DOI 10.18471/rbe.v38.56685

COMPUTER TOOLS FOR INFORMATION MANAGEMENT OF PATIENTS WITH RARE DISEASES: AN INTEGRATIVE REVIEW

FERRAMENTAS COMPUTACIONAIS PARA GESTÃO DA INFORMAÇÃO DE PACIENTES COM DOENÇAS RARAS: REVISÃO INTEGRATIVA

HERRAMIENTAS COMPUTACIONALES PARA LA GESTIÓN DE LA INFORMACIÓN DE PACIENTES CON ENFERMEDADES RARAS: REVISIÓN INTEGRADORA

Greici Capellari Fabrizzio[1]
Lincoln Moura de Oliveira[2]
Fred Jorge Oliveira Borges Junior[3]
Roberta Eduarda Grolli[4]
Gabriela Marcellino de Melo Lanzoni[5]
Alacoque Lorenzini Erdmann[6]
José Luís Guedes dos Santos[7]

How to cite this article: Fabrizzio GC, Oliveira LM, Borges Junior FJO, Grolli RE, Lanzoni GMM, Erdmann AL, et al. Computer tools for information management of patients with rare diseases: integrative review. Rev baiana enferm. 2024;38:e56685.

Objctive: identifying computer tools for information management of patients with rare diseases. Method: integrative literature review, carried out in November 2020, in the National Library of Medicine, Latin American and Caribbean Literature in Health Sciences, Scientific Electronic Library Online databases. The sample consisted of eight articles, after applying the selection criteria. Results: software tools, systems and portals were created to standardize language, identify genes, and aid in diagnosing and treating pathologies. These tools have made it possible to systematize data for information management, with clinical, epidemiological, and occurrence information. Conclusion: computational tools with an emphasis on rare diseases can help in the areas of management, clinical practice, and epidemiology, contributing to planning, evaluation, the development of new health policies, cost reduction, early diagnosis, decision-making, and disease monitoring.

Descriptors: Health Information Systems. Information Management. Rare Diseases. Biomedical Technology. Review.

Corresponding author: Greici Capellari Fabrizzio, greicicapellari@gmail.com

[1]Universidade Federal de Santa Catarina. Florianópolis, SC, Brasil. https://orcid.org/0000-0002-3848-5694.
[2] Universidade Federal do Ceará. Fortaleza, CE, Brasil. https://orcid.org/0000-0001-6016-745X.
[3] Universidade Federal da Bahia. Salvador, BA, Brasil. https://orcid.org/0000-0003-0718-2233.
[4] Universidade Federal de Santa Catarina. Florianópolis, SC, Brasil. https://orcid.org/0000-0002-5657-9280.
[5] Universidade Federal de Santa Catarina. Florianópolis, SC, Brasil. https://orcid.org/0000-0001-5935-8849.
[6] Universidade Federal de Santa Catarina. Florianópolis, SC, Brasil. https://orcid.org/0000-0003-4845-8515.
[7] Universidade Federal de Santa Catarina. Florianópolis, SC, Brasil. https://orcid.org/0000-0003-3186-8286.

Objetivo: identificar ferramentas computacionais para a gestão da informação de pacientes com doenças raras. Método: revisão integrativa de literatura, realizada em novembro de 2020, nas bases de dados National Library of Medicine, Literatura Latino-Americana e do Caribe em Ciências da Saúde, Scientific Electronic Library Online. A amostra foi composta por oito artigos, após aplicados os critérios de seleção. Resultados: evidenciaram-se ferramentas de softwares, sistemas e portais, criadas com intuito de padronização de linguagem, identificação de genes, auxílio ao diagnóstico e tratamento de patologias. Essas ferramentas viabilizaram a sistematização dos dados para a gestão da informação, contando com informações clínicas, epidemiológicas e de ocorrência. Conclusão: as ferramentas computacionais com ênfase em doenças raras podem auxiliar nos âmbitos da gestão, da clínica e epidemiologia, contribuindo para o planejamento, avaliação, elaboração de novas políticas de saúde, redução de custos, diagnóstico precoce, tomada de decisão e monitoramento das doenças.

Descritores: Sistemas de Informação em Saúde. Gestão da Informação. Doenças Raras. Tecnologia em Saúde. Revisão.

Objetivo: identificar herramientas computacionales para la gestión de información de pacientes con

enfermedades raras. Método: revisión integradora de la literatura, realizada en noviembre de 2020, en las bases de datos National Library of Medicine, Literatura Latinoamericana y del Caribe en Ciencias de la Salud, Scientific Electronic Library Online. La muestra estuvo compuesta por ocho artículos, luego de aplicar los criterios de selección. Resultados: se destacaron herramientas de software, sistemas y portales, creados con el objetivo de estandarizar el lenguaje, identificar genes, contribuir en el diagnóstico y tratamiento de patologías. Estas herramientas permitieron la sistematización de datos para la gestión de la información, sobre la base de información clínica, epidemiológica y reportes. Conclusión: las herramientas computacionales con énfasis en enfermedades raras pueden ayudar en las áreas de gestión, clínica y epidemiología, contribuyendo a la planificación, evaluación, desarrollo de nuevas políticas de salud, reducción de costos, diagnóstico temprano, toma de decisiones y seguimiento de enfermedades.

Descriptores: Sistemas de Información en Salud. Gestión de la Información. Enfermedades Raras. Tecnología Biomédica. Revisión.

Introduction

Rare diseases are those that affect up to 65 individuals per 100,000 people[1]. However, this figure can vary according to the criteria adopted in each country. In Brazil, it is estimated that 13 to 15 million Brazilians are affected by rare diseases[2].

In Brazil, there are health policies to increase the visibility of these diseases and patients' access to diagnosis and treatment. In 2014, Ordinance 199 of the Ministry of Health established the National Policy for Comprehensive Care for People with Rare Diseases within the Unified Health System (*Sistema Único de Saúde*, SUS) and instituted financial incentives[1]. In addition, the creation of a new national policy for the treatment of these pathologies is already being discussed, and it has been established that the Brazilian National Day for Rare Diseases will be celebrated on the last day of February.

Rare diseases, when looked at individually, can mistakenly be considered to affect a small portion of the population. However, when analyzing the data as a whole, it is possible to see the high number of individuals who face this health condition. Thus, the interest and capacity of researchers to develop research on these issues has a high impact on the health of the population[3].

In this context, managers, researchers and professionals working in this area have dedicated themselves to studying and developing new tools for managing information on rare diseases, with the help of Information and Communication Technologies (ICT). An integrated information system, structured in such a way as to help organize data, can help in the management of rare diseases and the sharing of experiences and information relating to these pathologies[4].

Caring for individuals with rare diseases requires multidisciplinary work and nursing is the category closest to patients and their families[5].The management and systematization of care is refined with the help of technology. Computerized systems make it possible to collect and process a large amount of data, enabling precise decision-making. The formulation of protocols and therapeutic itineraries are optimized with the

use of occupational tools for data management and grouping, providing the best therapeutic choice[6].

Based on the above, and in view of the impact that the flow of information has on the management of rare diseases, this study poses the following question as its research problem: *What computer tools for information management of patients with rare diseases are available?* The aim of this research is therefore to identify computer tools for information management of patients with rare diseases.

Method

This is an integrative literature review, carried out in November 2020, in the National Library of Medicine (PubMed), Latin American and Caribbean Health Sciences Literature (LILACS), and Scientific Electronic Library Online (SciELO) databases. The literature review comprised six sequential and independent stages, described below: Identification of the topic and elaboration of the research question; Establishment of inclusion and exclusion criteria for the articles and the literature search; Definition of the information to be collected from the articles and their categorization; Evaluation of the articles to be included; Interpretation of results; Synthesis of the review[7].

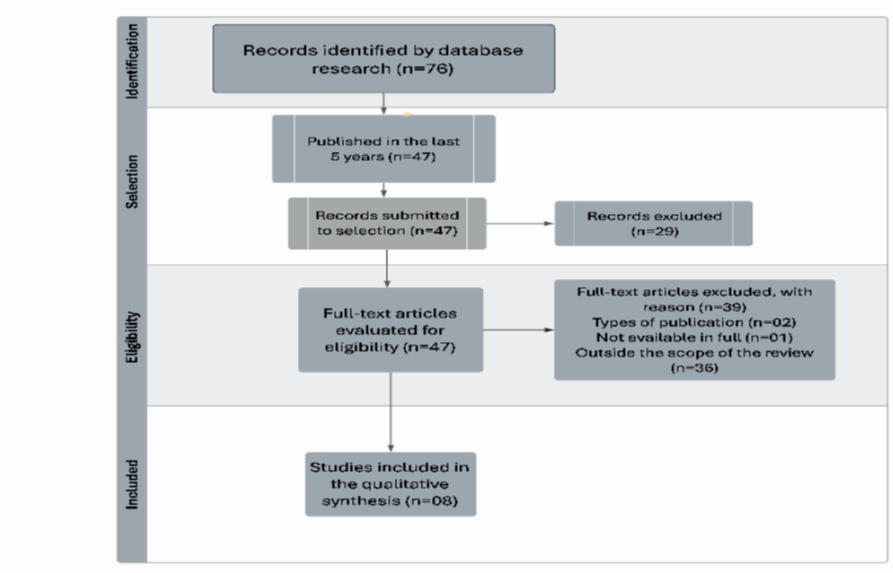
This study considered articles from primary studies published between 2015 and 2020. This time frame was established because it involves recent research on the topic and because of the increase in the use of computer tools in the health area in recent years.

The inclusion criteria for the articles were: articles published in the last five years, in English, Portuguese, or Spanish; articles from qualitative, quantitative, and mixed methods research. The exclusion criteria were: articles unavailable in full; annals of scientific events, review articles, dissertations, theses, books, and book chapters; duplicate studies or those that did not meet the scope of the research question.

The search strategy used in the databases was: health information systems AND information management AND rare diseases. The descriptors were extracted from the Medical Subject Headings (MeSH) and used in English in the PubMed; LILACS and SciELO databases.

When applying the search strategy, 76 articles were found; of these, 47 had been published in the last five years. After reading the titles and abstracts, one article was excluded because it was not entirely available, two articles were literature reviews and 36 articles did not fit the scope of this review. Thus, the corpus of analysis was composed of eight scientific articles, selected to compose the results, in line with the PRISMA recommendation, as shown in Figure 1. The data was processed and organized using *Lucidchart* diagramming and visual communication software and *Orange Data Mining* software. This literature review did not require acceptance by the Ethics Committee, as it consists of a theoretical and critical analysis of already published studies, and does not involve primary data collection or direct interaction with human or animal subjects.

Figure 1 – Flowchart of the article selection process



Source: prepared by the authors

Results

Chart 1 shows the results of the eight articles selected for this integrative review.

Chart 1 – Studies included in the sample with information on title, objective, and main results

(continued)

N.	Title of the article, professional affiliation of the authors	Objective and design of the study	Results (technology category)
1	An innovative electronic health records system for rare and complex diseases[8]. The authors are associated with the Computer Science Department of the Federal University of Minas Gerais.	Technological study, based on the workflow of health professionals aiming to propose the use of computer applications - Laboratory Information Management System (LIMS), for the diagnosis and treatment of rare diseases.	The FluxMED system is an electronic health record platform, capable of managing rare disease data in a highly specialized way, personalized for each disease, and assisting in diagnosis and treatment.
2	Registries for rare diseases: OSSE – An open-source framework for technical implementation[9]. The authors are researchers from different groups of medical informatics, epidemiology, and informatics, the reference center for rare diseases in Frankfurt and Heidelberg, Germany.	Software development study aiming to unify and streamline the process of establishing specific records for patients with rare diseases.	The study resulted in the development of a central Metadata Repository (MDR), which provides information on a specific disease, supports clinical research, and the quality and improvement of epidemiological links.

Chart 1 – Studies included in the sample with information on title, objective, and main results
(continued)

N.	Title of the article, professional affiliation of the authors	Objective and design of the study	Results (technology category)
3	Improving the visibility of rare diseases in health care systems by specific routine coding[10]. The authors are researchers at the German Institute for Medical Documentation and Information, Cologne, Germany.	Data analysis study, applying double coding to code rare diseases in a specific way.	The study resulted in the development of a dual coding system that integrates ICD-10 codes with Orphanet-specific codes (Orpha-Kennnummer) and retrieves rare disease concepts from ICD-11 through a unique identifier and a link to other classification systems.
4	The matchmaker Exchange: a plataforma for rare disease gene discovery[11]. The authors are affiliated with 36 institutes in the following countries: the United States, Spain, the United Kingdom, Canada, the Netherlands and Germany.	Technological study, for the development of an application programming interface (API) that provides a robust and systematic approach to the discovery of rare disease genes through the federated network, connecting databases of rare genotypes and phenotypes.	The study resulted in an application programming interface with matching algorithms for crossing genotypes and phenotypes. Its operation consists of innovative data sharing by the computational infrastructure, to support the scaling of genomics, improving human health.
5	Conceptualization and implementation of the Central Information Portal on Rare Diseases: Protocol for a qualitative study[12]. The authors are associated with research groups in medical informatics, health economics, genetics and rare disease institutes in Frankfurt, Hannover, Freiburg and Berlin, Germany.	Technological study, with the aim of conducting scientific research on how to design a portal to meet the needs of patients, families, professionals in the field and provide high quality information to those seeking information.	The study resulted in the implemented information portal – ZIPSE- providing high-quality information on rare diseases from a central access point.
6	Contribution of electronic medical records to the management of rare diseases[13]. The authors are associated with departments and medical centers of ophthalmology, cytogenetics, and medical informatics in Paris and Amiens, France.	Health technology evaluation study to determine whether electronic medical records in ophthalmology contribute to the management of rare eye diseases, either isolated or in syndromes.	The study resulted in the specific development of an electronic health record platform for ophthalmology, which made it possible to identify rare eye diseases through electronic medical records useful for clinical research.

Chart 1 – Studies included in the sample with information on title, objective, and main results (conclusion)

N.	Title of the article, professional affiliation of the authors	Objective and design of the study	Results (technology category)
7	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders[14]. The authors are associated with medical and genomic centers and university hospitals in Manchester, United Kingdom.	Experience report on the first 12 months of using European reference networks – ERN ITHACA – to facilitate clinical consultations between doctors and relevant specialists on complex or rare diseases.	Information technology that enabled multidisciplinary and personalized case management.
8	The role of primary care in the management of care diseases in Irland[15]. The authors are associated with national medical centers and clinical programs for rare disease research in Ireland.	Retrospective cross-sectional study to estimate the workload of general practitioners attributable to selectable rare diseases and evaluate the use of relevant information sources.	A pilot survey of general practitioners providing primary care to patients with rare diseases found that specific coding systems were inadequate for identifying patients with rare diseases.

Source: prepared by the authors.

Regarding the location of the eight studies, three were carried out in Germany, one in Brazil, one in France, one in Ireland and two multicenter studies involving Canada, the United States and the United Kingdom. Regarding language, six articles were published in English and two in German. Regarding the year of publication, three articles were published in 2015, two in 2020, two in 2017 and one in 2018.

In terms of the type of technology, two studies presented electronic registry platforms; one article demonstrated the development of a metadata repository; one article presented system development; one study described interoperability as central, with API development; one study registered an information portal; one article resulted in information technology and, finally, one last study developed a pilot survey to evolve in the evaluation of rare disease data registry technologies.

Although the studies are focused on recording and analyzing data, each one has a different methodological design. Regarding the design of the studies, three studies dealt with technological research, two reported on software development, one study was developed using data analysis, one article was a technology assessment, one research was developed using a retrospective cross-sectional design. This latest study, in particular, carried out in Ireland, showed that codification and sources of information on rare diseases are lacking in general practice.

The results showed the identification of computer tools for managing information on patients with rare diseases, essentially centered on the use of software, systems, and portals. The main aims of these tools are to standardize language, identify genes and assist in the diagnosis and treatment of these pathologies. The results also showed that most of the tools are recent, some of which are in the development and implementation phase, for later expansion and large-scale use.

Management is linked to the systematization of data on rare diseases, including clinical (signs and symptoms, diagnosis, treatment), epidemiological, and occurrence information.

Based on the formation of this rare disease database, it was identified that the purpose of the technologies is associated with the need to assist in clinical decision-making for professionals in the field and in identifying the experience of other cases, in addition to the dissemination of relevant and concise information to other interested parties.

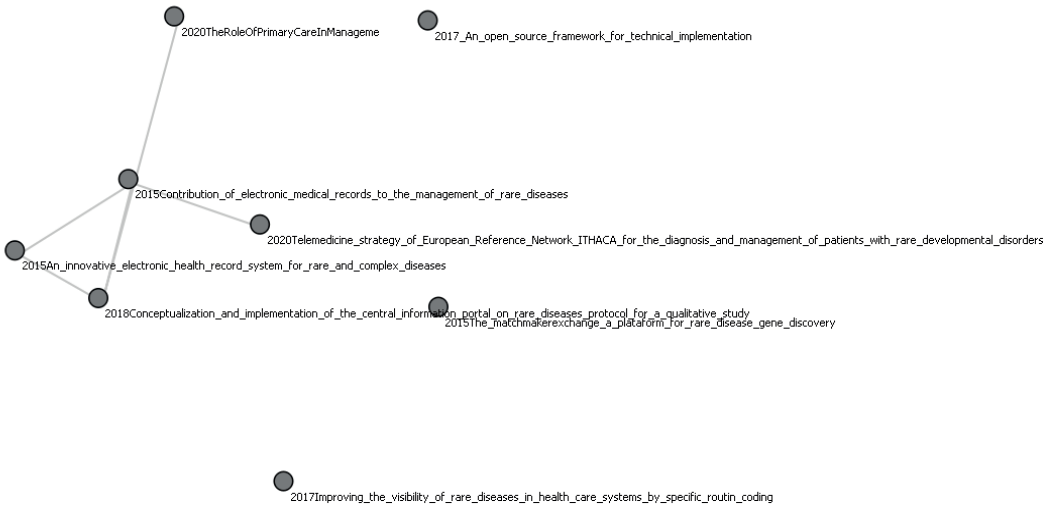
The result of the frequency of words, shown in Figure 2, indicates that the main discussions on the topic are directed towards computer tools, which refer to data and information systems. The words *disease* and *rare* are also frequent. Words related to patient care, such as medication, management, and diagnosis, are also frequent.

Figure 2 – Frequency of the most cited words in the articles

Source: prepared by the authors.

Figure 3 shows the multidimensional scaling for projecting points at certain distances. The technique seeks to adjust the distance between the articles (points), according to the relationship between each of them.

Figure 3 – Multidimensional scaling to adjust the distance between articles



Source: prepared by the authors.

Discussion

Based on the results obtained, Germany's leading role in the development of articles on management software for rare diseases was identified, making up half of the study sample for this research. Although there are already specific databases for rare diseases, such as the Information System on Rare Diseases (*Sistema de Informações sobre Doenças Raras*, SIDORA), which was developed by the Paraná State Health Department (*Secretaria de Estado da Saúde*, SESA), in partnership with the Paraná Information and Communication Technology Company (Celepar)[16], there is less research in this area in Brazil[8].

The results showed that most of the articles on the subject were published in the last five years. This may be due to the growing use of ICTs in the health area in recent years, since previously the scientific and technological apparatus of today was not available[17].

The management of rare diseases is still a challenge, directly related to the heterogeneity of the natural course of the disease and the limited number of individuals who can be selected to participate in research. Patients are still subjected to a long journey through health systems, passing through many health professionals and with little concrete scientific evidence[3,18].

In the field of technologies for rare diseases, economic investments for this specific population are not attractive to private investment, because they increase the risk of the research and development process and also because there is no high demand for this technology[2].

For this reason, some developed countries, such as the United States, which most often registers technologies for rare diseases, have specific policies for this purpose. The country encourages the development of technologies for rare diseases with strategies such as tax exemption, market exclusivity, credit, and a differentiated registration process. However, it is worth noting that these incentives are designed for a market health system. Other initiatives can be seen in Europe with the implementation of national plans for rare diseases(2,18) and incentives to create specific databases for this demand, such as Orphanet, a portal for information on rare

diseases and medicines, available in several languages, which presents Orpha Numbers for rare diseases that do not have an International Classification of Diseases number (ICD-10)[19].

The restricted sample size, associated with differences in severity, progression, presentation, exposure to previous treatment, and geographical dispersion, limit research in this context. The literature points to the following as the main challenges for the management of rare diseases: the limitation and geographical spread of specialists; the geographical spread of patients; limited knowledge; limited access to reliable sources of study; and the lack of robustness of research[3-4].

In this context, an integrated information system for rare diseases has emerged aiming to minimize these challenges arising from restricted access to information. However, this system must take into account standards, models, methods (taking into account collection processes), data storage, processing, retrieval and sharing. The system must be able to provide an overview of available data, with a pre-defined structure for information flow and sharing, to organize and coordinate direct healthcare through a secure network platform[4,14].

Shared access to Electronic Health Records (EHR) can improve communication and patient care. Through EHR, it is possible to identify which healthcare providers are actively managing the patient, which results in care being coordinated from one service to the next[15].

Therefore, this tool can break down the barrier of geographical distance by facilitating health surveillance of distant populations. In addition, other benefits resulting from its use are pointed out, such as health planning, creating health policies, evaluating the incorporation of new technologies, helping professionals with decision-making, managing rare diseases, and sharing information and experiences[4,17].

Through virtual interventions, it is possible to expand access to healthcare, include access to specialists and reduce waiting times. Based on these points, the results will be a reduction in costs, greater effectiveness, better interaction with the health team, accurate diagnoses, personalized care, and the qualification of health care, in order to make the principles described by the SUS a reality[17].

This study has some limitations, as the sample did not include articles unavailable in full, had access restrictions, or were paid for. Thus, this exclusion criterion may have contributed to a limitation in terms of sample size.

With the advance of medical technology, there has also been a significant increase in the diagnosis of individuals with rare diseases. Thus, by mapping which rare disease management tools are used globally, this study contributes to enhancing their use, connecting data, and enabling future improvements.

Final Considerations

The computer tools described for managing information on patients with rare diseases are recent and in the development and implementation phase. However, they are intended to contribute to three main areas: management, clinical and epidemiological.

In the management sphere, they favor health planning, the development of new policies, the evaluation of the incorporation of new technologies, and the reduction of rare disease costs. In the clinical scenario, they help with early and accurate diagnosis, professional decision-making, and access to specialists. Finally, in the epidemiological field, they contribute to monitoring diseases, and identifying the prevalence and incidence of diseases.

Therefore, an integrated system for the management of rare diseases has the potential to transcend the difficulties described for this knowledge area. Its functionalities and objectives point to new perspectives for care, and also for qualification in health. These findings corroborate the latent need for more technological studies for better development and evaluation of software that works with data recording and analysis.

Collaborations:

1 – conception and planning of the project: Greici Capellari Fabrizzio;

2 – analysis and interpretation of data: Greici Capellari Fabrizzio, Fred Jorge Oliveira Borges Junior e José Luís Guedes dos Santos;

3 – writing and/or critical review: Greici Capellari Fabrizzio, Lincoln Moura de Oliveira, Fred Jorge Oliveira Borges Junior, Roberta Eduarda Grolli, Gabriela Marcellino de Melo Lanzoni e Alacoque Lorenzini Erdmann;

4 – approval of the final version: Lincoln Moura de Oliveira, Roberta Eduarda Grolli, Gabriela Marcellino de Melo Lanzoni, Alacoque Lorenzini Erdmann e José Luís Guedes dos Santos.

Competing interests

There are no competing interests.

Acknowledgements

To the Coordination for the Improvement of Higher Education Personnel, for the master's degree scholarship.

To the National Council for Scientific and Technological Development, for research productivity scholarship.

References

1. Brasil. Ministério da Saúde. Portaria N° 199, de 30 de janeiro de 2014. Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras [Internet]. Brasília (DF); 2014 [cited Jan 30]. Available from: https://www.gov.br/saude/pt-br/composicao/saes/doencas-raras/legislacao#:~:text=Portaria%20n%C2%BA%20199%2C%20de%2030,institui%20incentivos%20financeiros%20de%20custeio
2. Silva EM, Sousa TRV. Avaliação econômica no âmbito das doenças raras: isto é possível? Cad Saúde Pública. 2015;31(3):496-506. https://doi.org/10.1590/0102-311X00213813
3. Whicher D, Philbin S, Aronson N. Na overview of the impacto of rare disease characteristics on research methodology. Orphanet J Rare Dis. 2018;13(1):14. DOI: https://doi.org/10.1186/s13023-017-0755-5
4. Derayeh S, Kazemi A, Rabiei R, Hosseini A, Moghaddasi H. National information system for rare diseases with a approach to data architecture: A systematic review. Intractable Rare Dis Res. 2018;7(3):156-63. DOI: 10.5582/irdr.2018.01065
5. Riegel BA, Schmitz J. ITINERÁRIO TERAPÊUTICO NA DOENÇA RARA E A IMPORTÂNCIA DA ENFERMAGEM NESSE PROCESSO. Enferm Foco. 2022;13:e-20228. DOI: https://dx.doi.org/10.21675/2357-707X.2022.v13.e-20228
6. Goncalves TLP, Tadeus VR, Campello TNC, Xavier JN, Oliveira NR, Rabito LBF, et al. Doença neurodegenerativa rara: sistematização da assistência de enfermagem em pacientes acometidos pela doença de Huntington. R Saúde Públ Paraná. 2022;5(2):1-17. DOI: https://doi.org/10.32811/25954482-2022v5n2.613
7. Mendes KDS, Silveira RCCP, Galvão CM. Revisão integrativa: método de pesquisa para a incorporação de evidências na saúde e na enfermagem. Texto contexto - enferm. 2008;17(4):758-64. DOI: https://doi.org/10.1590/S0104-07072008000400018
8. Faria-Campos AC, Hanke LA, Batista PHS, Garcia V, Campos SVA. An innovative electronic health records system for rare and complex diseases. BMC Bioinformatics. 2015;16(Suppl 19):S4. DOI: https://doi.org/10.1186%2F1471-2105-16-S19-S4
9. Storf H, Schaaf J, Kadioglu D, Göbel J, Wagner TOF, Ückert F. Registries for rare diseases: OSSE – An open source framework for technical implementation. Bundesgesundheitsblatt Gesundheitsforschung Gesundheitsschutz. 2017;60(5):523-31. DOI: 10.1007/s00103-017-2536-7
10. Marx MM, Dulas FM, Schumacher KM. Improving the visibility of rare diseases in health care systems by specific routine coding. Bundesgesundheitsblatt Gesundheitsforschung Gesundheitsschutz. 2017;60(5):532-6. DOI: 10.1007/s00103-017-2534-9
11. Philippakis AA, Azzariti DR, Beltran S, Brookes AJ, Brownstein CA, Brudno M, et al. The matchmaker exchange: a platform for rare disease gene Discovery. Hum Mutat. 2015;36(10):915-21. DOI: 10.1002/humu.22858

12. Litzkendorf S, Hartz T, Göbel J, Storf H, Pauer F, Babac A, et al. Conceptualization and Implementation of the Central Information Portal on Rare Diseases: Protocol for a Qualitative Study. JMIR Res Protoc. 2018;7(5):e112. DOI: 10.2196/resprot.7425

13. Bremond-Gignac D, Lewandowski E, Copin H. Contribution of electronic medical records to the management of rare diseases. Biomed Res Int. 2015;2015:954283. DOI: 10.1155/2015/954283

14. Smith M, Alexander E, Marcinkute R, Dan D, Rawson M, Banka S, et al. Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet J Rare Dis. 2020;15(1):103. https://doi.org/10.1186/s13023-020-1349-1

15. Byrne N, Turner J, Marron R, Lambert DM, Murphy DN, O'Sullivan G, et al. The role of primary care in management of rare diseases in Ireland. Ir J Med Sci. 2020;189(3):771-6. DOI: 10.1007/s11845-019-02168-4

16. Paraná. Sidora: Paraná lança sistema para mapear e apoiar pessoas sobre síndromes e doenças raras [Internet]. Curitiba: Agência Estadual de Notícias; 2023 [cited 2024 May 21]. Available from: https://www.aen.pr.gov.br/Audio/Sidora-Parana-lanca-sistema-para-mapear-e-apoiar-pessoas-sobre-sindromes-e-doencas-raras

17. Babac A, von Friedrichs V, Litzkendorf S, Zeidler J, Damm k, Graf von der Schulenburd JM. Integrating patient perspectives in medical decision making: a qualitative interview study examining potentials within the rare disease information Exchange process in practice. BMC Med Inform Decis Mak. 2019;19(1):188. DOI: 10.1186/s12911-019-0911-z

18. Souza ÍP, Androlage JS, Bellato R, Barsaglini RA. Doenças genéticas raras com abordagem qualitativa: revisão integrativa da literatura nacional e internacional. Ciênc saúde coletiva. 2019;24(10):3683-700. DOI: https://doi.org/10.1590/1413-812320182410.17822019

19. Ministério da Saúde. Departamento de Gestão e Incorporação de Tecnologias em Saúde da Secretaria de Ciência, Tecnologia e Insumos Estratégicos. Procedimentos laboratoriais para diagnóstico de doenças rarasassociadas a anomalias congênitas na tabela SUS [Internet]. Brasília (DF); 2014. Available from: https://www.gov.br/conitec/pt-br/midias/incorporados/doencasraras-eixosi-ii-iii-final.pdf

Received: September 24, 2023

Approved: June 29, 2024

Published: November 12, 2024