The journal's Editorial Board would consist of some of the most distinguished rare disease experts from different professional background and different countries. They would be able to identify journal collaborators that would have the task to seek rare disease high quality papers, as well as to form a short- and long-term development strategy of the journal in both sustainability and popularization aspects. They should be eased by the fact that different rare disease issues are

now in the centre of the contemporary health studies (-omics revolution, cost-effectiveness analysis in health care, etc.).

2. Journal preparatory activities

2.1. *Establishment of a working group and definition of the RARE Journal proposal*

A working group was established in September 2012 composed of the following project participants: Rumen Stefanov (task leader), Georgi Iskrov, Tsonka Miteva, Vanya Gyulina (BAPES), Domenica Taruscio (project leader, ISS) Cristina Morciano (WP6 dissemination leader, ISS), Holger Schünemann, UKFRL. Experts of publishing issues of the ISS, (Paola De Castro, Director of the ISS Publishing Unit, Federica Napolitani, editorial coordinator, ISS Publishing Unit) were also involved in the working group.

The objective of the working group was to define the journal proposal and to prepare the journal launch. A checklist of the activities to be carried out and of the critical issues to be addressed for the creation and launch of the journal was developed. Furthermore the group explored current journal practices such as editorial policy, author guidelines, standard of reporting, publishing platforms, copyright issues, promotional activities, etc. To perform this task the working group has proceeded through meetings, teleconferences and e-mail exchanges. The checklist has been used to prepare the Work Breakdown Structure of the task and the relative timeline.

ISS and BAPES team proposed the title and the subtitle of the journal “Rare diseases and orphan drugs.. An international journal of public health” (acronym RARE Journal) and initiated to compose the Editorial Board. Roles of the editors in chief were assigned (Domenica Taruscio and Holger Schünemann) as well as of the managing editor (Rumen Stefanov). ISS produced a first draft of the aim and scope, topics of the journal and proposed the sections and the subsections of the journal and roles and duties of the Editorial Board (editors in chiefs, section editors, associate editors, Advisory Board members).

This draft was reviewed by BAPES team and by Holger Schünemann (UKFRL). On that basis ISS team in collaboration with the graphics designer prepared the text of the first journal leaflet, the graphics of the leaflet (Figure 1), as well as of the graphics of the pdf format of the journal.

After that the working group initiated to draft the *journal policy content part I* (peer-review policy and guide for reviewers, copyright, open access and permission rules, standard of reporting) and *the journal policy content part II*: ethical guidelines, journal author guidelines, authorship and contribution, types of publications, publication-specific guidelines. In the figure 2 it is summarize the development process of the Journal.

RARE DISEASES AND ORPHAN DRUGS

An International Journal of Public Health

**Announcing
Call for Paper Submissions
First Issue in December 2013**

Co-Editors-in-Chief

Dr. Domenica Taruscio
*Director
National Centre for Rare Diseases
Istituto Superiore di Sanità
Italian National Institute of Health
Rome, Italy*

Prof. Holger Schünemann
*Professor and Chair
Department of Clinical
Epidemiology and Biostatistics
McMaster University
Hamilton, Canada*

www.rarejournal.org

RARE Journal (www.rarejournal.org) is a new international open access, online, peer-reviewed journal published three times per year, with **no publishing fees**.

The mission of **RARE Journal** is to provide an **advanced forum** on important aspects of **public health, health policy and clinical research** in ways that will improve health care and outcomes for persons suffering from rare diseases, as well as globally increase rare diseases experience sharing.

RARE Journal publishes original research articles, case reports, systematic reviews and meta-analyses, reports of clinical practice guidelines, HTA reports, epidemiological registry reports, commentaries, letters to Editors, meeting and project reports.

Selected examples of topics covered by the journal are given below:

- Health policies and services organization on rare diseases and orphan drugs
- Rare disease epidemiology research and registries
- Clinical research and methodology
- Rare disease best practices
- Comparative effectiveness research
- Practice guidelines
- Socio-economical analysis
- Primary prevention, risk factors and screening
- Health promotion
- Quality assurance (genetic testing, etc.)
- Legal and ethical issues
- Narrative medicine
- Patients' needs and health-related quality of life

RARE Journal also anticipates special issues dedicated to a specific topic, such as particular rare diseases or a group of rare diseases. Suggestions for topics are welcome. Accepted papers are published in electronic form and indexed for database reference.

RARE JOURNAL:

- No Publication Fees
- Timely Peer-Review
- Open Access to Articles Online
- Expert Editorial Assistance
- High Scientific Quality

For Further Information Please Visit:
WWW.RAREJOURNAL.ORG

RARE Journal Editorial Secretariat
e-mail: secretariat@rarejournal.org

FIRST ISSUE COMING IN DECEMBER 2013!

The RARE-Bestpractices project is funded by the European Union Seventh Framework Programme, Project Ref. n° 258933. Such responsibility lies with the Programme, and the European Commission cannot be held liable for any use of the information contained therein.
WWW.RAREBESTPRACTICES.EU

Figure 1. Journal Leaflet

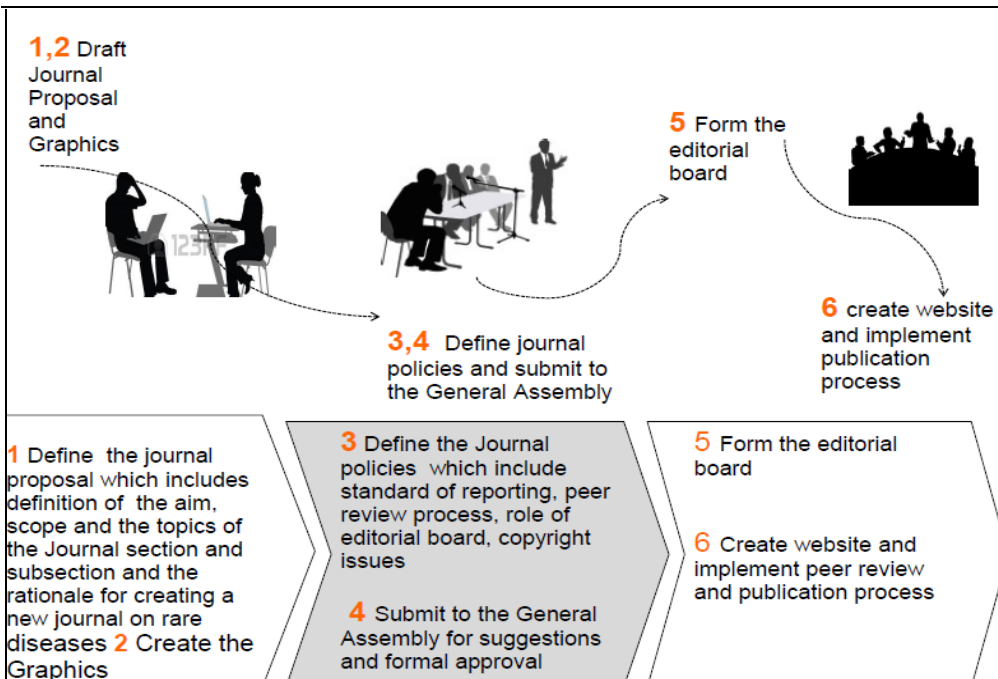


Figure 2. Journal development process

2.2. Presentation of the Work Break Down Structure (WBS) of the task 6.5 and of the preliminary policy of the journal at the RARE-Bestpractices kick-off Meeting

During the kick off meeting, BAPES and ISS team presented the Work Break Down Structure (WBS) of the task (summary in table 1):

Project partner	Subtask
BAPES in charge	Subtask 6.5.1 Journal website
ISS in charge	Subtask 6.5.2 Journal policy content part I: Journal mission, scope, sections and subsections, Peer-review policy and guide for reviewers, copyright, open access and permission rules, editorial staff and roles, standard of reporting
BAPES in charge	Subtask 6.5.3 Journal policy concept part II: Ethical guidelines Journal author guidelines, Authorship and contribution, types of publications, publication-specific guidelines
ISS in charge with the collaboration of UKFRL	Subtask 6.5.4 Forming Journal Editorial Board and enrolling Associate Editors, Advisory Board and Section Editors
BAPES in charge	Subtask 6.5.5 Journal management and workflow
ISS in charge	Subtask 6.5.6 Regulation of journal legal property rights
ISS in charge with the collaboration of the graphics designer and all the beneficiaries	Subtask 6.5.7 Journal graphic design (cover, inner page for printable version, etc) and journal launch

Table 1. Summary of WBS task 6.5. The timeline were established for all subtasks in the respect of the project schedule.

During the kick-off meeting ISS and BAPES team proposed the followings: -

- Rare Diseases and Orphan Drugs Journal (RARE Journal) is a new international open access, online, double-blind peer-reviewed journal published three times per year, with no publishing fees.
- The mission of RARE Journal is to provide an advanced forum on important aspects of public health, health policy and clinical research in ways that will improve health care and outcomes for persons suffering from rare diseases, as well as globally increase rare diseases experience sharing.
- RARE Journal serves the international rare diseases community by publishing high-quality articles from epidemiology, public health, health economics, social sciences, ethics and law, with a special accent on rare diseases best practices guidelines, rare diseases research recommendations, and rare diseases epidemiological reports.
- RARE Journal publishes several types of articles, including original research article, case report, systematic review and meta-analysis, report of clinical practice guidelines, HTA report, epidemiological registry report, commentary, letter to the editors, meeting and project reports. The journal also anticipates special issues dedicated to a specific topic, such as particular rare disease or a group of rare diseases. Suggestions for topics are welcome.
- Selected examples of the **topics** covered by the journal are given below:

- Health policies and services organization on rare diseases and orphan drugs
- Rare disease epidemiology research and registries
- Clinical research and methodology
- Rare disease best practices
- Comparative effectiveness research
- Practice guidelines
- Socio-economical analysis
- Primary prevention, risk factors and screening
- Health promotion
- Quality assurance (genetic testing, etc.)
- Legal and ethical issues
- Narrative medicine
- Patients' needs
- Quality of life
- **Sections**
 - **Epidemiology and Clinical Research**

This section guides epidemiologists, public health investigators and health professionals (physicians, nurses, and dentists) to what is new and important in epidemiological research including the latest principles and methods of data analysis.

 - Rare diseases epidemiology
 - Clinical research and methodology
 - **Guidelines, systematic reviews, health technology assessment, and horizon scanning**

This section aims to publish studies on comparison of treatments, tests, or health care services for rare diseases field to determine their effectiveness, benefits, and risks. Possible topics include methodology of rare diseases guidelines, systematic reviews, health technology assessment and horizon scanning.

 - Rare disease guidelines and systematic reviews
 - Health technology assessment and horizon scanning
 - **Prevention**

This section aims to publish studies on the protection of fetal and human well-being, as well as the prevention of long-term health consequences, that are related with rare diseases. Possible topics include methods to avoid occurrence of rare diseases, diagnosis of rare diseases, rare diseases rehabilitation and reintegration, health promotion in rare diseases field.

 - Primary prevention, risk factor and screening
 - Quality assurance (genetic testing, etc)
 - **Rare diseases policy**

This section aims to publish studies on political and societal interception of rare diseases and the human side of rare diseases issues. Possible topics include rare diseases care and access to it, procurement of rare diseases goods and services, health care interventions in rare diseases, decision-making for rare diseases medical goods and services; rare diseases interface between law, economics and health policy, narrative medicine and

integrated medical records of rare diseases, quality of life in rare diseases, freedom and human rights protection of rare diseases patients, rare diseases advocacy.

- Policy and decision-making
- Health economics
- Law and ethics
- Narrative medicine and patients' needs
- Quality of life

2.3. Approval of the Journal structure and policy

After the 2nd Project meeting (London, May 2013) a 2round consultation of the final version of the journal structure, policy documents, rules and procedures was conducted. project consortium members had the possibility to consult and comment the documents. The suggested changes were done and re-evaluated by the Project Consortium. After the 2nd round of consultation in June, consortiummembers were e-mailed with the final documents. Journal structure, policy documents, rules and procedures were approved by all members. Journal structure, policy documents, rules and procedures are attached to this deliverable (Appendix 1).

2.4. Editorial Board and Secretariat

Project participants with relevant expertise and reputation in rare diseases or in systematic reviews, guidelines, horizon scanning, health technology assessment were invited to be part of the Editorial Board by the Editors-in Chief. Editors-in-Chief invited experts external to the project consortium to be members of the Editorial Board as well. The Editorial Board member list and Secretariat is attached to this deliverable (Appendix I).

2.5. Launch of the Journal website and online tracking system

The journal website (www.rarejournal.org) and the online tracking system were created after the kick-off meeting. They underwent trial tests and were officially launched in June 2013. Workflow guidelines and instructions for Editorial Board members were elaborated and disseminated.

Workflow guidelines and instructions for Editorial Board members are attached to this deliverable report.

In September 2013 project consortium members agree that the first issue would consist of RARE-Bestpractices projectrelated papers. Work package leaders were invited to submit manuscripts, describing their specific objectives and activities. It was further agreed that the journal inaugural issue would include a supplement with the 2013 International Conference on Rare Diseases (ICORD), as a way to boost the Journal's visibility among global rare disease stakeholders. A general call for submissions was officially announced in September 2013.

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2.6. Copyright policy of the RARE journal

RARE-Journal provide immediate access to all its articles. Authors (or their author's institution or funder) do not pay an open access publishing fee. Articles are published by the ISS under the terms of the Creative Commons license Attribution-NonCommercial-NoDerivs 3.0 (<http://creativecommons.org/licenses/by-nc-nd/3.0/>).

2.7. Publication of the Journal inaugural issue

Journal inaugural issue was published in February 2014 on the website www.rarejournal.org.

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- *Editorial*
Domenica Taruscio, Holger Schünemann
- *RARE-Bestpractices: a platform for sharing best practices for the management of rare diseases*
Domenica Taruscio, Cristina Morciano, Paola Laricchiuta, Pierpaolo Mincarone, RARE-Bestpractices Consortium
- *Methodology for production of best practice guidelines for rare diseases*
Thomas Sejersen, Cinzia Del Giovane, Graziella Filippini, Carlo Giacomo Leo, Joerg J Meerpohl, Pierpaolo Mincarone, Silvia Minozzi, Sabina Saverio, Schünemann Holger, Juliette Senecat, Domenica Taruscio, RARE-Bestpractices consortium
- *Improving the Retrieval and Dissemination of Rare Disease Guidelines and Research Recommendations: A RARE-Bestpractices Initiative*
Michele Hilton Boon, Karen Ritchie, Jan Manson
- *Proceedings from the VIII International Conference on Rare Diseases and Orphan Drugs (ICORD). St. Petersburg (Russia), October 31-November 2, 2013*
Emilio José Antonio Roldán

Journal inaugural issue is attached to this deliverable.

The Journal inaugural issue was presented at the 2nd Annual Project meeting in March 2014. Ongoing issues were discussed such as reviewer recruitment, special calls for submissions, important deadlines and milestones. Inclusion to the major scientific reference databases was identified as a key issue for the sustainability of the Journal. It was agreed that the Journal should develop strategic partnerships with other EU-funded projects, as well as European and global rare disease stakeholders.

2 Document History

Date	Author	Changes
28/03/2014	Georgi Iskrov, Rumen Stefanov, Tsonka Miteva	First draft
1 April 2014	Cristina Morciano	Add some parts and made some corrections and comments
1 April 2014	Pierpaolo Mincarone	Minor revisions and formatting
2 April 2014	Cristina Morciano	Delete and amend some parts.
3 April 2014	Georgi Iskrov	Appendices 1 inserted
16 April 2014	Cristina Morciano	Other minor amendements (insert ISS secretariat, amend the guidelines for authors).

Appendix 1: Journal structure, policy documents, rules and procedures

Summary information

- *Acronym – RARE Journal*
- *Aim and scope*

Rare Diseases and Orphan Drugs Journal (RARE Journal) is a new international open access, online, peer-reviewed journal published three times per year, with no publishing fees.

The mission of RARE Journal is to provide an advanced forum on important aspects of public health, health policy and clinical research in ways that will improve health care and outcomes for persons suffering from rare diseases, as well as globally increase rare diseases experience sharing.

RARE Journal serves the international rare diseases community by publishing high-quality articles from epidemiology, public health, health economics, social sciences, ethics and law, with a special accent on rare diseases best practices guidelines, rare diseases research recommendations, and rare diseases epidemiological reports.

RARE Journal publishes original research articles, case reports, systematic reviews and meta-analyses, reports of clinical practice guidelines, HTA reports, epidemiological registry reports, commentaries, letters to Editors, meeting and project reports. The journal also anticipates special issues dedicated to a specific topic, such as particular rare disease or a group of rare diseases. Suggestions for topics are welcome.

All manuscripts are peer-reviewed. Accepted papers are published in electronic form.

Selected examples of topics covered by the journal are given below:

- Health policies and services organization on rare diseases and orphan drugs
- Rare disease epidemiology research and registries
- Clinical research and methodology
- Rare disease best practices
- Comparative effectiveness research
- Practice guidelines
- Socio-economical analysis
- Primary prevention, risk factors and screening
- Health promotion

- Quality assurance (genetic testing, etc.)
- Legal and ethical issues
- Narrative medicine
- Patients' needs and health-related quality of life

Journal sections

○ *Epidemiology and clinical research*

This section aims to publish studies on patterns, causes, and effects of rare disease conditions, epidemiology of rare diseases and the use of public health interventions for their control. Possible topics include rare diseases surveillance, registries and biomonitoring, as well as safety and effectiveness of medications, devices, diagnostic products and treatment regimens intended for prevention, treatment, diagnosis or for relieving symptoms of rare diseases.

- Subsection Rare diseases epidemiology
- Subsection Clinical research and methodology

○ *Guidelines, systematic reviews, health technology assessment, and horizon scanning*

This section aims to publish studies on comparison of treatments, tests, or health care services for rare diseases field to determine their effectiveness, benefits, and risks. Possible topics include methodology of rare diseases guidelines, systematic reviews, health technology assessment and horizon scanning.

- Subsection Rare disease guidelines and systematic reviews
- Subsection Health technology assessment and horizon scanning

○ *Prevention*

This section aims to publish studies on the protection of fetal and human well-being, as well as the prevention of long-term health consequences, that are related with rare diseases. Possible topics include methods to avoid occurrence of rare diseases, diagnosis of rare diseases, rare diseases rehabilitation and reintegration, health promotion in rare diseases field.

- Subsection Primary prevention, risk factor and screening
- Subsection Quality assurance (genetic testing, etc.)

○ *Rare diseases policy*

This section aims to publish studies on political and societal interception of rare diseases and the human side of rare diseases issues. Possible topics include rare diseases care and access to it, procurement of rare diseases goods and services, health care interventions in rare diseases, decision-making for rare diseases medical goods and services; rare diseases interface between law, economics and health policy, narrative medicine and integrated medical records of rare diseases, quality of life in rare diseases, freedom and human rights protection of rare diseases patients, rare diseases advocacy.

- Subsection Policy and decision-making
- Subsection Health economics
- Subsection Law and ethics
- Subsection Narrative medicine, patients' needs and quality of life

Editorial Board

Editors-in-Chief – Dr. Domenica Taruscio and Prof. Holger Schünemann

The Editors-in-Chief have the function of determine the editorial policy of the RARE Journal in consultation with the Editorial Board. They oversee the editorial process and define the communication strategy supported by the Managing Editor, the Associate Editors and the Advisory Board.

The Editors-in-Chief have the ultimate responsibility for the journal's content. They have specific functions in the peer review process, which include:

- initial evaluation of the submitted paper (determine if it is of sufficient quality and falls within the scope of the journal);
- forward manuscripts to the appropriate Section Editor(s) for review;
- make the final decision regarding publication and complete the correspondence with authors.

Specific duties also include:

- nominate Section Editors, Associate Editors and member of the Advisory Board;
- define strategy to increase the quality submission to the journal;
- promote the journal to broaden its visibility and diffusion;
- liaise closely with the Editorial Board and take a leading role in shaping future publishing strategy.

○ **Managing Editor (responsible for workflow process) – Prof. Rumen Stefanov**

The Managing Editor, who is responsible for the workflow process, oversees and coordinates the publications' editorial activities, ensuring punctuality and regularity of the journal by implementing an efficient workflow of paper from submission to publication. He coordinates the Technical Editor(s) who are expected to work on the accepted paper to ensure that is accurate and easily understandable.

The Managing Editor assists the Editors-in-Chief to increase the journal's visibility and readership and supports the Editorial Board in revisiting the journal's mission and publication strategy.

○ **Section Editors**

Section Editors will be responsible for own section, actively seeking high quality manuscripts to be submitted to the journal, as well as commissioning commentary for the journal. They work to maintain a positive image for the journal by ensuring regularity and punctuality.

Section Editors also manage the peer review process. Specific duties of the section editors in the peer review process include:

- identify appropriate external reviewers or among Associate Editors;
- correspond with authors and reviewers;
- deal with conflict of interest;
- consider reviewer's comments, make a decision concerning the manuscript and prepare a report for the Editors-in-Chief;
- identify potential reviewers, contributing to the ongoing expansion of the reviewers' database.

Section Editor may also act as a reviewer.

Associate Editors

Associate Editors are specialised in a particular section of the journal and may act as reviewers, as well as may suggest reviewers. They support the relevant Section Editors in seeking articles and commentaries for the journal. They contribute by providing contents and general structures of volumes, special issues, etc.

Associate Editors also support Sections Editors and Editors-in-Chief in deciding about publication of a manuscript and cooperate with Editors-in-Chief in ensuring that the policy of the journal is correctly implemented.

Advisory Board

The prominent role of the Advisory Board members is to support Editors in ensuring that the policy of the journal is correctly implemented (aim and scope, organization of the editorial process, impact). They provide recommendations and new ideas to the Editors to ensure the quality and credibility of the contents, efficiency of the editorial process, broad diffusion and high impact of the journal.

Members of the Editorial Board

Editors-in-Chief

- Domenica Taruscio, Istituto Superiore di Sanità, Rome (Italy)
- Holger Schünemann, McMaster University, Hamilton, ON (Canada)

Managing Editor

- Rumén Stefanov, Medical University of Plovdiv, Plovdiv (Bulgaria)

Section Editors

- **Epidemiology and Clinical Research**
 - Section Editor for Rare Diseases Epidemiology
 - Manuel Posada de la Paz, Instituto de Salud Carlos III, Madrid (Spain)
 - Section Editor for Clinical Research and Methodology
 - Alfonso Iorio, McMaster University, Hamilton, ON (Canada)
- **Guidelines, Systematic Reviews, Health technology Assessment, and Horizon Scanning**
 - Section Editor for Rare Diseases Guidelines and Systematic Reviews
 - Joerg Meerpohl, University Medical Centre Freiburg, Freiburg (Germany)
 - Section Editor for Health Technology Assessment and Horizon Scanning
 - Section editor, City (Country)
- **Prevention**
 - Section Editor for Primary Prevention, Risk Factors and Screening
 - Section editor, City (Country)
 - Section Editor for Quality Assurance (Genetic Testing, etc.)
 - Section editor, City (Country)
- **Rare Diseases Policy**
 - Section Editor for Policy and Decision Making
 - Benjamin Djulbegovic, University of South Florida, Tampa, FL (USA)
 - Section Editor for Health Economics
 - Section editor, City (Country)
 - Section Editor for Law and Ethics
 - Bartha Knoppers, McGill University, Montreal, QC (Canada)
 - Minh Thu Nguyen, McGill University, Montreal, QC (Canada)
 - Section Editor for Narrative Medicine
 - Section editor, City (Country)
 - Section Editor for Patients' Needs
 - Section editor, City (Country)
 - Section Editor for Quality of Life
 - Section editor, City (Country)

Associate Editors

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- Helen Dolk, Belfast (UK)
- Panos Kanavos, London (UK)
- Hanns Lochmuller, Newcastle (UK)
- Lucio Luzzatto, Florence (Italy)
- Federico Micheli, Buenos Aires (Argentina)
- Cristina Morciano, Rome (Italy)
- Karen Ritchie, Edinburgh (UK)
- Thomas Sejersen, Stockholm (Sweden)
- Pedro Serrano-Aguilar, Tenerife (Spain)
- Paolo Vineis, London (UK)

Advisory Board

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- Paola De Castro, Istituto Superiore di Sanità, Rome (Italy)
- Federica Napolitani, Istituto Superiore di Sanità, Rome (Italy)

Managing Editor

- Rumen Stefanov (BAPES)

Managing Editor Assistants

- Georgi Iskrov (BAPES)
- Tsonka Miteva-Katrandzhieva (BAPES)

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- Radostina Simeonova (BAPES)

Layout Designer

- Kristian Ivanov (BAPES)

Publishing Advisor

- Vanya Gyulina (BAPES)

Secretary ISS

- Linda Agresta (ISS)
- Sabina Tonon (ISS)
- Giorgio Vincenti (ISS)

Journal policy

Authorship criteria

An “author” is generally considered to be someone who has made substantive intellectual contributions to a published study. Scientific authorship implies to have important academic, social, and financial consequences. An author must take responsibility for at least one component of the work, should be able to identify who is responsible for each other component, and should ideally be confident in their co-authors’ ability and integrity. RARE Journal requests authors to provide information about contributions to studies from persons listed as authors in the acknowledgment section of the manuscript.

RARE Journal adheres to the ICJME criteria for authorship:

- substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data;
- drafting the article or revising it critically for important intellectual content;
- final approval of the version to be published.

Authors should meet all these conditions. All persons designated as authors should qualify for authorship, and all those who qualify should be listed. Each author should have participated sufficiently in the work to take public responsibility for appropriate portions of the content.

All contributors who do not meet the criteria for authorship should be listed in an acknowledgments section. Financial and material support should also be acknowledged.

- *Multicentre work*

If a multicentre group has conducted the work, the group should identify the individuals who accept direct responsibility for the manuscript. When submitting a manuscript authored by a group, the corresponding author should clearly indicate the preferred citation and identify all individual authors as well as the group name. Acquisition of funding, collection of data, or general supervision of the research group alone does not constitute authorship.

Peer review

RARE Journal believes unbiased, independent, critical assessment is an intrinsic part of all scholarly work, including the scientific process. Peer review helps RARE Journal decide which manuscripts are suitable for achieving the journal's objective, as well as helps authors to improve the quality of reporting (See above *Peer review policy*).

Peer review policy

Peer review process

The Editors-in-Chief make the initial decision whether a submitted manuscript is suitable for publication in RARE Journal. If they determine that the manuscript fall within the scope of the journal and meets the basic criteria of quality and potential interest and impact, then it is distributed to the relevant Section Editor(s), otherwise it is rejected with no further processing.

○ *Reviewers*

Section Editors prepare a list of expert reviewers who are either Associate Editors or are recruited externally. The manuscript is sent at least to two reviewers. Selection of reviewers is responsibility of the Section Editors, but authors may suggest reviewers. One Section Editor will usually take each article through from start to finish. All potential reviewers are contacted individually to determine availability. RARE Journal operates using a double blind review system. Submissions may also undergo statistical review, as appropriate.

Editors, authors and reviewers are informed that the manuscript must be considered confidential. Reviewers are asked to declare any competitive interest they may have in reviewing the manuscript. Any possible competitive interest (personal, financial, professional) with authors or related to the topic should be promptly reported by peer reviewers to the Section Editors. Section Editors identify another reviewer in their place when a conflict of interest exists. Authors can also indicate specific individuals whom they would like to have excluded as reviewers with conflict of interest. Requests to exclude certain potential reviewers may be honored at the discretion of the Section Editors. Editors, authors and reviewers are informed that the manuscript must be considered confidential.

Reviewers are asked to evaluate manuscripts within two weeks. There are four categories for decision: 1) accept; 2) consider with minor revision; 3) consider after major revision; 4) reject. Reviewers evaluate each submission within two weeks, and submit a written report to the Section Editors.

○ *Final decision*

The Section Editors consider the comments made by the reviewers, make a decision concerning the manuscript and prepare a report for the Editors-in-Chief. All revised papers are carefully reexamined

by the Editors-in-Chief who are the only persons who can officially accept a paper, in consultation with the Editorial Board and reviewers.

○ *Guide for Rare Diseases and Orphan Drugs reviewers*

RARE Journal uses a double blind review. This means that both author's and reviewer's identities remain anonymous. Manuscripts are evaluated by at least two reviewers who are either member of the Editorial Board or external experts.

Reviewers should maintain the confidentiality about the content of the manuscript. Comments elaborated by the reviewers and any communications about the manuscript should not be disclosed outside the editorial environment. Any personal use is also not permitted.

Reviewers are asked to inform the Section Editors about any possible competitive interest related to the topic. They should evaluate manuscripts within two weeks and recommend whether the manuscript should be: 1) accepted; 2) considered with minor revision; 3) considered after major revision; 4) rejected.

They have the primary responsibility to verify the validity of the article and provide a written report with clear and constructive comments that allow the Editors to decide whether to publish the submitted article and help the authors to improve the manuscript and then resubmit. Assessing the manuscript, reviewers should consider if the author has met or not the Editorial Policy of RARE Journal and the Standard of Reporting initiatives are supported by the Journal.

Reviewing the manuscript, they should also detect and report any ethical issues or possible misconduct (plagiarism, failure to disclose conflict of interest, forged results, authorship, etc.) as well as check for the clarity and readability of the written English.

Reviewers are encouraged/should consider the following general guidance when they appraise research articles:

- Research question: is it important, clearly defined?
- Design of the study: is it appropriate to the research question?
- Method: is it clearly described and sufficient detailed to be reproducible and in compliance with the relevant reporting guidelines/checklist as recommended by RARE Journal? Ethics of the study?
- Statistical analyses: do they need to be assessed by a statistical expert?
- Results: are they original and relevant?
- Interpretation and conclusions: are they consistent with the results provided and give an appropriate answer to the research question?
- Study description and organisation: is the study well presented, the written English readable and the scientific terminology used appropriately without ambiguity? Does the information organized in a text that shows a logical sequence? Are tables and figures adequate to explain and support the description of the study?

- Abstract: is it clear and does it summarize properly the more important information of the article?

Standard of health research reporting

RARE Journal supports initiatives aimed at improving the reporting of health research since it considers that:

- good reporting is an integral part of good research;
- good reporting increases the usability of research;
- good reporting facilitates the evaluation of the manuscript;
- good reporting increase the probability of a successful acceptance;

Therefore authors should use the following guidelines when drafting their manuscript:

- [CONSORT Statement](#) if reporting of randomized controlled trials;
- [STROBE](#) if reporting of observational studies;
- [STARD](#) reporting of diagnostic accuracy studies;
- [PRISMA](#) reporting of systematic reviews systematic review or meta-analysis;
- [MOOSE](#) reporting of meta-analyses of observational studies.

The above mentioned guidelines are collected and available at the Equator network website (www.equator-network.org).

○ *Competing interest*

A competing interest or conflict of interest is a set of circumstances that creates a risk that professional judgment will be unduly influenced by secondary interests. It means that the interpretation and presentation of data may be influenced by your personal or financial relationship with other people or organisations. That is why public trust in the peer-review process and the credibility of RARE Journal published articles depends on removing any form of competing interest during the processes of peer-review, and editorial decision-making.

All authors and reviewers are requested to disclose any actual or potential conflict of interest including any financial, personal or other relationships within three years of beginning the submitted work that could inappropriately influence, or be perceived to influence, their work. To prevent ambiguity, authors must state explicitly whether potential conflicts do or do not exist.

RARE Journal is making its final decisions about manuscripts on no personal, professional, or financial ground that may influence the judgment. RARE Journal Editorial staff provides editors a regular declaration of financial interests and is asked to reclude from any decisions in which a conflict of

interest exists. Editorial staff will not use information gained through working with manuscripts for private gain.

- *Mandatory conflict-of-interest disclosure declaration*

RARE Journal adopts the ICMJE uniform form for conflict-of-interest disclosure (http://www.icmje.org/coi_instructions.html). All authors must provide a signed copy of it upon the manuscript's submission.

- *Privacy and confidentiality*

Manuscripts will be reviewed with due respect for authors' confidentiality. Reviewers also have rights to confidentiality, which must be respected by the editor. Confidentiality may have to be breached if dishonesty or fraud is alleged but otherwise must be honored. RARE Journal editors won't disclose information about manuscripts (including their receipt, content, status in the reviewing process, criticism by reviewers, or ultimate fate) to anyone other than the authors and reviewers. This includes requests to use the materials for legal proceedings.

RARE Journal makes clear to the reviewers that manuscripts sent for review are privileged communications and are the private property of the authors. Therefore, reviewers and members of the editorial staff will respect the authors' rights by not publicly discussing the authors' work or appropriating their ideas before the manuscript is published. Reviewers are not allowed to make copies of the manuscript for their files and are prohibited from sharing it with others, except with the editor's permission. Reviewers destroy copies of manuscripts after submitting reviews. Editors are not keeping copies of rejected manuscripts.

Reviewer comments are not be published without permission of the reviewer, author, and editor.

Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

- *Protection of human subjects and animals in research*

When reporting experiments on human subjects, authors should indicate whether the procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2008. Authors must explain the rationale for their approach and demonstrate that the institutional review body explicitly approved the doubtful aspects of the study. When reporting experiments on animals, authors should indicate whether the institutional and national guide for the care and use of laboratory animals was followed.

- *Overlapping publications*

RARE Journal is not accepting for review manuscripts that are simultaneously being considered by other journals. Authors are asked to declare that the submitted material is not either considered, or being already published in another journal.

Duplicate publication of original research is not allowed. RARE Journal is not publishing manuscripts that have already been reported in large part in a published article or are contained in another paper that has been submitted or accepted for publication elsewhere, in print or in electronic media.

Nevertheless, RARE Journal is accepting for consideration papers that have been rejected by another journal, or a complete report that follows publication of a preliminary report, such as an abstract or poster displayed at a professional meeting. In the last case, the manuscript could have been presented at a scientific meeting but has not been published in full. Brief press reports of scheduled meetings are exempt from this rule, but they are required to contain additional data, tables or figures. RARE Journal is following the ICMJE decision not to consider results posted in clinical trial registries as previous publication if the results are presented in the same. The registry should either cite full publications of the results when available or include a statement that indicates that the results have not yet been published in a peer-reviewed journal.

- *Previous submission*

Authors are asked to make a complete statement to the editor about all submissions and previous reports (including meeting presentations and posting of results in registries) that might be regarded as redundant or duplicate publication. The author must alert the editor if the manuscript includes subjects about which the authors have published a previous report or have submitted a related report to another publication. Any such report must be referred to and referenced in the new paper. Copies of such material should be included with the submitted manuscript to help the editor decide how to handle the matter.

If RARE Journal was not aware of the violations and the article has already been published, then a notice of duplicate publication will be published with or without the author's explanation or approval.

- *Preliminary reporting*

Preliminary reporting to public media, governmental agencies, or manufacturers of scientific information described in a paper or a letter to the editor that has been accepted but not yet published violates the ethic policies of RARE Journal. Such reporting should be discussed with and agreed upon by the editor in advance.

- *Translations*

RARE Journal does not consider translations to be “republications” and does accept them.

- *Co-authors' disputes*

RARE Journal is not considering competing manuscripts based on same study and does not take any responsibility in dealing with co-investigators' disputes. Authors when submitting a manuscript should declare that they are the legitimate owners of the papers' rights. If RARE Journal was not aware of such violations and the article has already been published, then a notice of competing manuscript will be published with or without the author's explanation or approval.

If co-authors have differences in analysis or interpretation, they may submit a manuscript that clearly presents both versions. The difference of opinion should be explained in a cover letter. The normal process of peer and editorial review may help to resolve this disagreement. If the dispute cannot be

resolved and the study merits publication, both versions should be published. Options include publishing two papers on the same study, or a single paper with two analyses or interpretations. In such cases, it would be appropriate for the editor to publish a statement outlining the disagreement and the journal's involvement in attempts to resolve it.

- *Corrections, retractions and expressions of concern*

RARE Journal editors assume initially that authors are reporting work based on honest observations. Nevertheless, different types of problems may arise.

- *Technical errors*

Technical errors may be noted in published articles that require the publication of a correction or erratum on part of the work. The corrections would appear on a numbered page, be listed in the Table of Contents, include the complete original citation, and link to the original article. If an error is conceived as serious to vitiate the entire body of the work, RARE Journal may require a special correction or even a withdrawal of the material. Such an error should not be confused with inadequacies exposed by the emergence of new scientific information in the normal course of research. The latter requires no corrections or withdrawals.

- *Scientific fraud and plagiarism*

RARE Journal strongly condemns scientific fraud and plagiarism. If substantial doubt arises about the honesty, integrity or authorship of work, either submitted or published, the Journal ensures that the question is appropriately pursued and responded. However, it is not the responsibility of RARE Journal to conduct a full investigation or to make a determination – this responsibility lies with the institution where the work was done or with the funding agency. If such paper has been published, RARE Journal will print a retraction. If this method of investigation does not result in a satisfactory conclusion, the editor may choose to conduct his or her own investigation. As an alternative to retraction, the editor may choose to publish an expression of concern about aspects of the conduct or integrity of the work. The Journal may ask the author's institution to assure them of the validity of earlier work published in the journals or to retract it. If this is not done, RARE Journal may choose to publish an announcement expressing concern that the validity of previously published work is uncertain.

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Authors Guidelines

- *Authorship (See Journal policy)*

- *Submission*

RARE Journal is electronic journal and all articles are only online published. Submission is only online and stepwise guided. All journal correspondence takes place only by e-mail. Manuscripts are submitted by one of the authors. He/she takes responsibility for the article during submission and peer review.

- *File formats*

The following file formats are acceptable for the manuscript documents:

- Microsoft word (DOC, DOCX)
- Open office writer (ODT)
- Rich text format (RTF)

- *Submission declaration*

RARE Journal submission implies that the material described has not been published previously (except as part of a published lecture, academic thesis, or in the cases described in the Overlapping publications section of Ethical Policy), that it is not under consideration for publication elsewhere, that its publication is approved by all authors and tacitly or explicitly by the responsible authorities where the work was carried out, and that, if accepted, it will not be published elsewhere in any form, in English or in any other language, without the written consent of the copyright-holder.

The submission implies that all listed authors have read and agreed to the manuscript content, and that any experimental research that is reported has been performed with the approval of an appropriate ethics committee. Any experimental research under any circumstances must follow both national laws and internationally recognised guidelines. Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

- *Changes to authorship*

After being accepted for publication, the material can not be changed in terms of addition, deletion, or rearrangement of author names in the authorship.

- *Submission cover letter*

Corresponding author is requested to include a short cover letter when initially submitting the manuscript for publication. It should explain why the manuscript should be published in the journal, as well as elaborating on any issues relating to the editorial policies, and declaring any potential competing interests.

Authors may provide contact details (e-mail addresses) of potential peer reviewers for their manuscript. Any suggested peer reviewers should not have published with any of the authors of the manuscript within the past five years, should not be current collaborators, and should not be members of the same research institution. All suggested reviewers will be considered alongside potential reviewers recommended by the RARE Journal Editorial staff.

- *Competing Interest (See Journal Policy)*

Types of Publications

RARE Journal considers articles in the following categories. In the process of submission, authors should indicate the category under which they wish their paper to be considered. All submissions will be considered for peer-review prior to publication, with the exception of 'Editorials' and 'Letters to the Editor', which will be reviewed by the Editors-in-Chief only.

- **Original research articles**

This type of publication reports the findings of original research. It may contain the results of empirical analysis, instrument development or policy analysis. They may be elaborated for proposed or ongoing research, and should provide a detailed account of the hypothesis, methodology and conclusion of the study. These papers can be up to 5 000 words long, excluding references, and may have up to 5 figures or tables and up to 50 references.

- **Systematic reviews and meta-analyses**

This type of publication reports on reviews of empirical studies consistent with the methods of systematic review proposed by the Cochrane Collaboration. It identifies, appraises, selects and synthesises high quality research evidence relevant to one or more focus topics. It may present reviews of randomised controlled trials, observational studies, economic evaluations, outcomes research studies and preference-based assessments. These papers can be up to 6 000 words long, excluding references, and may have up to 5 figures or tables, no limit of references.

- **Review articles**

This type of publication reports important rare diseases topics within the scope of the journal. They may reflect conceptual pieces or reviews of the literature. These papers can be up to 3 000 words long, excluding references, and may have up to 4 figures or tables.

- **Methodology articles**

This type of publication reports on methodological issues in any of the topic areas within the rare diseases field. It presents a new experimental or computational method, test or procedure (e.g., epidemiological methods, survey methods and health impact assessment methods). The method described may either be completely new, or may offer a better version of an existing method. It can include data if these are required to illustrate the importance of particular methodological points. These papers can be up to 2 000 words, excluding references, and may have up to 3 figures or tables and up to 20 references.

- **Case reports**

This type of publication reports on empirical research outcomes with a more narrow focus than original research articles and generally a single aim. It usually represents a description of new or unusual events, relating to a clinical condition, association, reaction, treatment, etc, in one or more patients that can advance basic comprehension of a medical condition, increase scientific knowledge, or draw proposals for further investigation. It is one of the most common epidemiological tools for studying rare diseases and rare forms of common disorders. These papers can be up to 2 000 words, excluding references, and may have up to 2 figures or tables and up to 20 references.

- **Letters to the Editors**

This type of publication represents a short communication allowing quick feedback or comments from the Journal's readership. It may offer substantial re-analysis or a brief report of research findings that are relevant for a specific article, published in RARE Journal. The letters to Editors are accepted for consideration and publication only in a 30-day period after the concerned material has been published. In instances where the letter contains a critique of a published paper, the authors will be given the right to reply. These papers can be up to 1 000 words, 1 table or 1 figure, 10 references.

- **Editorials**

Editorials are commissioned by the Editorial Board and report on issues of current interest in the field of rare diseases. It covers an aspect of an issue that is relevant to the RARE Journal's scope. Examples of this type of commentary could be a discussion of the impact of new technology on research and treatment, or a discussion of changes in peer review or grant application procedures and their effect on research. These papers can be up to 1 000 words long, but may be longer, with agreement from the Editors.

RARE Journal particularly welcomes articles in the following rare diseases topic areas:

1 Reports of clinical practice guidelines – this type of publication represents a summary of evidence-based clinical practice guidelines that are issued by health organisations, scientific societies, academic institutions, patient advocacy groups, etc. Maximum volume of 5 000 – 6 000 words, 5 tables and figures in total, no limit for references.

2 Reports of health technology assessment (HTA) – this type of publication presents summarized results of the examination process of medical, economic, social and ethical implications of the incremental value, diffusion and use of a medical technology in healthcare. Such reports support management, clinical, and policy decisions by providing scientifically rigorous tools to address a focused policy question. Maximum volume of 4 000 words, 7 tables and figures in total, no limit of references.

3 Reports of epidemiological registries – this type of publication presents summarized results and conclusions of epidemiological registries for rare diseases. These reports are an important source of information for medical professionals, healthcare providers and public health officials. Epidemiological registries reports are useful for preventing and controlling rare diseases and improving treatment and patient care. Data from these publications may share and compare prevalence and incidence rates, describe disease patterns in the population, and discuss planning programmes for people affected with rare diseases. Maximum volume of 4 000 words, 7 tables and figures in total, 25 references.

4 Meeting and project reports – this type of publication aims to improve visibility and increase dissemination of specific events and activities. It encourages to present scientific and public health initiatives that may be important for specific segments of Journal’s readership. A meeting or project reports generally features communication details, progress update, specific conclusions, expertise and advice. Meeting reports are accepted for consideration and publication only in a 30-day period after the concerned event has took place, while project reports – in a 3-month period after the concerned project’s closure. A meeting report can be only submitted by meeting organisers, while project reports should be submitted by project coordinator. Both authors are encouraged to contact the Editors-in-Chief with proposals for such publications. Maximum volume of 1 500 words, 1 table or 1 figure, 10 references.

- *General Requirements for Authors*
- *Bibliography*

All references, including URLs, in the manuscript have to be done as numbers in square brackets, followed by any in tables or legends. All references must be listed at the end of the paper on a separate page, arranged in numerical order of their appearance in the text. There should be clear correspondence between the names and years in the text and those on the bibliography. Authors should provide direct references to original research sources whenever possible. Authors are also kindly asked to avoid excessive referencing.

Authors should avoid using abstracts as references. References to papers accepted but not yet published should be designated as “in press” or “forthcoming”. Authors may be asked to provide verification that these have been accepted for publication. Authors should avoid citing unpublished abstracts, unpublished data and personal communications. When such are used, authors are requested to obtain and provide a written permission and confirmation of accuracy from the source.

Citations in the reference list should include all named authors, up to the first 3 before adding '*et al.*'.

All web links and URLs should be given a reference number and included in the reference list. They should be provided in full, including the title of the site, the URL and the date of last access.

Authors should consult and use the US National Library of Medicine’s (NLM) Citing Medicine for information on its recommended formats for reference types (http://www.nlm.nih.gov/bsd/uniform_requirements.html). The titles of journals should be abbreviated according to the style used in the list of Journals Indexed for MEDLINE, posted by the NLM on the Library’s Web site.

- *Figures and tables*

Include only figures and tables that are essential to clarify arguments. Figures and tables should be easy to understand, with proper title, labels, and legend. All supporting tables, figures and illustrations should be as self-explanatory as possible. Figures and tables should be provided as separate files, not embedded in the text file. Titles and detailed explanations belong in the legends, not on the illustrations themselves.

The following minimum resolution requirements for figures and other graphic illustrations are recommended:

1. TIFF format – minimum of 300 dpi for color or grayscale photographs (halftones), and a minimum of 1000 dpi for bitmapped line drawings
2. Microsoft Office application (DOC/DOCX, PPT/PPTX, XLS/XLSX) should be submitted in original version.

For x-ray films, scans, and other diagnostic images, as well as pictures of pathology specimens or photomicrographs, authors should send sharp, glossy, black-and-white or color photographic scans, 127 x 173 mm (5 x 7 inches). Photomicrographs should have internal scale markers. Symbols, arrows, or letters used in photomicrographs should contrast with the background.

RARE Journal will publish only author-owned graphic illustrations. Authors have the responsibility to respect the copyright and intellectual property rights. Photographs of potentially identifiable people must be accompanied by written permission to use the photograph.

All figures and tables should be explicitly referred in the manuscript in a numbered way according to their appearance.

- *Appendices*

Appendices should be generally avoided. Technical details (samples, questionnaires, etc.) may be needed for the review process, but won't appear in the final version for publication. They could be available from the authors upon request.

- *Style and language*

RARE Journal only accepts manuscripts written in English. Spelling should be US English or British English, but not a mixture. Spelling and style editing is a responsibility of the authors. Submitted manuscripts would not be edited by the Journal. However, reviewers may advise rejection of a manuscript if it is compromised by grammatical errors.

- *Typography*

Authors are kindly asked to observe the following requirements:

- Text should be double line spaced.
- Text should be unjustified, without hyphenating words at line breaks.
- Hard returns can be only used to end headings and paragraphs, not to rearrange lines.
- Manuscripts should include a line count.
- Only the first word and proper nouns in the title should be capitalised.
- All pages should be numbered.
- *Abbreviations and symbols*

Authors are recommended to use only standard abbreviations. The spelled-out abbreviation followed by the abbreviation in parenthesis should be used on first mention unless the abbreviation is a standard unit of measurement.

- *Units of measurement*

Authors should report measurements of length, height, weight, and volume in metric units (meter, kilogram, or liter) or their decimal multiples. Temperatures should be in degrees Celsius. Blood pressures should be in millimeters of mercury. Drug concentrations may be reported in either SI or mass units.

- *Publication-Specific Guidelines*

- Research articles

RARE Journal demands as a condition of consideration for publication of research articles a registration in a public trials registry (http://www.icmje.org/publishing_10register.html). Authors are required to follow the CONSORT reporting guidelines for randomized controlled trials and STROBE for observational studies (<http://www.cochrane.org/about-us/evidence-based-health-care/webliography/books/reporting>) (see also *Standard of reporting*) Authors are kindly asked to adhere to these guidelines, when submitting research articles to the Journal.

Manuscript structure for research articles

Manuscripts for research articles should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Material and methods
- Results and discussion
- Conclusions
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
- Tables (if any)

Title page

Title page should include:

Title

The manuscript's title should be appropriate and informative. It is a very important detail of the work, as it essential for information retrieving. Authors should include all information in the title that will make electronic retrieval of the article both sensitive and specific.

Authors' names and affiliations

Authors' affiliation addresses should be presented below the names. All affiliations must be indicated with a lower-case superscript letter immediately after the author's name and in front of the appropriate address.

Disclaimers, if any

Corresponding author

Provide phone number, e-mail and complete postal address of the designated corresponding author. He/she should handle correspondence with RARE Journal at all publishing stages. The corresponding author should indicate clearly whether his or her e-mail address can be published.

Source(s) of support in the form of grants, equipment, drugs, or all of these

Word count (excluding abstract, acknowledgments, figures, tables and references)

Number of figures and tables

Abstract

This section should not exceed 350 words and must follow the structure of the manuscript itself. As the abstract is most likely to be read, authors should formulate it in informative and attractive manner. Indicate clearly the purpose, the principal findings and major conclusions of the submitted paper. It should emphasise new and important aspects of the study or observations. It is recommended to mention the country, as well as methods and dates. Abbreviations, acronyms and references within the abstract should be avoided.

Keywords

Authors are asked to include 3 to 6 keywords representing the main content of the article, preferably from the Medical Subject Headings from Index Medicus.

Introduction

This section should be written in an accessible style to researchers without specialist knowledge in that area and must clearly state the background to the research and its aims. If appropriate, it should include a summary of a literature search and a state-of-the-art analysis to indicate why this study was necessary and how it contributes to increasing the body of knowledge in this field.

Material and methods

This section should include information about the study's design, settings, participants or materials involved, description of all interventions and comparisons. Authors should describe the selection of observational or experimental participants clearly, including eligibility and exclusion criteria and a description of the source population. References should be provided to established methods, and brief descriptions should be given for methods that have been published but are not well-known. It should be notified if new or substantially modified methods are used. All drugs and chemicals used, including generic name, dose, and route of administration have to be precisely specified.

Authors should inform about the statistical methods they used. When possible, findings should be quantified and presented with appropriate indicators of measurement error or uncertainty.

A statement detailing ethical approval and consent should be included in the methods section for studies involving human participants. Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

Results and discussion

The results and discussion may be combined into a single section or presented separately.

Results are best presented in tables with accompanying diagrams whenever possible. Statistical analysis (if appropriate) should include relative and absolute risks or risk reductions, and confidence intervals. Most important findings could be emphasised or summarised in text. If the section is too long, subsections and subheadings may be used.

The discussion part should not repeat in detail the results section's data. It should outline and critically assess important aspects of the study, their practical relevance, implications and impact in the specific field of knowledge. Results should be related and compared to other published research outcomes. Any study limitations should be also considered.

Conclusions

This section should state clearly and briefly the main conclusions of the research and give a clear explanation of their importance and relevance. It should assess whether the initial objectives are met and propose future areas for research.

Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

Acknowledgements

Anyone who contributed towards the article by making substantial contributions, but who does not meet the criteria for authorship, should be mentioned in this section.

Authors are also required to include information about the source(s) of funding for the study, and for the manuscript preparation. Funding body's role should be also described.

- Case reports

Manuscript structure for case reports

Manuscripts for case reports should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Case presentation
- Conclusions
- Consent
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
- Tables (if any)

Title page

Title page should include:

Title

The manuscript's title should be appropriate and informative. The title should clearly identify the manuscript as a case report. It is a very important detail of the work, as it essential for information retrieving. Authors should include all information in the title that will make electronic retrieval of the article both sensitive and specific.

Authors' names and affiliations

Authors' affiliation addresses should be presented below the names. All affiliations must be indicated with a lower-case superscript letter immediately after the author's name and in front of the appropriate address.

Disclaimers, if any

Corresponding author

Provide phone number, e-mail and complete postal address of the designated corresponding author. He/she should handle correspondence with RARE Journal at all publishing stages. The corresponding author should indicate clearly whether his or her e-mail address can be published.

Source(s) of support in the form of grants, equipment, drugs, or all of these

Word count (excluding abstract, acknowledgments, figures, tables and references)

Number of figures and tables

Abstract

This section should not exceed 200 words and must follow the structure of the manuscript itself. As the abstract is most likely to be read, authors should formulate it in an informative and attractive manner. Indicate clearly the purpose, the principal findings and major conclusions of the submitted paper. It should emphasise new and important aspects of the case(s) or observations. It is recommended to mention the patient's characteristics (sex, age, ethnicity, etc), country, as well as methods and dates. Abbreviations, acronyms and references within the abstract should be avoided.

Keywords

Authors are asked to include 3 to 6 keywords representing the main content of the article, preferably from the Medical Subject Headings from Index Medicus.

Introduction

This section should be written in an accessible style to researchers without specialist knowledge in that area and must clearly state and introduce the case's background. If appropriate, it should include a summary of a literature search and a state-of-the-art analysis to indicate why this study was necessary and how it contributes to increasing the body of knowledge in this field.

Case presentation

This section should include information about the patient's demographic information (any details that could lead to the identification of the patient must be removed), medical history, symptoms and signs, tests, undergone treatment and/or interventions. References should be provided to established methods, and brief descriptions should be given for methods that have been published but are not well-known. It should be notified if new or substantially modified methods are used. All drugs and chemicals used, including generic name, dose, and route of administration have to be precisely specified. If the section is too long, subsections and subheadings may be used.

Conclusions

This section should state clearly and briefly the main conclusions of the presented case and give a clear explanation of their importance and relevance. It should assess whether the initial objectives are met and propose future areas for research.

Consent

Authors are required to provide a confirming statement that the patient(s) has (have) given written consent for the case report to be published. This document may be further requested by the RARE Journal at any time. If the patient has died, then consent must be sought from the patient's relatives. If the patient is a minor or unable to provide consent, then consent must be sought from the parents or legal guardians of the patient.

Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

Acknowledgements

Anyone who contributed towards the article by making substantial contributions, but who does not meet the criteria for authorship, should be mentioned in this section.

Authors are also required to include information about the source(s) of funding for the study, and for the manuscript preparation. Funding body's role should be also described.

- Systematic reviews and meta-analyses

RARE Journals adopts the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) and MOOSE (Proposal for Reporting Meta-Analyses of Observational Studies in Epidemiology) statements (<http://www.cochrane.org/about-us/evidence-based-health-care/webliography/books/reporting>). Authors are kindly asked to adhere to these guidelines, when submitting systematic reviews and meta-analyses to the Journal.

Manuscript structure for systematic reviews and meta-analyses

Manuscripts for systematic reviews and meta-analyses should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Material and methods
- Results and discussion
- Conclusions

- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
- Tables (if any)

Title page

Title page should include:

Title

The manuscript's title should be appropriate and informative. It is a very important detail of the work, as it essential for information retrieving. The title should clearly identify the manuscript as a systematic review, meta-analysis, or both. Authors should include all information in the title that will make electronic retrieval of the article both sensitive and specific.

Authors' names and affiliations

Authors' affiliation addresses should be presented below the names. All affiliations must be indicated with a lower-case superscript letter immediately after the author's name and in front of the appropriate address.

Disclaimers, if any

Corresponding author

Provide phone number, e-mail and complete postal address of the designated corresponding author. He/she should handle correspondence with RARE Journal at all publishing stages. The corresponding author should indicate clearly whether his or her e-mail address can be published.

Source(s) of support in the form of grants, equipment, drugs, or all of these

Word count (excluding abstract, acknowledgments, figures, tables and references)

Number of figures and tables

Abstract

This section should not exceed 350 words and must follow the structure of the manuscript itself. As the abstract is most likely to be read, authors should formulate it in informative and attractive manner. Indicate clearly the purpose, the principal findings and major conclusions of the submitted paper. It should emphasise new and important aspects of the study or observations. It is recommended to

mention the methods and dates. Abbreviations, aconyms and references within the abstract should be avoided.

Keywords

Authors are asked to include 3 to 6 keywords representing the main content of the article, preferably from the Medical Subject Headings from Index Medicus.

Introduction

This section should be written in an accessible style to researchers without specialist knowledge in that area and must clearly state the rationale to the research and its aims. If appropriate, it should include a summary of a literature search and a state-of-the-art analysis to indicate why this study was necessary and how it contributes to increasing the body of knowledge in this field.

Material and methods

This section should include information about the review's protocol and registration (if appropriate), eligibility criteria and specific characteristics, information sources used, search strategy and data collection process. Risk management should be specifically addressed – risk of bias of individual studies and summary measures (e.g., risk ratio, difference in means), risk of bias across studies, methods of additional analyses (e.g., sensitivity or subgroup analyses, meta-regression).

Results and discussion

The results and discussion may be combined into a single section or presented separately.

Results are best presented in tables with accompanying diagrams whenever possible. Statistical analysis (if appropriate) should include relative and absolute risks or risk reductions, and confidence intervals. Most important findings could be emphasised or summarised in text. If the section is too long, subsections and subheadings may be used.

The discussion part should not repeat in detail the results section's data. It should outline and critically assess important aspects of the review, its practical relevance, implications and impact in the specific field of knowledge. Results should summarise the main findings including the strength of evidence and relevance to key groups. Any study limitations should be also considered.

Conclusions

This section should state clearly and briefly the main conclusions of the research and give a clear explanation of their importance and relevance. It should assess whether the initial objectives are met and propose future areas for research.

Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

Acknowledgements

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- Reports of clinical practice guidelines

Manuscript structure for reports of clinical practice guidelines

Manuscripts for reports of clinical practice guidelines should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Material and methods
- Results and discussion
- Conclusions
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
- Tables (if any)

Title page

Title page should include:

Title

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Authors' names and affiliations

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Material and methods

This section should include information about the clinical practices guideline's process of development and information about the organisation that has produced the guideline.

Results and discussion

The results and discussion may be combined into a single section or presented separately. If the section is too long, subsections and subheadings may be used.

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- Reports of health technology assessment

Manuscript structure for reports of health technology assessment

Manuscripts for reports of clinical practice guidelines should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Material and methods
- Results and discussion
- Conclusions
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
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- Tables (if any)

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Material and methods

This section should include information about the health technology assessment report's protocol and registration (if appropriate), eligibility criteria and specific characteristics, elaboration and adoption chronology, information about the organisation that has produced this report, its practical application and outcomes so far.

Results and discussion

The results and discussion may be combined into a single section or presented separately.

Results are best presented in tables with accompanying diagrams whenever possible. Statistical analysis (if appropriate) should include relative and absolute risks or risk reductions, and confidence intervals. Most important findings could be emphasised or summarised in text. If the section is too long, subsections and subheadings may be used.

The discussion part should not repeat in detail the results section's data. It should outline and critically assess important aspects of the report, its practical relevance, implications and impact in the specific field of knowledge. Results should summarise the main findings including the strength of evidence and relevance to key groups. Any study limitations should be also considered.

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Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

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- Reports of epidemiological registries

Manuscript structure for reports of epidemiological registries

Manuscripts for reports of epidemiological registries should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Construction and content
- Results and discussion
- Conclusions
- Availability and requirements
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
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Abstract

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Keywords

Authors are asked to include 3 to 6 keywords representing the main content of the article, preferably from the Medical Subject Headings from Index Medicus.

Introduction

This section should be written in an accessible style to researchers without specialist knowledge in that area and must clearly state the rationale to the research and its aims. If appropriate, it should include a summary of a literature search and a state-of-the-art analysis to indicate why this registry was necessary and how it contributes to increasing the body of knowledge in this field.

Construction and content

This section should include information about the epidemiological registry's schema and registration (if appropriate), eligibility criteria and specific characteristics, data sources and data generation, quality control, preparation and implementation chronology, information about the organisation that is managing the registry, its practical application and outcomes so far.

A statement detailing ethical approval and consent should be included in the methods section for studies involving human participants. Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

Results and discussion

The results and discussion may be combined into a single section or presented separately.

Results are best presented in tables with accompanying diagrams whenever possible. Statistical analysis (if appropriate) should include relative and absolute risks or risk reductions, and confidence intervals. Most important findings could be emphasised or summarised in text. If the section is too long, subsections and subheadings may be used.

The discussion part should not repeat in detail the results section's data. Intended uses and benefits of the registry's implementation should be presented, together with data on how its performance and functionality, comparison with similar registries. A case study of the use of the registry may be included. Results should summarise the main findings including the strength of evidence and relevance to key groups. Any study limitations should be also considered.

Conclusions

This section should state clearly and briefly the main conclusions of the registry and give a clear explanation of their importance and relevance. It should assess whether the initial objectives are met and propose future areas for development.

Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

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- Methodologies

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- Introduction
- Material and methods
- Results and discussion
- Conclusions
- Competing interests

- Authors' contributions
- Acknowledgements (if any)
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Keywords

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Introduction

This section should be written in an accessible style to researchers without specialist knowledge in that area and must clearly state the rationale to the research and its aims. If appropriate, it should include a summary of a literature search and a state-of-the-art analysis to indicate why this methodology was necessary and how it contributes to increasing the body of knowledge in this field.

Material and methods

This section should include information about the design of the study, the type of materials involved, a clear description of all comparisons, and the type of analysis used, to enable replication, elaboration and adoption chronology, information about the organisation that has produced this methodology, its practical application and outcomes so far.

Authors should describe the selection of observational or experimental participants clearly, including eligibility and exclusion criteria and a description of the source population. References should be provided to established methods, and brief descriptions should be given for methods that have been published but are not well-known. It should be notified if new or substantially modified methods are used. All drugs and chemicals used, including generic name, dose, and route of administration have to be precisely specified.

Authors should inform about the statistical methods they used. When possible, findings should be quantified and presented with appropriate indicators of measurement error or uncertainty.

A statement detailing ethical approval and consent should be included in the methods section for studies involving human participants. Identifying information (including names, initials, or hospital numbers) of study participants should not be published under any circumstances. It is the authors' responsibility to respect the patients' right of privacy, as well as the national and international legal framework on this issue.

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Conclusions

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Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

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- Commentaries

Manuscript structure for commentaries

Manuscripts for commentaries should be divided into the following sections in this order:

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- Abstract
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- Main text
- Conclusions
- Competing interests
- Authors' contributions
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- Bibliography
- Figures (if any)
- Tables (if any)

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Keywords

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Introduction

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Main text

This section should contain the body of the article. If it is too long, subsections and subheadings may be used. It should outline and critically assess important aspects of a specific field, their practical relevance, implications and impact in the specific field of knowledge.

Conclusions

This section should state clearly and briefly the main conclusions of the commentary and give a clear explanation of its importance and relevance. It should assess the current achievements in a specific field and propose future areas for research.

Competing interests (see Journal policy)

Authors' contributions (see Journal policy)

Acknowledgements

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- Letters to Editors

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- Abstract
- Keywords
- Correspondence
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- Acknowledgements (if any)
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- Figures (if any)
- Tables (if any)

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Competing interests (see Journal policy)

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- Meeting and project reports

Manuscript structure for meeting and project reports

Manuscripts for ***for meeting and project reports*** should be divided into the following sections in this order:

- Title page
- Abstract
- Keywords
- Introduction
- Main text
- Conclusions
- Competing interests
- Authors' contributions
- Acknowledgements (if any)
- Bibliography
- Figures (if any)
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Title page

Title page should include:

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Introduction

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Main text

This section should include information about the meeting's/project's framework – objectives, tasks, activities, deliverables, milestones, participants, funding, duration, scope, dissemination and evaluation.

Meeting/project results are best presented in tables with accompanying diagrams whenever possible. Statistical analysis (if appropriate) should include relative and absolute risks or risk reductions, and confidence intervals. Most important findings could be emphasised or summarised in text. Intended uses and benefits of the meeting/project should be presented, together with data and comparison with

similar initiatives. Outcomes should summarise the main findings including the strength of evidence and relevance to key groups.

Finally, it should assess whether the initial objectives are met and propose future areas for development.

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