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A SYSTEMATIC ANALYSIS OF ROLE OF ARTIFICIAL INTELLIGENCE IN MEDICAL IMAGING: IMPACTS AND THREATS ON RARE DISEASE COMMUNITY

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ABSTRACT— The current study aims to systematically review the role of artificial intelligence in medical imaging as well as impacts and threats on rare disease community as well as aims to present a concise opinion on current research in this area and forward proposal for further studies. Based on a comprehensive assessment of the literature, this study gathers, synthesises, and evaluates data. Thirty-five research articles on the community and its dimensions related to rare diseases, medical imaging, and artificial intelligence were selected from seven databases between 2014 and 2024, in accordance with a prearranged review methodology. The chosen works have been assessed in order to compile pertinent information for a number of academic topics. We have observed expanding tendencies in the articles in 2022. The majority of papers have looked at the community, medical, and artificial intelligence aspects of people with uncommon community diseases. The information extraction process yielded the following results: "artificial intelligence," "threats," "medical imaging," "impacts," and uncommon community diseases" are the terms that appear most frequently in the literature individually. "Combining all these data variables" is still a step, but it is less iterative. As a result, the review's findings can aid in the effective implementation and development of artificial intelligence in medical imaging by academics.

KEYWORDS: artificial intelligence, medical imaging, impacts, threats, rare disease community

1. Introduction

Using scientific knowledge and the greatest resources available, medicine is the art of providing compassionate care [1]. A doctor views every patient as an individual human being with a distinct past, present, and future. Every patient is an individual human being; none are "rare patients" or "common patients." However, it is necessary to classify the constellation of clinical observations into certain diseases or syndromes (phenotyping) due to modern nosologic taxonomy [2- 4]. Here, we put several people into a single category. This is a useful strategy because it makes it possible to identify and apply appropriate

treatments by comprehending the molecular underpinnings of these phenotypic categories. Because of their distinct molecular aetiology, the majority of pharmaceutical treatments are meant to be used with a "group of subjects with a common denominator." [5]. Clinical medicine involves a cognitive job that involves meticulous observation and phenotyping of patients based on a comprehensive assessment of their physical, biochemical, radiological, histological, and genetic results. The right diagnosis is the outcome of this procedure. Moving between the "unique human-being" and "grouped disease categories"—that is, being both separate and related at the same time—is the fundamental source of innate cognitive dissonance. If this paradoxical condition is left unchecked, it might lead to a variety of "diagnostic cognitive biases" [6]. Every patient should be seen as distinct and separate, but also as connected to and included in the right categories as feasible. Designating a set of illnesses as "rare diseases" is an effort to make them easier to "include" into the right category when necessary (make the connections easier). An effort to promote "individualized medicine" and acknowledge individual distinctions is to facilitate uniqueness, or individuation. Even though they appear to be at odds, these two endeavours are really only aspects of one reality. Here, we'll talk about how the development of artificial intelligence (AI) in medicine may impact the "inclusion" issue. Our goal is to demonstrate how rare diseases (RDs) and AI technologies interact in a way that has the potential to have both terrible outcomes and enormous advancements. The future is hard to foretell, but it may be created through active participation. The clinical efficacy and integration of AI medical devices into standard clinical practice can be severely impeded by subpar performance of AI technologies in patients with rare diseases [7- 10]. Less is distinct. Drug development on an even smaller scale is not the same as orphan drug development. The market's limitations are not the only difficulties [11- 13]. Less is different in this case. Translation of preclinical targets to clinical phase research requires particular effort (precise translation) because of the small number of affected patients eligible for clinical trials [12]. Since fewer than 9% of investigational medications enrolled in phase 1 clinical trials receive FDA approval, artificial intelligence (AI) generally helps drug development by facilitating more successful target selection [13], [14]. This is a step in the right direction for CDs, but it's crucial for RDs as well. Population-based data may not be sufficient for physiologically based pharmacokinetic modelling, and the small pool of candidates increases the risk of inaccuracies in pharmacokinetic estimations because of sparser and smaller sample sizes. By using TB-PET dynamic imaging, AI-based PET kinetic modelings can yield useful information about the pharmacokinetics of investigational pharmaceuticals with a small number of participants (Figure 1) [15]. The comprehensiveness of this data, together with AI-enhanced pharmacochemistry, offers a platform for precise structural optimisation of the chosen target [16- 20].

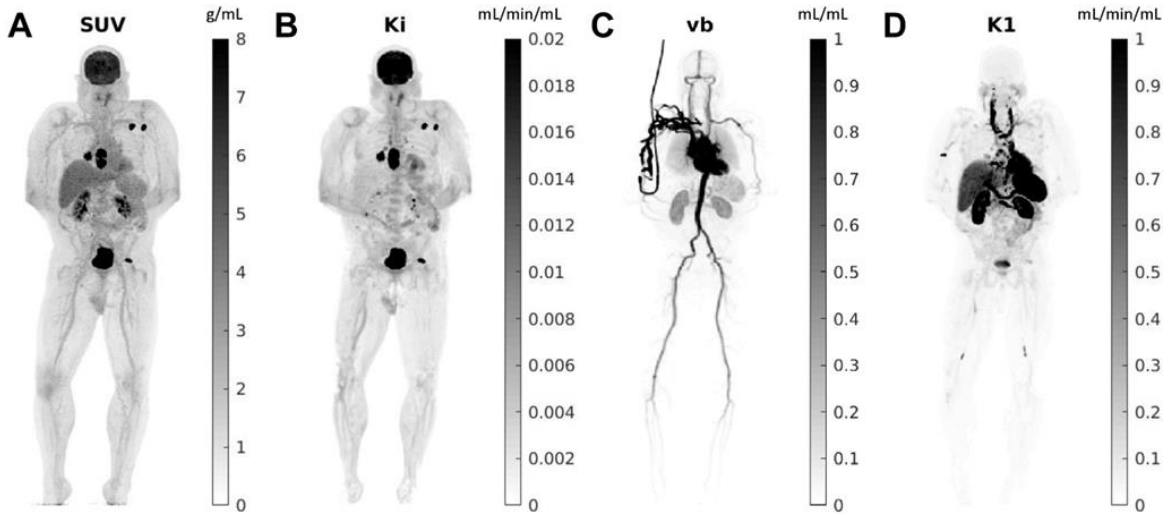

Figure 1: Estimated total-body parametric pictures from a 60-minute dynamic 18F-FDG scan on the uEXPLORER of a patient with metastatic cancer: SUV, FDG net influx rate Ki, fractional blood volume

(vb), FDG delivery rate K1, and C

In summary, advanced computational techniques in systems biology and multi-scale modeling in addition to the novel technology of dynamic TB-PET imaging and AI-empowered complex systems modeling can aid in the creation of virtual replicates of patients, also known as digital twins [21- 25]. Therefore, it is ethically required to address opportunities and issues linked to the diagnosis of RD with AI-based molecular imaging in order to properly integrate AI into clinical practice. Medical AI may have two negative effects on patients with RDs in terms of society, economy, and health: (1) marginalizing and excluding people with RDs (discrimination by omission); and (2) overt or covert unregulated post-deployment discrimination or malevolent uses of AI technologies (discrimination by commission). The purpose of this work is to thoroughly examine the medical, ethical, and social implications of implementing RDs-aware AI in the healthcare system.

2. RESEARCH METHODLOGY

2.1 Review Approach

The systematic literature review is a frequently employed method for integrating recently established research fields [26–30]. One of the five types of systematic literature reviews [31] that [32] identified in this instance is the domain-based review strategy, which is used in this study to examine the function of artificial intelligence in medical imaging as well as the implications and risks for the community of rare diseases. The choice of the methodology that the systematic literature review should use is just as important as the choice of the organising framework. To investigate the function of artificial intelligence in medical imaging as well as the implications and risks for the community of rare diseases, a systematic literature review (SLR) was carried out. These phases are described in the headings below.

2.2 Research Questions

The current study examines the function of artificial intelligence in medical imaging, as well as the implications and risks for the community affected by uncommon diseases. The findings are expected to have an impact on future research and development. To this end, a number of research questions have been proposed and explored. This SLR attempted to determine the extent of research being conducted in this field by responding to the study's question: To what extent is the science of artificial intelligence being applied to medical imaging, and what are the implications and risks for the community of rare diseases? Scholars have used a range of investigative methods to investigate the question. In this study, articles were analyzed with respect to their individual research approaches using the following research question: Which approaches are used in these specific studies?

2.3 SLR Protocol

Recent years have seen an increase in the number of systematic literature reviews published as well as an increase in the number of articles discussing the systematic literature review methodology. Every technique step is numbered and categorized in a similar manner in the many instructions that are now available. In order to resolve this methodological issue, [33] created a Systematic Review of Literature, a thorough strategy that incorporates continuous education into a demanding procedure [34], [35].

2.4 Search process

The aim of the two-phase search process coordinate is to locate data using the selection criteria for study. Phase 1: Gather study data and investigate the seven important websites. Phase 2: In order to identify which publications are especially relevant, go over the list of references for each inquiry on the role of artificial

intelligence in medical imaging as well as impacts and threats on the community of rare diseases that has been selected. This investigation's computerised search approach made use of intellectual e-databases. The following methods were used to conduct the keyword searches:

(a) We removed significant terms from the research questions.

(b) As was already established, we are able to recognise synonyms and other forms of the words.

(c) To build the search strings, we use the Boolean operators "and" and "or".

These phases have resulted in the role of artificial intelligence in medical imaging as well as impacts and dangers on the community of rare diseases. The main resource for conducting methodical keyword searches was the Web of Science (WOS). Scopus, which is also commonly utilised in reviews that are centred around models, was utilised as an additional resource for cross-referencing. We selected seven important electronic databases—Science Direct, Taylor & Francis, Wiley, Elsevier, Emerald, Routledge, and Springer—in order to compile the pertinent literature. The aforementioned databases were chosen due to their perceived applicability, ability to offer the best conference papers and influential publications about the application of artificial intelligence in medical imaging, as well as their potential effects and dangers to the rare disease community. The research's source publications might be found in the electronic databases indicated in Table 1.

TABLE 1 Resources for Systematic Literature Reviews

Electronic Databases	URL
Springer	http://link.springer.com/
Science Direct	http://www.sciencedirect.com/
Wiley	https://onlinelibrary.wiley.com
Taylor and Francis	https://www.tandfonline.com/
Elsevier	https://www.elsevier.com/
Emerald	https://www.emerald.com/insight/
Routledge	https://www.routledge.com/

2.5 Study selection

The steps that follow the exploration process, which produced pertinent publications, make up the article selection process. (A) Selecting the study based on its title and subject. (b) Documents having limited or no access are removed. (b) Duplicate or academically pointless articles are eliminated. (d) Once the investigation's guidelines have been followed for outcomes examination, it is crucial to identify a small number of noteworthy outcome data that might firmly support the review worries. The following criteria for inclusion and exclusion are defined in the research paper. Read the abstracts, introductions, study methods, conclusions, and findings of the papers. The appropriate articles were selected based on the quality evaluation standards covered in the next section, as well as the exclusion and inclusion criteria.

Inclusion criteria:

1. Only papers that discuss role of artificial intelligence in medical imaging as well as impacts and threats on rare disease community.
2. Studies with a proposed framework and a clear research technique.
3. The journal version was chosen for the research, including a conference version.
4. In identical papers, the most recent one was chosen. The papers that used the review of literature as a foundation for addressing related literature and a starting point for expanding this review were considered.

Exclusion criteria:

1. Research projects lacking a conceptual or methodological foundation.
2. Publications not addressing role of artificial intelligence in medical imaging as well as impacts and

threats on rare disease community.
3. Papers from predatory journals or databases.
4. Papers in languages other than English.

2.6 Quality assessment

Each study paper was examined more thoroughly using the following quality assessment questions: 1. Is the field or category accurately described? 2. Is the article supported by research, or is it just an expert-based synopsis of learned insights? 3. Are the goals of the research clearly stated? 4. Does the study's setting provide a sufficient description? 5. Does the research design effectively achieve the goals of the study? 6. Is the concluding message succinct and clear? Does the research address all the facets of artificial intelligence's function in medical imaging, including its effects and potential risks to the community of rare diseases? 8. Does the research provide an overview of the topic? 9. Does the study utilise a well-established design to support its analysis? N (No) = 0 or ambiguous (i.e., the data wasn't made explicit), P (Partially) equals 0.5, and Y (Yes) equals 1. This is how the rating system for the article was created. Although the question scoring helped to analyses the literature, it also made it easier to determine the importance of a certain study when taking the findings into account. Furthermore, scoring provided more precise inclusion and exclusion standards.

2.7 PRISMA Flow Chart of Studies

Out of a total of slightly more than 82 publications, one hundred papers were selected for the initial search or identification procedure. To undertake this kind of task, a systematic review of all the study database lists is conducted. Subject-matter analysts were categorized as contributors and researchers. The 65 articles have undergone a full-text assessment in order to extract information from correspondence during the eligibility process. Twelve more studies were left out of this review because the recommendations for actual research data were too vague or generic. 53 papers were identified as appropriate as a result of this procedure. Finally, additional checks were made on the final collection of articles to exclude any relevant modification data discovered in the design guidelines during the information withdrawal process (as indicated by the term "Contained" in the PRISMA chart). After screening the titles and abstracts, 35 possibly valuable studies were left after excluding studies conducted outside of the country. The process we used in our study to identify relevant publications in PRISMA diagrams is depicted in Figure 2.

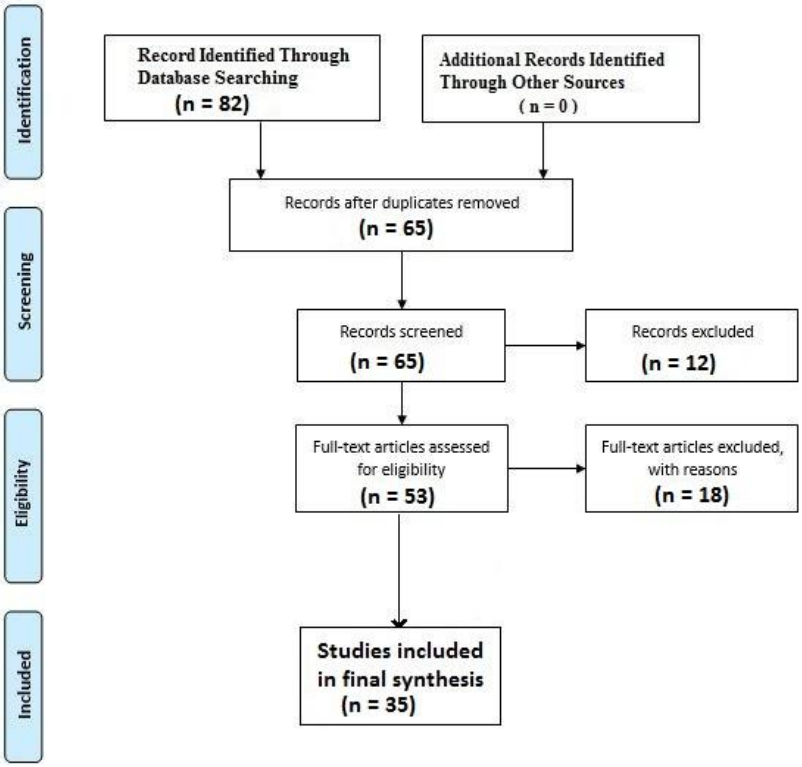

Figure 2: PRISMA Flow Diagram Shown Study Selection

3. Review Analysis and Results

We provide a summary of the SLR's output in the following paragraphs. There is a list of the chosen articles with some information. Next, we present the findings of the quality assessment of the chosen articles.

3.1 Search Results

Parameters from each selected publication were to be extracted during the data extraction stage. The basic information about each publication, the research, the approach, and the objective are included in the extracted data. Five important electronic databases have been identified. For this evaluation, the search process produced 35 publications. The number of research we select from electronic databases is shown in Figure 3. Just one paper was chosen from the Taylor & Francis databases, while most papers (n=10) were taken from the Wiley database. Additionally, Figure 4 shows the chronological order of studies over time. According to the graph, most papers were released between 2014 and 2024, and from 2022 there has been an indication of increased focus in the field. We used nine quality assessment questions to evaluate the overall quality of the included research. Each question can receive a maximum score of 1, with an overall score of 9. Depending on how well it performed, each paper was given points for the questions listed in the methodology section as high, medium, or low. A paper is given a score of 1 if it fully satisfies the requirements, a score of 0.5 if it fulfills some of the requirements, and a score of 0 if it does not fully satisfies the requirements. The total value of a study is rated as high, medium, or low depending on whether it is greater than or equal to 10, 5, or less than 5. Figure 5 illustrates the high worth of the selected research because their quality score is above 5. Figure 6 presents a summary of the findings from this study, which examined the research methodologies used in a few investigations.

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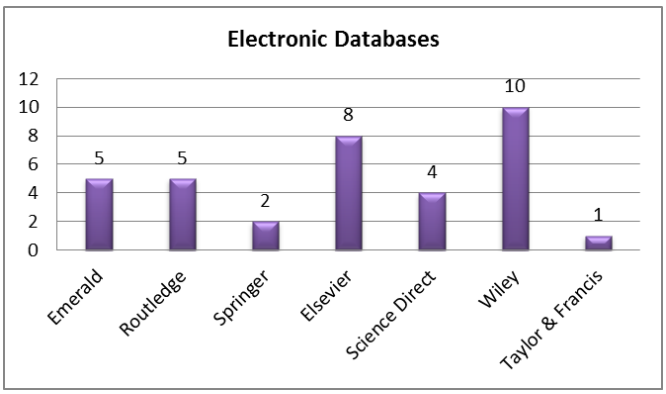

Figure 3: Papers in five selected electronic databases

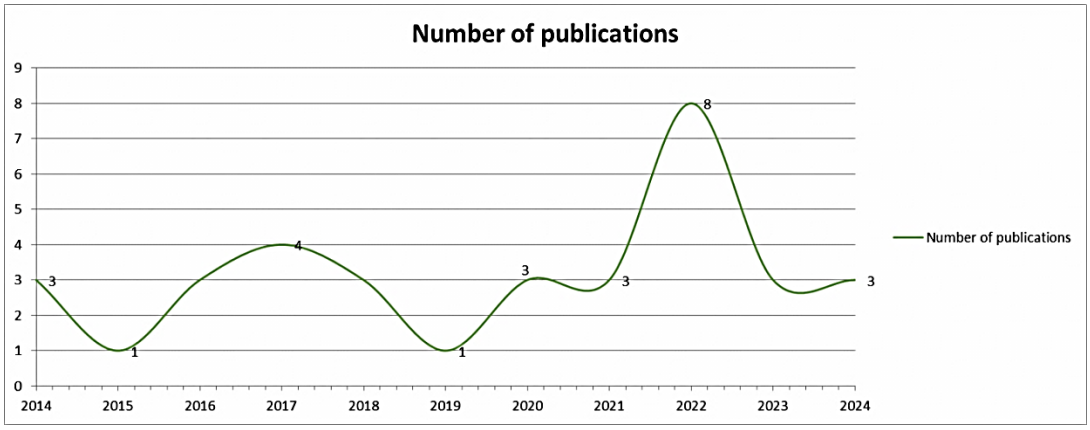

Figure 4: Chronological order of studies over time

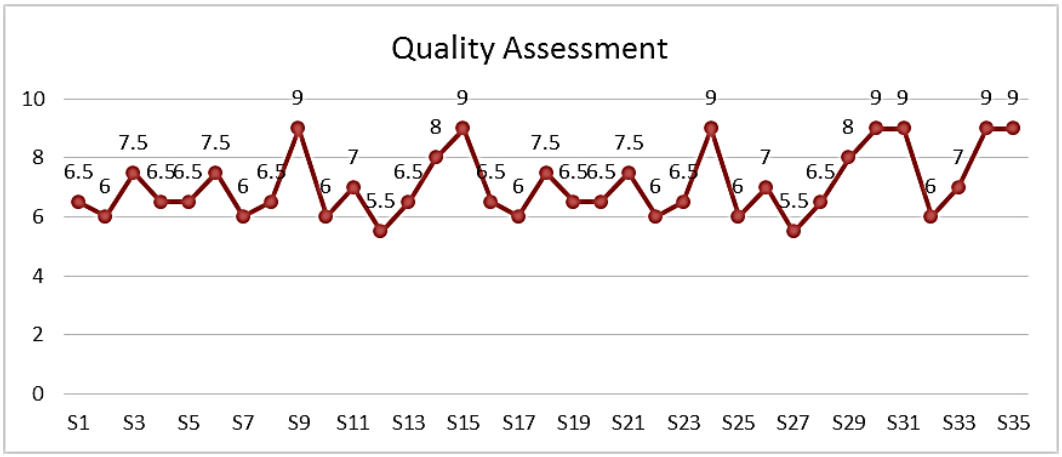

Figure 5: Quality assessment of all included studies

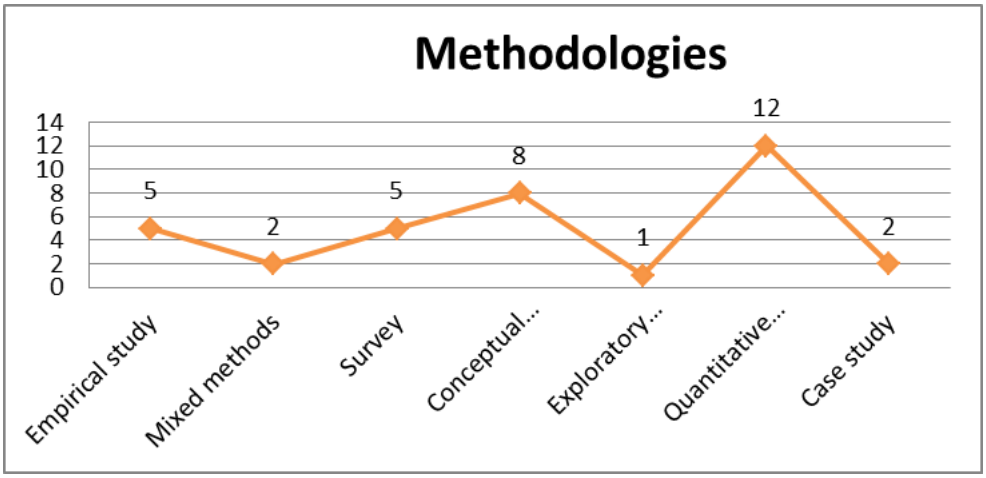

Figure 6: Methodologies of selected studies

4. DISCUSSION AND CONCLUSION

For this review, relevant works that highlight the intersection of artificial intelligence and medical imaging were selected. Seven databases were examined. In addition, the articles were selected by an examination of the abstracts, the application of inclusion and exclusion criteria, and an analysis of the results based on the keywords and title. According to our estimate, there will be a noticeable increase in stories regarding studies beginning in 2022. Thus, new trends in the field illustrated study about the role of artificial intelligence in medical imaging as well as impacts and dangers on the community of rare diseases. Less emphasis has been paid to research that combine artificial intelligence with medical imaging to examine the effects and hazards rare diseases pose to the population. Thus, a comprehensive SLR was conducted. 35 articles covering the period from 2014 to 2024 were selected. Following examination, only the papers' discussion of artificial intelligence's function in medical imaging and their implications and risks to the community of rare diseases were retained for further study in seventeen different countries. Scholars in the discipline must focus more on these industries and other scenarios that aren't covered. Most of the selected papers had no background at all. As such, we consider them to be the entire context. Scholars ought to consider these more, for this reason. It is therefore recommended that researchers take emerging countries into account. Next, the selected articles' research methodologies were scrutinised. The majority of the publications used the survey strategy, whereas other approaches were used less frequently. A major component of such goals is one of its essential elements. Through these contributions and our study of the subject, we want to advance the state of knowledge in this emerging topic by providing a future research agenda based on a thorough examination of the last ten years' worth of literature. We raise awareness within the research audience about the growing potential for further study, as well as the effects and dangers on the community of rare diseases literature and the role of artificial intelligence in medical imaging. Even though research in this area is still in its early stages, more thorough conceptual and empirical work is desperately needed to keep up with the quick pace of technological advancement and the evolving field of medical imaging in artificial intelligence. The majority of papers have looked at the community, medical, and artificial intelligence aspects of people with uncommon community diseases. The information extraction process yielded the following results: "artificial intelligence," "threats," "medical imaging," "impacts," and uncommon community diseases" are the terms that appear most frequently in the literature individually. "Combining all these data variables" is still a step, but it is less iterative. As a result, the results of this review can help scholars apply and improve artificial intelligence in medical imaging more successfully.

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