DOI 10.18.276/978-83-8419-053-1-23

28th European Conference on Artificial Intelligence (ECAI 2025)

AI-POWERED KNOWLEDGE MANAGEMENT AND DECISION SUPPORT FOR IVF WITH PGT: LITERATURE REVIEW AND MODEL PROPOSAL

Małgorzata Skweres-Kuchta[a], Agnieszka Skweres[b], Elżbieta Szaruga[c], Krzysztof Łukaszuk[d]

[a] *University of Szczecin, Institute of Management, Szczecin Poland*
[b] *Pomeranian University in Słupsk, Institute of Health Sciences, Słupsk, Poland*
[c] *University of Szczecin, Institute of Management, Szczecin, Poland*
[d] *Medical University of Gdańsk, Institute of Nursing and Midwifery, Gdańsk, Poland*

ABSTRACT

Purpose: *This study aims to apply machine learning as an AI-based tool for comprehensive literature review and to propose an Integrated Clinical Management Model (ICMM) that leverages AI-driven decision support, data integration, and change management to optimize decision-making in reproductive medicine. The medical context is exemplified by Preimplantation Genetic Testing-In Vitro Fertilization (PGT-IVF) for rare monogenic diseases, focusing on improving clinical outcomes and patient-centered care through personalized and transparent support systems.*

Need for the study: *As PGT-IVF procedures become more common in reproductive medicine, there is a need to better assess their effectiveness in monogenic diseases. Machine learning can enhance literature analysis and support decision-making, leading to more informed choices for both doctors and patients.*

Methodology: *Literature review based on the Machine Learning Perspective.*

Findings: *Machine learning has enabled an in-depth analysis of available knowledge and the identification of research gaps in the context of implementing new AI-driven solutions in reproductive medicine using PGT-IVF: The scientific evidence is based mainly on case studies with a lack of comprehensive population-level data (1); PGT-IVF in couples at risk of genetic disease is justified, but the procedure does not guarantee a healthy birth (2); Effectiveness analysis boils down to demonstrating a birth to a healthy child (3); The possibilities of implementing the solution differ between countries, Poland is an example (4).*

Practical Implications: *The effective implementation of the proposed AI-powered ICMM requires comprehensive access to clinical, genetic, financial, and psychosocial data across all stages of the PGT-IVF process. This integrated approach supports complex, multi-dimensional decision-making, enabling personalized treatment pathways that balance medical effectiveness, cost-efficiency, and emotional well-being for couples undergoing treatment.*

Keywords: machine learning, decision support system, predictive analytics, preimplantation genetic testing (PGT), in vitro fertilization (IVF), knowledge management, personalized medicine

Jel codes: I10, I18, C63, C80, D81, D83

1. INTRODUCTION

The use of AI-based methods enhances the understanding of the dynamics of scientific discourse across many fields – here, focusing on rare diseases – by revealing patterns of knowledge and the evolution of terminology. Integrating these advanced computational techniques in research underscores the crucial role of machine learning in knowledge discovery. This study contributes to the broader discussion on artificial intelligence applications in text mining, reflecting on how computational models can optimize literature synthesis and research assessment in highly specialized and rapidly evolving domains. These methods enable identification of key research areas and conceptual frameworks. Additionally, hierarchical clustering (using Manhattan and Ward metrics) and word enrichment analysis facilitate classification of thematic clusters and detection of semantic relationships between concepts.

Our literature review highlights opportunities to apply AI tools for predictive analytics and decision support in reproductive medicine, particularly regarding in vitro fertilization combined with preimplantation genetic testing (PGT) for couples at risk of transmitting rare monogenic diseases.

Rare diseases, often chronic and genetically determined, affect approximately 6–8% of the global population – around 400 million people worldwide, including 27–36 million in the European Union (Nguengang Wakap et al., 2020; Pieklak et al., 2023). According to Orphanet, nearly 72% of these conditions have a genetic origin (Orphanet, 2024). In reproductive medicine, 2–4% of couples are at risk of passing autosomal recessive or X-linked genetic disorders to their offspring (Capalbo et al., 2021).

Modern reproductive medicine offers two main diagnostic strategies for such couples: prenatal diagnosis (PND) and preimplantation genetic testing (PGT). While PND is performed during pregnancy, PGT allows early selection of unaffected embryos prior to implantation, reducing emotional and legal complexities related to pregnancy termination (De Wert et al., 2014; Klein & Mahoney, 2007).

The aims of this study are:

- To apply machine learning in literature review and identify potential research gaps in the field of reproductive medicine, with a focus on Preimplantation Genetic Testing-In Vitro Fertilization (PGT-IVF) in the context of rare monogenic diseases.
- To apply machine learning for literature review and identify research gaps in reproductive medicine, focusing on PGT-IVF in the context of rare monogenic diseases.
- To evaluate the legitimacy and effectiveness of PGT-IVF for couples at risk of transmitting rare monogenic diseases, defining effectiveness as the ratio of benefits to all associated financial, non-financial, and psychological costs related to choosing this strategy to have a healthy child, thereby supporting informed decision-making.
- To explore the potential of intelligent knowledge management and artificial intelligence in supporting decision-making processes in PGT-IVF from both medical and patient perspectives.
- To present Poland as a case study illustrating challenges related to the implementation of PGT-IVF under specific legal conditions that vary internationally.

The literature review highlights a clear need for decision support systems integrating medical, genetic, economic, and psychosocial data to enable patients and clinicians to make informed, personalized, and ethical therapeutic decisions (Khalfallah et al., 2023; Haude et al., 2017).

2. LITERATURE REVIEW

Intelligent management in healthcare involves the deployment of artificial intelligence (AI) systems designed to assist clinicians in their decision-making processes. These systems are responsible for aggregating, storing, organizing, retrieving, and analyzing medical data to support informed clinical judgments. By streamlining data workflows, intelligent management enhances the efficiency of healthcare delivery for both practitioners (doctors) and service providers (Kuo, 2023, p. 3).

Central to intelligent management is the application of machine learning, which is becoming increasingly embedded in clinical environments. This integration bridges preclinical data analysis with real-time bedside diagnostics, enables patient stratification (e.g., clustering based on clinical profiles), and supports decisions related to treatment planning or early intervention. These capabilities are essential for both primary and secondary prevention and contribute to improved decision accuracy and treatment effectiveness (Belciug & Gorunescu, 2020, pp. 642, 659).

Such management systems often rely on intelligent decision support systems (i-DSS), a category of AI-driven tools that facilitate both administrative and clinical processes. In clinical contexts, these are referred to as Clinical Decision Support Systems (CDSS) – platforms that provide tailored recommendations and predictive insights based on individual patient data. These tools assist clinicians in diagnosing conditions, selecting interventions, and reducing the likelihood of errors. By leveraging extensive clinical and biological datasets, CDSS apply algorithmic models to correlate patient data with phenotypic patterns, ultimately enabling more precise and personalized care (Khalfallah et al., 2023, p. 28).

The broader notion of digital health management encompasses the integration of digital technologies – including mobile health apps, health information systems, the Internet of Things (IoT), and AI – into the organization and delivery of care. This approach seeks to enhance decision-making, foster patient and caregiver engagement, and improve overall health outcomes (Pyper, McKeown, Hartmann-Boyce, & Powell, 2023; Zhou et al., 2024; Zajac, Braun, Klein, & Kolbe, 2024; Oshni Alvandi & and Bain, 2023).

Knowledge-based management, by contrast, is an approach grounded in the systematic use of existing knowledge to guide decision-making, particularly in complex or uncertain clinical scenarios such as rare diseases. This method depends on solid infrastructural and technological foundations and should be a core element of digital health strategies, especially those aimed at supporting vulnerable populations (Montani & Bellazzi, 2002; Qiu, Gorantla, Rajan, & Tan, 2021; Oshni Alvandi et al., 2023; Zajac et al., 2024; Zhou et al., 2024).

Preimplantation genetic testing (PGT) is a multi-step, highly specialized procedure that enables the identification of genetic disorders in embryos before implantation. This paper focuses on PGT-M, which targets monogenic conditions (De Rycke & Berckmoes, 2020). The process is medically and emotionally demanding for patients, involving strict clinical protocols, significant financial costs, and complex decision-making. The emotional burden associated with repeated procedures and uncertain outcomes further highlights the need for decision-support tools and personalized guidance (Harper et al., 2012; Takeuchi, 2021; Haude et al., 2017).

3. METHODOLOGY

Initially, 275 publications on PGT and rare diseases were searched in the most popular databases: 174 items in Scopus, 26 items in Web of Sciences, 28 items in PubMed, and 47 items in EBSCO. A query string such as '(ALL (rare AND disease*) AND ALL (PGT))' was used (in different databases, the query syntax could have slightly different field tags, but the syntax expressed the same query). 46 duplicates were then eliminated after being assessed for topical relevance. Many important papers in other languages, those indexed in other databases, or those that weren't indexed were not included in the analysis. It was required to embrace these limitations to tokenize and alter the text, including lemmatization. Finally, 229 publications [Appendix A1] in English and German were selected for the analysis of text mining and machine learning.

The analysis used such techniques in the field of text mining and machine learning, i.e. word cloud (Gárate-Escamilla et al., 2022; Qu et al., 2022; Sun & Zhao, 2023), a bag of words (with L1) (Bagherzadeh et al., 2021; Castro et al., 2022), topic modeling by Latent Semantic Index (LSI) (Lossio-Ventura et al., 2021; Venugopal et al., 2023) and keyword extraction by TF-IDF scoring (Veisi et al., 2019; Yahya Saeed et al., 2021), hierarchical clustering (with Manhattan Metric and Ward Metrics) (Amine et al., 2010), word enrichment (Kuhr et al., 2020; Ning et al., 2018) and sentimental analysis (with SentiArt method) (Jacobs, 2019, 2022; Jacobs & Kinder, 2022).

The research process was divided into eight steps (Figure 1). The approach is multi-dimensional. The pre-processing of the text took place in the first stage. This stage included transformation (lowercase, remove URLs), tokenization (regexp) and normalization (LemmagenLematizer), and filtering (stopwords, lexicon, regexp). The second stage involved generating the word cloud. The corpus included: 1388 tokens and 20 types. Parallel to the second stage was the third stage. The third stage used the Bag of Words (term frequency - count, regularization – L1 (sum of elements)).

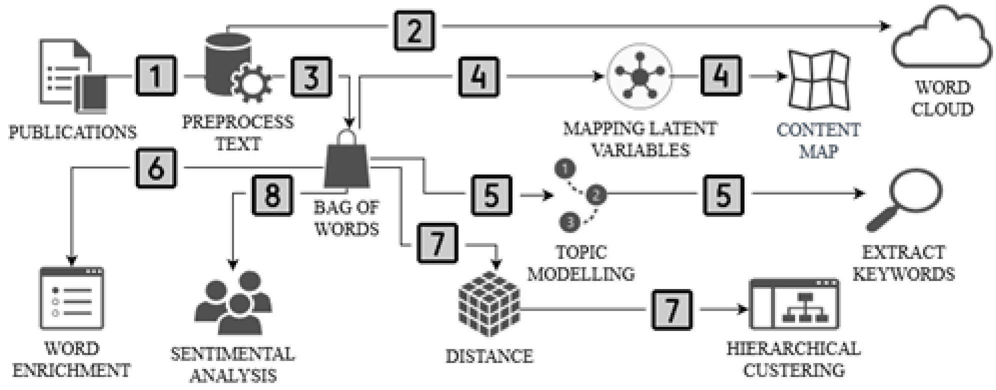


Figure 1. The framework of the authors' research procedure (methodology)
Source: own elaboration.

Stages 4 –8 continued the third stage and were interconnected. In step 4, mapping latent variables was conducted with the presentation of the publications' content map. Stage 5 involved topic modeling (Latent Dirichlet Allocation) and the extraction of 10 keywords.

In step 6, word enrichment analysis was employed to evaluate significance in the corpus's studied subset (i.e. it is not a random word, but a word closely related to the examined term). Later, hierarchical clustering was performed (using Ward linkages). Stage 7 uses the SentiArt method for sentimental analysis to recognize moods: happiness, surprise, disgust, anger, fear, and sadness.

Using the aforementioned methods, we can evaluate how the automated process aids in choosing the right publications for an extensive literature review. It also provides a suggestion as to which topics are worthwhile researching and which publications are more similar in terms of their themes. The approach is multi-dimensional. The results are shown in the next section.

The literature analysis revealed significant gaps in the systematic evaluation of effectiveness and comprehensive decision support in IVF with PGT procedures. In response to these challenges, we propose developing an AI-based decision support model that combines clinical information, treatment success predictions, financial aspects, and emotional and psychosocial factors that are integral to patient experience (Takeuchi, 2021; Capalbo et al., 2021).

4. RESULTS

Figure 2 shows word clouds based on the examined corpus, which included the title and keywords. It illustrates the most common terms around the studied issue. In the initial examination, it could be noticed that the most important words were: genetic (weight 541), PGT (weight 305), testing (weight 301), preimplantation (weight 217), disease (weight 183), gene (weight 181), clinical (weight 180), sequencing (weight 170), diagnosis (weight 164), patients (weight 164).

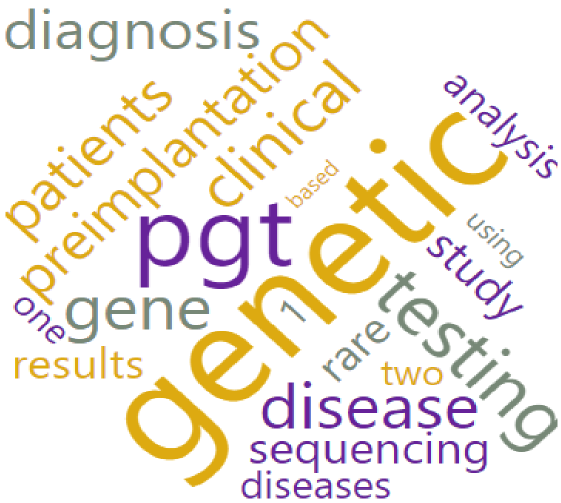


Figure 2. Word clouds of terms. Limitations: The authors did not take into account many valuable publications in the study, instead taking into account the need to narrow the thematic scope of the described issues

Source: own elaboration based on 229 references [A1].

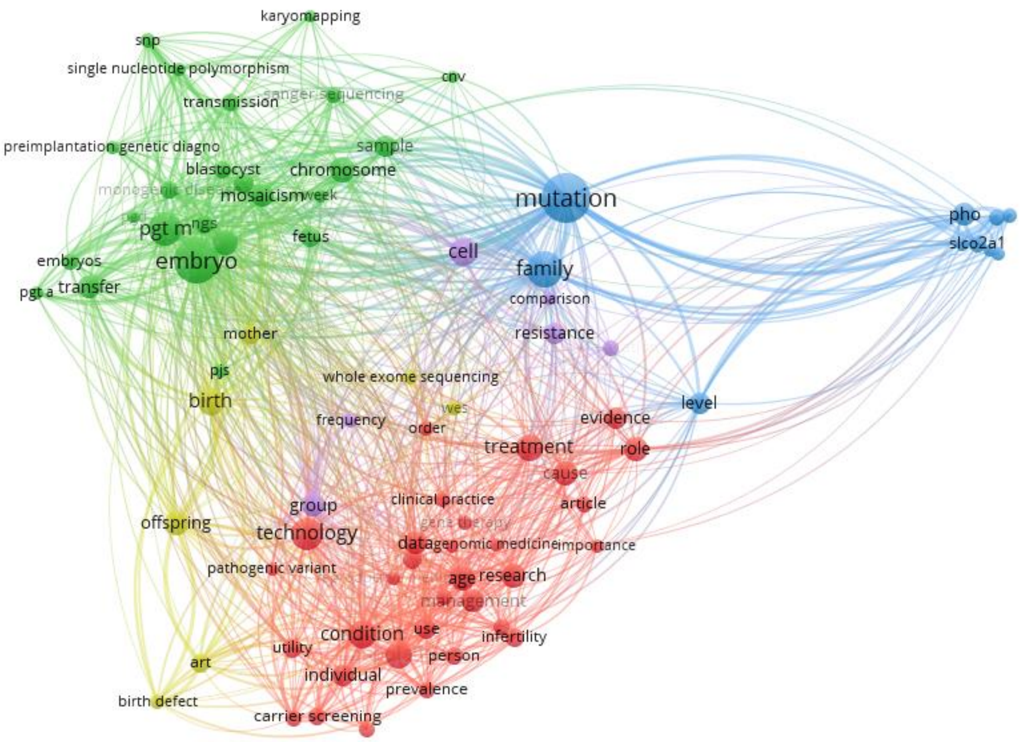


Figure 3. Mapping latent variables from titles and abstracts of the publications on issues connected with rare diseases and PGT. Note: Technical publication on mapping and clustering using VOSviewer (Ding & van Eck, 2014; Szaruga & Załoga, 2022). Limitations: The authors did not take into account many valuable publications in the study, instead taking into account the need to narrow the thematic scope of the described issues

Source: own elaboration based on 229 references [A1].

While exploring the studied publications in-depth, particular attention was paid to mapping latent variables in the network connections map (Figure 3 and Figure 4). The larger the ball, the more often the latent variable in the analyzed texts appeared. The shorter the bond line, the stronger the bond, and the longer, the weaker the interdependence of the latent variables.

Based on the titles and abstracts of the analyzed publications, 5,991 terms (latent variables) were found, 131 of which appeared 10 times in the text. On their basis, using the "association length" method, 5 clusters were found in the connection network, with a total number of 1657 links (Figure 3):

- cluster 1 - red (32 items),
- cluster 2 - green (24 items),
- cluster 3 - blue (10 items),
- cluster 4 - yellow (7 items),
- cluster 5 - purple (6 items).

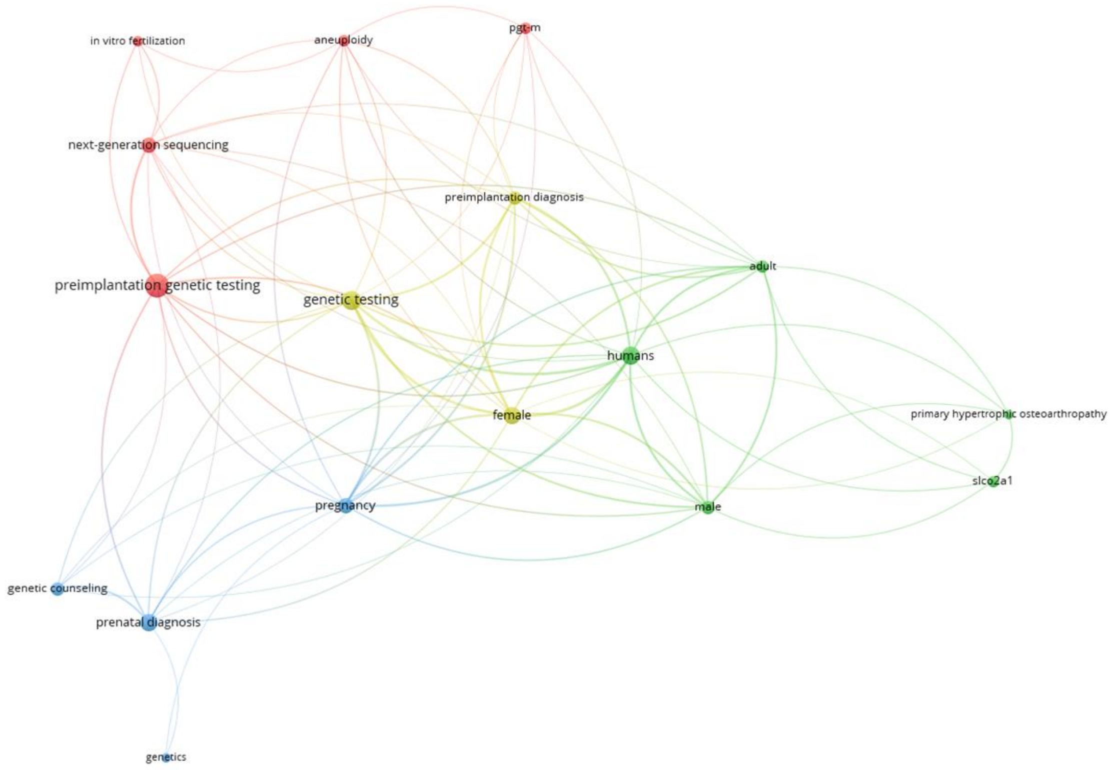


Figure 4. Mapping latent variables from keywords of the publications on issues connected with rare diseases and PGT. Note: Technical publication on mapping and clustering using VOSviewer (Ding & van Eck, 2014; Szaruga & Załoga, 2022). Limitations: The authors did not take into account many valuable publications in the study, instead taking into account the need to narrow the thematic scope of the described issues

Source: own elaboration based on 229 references [A1].

Using bibliographic data and analyzing only 'keywords', 17 keywords were selected (out of 776) that appeared at least 5 times in the text (full counting). 4 clusters with 174 connections were obtained (Figure 4):

- Cluster 1 (red) includes 5 latent variables: aneuploidy, in vitro fertilization, next-generation sequencing, PGT-M, and preimplantation genetic testing.
- Cluster 2 (green) includes 5 latent variables: adult, humans, male, and primary hypertrophic osteoarthropathy.

- Cluster 3 (blue) includes 4 latent variables: genetic counseling, genetics, pregnancy, and prenatal diagnosis.
- Cluster 4 (yellow) includes 3 latent variables: female, genetic testing, and preimplantation diagnosis.

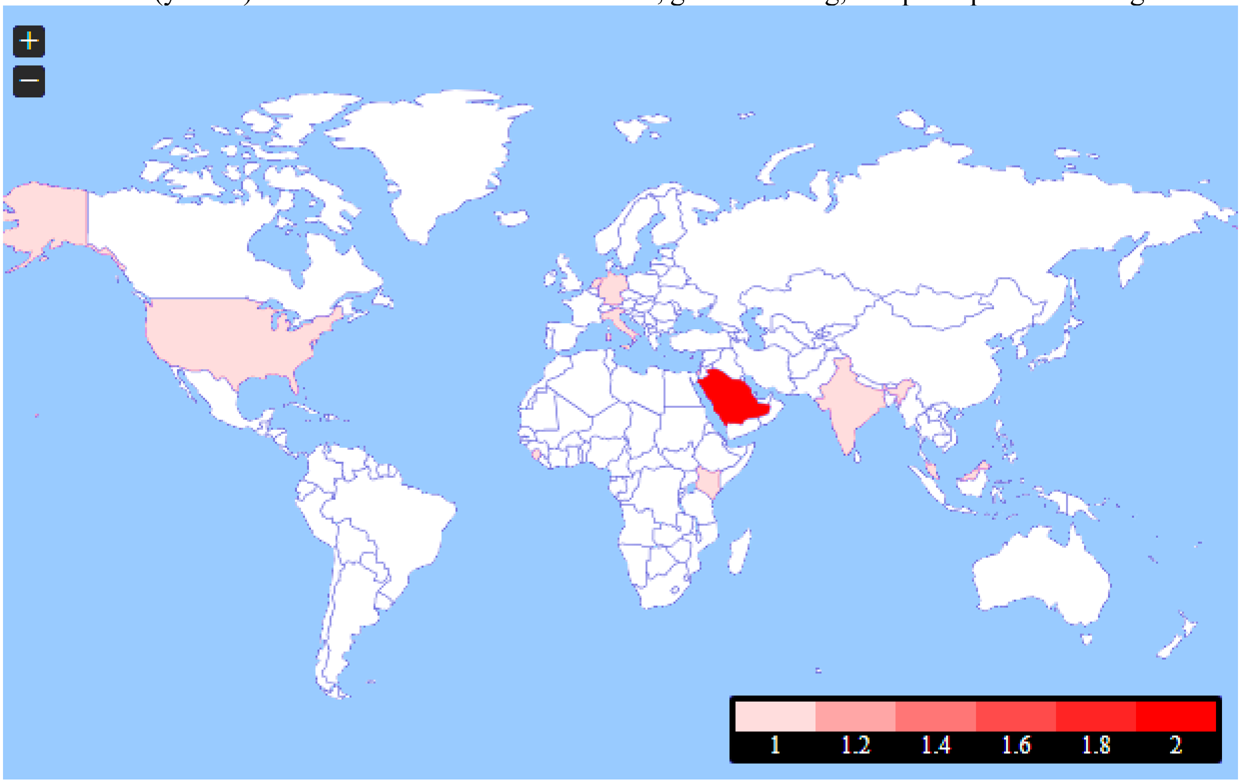


Figure 5. Publications' Content Maps from titles, abstracts, and keywords. Note: The more intense the color in the legend, the more often the geographic location is mentioned
Source: own elaboration based on 229 references [A1].

Figure 5. shows which locations were most often referred to by the authors in the context of rare diseases and PGT: United States, Netherlands, Germany, Italy, Cyprus, Israel, Saudi Arabia, Kenya, Sierra Leone, India, and Malaysia.

Words closely related to the term PGT are presented in Table 2.

From the results of thematic modeling (Latent Semantic Indexing; log perplexity = 8.0531; topic coherence = 0.8935) keywords were extracted using aggregation by average and the TF-IDF scoring method (Table 1).

Table 1. Extract keywords by TF-IDF scoring

Word	TF-IDF scoring	Word	TF-IDF scoring
genetic	0.247	preimplantation	0.122
PGT	0.147	disease	0.121
testing	0.146	patients	0.118
clinical	0.133	diagnosis	0.109
gene	0.125	sequencing	0.107

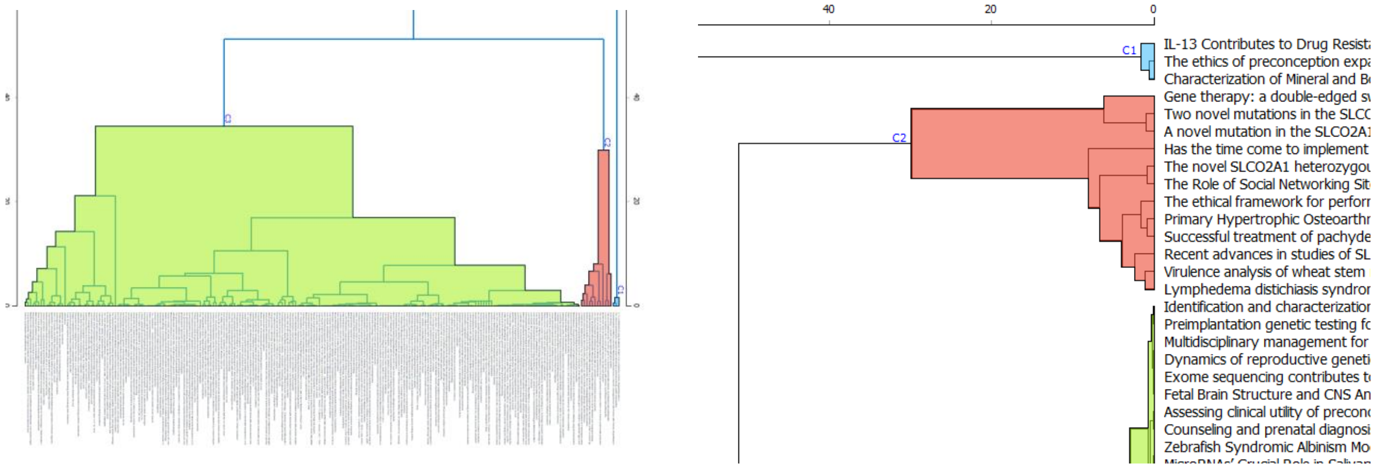
Source: own elaboration based on 229 references [A1].

Table 2. Word enrichment

Related word with PGT	p-value	FDR	Related word with PGT	p-value	FDR
preimplantation	1.8×10^{-22}	1.8×10^{-21}	results	1.2×10^{-4}	2.9×10^{-4}
testing	2.2×10^{-17}	1.5×10^{-16}	gene	5.1×10^{-3}	7.8×10^{-3}
genetic	1.7×10^{-9}	8.3×10^{-9}	diagnosis	7.4×10^{-3}	0.0106
analysis	6×10^{-6}	2×10^{-5}	rare	8.4×10^{-3}	0.0112
sequencing	1.6×10^{-6}	2.7×10^{-5}	diseases	9×10^{-3}	0.0113

Source: own elaboration based on 229 references [A1].

Table 2 presents statistically significant relationships (p-value < 0.05, FDR < 0.2). If publications are concerned with PGT, important in this context are preimplantation, rare diseases, gene diagnosis or genetic testing/analysis, sequencing, and results. Figure 6 shows the classification of publications into three thematic clusters. We can distinguish one cluster containing 3 publications (blue cluster), the second - containing 12 publications (red cluster), and the most numerous (green) - containing 214 publications.



(a) (b)

Figure 6. Hierarchical clustering. Note: (a) all publications, (b) zoom of clusters
Source: own elaboration based on 229 references [A1].

The presented method of clustering (grouping), called taxonometrics or hierarchical classification, suggests that green thematic clusters should be analyzed further, while blue and red clusters should be rejected.

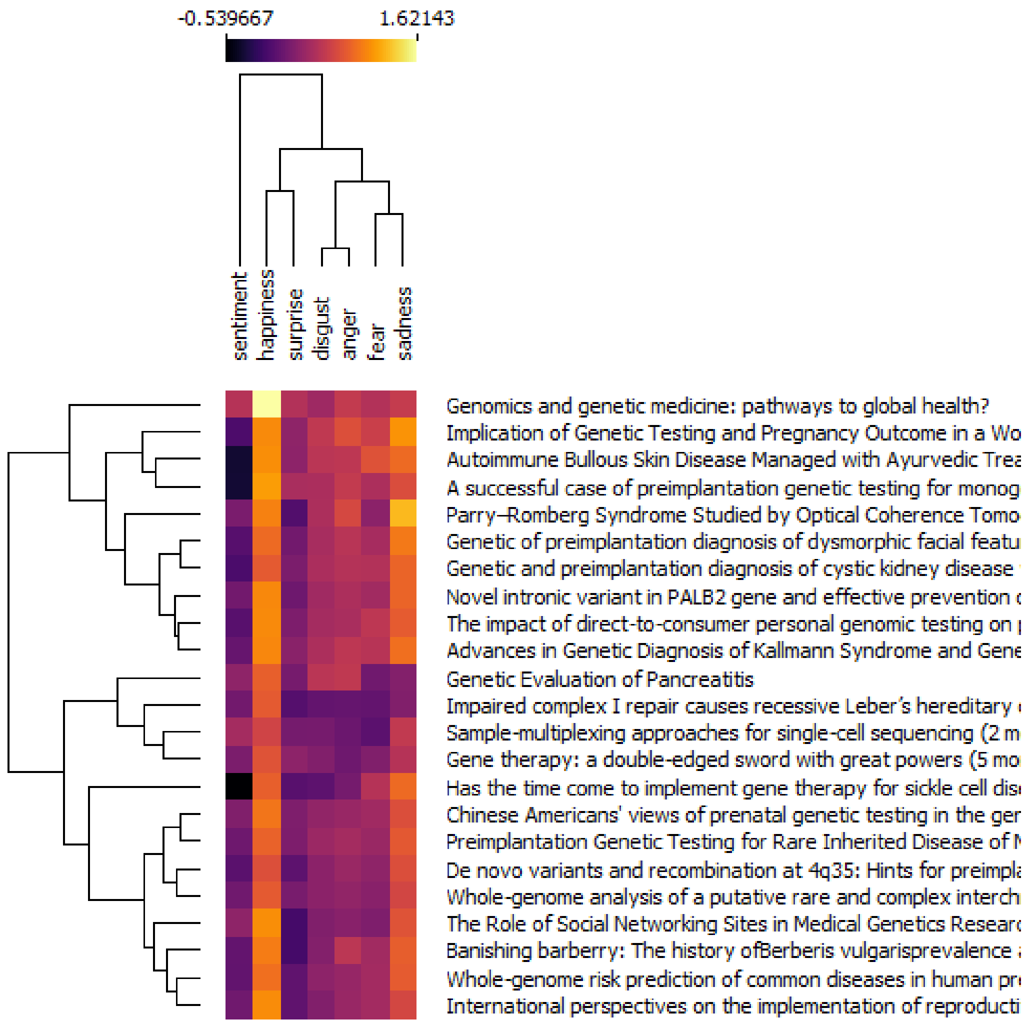


Figure 7. Sentiment analysis
Source: own elaboration based on 229 references [A1].

The last step was to conduct a sentimental analysis (Figure 7). Sentiment map using the method (SentiArt), sample type: bootstrap. Figure 7 presents 23 publications (representing 10% of all; merge by k-means). Black color represents negative values and light yellow - positive values. The presented heat map visualizes the sentiment in a collection of publications, which calculates sentiment scores for each of them. The analysis results include 7 columns. The first column 'sentiment' aggregates the remaining types of sentiment and is the resultant of all sentiments in the publication. Column 2 shows happiness, third - surprise, fourth - disgust, fifth - anger, sixth - fear, and seventh - sadness. Clustering was used to group moods. Fear and sadness formed one group of moods, anger and disgust another. The first and second groups of moods formed the third group of moods: sadness, fear, anger, and disgust together. The fourth group of moods consisted of happiness and surprise. It contrasted with the third group of moods. Generally, it can be noticed that the publications had a negative expression of moods (dark colors), although it is also possible to identify those in which negative and positive moods were balanced (sustained). In general, happiness was associated through the prism of a positive mood, surprise, disgust, and fear mainly as negative strong moods, and sadness dichotomously positive and negative (although theoretically, it could be a negative mood, in combination with hope or other moods - it could take the form of a positive attitude).

The frequency of terms related to the effectiveness of IVF with PGT in the analyzed publications was low, and these terms were excluded from the clustering analysis (Table 3). Among the identified records, some studies were directly relevant to the aim of this research. The results suggest that preimplantation genetic testing (PGT) may be an effective tool to help couples with hereditary disorders achieve healthy offspring. However, available data often focus on successful cases, and the overall effectiveness of IVF with PGT varies widely. Couples should understand that the number of eggs retrieved, embryos obtained, and unaffected embryos selected after PGT can vary greatly, and the procedure is not always successful.

Effectiveness rates reported in the literature depend on the condition and patient population, with live birth rates ranging broadly from approximately 10% to 80%. Additionally, economic evaluations indicate that IVF with PGT may be more cost-effective than natural conception with prenatal diagnosis for certain genetic diseases.

These findings highlight the complexity of treatment outcomes and emphasize the importance of individualized counseling and decision support for couples considering IVF with PGT.

Table 3. Terms related to the effectiveness of the IVF+PGT procedure are mentioned in the analyzed publications

Term	Number of articles	Relevant to our study	Case study	Other than case study
efficiency/effectiveness/effective/efficient	53	9	2	7
success/successfully	40	12	7	5
goal/aim/objective achievement	0	0	0	0
total	93	21	9	12

Source: own elaboration based on 229 references [A1].

Our review revealed a research gap in the assessment of the effectiveness of IVF with PGT, which has serious implications for making an informed decision by couples at risk of developing a rare monogenic disease in their offspring. Considering the seven defined research gaps presented by Miles (Miles, 2017), we suggest possibilities to develop a research agenda in the following areas:

- Knowledge gap – available reports focus on the reliability and effectiveness of IVF with PGT measured by the successful detection of selected disorders in embryos, the birth of healthy babies, and optimized time and financial cost of testing procedures. However, for many couples, it would also be important to answer other questions regarding the survival of embryos obtained through IVF, the number of stimulation cycles necessary to achieve the goal, and financial and non-financial costs, including side effects. This knowledge is important for couples who request IVF not because of fertility problems but because the procedure offers a chance to stop the occurrence of the disease in their family. To achieve their goal, they need a larger number of embryos than in the standard IVF without PGT. It should be noted, however, that in autosomal recessive disorders, embryos with a carrier status can be transferred because they are usually phenotypically normal.
- Methodological and theoretical gap – currently, the effectiveness of the presented solutions is measured with a successful live birth of a healthy baby and there is no analytical model that would estimate the relation between all benefits and all costs (financial and non-financial). The complexity and multi-stage character of the PGT-IVF procedure, particularly the uncontrollable randomness involved in the combination of parental alleles, contributes to considerable psychological burden experienced by patients. Couples appear to downplay this factor at the outset of their journey with the procedure, being highly focused on achieving success. As a result, any failures become especially psychologically distressing. The results of the sentiment analysis appear to confirm this.
- Population gap – available publications concern case studies describing single couples or cohorts of couples affected by a specific genetic disease. The results of such a scale only indicate the potential for success of the PGT-IVF procedure. They do not provide insight into the overall effectiveness of the procedure or the effectiveness at its individual stages – hormonal stimulation, embryo testing, embryo transfer, pregnancy, and birth. The absence of statistically reliable, population-level data

presents a significant challenge to the effective application of artificial intelligence–based management tools, such as: integrated clinical and genetic data management across all stages of the procedure; the use of AI and machine learning for outcome prediction; decision support systems for both physicians and patients; standardization and knowledge sharing; data security, privacy, and ethical compliance; and the automation of laboratory processes and procedural workflows.

Detailed problems associated with the definition of effectiveness in the context specific to IVF with PGT and components considered in the assessment of this procedure are presented in the Discussion.

5. DISCUSSION

5.1. The Future of PGT-IVF in the Context of Intelligent Management

Intelligent management in reproductive medicine is grounded in key theories such as the Knowledge-Based View (Grant, 1996; Nonaka & Takeuchi, 1995), Clinical Decision Support Systems (Belciug & Gorunescu, 2020; Khalfallah et al., 2023), Decision Theory under Uncertainty and Multi-Criteria Decision Analysis (Keeney & Raiffa, 1993; Dyer & Forman, 1992), and Systems Thinking (Ackoff, 1999; Senge, 1990). These frameworks support the use of AI tools tailored to patient contexts and ethical considerations.

Families with a child affected by genetic disease face complex decisions, including relationship strain and family planning choices. IVF with preimplantation genetic testing for monogenic disorders (PGT-M) offers a potential solution, though it raises ethical and systemic challenges (Kruse et al., 2022). Genetic counseling and education remain critical to address fears and improve awareness.

Integrating databases and intelligent management tools can reveal patterns to improve success rates, reduce failures, and lower financial and psychological burdens. Proposed directions include machine learning to predict IVF success based on clinical data, AI-powered decision support for clinicians and patients, automation in laboratory processes, and psychological well-being assessment throughout IVF cycles.

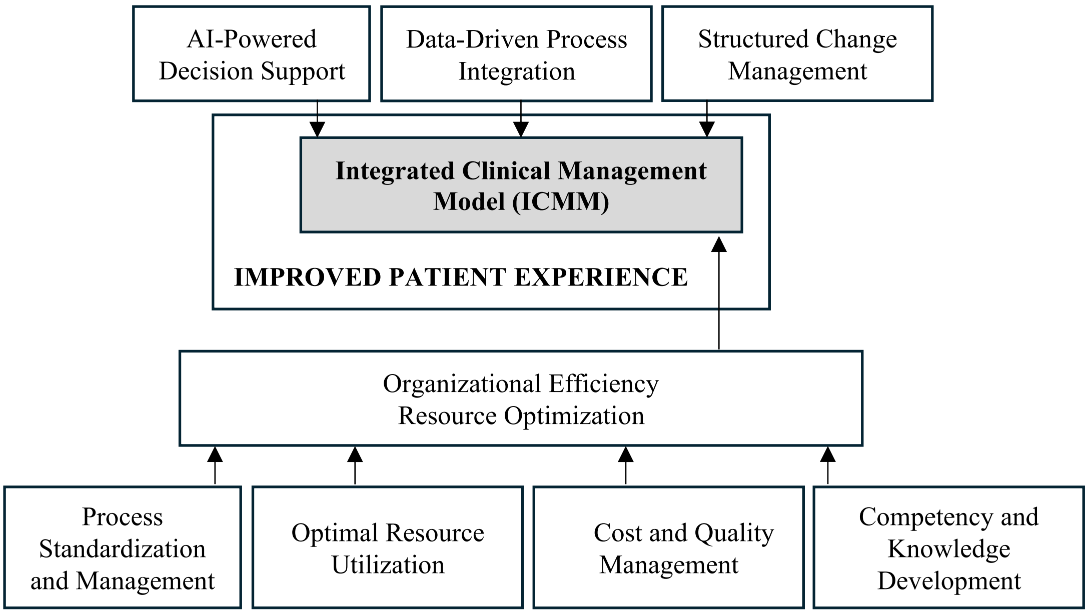


Figure 8. Integrated Clinical Management Model (ICMM) for IVF with PGT
Source: own elaboration based on Khalfallah et al. (2023), Belciug & Gorunescu (2020), and Greenhalgh et al. (2017).

We propose the ICMM, combining AI-powered decision support, data-driven integration, and structured change management (Figure 8). The model improves clinical precision and patient experience by enabling transparent, personalized pathways and fostering trust and emotional resilience. This improved patient experience is a core outcome of integrating AI-powered decision support, data-driven processes, and organizational efficiency, ensuring patients feel more supported and engaged throughout their reproductive journey.

ICMM integrates clinical, genetic, economic, and emotional data to simulate treatment scenarios, supporting informed decisions (Belciug & Gorunescu, 2020; Khalfallah et al., 2023). It is flexible, scalable, integrates with EHR and LIS, and continuously learns from data, improving decision efficiency, reducing unnecessary interventions, and optimizing resources.

Beyond patient support, the ICMM enhances organizational efficiency by automating decision processes, standardizing care pathways, reducing clinical workload, and improving resource planning. By optimizing resource allocation and standardizing care, ICMM not only enhances organizational performance but also contributes to a more positive and supportive patient journey. Predictive components within the system support staffing allocation and laboratory scheduling, which is crucial for the time- and resource-sensitive IVF clinics (Topol, 2019; Davenport & Kalakota, 2019; Jiang et al., 2017). This dual focus on individual patient care and organizational effectiveness makes the model well-suited for reproductive healthcare settings where precision and efficient resource management are critical.

5.2 Efficacy and Efficiency in IVF with PGT

Effectiveness in IVF with PGT refers to achieving the birth of a healthy child, while efficiency concerns optimizing resources such as hormonal stimulations and embryos used to maximize success (Drucker, 2024; Gasparski, 2022; Nowosielski, 2008; Polski Komitet Normalizacyjny, 2001). These concepts overlap but differ: effectiveness means ‘doing the right things’, whereas efficiency means ‘doing things right’ (Drucker, 2024).

Depending on patient goals – avoiding genetic defects or successfully having a healthy child – success criteria vary. AI-powered decision-support tools help clinicians and patients set realistic, personalized goals by integrating medical, genetic, emotional, and financial data, thereby improving transparency and informed decision-making (Khalfallah et al., 2023).

For example, AI platforms like AIVF’s EMA analyze embryo viability and genetics in real time to guide embryo selection, illustrating AI’s potential to enhance IVF success rates (Chavez-Badiola et al., 2019; AIVF, 2024).

5.3. Financial and Emotional Costs of IVF with PGT

IVF with PGT is often chosen not due to infertility but to ensure the birth of a genetically healthy child. This procedure entails significant financial burdens, especially where public funding is limited by age or other criteria (BGH, 2019). Emotional costs are also substantial, as failed stimulation cycles, embryo loss, or embryo freezing for donation cause considerable stress. Despite these challenges, couples persist in pursuing IVF with PGT to maximize their chances of a healthy child.

Support systems for couples vary across countries, influencing the financial and emotional experience during treatment. AI-driven digital health tools like the iYoni mobile application offer valuable support in early reproductive planning. Although iYoni is not directly integrated with PGT procedures, it provides functionalities such as menstrual cycle tracking, infertility treatment assistance, and reproductive health education (iYoni, 2024). By delivering personalized insights based on user data, the app empowers users to better understand their reproductive health, anticipate fertility windows, and make informed decisions. This helps reduce emotional uncertainty and stress throughout the complex reproductive journey, especially for couples at genetic risk who face heightened psychological challenges. iYoni exemplifies how AI-based digital solutions can extend support beyond clinical settings, promoting well-being throughout the continuum of reproductive care.

5.4. Dynamic Efficiency and Ethical Considerations

Effectiveness and efficiency in IVF with PGT must be considered alongside ethical and social dimensions. The ‘3Es’ model – effectiveness, efficiency, and ethics – provides a comprehensive framework for evaluating clinical performance (Dietl & Gasparski, 2000). Static efficiency involves minimizing waste and optimizing processes, while dynamic efficiency focuses on long-term benefits such as family well-being and risk reduction (Górecka et al., 2021; Kotarbiński & Kleszcz, 2019; McGrath, 1970).

This approach recognizes couples as clients within a reproductive medicine business model, emphasizing a holistic perspective that balances immediate clinical outcomes with psychological and social impacts. IVF with PGT strives to achieve both a healthy live birth and the efficient use of resources, considering emotional and ethical implications for families facing genetic risks (Gasparski, 2022; Nowosielski, 2008).

6. CONCLUSION

A key feature of the proposed Integrated Clinical Management Model (ICMM) is its capacity for continuous learning from clinical data and patient feedback. This enables increasingly personalized recommendations, fostering more effective and empathetic decision support for couples undergoing IVF with PGT. The next steps involve pilot collaborations with clinics and further research to validate the model in clinical practice (Greenhalgh et al., 2017; Hughes et al., 2021).

This study employed a range of AI-based techniques, including word cloud visualization, bag of words with L1 regularization, Latent Semantic Indexing (LSI), and keyword extraction using TF-IDF scoring. Sentiment analysis via the SentiArt method added contextual interpretation of the emotional dimensions associated with different research areas.

In the context of PGT for rare diseases, text mining and machine learning provide a systematic means of extracting, structuring, and interpreting large volumes of unstructured data. IVF with PGT offers at-risk couples an alternative to prenatal testing and potential pregnancy termination, supporting the goal of having a healthy child while minimizing physical, emotional, and financial burdens.

Our interdisciplinary literature review revealed thematic clusters related to IVF with PGT and highlighted a major gap: the lack of large-scale, population-level studies capturing both successes and failures. These limits understanding of real-world effectiveness and obscures the full scope of emotional and financial burdens carried by patients. Defining standardized success criteria remains a challenge, particularly given that couples at risk of rare diseases differ significantly from traditional infertility patients.

The sentiment analysis confirmed the high emotional toll experienced by these couples. For many, success is defined strictly as the birth of a healthy, unaffected child, often requiring more embryos and higher emotional and financial investments. Current systems of support are not well-equipped to address these unique needs, and cost-effectiveness models tailored to this population are largely absent.

Several limitations affect this study. The available evidence is based mainly on case reports and small-sample research. Publication bias and limited jurisdictional context (focused on Poland) further constrain generalizability. Machine learning was used solely for literature analysis, not clinical prediction. Real-time, patient-level clinical data were unavailable due to access restrictions. The literature review covered four databases, and keyword selection—though expert-informed—may have overlooked relevant studies. The focus on monogenic disorders points to the need for future research into polygenic and multifactorial conditions.

Future work should expand the assessment model to include economic and psychological variables, moving toward a multi-criteria framework. This expanded framework is embodied in the Integrated Clinical Management Model (ICMM), which integrates clinical, genetic, economic, and psychosocial data to provide comprehensive, personalized, and ethically sound decision support. The model's flexibility and scalability allow adaptation across different healthcare settings, supporting both patient-centered care and organizational efficiency. Broader collaborations with clinics and patient advocacy organizations are planned to gather more representative data across various populations. These efforts will support the use of AI not only for knowledge management but also for the development of predictive tools to enhance PGT-IVF effectiveness.

In addition to personalized decision support, the ICMM contributes to organizational performance. The expanded ICMM integrates multiple data dimensions and emphasizes continuous learning and adaptation, positioning it as an innovative solution for the complex needs of IVF with PGT. By streamlining workflows, supporting resource allocation, and enhancing scalability through digital infrastructure, it promotes both clinical precision and operational efficiency (Kellermann & Jones, 2013). As AI becomes more integrated into healthcare systems, its dual role in improving patient care and organizational function becomes increasingly important (Topol, 2019; Davenport & Kalakota, 2019; Jiang et al., 2017).

Funding: Co-financed by the Minister of Science under the "Regional Excellence Initiative".

Regionalna Inicjatywa Doskonałości

Ministry of Science and Higher Education Republic of Poland

REFERENCES

Ackoff, R. L. (1999). Ackoff's Best: His Classic Writings on Management. Wiley.

AIVF. (2024). *EMA: AI-powered embryo evaluation platform*. Retrieved from: https://aivf.com. (10.04.2025).

Amine, A., Simonet, M., & Elberrichi, Z. (2010). Evaluation of Text Clustering Methods Using WordNet. *The International Arab Journal of Information Technology*, *7(4):349-357*.

Bagherzadeh, S., Shokouhyar, S., Jahani, H., & Sigala, M. (2021). A generalizable sentiment analysis method for creating a hotel dictionary: Using big data on TripAdvisor hotel reviews. *Journal of Hospitality and Tourism Technology*, *12*(2), 210–238. https://doi.org/10.1108/JHTT-02-2020-0034.

Belciug, S., & Gorunescu, F. (2020). Intelligent decision support systems: A journey to smarter healthcare. Springer. https://doi.org/10.1007/978-3-030-14354-1.

BGH. (2019). *Urteil des Bundesgerichtshofs (BGH) vom 4. Dezember 2019*.

Capalbo, A., Fabiani, M., Caroselli, S., Poli, M., Girardi, L., Patassini, C., Favero, F., Cimadomo, D., Vaiarelli, A., Simon, C., Rienzi, L. F., & Ubaldi, F. M. (2021). Clinical validity and utility of preconception expanded carrier screening for the management of reproductive genetic risk in IVF and general population. *Human Reproduction*, *36*(7), 2050–2061. https://doi.org/10.1093/humrep/deab087.

Castro, A. P., Wainer, G. A., & Calixto, W. P. (2022). Weighting construction by bag-of-words with similarity-learning and supervised training for classification models in court text documents. *Applied Soft Computing*, *124*, 108987. https://doi.org/10.1016/j.asoc.2022.108987.

Chavez-Badiola, A., Flores-Saiffe Farias, A., Mendizabal-Ruiz, G., Garcia-Sanchez, R., & Drakeley, A. J. (2019). Artificial vision and machine learning designed to predict PGT-A results. Fertility and Sterility, 112(3), e1–e2.

Davenport, T., & Kalakota, R. (2019). The potential for artificial intelligence in healthcare. Future Healthcare Journal, 6(2), 94–98. https://doi.org/10.7861/futurehosp.6-2-94.

De Rycke, M., & Berckmoes, V. (2020). Preimplantation Genetic Testing for Monogenic Disorders. *Genes*, *11*(8), 871. https://doi.org/10.3390/genes11080871.

De Wert, G., Dondorp, W., Shenfield, F., Devroey, P., Tarlatzis, B., Barri, P., Diedrich, K., Provoost, V., & Pennings, G. (2014). ESHRE Task Force on Ethics and Law22: Preimplantation Genetic Diagnosis. *Human Reproduction*, *29*(8), 1610–1617. https://doi.org/10.1093/humrep/deu132.

Dietl, J., & Gasparski, W. (Eds.). (2000). *Etyka biznesu* (Wyd. 1). Wydawn. Naukowe PWN.

Ding, Y., & van Eck, N. J. (2014). Visualizing bibliometric networks. In L. Waltman, R. Rousseau, & D. Wolfram (Eds.), *Measuring scholarly impact: Methods and practice*. Springer.

Drucker, P. F. (2024). *Praktyka zarządzania* (J. W. Gołębiowski, T. Basiuk, & Z. Broniarek, Trans.). MT Biznes.

Dyer, J. S., & Forman, E. H. (1992). Group decision support with the Analytic Hierarchy Process. Decision Support Systems, 8(2), 99–124.

Gárate-Escamilla, A. K., Amaya, I., Cruz-Duarte, J. M., Terashima-Marín, H., & Ortiz-Bayliss, J. C. (2022). Identifying Hyper-Heuristic Trends through a Text Mining Approach on the Current Literature. *Applied Sciences*, *12*(20), 10576. https://doi.org/10.3390/app122010576.

Gasparski, W. (2022). *Decyzje i etyka: Co o etyce każdy decydent wiedzieć powinien?: 75+ minitekstów o maxisprawach* (Wydanie rozszerzone i uzupełnione). Poltext.

Grant, R. M. (1996). Toward a knowledge-based theory of the firm. Strategic Management Journal, 17(S2), 109–122.

Greenhalgh, T., Wherton, J., Papoutsi, C., et al. (2017). Beyond adoption: A new framework for theorizing and evaluating nonadoption, abandonment, and challenges to the scale-up, spread, and sustainability of health and care technologies. Journal of Medical Internet Research, 19(11), e367. https://doi.org/10.2196/jmir.8775.

Górecka, D., Brzezicki, Ł., Miszczyńska, K., Piontek, T., & Prędki, A. (2021). *W poszukiwaniu efektywności*. Wydawnictwo Naukowe Uniwersytetu Mikołaja Kopernika.

Harper, J. C., Wilton, L., Traeger-Synodinos, J., Goossens, V., Moutou, C., SenGupta, S. B., Pehlivan Budak, T., Renwick, P., De Rycke, M., Geraedts, J. P. M., & Harton, G. (2012). The ESHRE PGD Consortium: 10

years of data collection. *Human Reproduction Update*, *18*(3), 234–247. https://doi.org/10.1093/humupd/dmr052.

Haude, K., McCarthy Veach, P., LeRoy, B., & Zierhut, H. (2017). Factors Influencing the Decision-Making Process and Long-Term Interpersonal Outcomes for Parents Who Undergo Preimplantation Genetic Diagnosis for Fanconi Anemia: A Qualitative Investigation. *Journal of Genetic Counseling*, *26*(3), 640–655. https://doi.org/10.1007/s10897-016-0032-0.

iYoni. (2024). *Aplikacja mobilna do planowania ciąży i wspierania zdrowia reprodukcyjnego* [Mobile application]. Retrieved from: https://www.iyoni.pl (15.04.02025).

Jacobs, A. M. (2019). Sentiment Analysis for Words and Fiction Characters From the Perspective of Computational (Neuro-)Poetics. *Frontiers in Robotics and AI*, *6*, 53. https://doi.org/10.3389/frobt.2019.00053.

Jacobs, A. M. (2022). The neurocognitive poetics model of literary reading 10 years after. In Chatterjee & Cardillo (Eds.), *Brain, beauty, and art: Essays bringing neuroaesthetics into focus, Chatterjee, A., & Cardillo, E.R. (Eds.)*. Oxford University press.

Jacobs, A. M., & Kinder, A. (2022). Computational Models of Readers' Apperceptive Mass. *Frontiers in Artificial Intelligence*, *5*, 718690. https://doi.org/10.3389/frai.2022.718690.

Jiang, F., Jiang, Y., Zhi, H., Dong, Y., Li, H., Ma, S., ... & Wang, Y. (2017). Artificial intelligence in healthcare: Past, present and future. Stroke and Vascular Neurology, 2(4), 230–243. https://doi.org/10.1136/svn-2017-000101.

Keeney, R. L., & Raiffa, H. (1993). *Decisions with multiple objectives: Preferences and value trade-offs*. Cambridge University Press.

Kellermann, A. L., & Jones, S. S. (2013). What it will take to achieve the as-yet-unfulfilled promises of health information technology. Health Affairs, 32(1), 63–68. https://doi.org/10.1377/hlthaff.2012.0693.

Khalfallah, M., et al. (2023). Artificial Intelligence, Clinical Decision Support Algorithms, and Assisted Reproductive Technologies: A Systematic Review. MCP Digital Health, 1(1), 100082. https://doi.org/10.1016/j.mcpd.2023.100082.

Klein, R. D., & Mahoney, M. J. (2007). Medical Legal Issues in Prenatal Diagnosis. *Clinics in Perinatology*, *34*(2), 287–297. https://doi.org/10.1016/j.clp.2007.03.006.

Kotarbiński, T., & Kleszcz, R. (2019). *Traktat o dobrej robocie* (Wydanie I). Wydawnictwo Uniwersytetu Łódzkiego.

Kruse, C., Pollex, H., & McKay, S. (2022). Ethical challenges of IVF and preimplantation genetic testing: A review. Bioethics Today, 15(1), 45-60.

Kruse, J., Mueller, R., Aghdassi, A. A., Lerch, M. M., & Salloch, S. (2022). Genetic Testing for Rare Diseases: A Systematic Review of Ethical Aspects. *Frontiers in Genetics*, *12*, 701988. https://doi.org/10.3389/fgene.2021.701988.

Kuhr, F., Braun, T., Bender, M., & Möller, R. (2020). Augmenting and Automating Corpus Enrichment. *International Journal of Semantic Computing*, *14*(02), 173–197. https://doi.org/10.1142/S1793351X20400061.

Kuo, C.-L. (2023). Revolutionizing Healthcare Paradigms: The Integral Role of Artificial Intelligence in Advancing Diagnostic and Treatment Modalities. International Microsurgery Journal (IMJ). doi: 10.24983/scitemed.imj.2023.00177.

Lossio-Ventura, J. A., Gonzales, S., Morzan, J., Alatrista-Salas, H., Hernandez-Boussard, T., & Bian, J. (2021). Evaluation of clustering and topic modeling methods over health-related tweets and emails. *Artificial Intelligence in Medicine*, *117*, 102096. https://doi.org/10.1016/j.artmed.2021.102096.

McGrath, J. E. (1970). *Social psychology: A brief introduction*. Holt, Rinehart [and] Winston.

Miles, D. A. (2017). *A taxonomy of research gaps: Identifying and defining the seven research gaps.* Doctoral Student Workshop: Finding Research Gaps - Research Methods and Strategies, Dallas, TX, USA.

Montani, S., & Bellazzi, R. (2002). Supporting decisions in medical applications: The knowledge management perspective. International Journal of Medical Informatics, 68(1), 79–90. doi: 10.1016/S1386-5056(02)00067-9.

Nonaka, I., & Takeuchi, H. (1995). The knowledge-creating company: How Japanese companies create the dynamics of innovation. Oxford University Press.

Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y., & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: Analysis of the Orphanet database. *European Journal of Human Genetics*, *28*(2), 165–173. https://doi.org/10.1038/s41431-019-0508-0.

Ning, W., Lin, S., Zhou, J., Guo, Y., Zhang, Y., Peng, D., Deng, W., & Xue, Y. (2018). WocEA: The visualization of functional enrichment results in word clouds. *Journal of Genetics and Genomics*, *45*(7), 415–417. https://doi.org/10.1016/j.jgg.2018.02.008.

Nowosielski, S. (2008). Skuteczność i efektywność realizacji procesów gospodarczych. In T. Dudycz & Z.

Wilimowska (Eds.), *Efektywność funkcjonowania szkół wyższych*. Politechnika Wrocławska : Indygo Zahir Media.

Oshni Alvandi, A., Burstein, F., & and Bain, C. (2023). A digital health ecosystem ontology from the perspective of Australian consumers: A mixed-method literature analysis. Informatics for Health and Social Care, 48(1), 13–29. doi: 10.1080/17538157.2022.2049273.

Parikh, F., Athalye, A., Kulkarni, D., Sanap, R., Dhumal, S., Warang, D., Naik, D., & Madon, P. (2021). Evolution and utility of preimplantation genetic testing for monogenic disorders in assisted reproduction—A narrative review. *Journal of Human Reproductive Sciences*, *14*(4), 329. https://doi.org/10.4103/jhrs.jhrs_148_21.

Pieklak, M., Pałka, K., Frelas, K., Rumiancew, K., Kiełczewski, T., Kamińska, E., Marciniak, A., & Smaga, A. (2023). *Choroby rzadkie dostępność terapii: Przegląd rozwiązań i rekomendacje dla Polski*. Domański Zakrzewski Palinka sp. k. & PEX PharmaSequence Sp. z o.o.

Polski Komitet Normalizacyjny. (2001). *Systemy zarządzania jakością—Podstawy i terminologia PN-EN ISO 9000*.

Pyper, E., McKeown, S., Hartmann-Boyce, J., & Powell, J. (2023). Digital Health Technology for Real-World Clinical Outcome Measurement Using Patient-Generated Data: Systematic Scoping Review. Journal of Medical Internet Research, 25(1), e46992. doi: 10.2196/46992.

Qiu, L., Gorantla, S., Rajan, V., & Tan, B. C. Y. (2021). Multi-disease Predictive Analytics: A Clinical Knowledge-aware Approach. ACM Trans. Manage. Inf. Syst., 12(3), 19:1-19:34. doi: 10.1145/3447942.

Qu, X., Walsh, E., Cherbuin, N., & Black, L. (2022). Mapping the Literature on Diet and Multiple Sclerosis: A Data-Driven Approach. *Nutrients*, *14*(22), 4820. https://doi.org/10.3390/nu14224820.

Senge, P. M. (1990). The Fifth Discipline: The Art & Practice of The Learning Organization. Doubleday.

Sun, Y., & Zhao, X. (2023). Effective Data Science Leadership Based on Text Mining and Machine Learning Model. In T. Wang, S. Patnaik, W. C. Ho Jack, & M. L. Rocha Varela (Eds.), *Applications of Decision Science in Management*, 260, 181–193. Springer Nature Singapore. https://doi.org/10.1007/978-981-19-2768-3_17.

Szaruga, E., & Załoga, E. (2022). Qualitative–Quantitative Warning Modeling of Energy Consumption Processes in Inland Waterway Freight Transport on River Sections for Environmental Management. *Energies*, *15*(13), 4660. https://doi.org/10.3390/en15134660.

Takeuchi, K. (2021). Pre-implantation genetic testing: Past, present, future. *Reproductive Medicine and Biology*, *20*(1), 27–40. https://doi.org/10.1002/rmb2.12352.

Topol, E. J. (2019). High-performance medicine: The convergence of human and artificial intelligence. Nature Medicine, 25(1), 44-56. https://doi.org/10.1038/s41591-018-0300-7.

Veisi, H., Aflaki, N., & Parsafard, P. (2019). Variance-based Features for Keyword Extraction in Persian and English Text Documents. *Scientia Iranica*, *0*(0), 0–0. https://doi.org/10.24200/sci.2019.50426.1685.

Venugopal, M., Sharma, V. K., & Sharma, K. (2023). Web information mining and semantic analysis in heterogeneous unstructured text data using enhanced latent Dirichlet allocation. *Concurrency and Computation: Practice and Experience*, *35*(1), e7410. https://doi.org/10.1002/cpe.7410.

Yahya Saeed, M., Awais, M., Younas, M., Arif Shah, M., Khan, A., Irfan Uddin, M., & Mahmoud, M. (2021). An Abstractive Summarization Technique with Variable Length Keywords as per Document Diversity. *Computers, Materials & Continua*, *66*(3), 2409–2423. https://doi.org/10.32604/cmc.2021.014330.

Zajac, K., Braun, M., Klein, J., & Kolbe, L. M. (2024). Towards Digital Health Equity? – Assessing the Inclusivity of National Digital Health Strategies for Vulnerable Groups. ECIS 2024 Proceedings. Retrieved from: https://aisel.aisnet.org/ecis2024/track01_peoplefirst/track01_peoplefirst/7 (15.04.2025).

Zhou, Z., Jin, D., He, J., Zhou, S., Wu, J., Wang, S., … Feng, T. (2024). Digital Health Platform for Improving the Effect of the Active Health Management of Chronic Diseases in the Community: Mixed Methods Exploratory Study. Journal of Medical Internet Research, 26(1), e50959. https://doi.org/10.2196/50959.