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Integrating AI and ML in Myelodysplastic Syndrome Diagnosis: State of the Art and Future Prospects

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Introduction:

Myelodysplastic syndrome (MDS) is a diverse group of hematological malignancies characterized by dysfunctional pluripotent stem cells, leading to abnormal hematopoiesis and cytopenia. Early diagnosis is crucial for improving patient outcomes. Artificial Intelligence (AI) and Machine Learning (ML) have emerged as powerful tools in healthcare for disease diagnosis and monitoring. This review aims to summarize the current state of AI application in the diagnosis of MDS, discussing various ML models, their advantages, disadvantages, and performance metrics.

Methods:

A comprehensive search strategy was developed using Medical Subject Headings (MeSH) terms and relevant keywords related to MDS and AI/ML. Pubmed, Embase, and Scopus were searched, and duplicates were removed. Only original research articles studying ML algorithms in the diagnosis of MDS in human subjects were included. Studies on animals, reviews, non-original articles, and non-English papers were excluded. For each article, data on the area under the receiver operating curve, sensitivity, specificity, and accuracy for the best performing model were collected.

Results:

The search yielded 313 articles, and after screening, 16 articles met the inclusion criteria. These articles were grouped based on the data utilized in the training of their models. Five articles utilized bone marrow smears for the training of their data. Their models aimed to detect dysplastic cells and blasts in bone marrow smears. The majority of these studies utilized convolutional neural network (CNN), a ML algorithm, for image recognition. One study utilized decision tree models. The AUC of these models ranged between 0.8 and 0.996. All the models were internally validated but were not externally validated. Three studies utilized peripheral blood smears for the training of their ML models. Two of these studies also utilized CNN for the detection of dysplasia and subsequent diagnosis of MDS. One study utilized CART to create a decision tree algorithm. The AUC for these models ranged between 0.982-0.99. Four studies utilized flowcytometry data from bone marrow samples for the training of their AI models. These models achieved AUC ranging between 0.935-0.97. Two of these models were both internally and externally validated, one of which was on a prospective cohort.

Conclusion:

The integration of AI and ML in MDS diagnosis holds promise for early detection and improved patient care. The reviewed studies showed encouraging results, indicating that AI-based approaches can complement traditional diagnostic methods and aid in making more informed clinical decisions especially when it comes to analysing blood/bone smears. However, many of these models need to be externally validated in large prospective cohorts to establish AI's role in routine clinical practice.

Disclosures No relevant conflicts of interest to declare.

Table 1. performance metrics for the best models in the included full-text articles.

Study	Outcomes	Best Models	Validation	AUC	ACC	SEN	SPE
Wang, M., et al.	Diagnosing MDS	CNN	Internal	0.985	0.914	0.992	0.881
			External	0.942	0.921	0.886	0.938
	Distinguishing MDS from AA and AML	CNN	Internal	0.968	0.929	0.857	0.967
			External	0.948	0.915	0.887	0.929
Lee, N.	Detecting dysplastic Erythrocytes	CNN	Internal	0.972	0.988	0.790	0.992
	Detecting dysplastic Granulocytes	CNN	Internal	0.996	0.993	0.900	0.999
	Detecting dysplastic Megakaryocytes	CNN	Internal	0.971	0.931	0.899	0.948
	Detecting Blasts	CNN	Internal	0.973	0.932	0.831	0.951
Mori, J.	Diagnosis of MDS using severe dysplasia (DG-3)	CNN	Internal	0.944	0.972	0.910	0.977
	Diagnosis of MDS using dysplasia and severe dysplasia	CNN	Internal	0.921	0.982	0.852	0.989
Wu, J.	Diagnosis of hypocellular MDS and distinguishing from AA	Decision Tree	Internal	0.800	0.805	0.765	0.837
Wu, Y.	Detection of >5% blasts	CNN: BMSnet	Internal	0.948	NR	NR	NR
Acevedo, A.	Detection of hypogranulated dysplastic neutrophils	CNN: model M1	Internal	0.982	0.949	0.955	0.943
Kimura, K.	Diagnosis of MDS and distinguishing from AA	CNN with Xgboost	Internal	0.990	>0.900	0.962	1.000
Zhu, J.	Diagnosis of MDS	CART	Internal	NR	NR	0.845	0.978
Radhachandran, A.	Predicting Risk of MDS	Xgboost	Internal	0.872	NR	0.785	0.804
Radakovich, N.	Diagnosis of MDS	GBM	Internal	0.951	NR	NR	NR
Pozdnyakova, O.	Detection of cytopenia related to MDS	Random Forest	Internal	0.930	NR	0.890	0.840
Xiang, P.	Diagnosis of MDS and distinguishing from AA	IDEAL-IQ-based SVM	Internal	0.930	0.922	NR	NR
Clichet, V.	Diagnosis of MDS	Elasticnet (LinearR)	External	0.935	NR	0.918	0.925
Duetz, C.	Diagnosis of MDS in suspected patients	Random Forest	Internal	0.964	NR	0.850	0.950
			External	NR	NR	0.970	0.950
Herbig, M.	Prediction of MDS	Random forest	Internal	0.950	0.910	0.860	1.000
Li, J. L.	Classification of MDS vs Normal	LogR using AGF-P	Internal	0.956	0.960	NR	NR
	Classification of MDS vs AML	LogR using AGF-P	Internal	0.911	0.875	NR	NR

Figure 1

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