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Advances in current state of diagnostic approaches towards investigating pervasive development disorder

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ABSTRACT

Pervasive development disorder (PDD) is characterized by various types of neurodevelopment conditions influencing communication skill, behavioral, and social skills. Current state of screening methods mainly uses neurophysiological and clinical assessments that are reliable but suffer from challenges e.g., scalability, increased cost, and subjectivity. Artificial intelligence (AI) based solution is witnessed to have increased adoption in this regard also suffers from clinical integration, data bias, and explainability-oriented problems despite its potential contribution till date. Therefore, the proposed study presents a systematic review of recent literature with a closer emphasis on AI and non-AI-based methodologies adopted to diagnose PDD. The study outcome also exhibits similar facts. The novelty of this work resides in its comparative analysis, temporal mapping of current trends, and its integrative framework. It also contributes towards identifying certain underexplored areas, thereby facilitating practically viable insight towards the future direction of implementation. The idea is to understand its viability, which is not only clinically relevant but also offers sustainable diagnostics.

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1. INTRODUCTION

Pervasive development disorder (PDD) can be termed as a spectrum of neurodevelopment disorders that is featured by delays in behavior, communication, and socialization. PDD also involves multiple related disorders, e.g., Rett's disorder, childhood disintegrative disorder (CDD), Asperger's syndrome, autistic disorder. At present, there are various screening techniques that is divided into various classes as artificial intelligence (AI)-assisted computational method, clinical (behavioral), and neuropsychological method. The behavioral and clinical screening tool involves the use of the modified checklist for autism in toddlers-revised with follow-up (M-CHAT-R/F), autism diagnostic observation schedule, 2nd edition (ADOS-2), autism diagnostic interview-revised (ADI-R), childhood diagnostic interview-revised (CDI-R), the childhood autism rating scale (CARS-2), and social communication questionnaire (SCQ) [1]. The neurophysiological and neuroimaging approaches involve eye-tracking and gaze analysis, magnetic resonance imaging (MRI) or functional magnetic resonance imaging (fMRI), and brain activity pattern screening [2]. The AI-based computational screening approaches involves machine learning (ML), deep

learning (DL), and hybrid models. From the viewpoint of efficiency, clinical screening tool are highly reliable but they are subjective and time-intensive, while accuracy is strongly dependent on professional expertise. AI screening tools also offer higher accuracy in a controlled research environment; however, they have lack of explainability and data bias resulting in restricted clinical translation. Neurophysiological tools provide highly objective insight but they have increased cost and reduced scalability [3]–[5]. Hence, the shortcomings of the current state of screening tools are explainability issues in AI models, inferior integration between clinical practices and AI, diversity gaps, data scarcity, limited early detection, and human bias. It was also noted that the adoption of AI-based solutions is increasing in its pace towards diagnosis of PDD. Screening platforms using AI models is known to play a role of early screening and detection, feature extraction and pattern recognition, predictive diagnosis, decision support for clinicians, personalized intervention planning. In contrast to conventional diagnosis methods, ML contributes towards learning discriminative features from behavioral/clinical data.

DL contributes towards the automatic extraction of complex representation of clinical data of various forms. It is also seen that natural language processing (NLP) contributes towards analyzing linguistic patterns as well as speech semantics from various communication samples while multimodal fusion models offer integration of behavioral features with multimedia for better profiling scope [6]. Hence, an AI-based solution is known to offer increased predictive accuracy along with better scalability and automation. It is also known to address issues pertaining to subjective bias that is frequently seen in clinician-reported or parent-reported scales. Studies using AI models can enhance over period of time with increasing exposure to unseen data, while vision-based and NLP-based solutions are known for its cost effectiveness. AI-based diagnostic models encounter various constraints too viz. integration with the healthcare system, privacy and ethical concern, clinical validation gap, transparency and explainability, imbalance and data scarcity. In a nutshell, AI acts as proactive solution to bridge the gap between computational intelligence and behavioral science to offer adaptive, objective, and scalable diagnostic solution. In order to offer comprehensive insights towards the current forms of diagnostics, it is essential to understand the scale of effectiveness along with their possible constraints or shortcomings associated with the current methods associated with diagnosis of PDD.

Various related works have been studied in this regard to find the orientation of different methodologies. Huda *et al.* [7] have presented discussion towards varied screening tools associated with identification of such disorders. Similar direction of study with specific emphasis on Indian population highlighting indigenous resources with potential information has been carried out by Sreelakshmi *et al*. [8]. Gkintoni *et al.* [9] have studied various DL-based approaches towards prediction of such form of disorder empirically evaluated on multiple standard datasets. The work carried out by Liu and Ma [10] have presented a discussion towards varied forms of screening and evaluation methods considering multiple psychometric properties to offer significant insights towards varied perspective of diagnosis. The study carried out by Speyer *et al.* [11] have presented various non-pharmacological interventions to find its effectiveness.

After studying the above related work, following research problems have been arrived viz. i) existing review papers is noted to either focus on AI or clinical methods separately leading to fragmented insights, ii) almost all the current models doesn't report of uniform benchmarks thereby rendering it challenging to evaluate and do comparison of various models, iii) the temporal evaluations associated with diagnostic methods have not been captured effectively in existing studies, and iv) there are only few studies that has ever attempted to correlated scientific-centric outcomes with clinical interpretability. Hence, it is clear from existing studies that AI-based methods are gaining pace while non-AI based methods also exist. However, it is not clear which one is dominant and effective towards diagnosis of PDD. These issues and challenges are addressed in the proposed study by integrating AI-based methodology with non-AI-based diagnostic approaches, thereby facilitating a holistic and universally relevant understanding of the present state of diagnostic methods.

The aim of the proposed study is to carry out a comprehensive and updated insight into AI and non-AI-based research approaches adopted for the diagnosis of PDD. The notion of this study is to draw a conclusive remark associated with strength and shortcomings of the current state of research models. The contributions of the current study are as follows:

- The paper reviews dual-domain methods associated with both AI and non-AI techniques, complying with the analytical framework.
- Adopting a standard and universally accepted research method, the study offers a quantitative insight that involves trends of dataset adoptions and practical ranges of accuracy.
- The current study offers a temporal mapping where research trends of last 5-years publications are reviewed and analyzed along with distinction between implementation and review-oriented studies.
- The study offers a unique learning outcome identifying certain underexplored areas along with highlights of gaps and answers to crafted research questions (RQ) in line of diagnostic methods for PDD.

The organization of the manuscript is as follows. Section 2 presents a discussion of the adopted research method. Section 3 presents the results and discussion of the study. Finally, the conclusions of this study are discussed in section 4.

2. METHOD

The present review work is carried out considering systematic literature review (SLR). Figure 1 presents a detailed overview of the process. Each of the main steps is clearly illustrated for clarity and understanding in the respective sections. A systematic search was conducted across IEEE Xplore, SpringerLink, Elsevier, MDPI, and PubMed using user desired search keywords (PDD, autism spectrum disorder (ASD), diagnosis, ML, DL, multimodal, and audio analysis). In order to have accessibility only open-access journal articles are taken into consideration. The initial search yielded a total of 1,516 records. After removing 706 duplicates, 810 studies remained for title and abstract screening. At this stage, 515 records were excluded for being non-relevant, non-peer-reviewed, or purely clinical in nature without computational modelling. The full texts of 295 studies were then assessed for eligibility based on the inclusion criteria (peer-reviewed, English language, published between 2021 and 2025, and focusing on computational approaches to PDD/ASD diagnosis). Following this assessment, 268 articles were excluded due to insufficient methodological details or lack of computational analysis, resulting in a final set of 27 studies included in the review. The selection process of the information is carried out considering preferred reporting items for systematic reviews and meta-analyses (PRISMA) based methodology for ensuring reproducible technique.

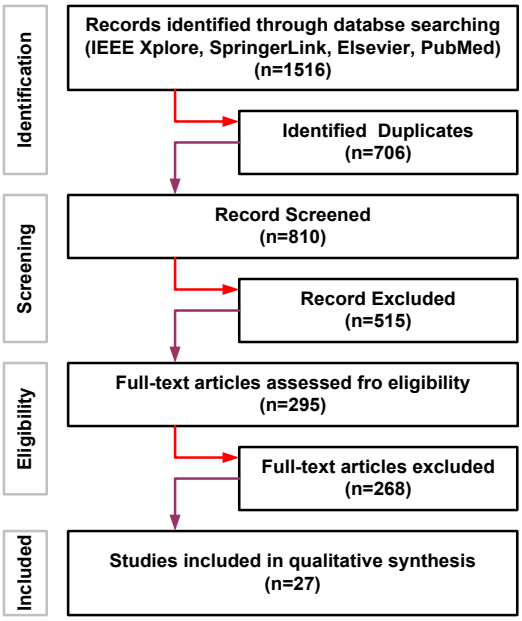


Figure 1. PRISMA flow diagram of study selection process for the systematic review

Adopting the methodology, it is noted that approaches towards diagnosis of PDD is eventually of two types viz. AI-based and non-AI based. Although, AI-based methodology is increasing yet the significance of non-AI based methodology can never be discarded. Both has their own significance and issues too. Hence, it is anticipated that adoption of this methodology should provide answers to various ongoing unsolved questions. The present review is guided by the following RQ:

- RQ_1: what computational approaches currently exist for diagnosing PDD, and how have they evolved across modalities such as text, audio, image, and multimodal frameworks?
- RQ_2: what are the primary challenges and limitations associated with existing models, particularly regarding dataset availability, generalizability, interpretability, and computational cost?
- RQ_3: what opportunities can be identified for future research to enhance diagnostic accuracy, improve model robustness, and address gaps in cross-lingual, demographic, and multimodal applicability?

3. RESULTS AND DISCUSSION

This section presents the outcomes of the presented review findings that offers a comprehensive insight towards the present state of research work for PDD. Adopting the research methodology discussed in prior section, the information collected was contextualized with a notion to withdraw the essential insights. The ideation of this review findings is towards understanding the effectiveness of current state-of-the-art methods for the diagnosis of PDD.

3.1. Review findings

Table 1 presents the research trends towards current publications associated with implementation papers (IP) and review-paper (RP). The tabulated outcome showcases consistent growth towards research activity in last 5 years. A closer look shows that number of IP have significantly increased in every aspect.

Table 1. Research trends towards publications related to diagnosis of PDD

Year	Database	Journal		Open access		Conference		Total	
		IP	RP	IP	RP	IP	RP	IP	RP
2020	Scopus	85	22	42	10	60	12	187	44
2021	IEEE Xplore	73	25	38	13	78	15	189	53
2022	SpringerLink	96	28	51	15	70	18	217	61
2023	ScienceDirect	108	31	64	18	88	22	260	71
2024	PubMed	122	34	75	20	92	25	289	79
2025	ACM digital library	135	39	80	24	100	28	315	91

Table 2 highlights that autism brain imaging data exchange (ABIDE) is one of the most dominant adopted in research studies till date due to its potential community support, highly structured neuroimaging data, and accessibility. There are also increasing trend noted for private/clinical dataset as well as Simons simplex collection (SSC) dataset too in recent times. However, its adoption is very less compared to ABIDE. Similar pattern is also noted for national databased for autism research (NDAR) and synthetic dataset.

Table 2. Dataset adoption trend

Year	ABIDE	SSC	NDAR	Private/clinical datasets	Synthetic datasets
2020	62	35	29	41	18
2021	71	39	34	46	22
2022	85	44	38	54	29
2023	97	51	43	60	36
2024	108	57	48	67	41
2025	122	63	54	73	48

Table 3 showcases that DL approaches are gaining increasing pace in contrast to ML models as found in the last 5 years. The adoption rate for DL is higher because of its capability towards handling multimodal dataset and complex scenarios mainly. At the same time, there is increasing adoption for explainable artificial intelligence (XAI) too due to its capability of offering interpretability in healthcare diagnostics. It is also noted that adoption of hybrid models and reinforcement learning (RL) models are quite less in contrast to other AI schemes.

Table 3. Adoption of AI models in PDD research

Year	ML	DL	XAI	RL	Hybrid/ensemble models	Total publications using AI
2020	58	47	16	8	11	140
2021	64	55	20	10	15	164
2022	71	68	26	13	19	197
2023	78	82	34	16	24	234
2024	85	96	42	21	29	273
2025	91	110	51	27	34	313

An investigation is also carried out to understand the influence of two equally dominating techniques i.e., AI-based and non-AI based approaches. A closer look in Table 4 shows that accuracy accomplished is quite higher by AI-based approaches in contrast to non-AI-based approaches. Specifically, hybrid and DL approaches are found to accomplish 80-90% range of accuracy in existing research works that is highly popular for automated assessment of PDD. Yet, there are issues pertaining to interoperability and data that restrict clinical deployment in routine manner.

Table 4. Comparative overview of AI and non-AI methods towards PDD

Type	Approaches	Identified study contribution along with accuracy range	Identified shortcomings of models demanding attention
AI-based methods	ML [12]	– Supports classification of neuroimages. – Early diagnosis and pattern recognition. – Accuracy range: 76–89%.	– Demands feature engineering. – limited scalability for large heterogeneous datasets.
	DL [13]	– Learns complex representations. – Enhance diagnostic precision. – Accuracy range: 83–96%.	– Limited interpretability in clinical use. – Computationally expensive.
	NLP [14]	– Analyzes complex communication patterns -Identifies atypical speech and text cues. – Accuracy range: 71–86%.	Performance depends on linguistic nuances and dataset diversity.
	XAI [15]	– Improves interpretability. – Key features driving decisions. – Accuracy range: 79–89%.	– Lacks universal evaluation standards. – Explanations are still abstract.
	RL [16]	– Deployed for adaptive therapy. – Use in personalized interventions. – Accuracy range: 69–81%.	– Ethical issues in live adaptation. – Demands large sequential interaction data.
	Hybrid/ensemble AI models [17], [18]	– Integrates multiple models. – Robust and generalizable diagnosis. – Accuracy range: 86–95%.	Increased computation time and model complexity.
	Graph neural networks [19]	– Frame brain connectivity patterns. – Identify atypical neural links. – Accuracy range: 81–91%.	– Interpretability challenges. – Demands well-structured graph data.
	Transfer learning [20]	– Supports small PDD datasets. – Accuracy range: 84–93%.	Domain bias.
Non-AI methods	Statistical modelling [21]	– Determines potential group differences. – Accuracy range: 61–76%.	Limited in handling complex, non-linear relationships.
	Neuroimaging-based models [22]	– Reveals brain structure. – Study activity differences. – Accuracy range: 71–86%.	High cost, small samples, and technical expertise demanded.
	Behavioral observation-based models [23]	– Tracks social and communication behaviors. – Accuracy range: 65–80%.	Subjective scoring and variability among raters.
	Genetic and biomarker-based models [24]	– Determines biomarkers and genetic correlations for PDD risk. – Accuracy range: 61–71%.	– Complicated patterns of inheritance. – Low diagnostic specificity.
	Survey-based and qualitative methods [25]	– Fuses perceptions from caregivers. – Reliability: 56–71%.	– Bias. – Lacks objective quantification.
	Cognitive/psychometric testing-based models [26]	– Evaluates attention in PDD individuals. – Screening validity: 66–79%.	– Cultural and linguistic variability affects generalization.
	Neurofeedback-based experiments [27]	– Behavior regulation and emotional control. – Effectiveness: 71–84%.	– Variable outcomes across individuals. – Time-intensive.

3.2. Learning outcomes

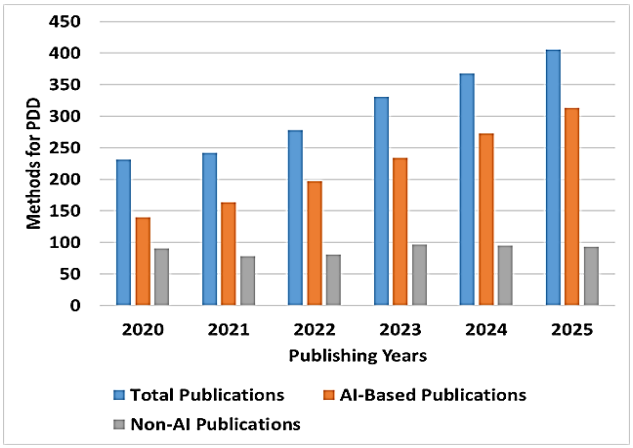
One of the key outcomes of the study is shown in Figure 2, which eventually showcases the adoption of AI-approaches is growing significantly higher in contrast to non-AI methods (Figure 2(a)). At the same time, adoption of ABIDE dataset is found consistently to be adopted in the last 5 years (Figure 2(b)). Upon reviewing various research methodologies and studying their effectiveness, the extracted learning outcomes of the study are as follows:

- The number of research works towards non-AI-based approaches is very less in contrast to AI-based approaches, while this unbalanced ecosystem offers a scope of intervention-oriented AI too.
- The area of hybrid models and multi-modal approaches is increasing in implementation works that is noted to combine feature related to language, behavior, and neuroimaging in order to establish comprehensive profiling of PDD.
- ABIDE and NDAR is one of the most widely adopted datasets, and still its adoption rate remains the same in current state of research publication in contrast to other forms or types of datasets.
- At present, the dataset is highly specific of geographic regions from where the samples have been collected and curated. Hence, its research papers considering such region-specific dataset cannot claim for generalization or extensive applicability of their work.

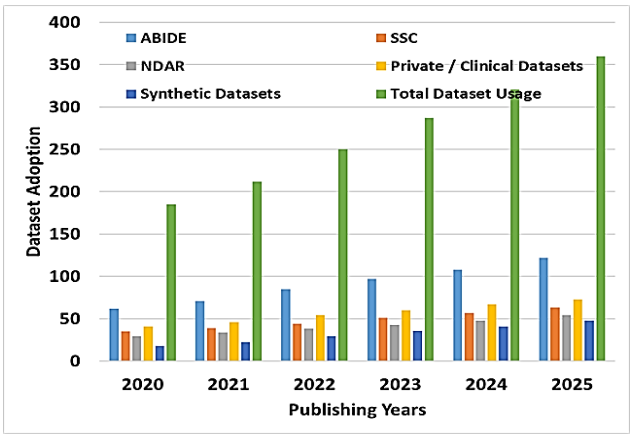
The identified research gaps are as follows:

- All the existing open dataset are quite small in number and yet they are found to potentially dominant the IP work towards PDD. Hence, dataset biasing is quite inevitable while there is restricted diversity in the dataset rending the findings less applicable to larger scale.

- The approaches e.g., graph neural network and RL have quite a better potential towards boosting detection and classification of PDD; however, current state of publications in this regard are very few to find benchmarking outcomes.
- There is a potential gap between what is presented in research work and what is really demanded on real-time towards PDD diagnosis. With less research papers on XAI, the innovativeness towards validation and interpretability is still less.
- The research publications for AI-based approaches are highly disproportionate to non-AI-based approaches. Although, it is encouraging for proactive diagnostic methods in PDD diagnosis, yet almost all AI methods are not without flaws (unreliable prediction, overfitting, increased response time, and extensive resource dependencies) that are yet to be investigated.



(a)



(b)

Figure 2. Core study findings for trends of (a) AI and non-AI based methods and (b) dataset adoption

3.3. Answers to research question

The identified solution to the RQ_1 is as: the computational approaches towards diagnosis of PDD are evolutionary trends, multimodal approaches, image-based approaches, audio-based approach, and text-based approach. Further, it is noted that computational methods have slowly transformed itself to DL and multimodal models from simple and manual-operation based conventional model. This newly evolved model is capable of integrating neuroimaging data, linguistic traits, and complex behavior for more scalable and accurate diagnosis of PDD.

The identified solution to RQ_2 is as: the primary challenges as well as limitation associated with the conventional models are mainly related to increased computational cost, reduced interpretability, poor generalization, and limited dataset availability. There are also few studies that have addressed issues pertaining to longitudinal data neither sufficient work is carried out towards adaptive learning framework.

The evolving approaches related to large transformer-oriented NLP models, analysis of neuroimaging, or multimodal fusion demands significantly high computational resources.

The identified solution to RQ_3 is: there are various quantified opportunities towards future research mainly in direction of emerging methodologies e.g., AI-assisted therapy, frameworks designed using RF and graph neural network. There is also enough scope towards exploration of personalized and adaptive interventions by AI-predictive modelling. Future work can also lay foundation towards clinical applicability and interpretability as well as by addressing data gaps by extending public dataset to involve languages, age groups, and population. Methodologies adopting validations using cross-demographics, cross-lingual, and cross-sit can offer broader generalization towards diagnostic model of PDD.

Therefore, the study synthesizes both the current and future state of varied approaches (especially computational) towards investigating PDD. The answer of RQ_1 suggest contextually to continue improvising DL models as it has higher scope and potential. The answer of RQ_2 suggests certain shortcomings and critical challenges that demand attention for addressing in the upcoming modelling perspective, while answers to RQ_3 state the higher scope and opportunities residing in varied evolving modelling perspectives.

3.4. Novelty of proposed study

Table 5 showcases the novelty and contribution associated with the presented work. It is noted that a potential establishment if an integrated framework connecting publication dynamics, dataset evolution, and methodological advances in PDD research. The outcome of the study also explored DL methodologies to be leading in contrast to other AI-centric models.

There is a requirement of ethical validations and explainability by these models that is not found to be potentially established as ground work by any researchers till date with an applicability on PDD. The proposed investigation has underscored hybrid clinical-AI integration as the upcoming frontiers towards enhancing therapeutic personalization and diagnostic reliability. Different from existing review work towards related topic, it is noted that proposed review work presents a temporal dimension by mapping research trends over 2020-2025. This potentially contributes novelty attributes introduced by proposed review work.

Table 5. Novelty and contribution of the proposed review

Items	Proposed review	Existing reviews
Coverage scope	Both AI and non-AI methods with illustrative comparison of their accuracy, contribution, and shortcomings.	Only deals with AI-based diagnostic tools or clinical methods.
Taxonomy of methods	Multi-level classification of AI and non-AI techniques.	Lacks fine-grained taxonomy.
Quantitative insight	Involve practical accuracy ranges, updated dataset trends	– Lacks numerical trend data. – Primarily qualitative summaries.
Dataset adoption	– Assesses open-access and private datasets. – Highlights research bias and dataset dominance	No systematic dataset analysis.
Mapping temporal trend	– Five-year publication. – Differentiating experimental vs. survey papers.	Most reviews cover shorter or undefined periods.
AI variant comparison	– Evaluates performance and popularity of AI variants.	Usually limited to a single AI family
Integration perspective	Focuses on cross-modal and hybrid integration.	Rarely addresses multi-modal fusion approaches.
Clinical relevance and explainability	– Discusses challenges related to clinical deployment, interpretability, XAI.	Less prior reviews engage with clinical usability or explainability
Novel contribution	– Balanced, end-to-end synthesis, datasets, publication dynamics, emerging directions and research gaps.	Lacks integrative synthesis across AI and non-AI domains, domain-restricted.

4. CONCLUSION

The study outcome of the present work showcases increasing pave of adoption of data-centric AI methods that have evolved from conventional neurophysical and behavioral methods. The study finds that DL and hybrid approaches are very much emerging solutions towards pattern recognition and diagnostic accuracy. Another interesting finding of the study is potentially higher adoption of NDAR and ABIDE datasets despite the fact that both of these datasets have lower degree of diversity and has biasness factors. This eventually remains an open-end challenge. The study findings also noted that AI-centric modelling suffers from ethical concerns, clinical applicability, and interoperability, even if they are claimed for certain set of accomplished contributions in diagnosis of PDD and its related disease. Future direction of work can be carried out towards adopting federated learning with a multimodal-oriented framework that can integrate linguistic data, behavioral, and neuroimaging. from diverse set of populations. Study can also be carried out

towards improvising interpretability and clinical trust in making decision. Finally, a modelling perspective considering graph neural network and RL can also be assessed to see certain innovative findings.

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C : **C**onceptualization
M : **M**ethodology
So : **So**ftware
Va : **Va**lidation
Fo : **Fo**rmal analysis
I : **I**nvestigation
R : **R**esources
D : **D**ata Curation
O : Writing - **O**riginal Draft
E : Writing - Review & **E**diting
Vi : **Vi**sualization
Su : **Su**pervision
P : **P**roject administration
Fu : **Fu**nding acquisition

CONFLICT OF INTEREST STATEMENT

Authors state no conflict of interest.

DATA AVAILABILITY

Data availability is not applicable to this paper as no new data were created or analyzed in this study.

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