

NEURODEVELOPMENTAL DISORDERS: SELECTED STUDIES USING MACHINE LEARNING

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**DEPARTMENT OF MANAGEMENT STUDIES
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by

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DEPARTMENT OF MANAGEMENT STUDIES

submitted

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This dissertation is dedicated to my cherished son, Ekagrah Kohli, who, despite being diagnosed with Autism Spectrum Disorder, has displayed remarkable resilience and unwavering strength. He has been an endless source of inspiration, teaching me the profound meaning of unconditional love and the boundless power of determination. This work stands as a tribute to his indomitable spirit and serves as a testament to the significance of early intervention and support for individuals with neurodevelopmental disorders.

I also extend this dedication to my wife, Swati Kohli, a dedicated special education and Behavior Analyst. Swati's steadfast commitment to comprehending and bolstering Ekagrah's journey has been immeasurable. Her expertise, empathy, and ceaseless advocacy for our son's needs have been pivotal in his progress. This research would not have come to fruition without her unswerving support and unwavering belief in Ekagrah's potential.

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Certificate

This is to certify that the dissertation entitled “**Neurodevelopmental Disorders: Selected Studies Using Machine Learning**”, submitted by **Manu Kohli** to the Indian Institute of Technology Delhi, for the award of the degree of **Doctor of Philosophy** in the Department of Management Studies, is a record of the original, bonafide research work carried out by him under our supervision and guidance. The dissertation has reached the standards fulfilling the requirements of the regulations related to the award of the degree.

The results contained in this dissertation have not been submitted in part or in full to any other University or Institute for the award of any degree or diploma to the best of our knowledge.

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Abstract

The prevalence of Autism Spectrum Disorder (ASD) is estimated to be 1% in India ([Arora et al., 2018](#)) and 1.5% in the US ([CDC, 2021](#)). However, in low- and middle-income countries (LMICs) like India, the healthcare and social care infrastructure for detecting and managing ASD and Other Developmental Disorders (ODD) is insufficient. Existing clinical methods face challenges that can lead to delays in early detection and access to early intervention, ultimately affecting the social, intellectual, and vocational development of children with ASD. These limitations hinder the growth and inclusion of ASD children. Fortunately, technological advancements and data science methods offer a potential solution to address these inefficiencies.

The dissertation titled “Neurodevelopmental Disorders: Selected Studies Using Machine Learning” explores the role of machine learning and data science in diagnosing and treating neurodevelopmental disorders, particularly Autism Spectrum Disorder (ASD).

This dissertation comprises eight chapters encompassing five distinct studies, addressing three research questions and five research objectives. The studies involve experiments, reviews, and stakeholder analyses focusing on the role of technology in ASD detection and intervention.

Chapter 1, the introduction, establishes the foundational aspects by providing background information and conducting a literature review that serves as the basis for the subsequent studies (1-5) and chapters.

Chapter 2, Research Methods, covers machine learning techniques such as supervised and unsupervised learning. It also explores the role of patient similarity and collaborative filtering methods to recommend and personalize intervention for ASD participants. This chapter also includes the PRISMA methodology used to conduct two scoping reviews (Chapters 3 and 6).

Chapters 3 and 4 cover scoping reviews and human action recognition experiments. These chapters assess the capabilities of machine learning models in detecting and assessing Autism Spectrum Disorder (ASD) in its early stages using multimodal structured and unstructured datasets.

Chapter 5 involves experiments that employ machine learning algorithms to provide treatment recommendations and personalization systems for ASD. It utilizes patient similarity and collaborative filtering machine learning methods to recommend and personalize ABA (Applied Behavior Analysis) treatment.

Chapter 6 covers a scoping review to examine the role of robot-mediated interventions for children with ASD. Various robotic technologies are highlighted, showcasing diverse roles played by Robots in improving the skills of children with ASD across twelve areas.

Chapter 7 captures stakeholder perspectives on using machine learning integrated information and communication technology for ASD and ODD detection and intervention during and after the COVID-19 pandemic. The examination reveals a perceptual paradox within the stakeholder community, shedding light on fundamental causative factors that emerged during the COVID-19 pandemic. These factors

encompass techno-stress, stress induced by the pandemic itself, the multifaceted roles assumed by stakeholders surrounding caregiving and parenthood, and concerns related to technology trust and literacy.

Chapter 8, the concluding chapter, provides a comprehensive overview of the research, emphasizing its contributions, limitations, and potential future directions.

The dissertation demonstrates the potential of machine learning techniques in improving early detection, personalizing interventions, and ultimately enhancing outcomes, affordability, access, and quality of care for individuals and families with neurodevelopmental disorders.

The dissertation makes noteworthy contributions to the literature, paving the way towards developing an end-to-end digital framework for detection and intervention, serving families and clinicians in remote locations.

संक्षेप

ऑटिज़म स्पेक्ट्रम डिसऑर्डर (ASD) की प्रसार भारत में लगभग 1% है (Arora et al., 2018) और संयुक्त राज्य अमेरिका में लगभग 1.5% है (CDC, 2021)। हालांकि, भारत जैसे कम- और मध्य-आय देशों में (LMICs) ऑटिज़म स्पेक्ट्रम डिसऑर्डर (ASD) और अन्य विकासात्मक विकारों (ODD) के लिए स्वास्थ्य और सामाजिक देखभाल की क्लिनिकल बुनियादी ढांचा अपर्याप्त है। मौजूदा क्लिनिकल तरीकों का सामना चुनौतियों से करना पड़ता है जो पहले डिटैक्शन और प्रारंभिक हस्तक्षेप (इंटरवेंशन) में देरी और पहुंच में बाधा डालती है, अंततः ऑटिज़म स्पेक्ट्रम डिसऑर्डर (ASD) वाले बच्चों के सामाजिक, बौद्धिक, और व्यावसायिक विकास पर प्रभाव डाल सकती है। ये सीमाएं एएसडी बच्चों की वृद्धि और समावेश को बाधित करती हैं। भाग्यशाली रूप से, तकनीकी उन्नति और डेटा विज्ञान के प्रगति इन कमीओं का समाधान प्रस्तुत कर सकते हैं। यह थीसिस "न्यूरोडेवलपमेंटल डिसऑर्डर्स: मशीन लर्निंग का उपयोग करते हुए चयनित अध्ययन" मशीन लर्निंग और डेटा विज्ञान की भूमिका को न्यूरोडेवलपमेंटल डिसऑर्डर्स, विशेष रूप से ऑटिज़म स्पेक्ट्रम डिसऑर्डर (ASD) के पहचान और इंटरवेंशन में अन्वेषण करता है।

इस थीसिस में आठ अध्याय और पांच विभिन्न अध्ययनों का समावेश है, जिसमें तीन शोध प्रश्न और पांच शोध उद्देश्यों का ध्यान दिया गया है। यह अध्ययन प्रयोग, साहित्य की समीक्षा, और हितधारक (स्टेकहोल्डर) दृष्टिकोण को शामिल करता है जो ऑटिज़म स्पेक्ट्रम डिसऑर्डर (ASD) के डिटैक्शन और इंटरवेंशन में तकनीकी इंटीग्रेशन की भूमिका पर ध्यान केंद्रित करता है।

अध्याय 1, इंटीडक्शन, बुनियादी पहलुओं को स्थापित करता है जिसमें आवश्यक बैकग्राउंड जानकारी प्रदान करना और साहित्य समीक्षा करना शामिल है, जो आगामी अध्ययनों (1-5) और अध्यायों के आधार के रूप में कार्य करती है।

अध्याय 2, रिसर्च मेथड, सुपरवाइज्ड और अनुपेक्षित शिक्षा जैसी मशीन लर्निंग तकनीकों को शामिल करता है। अध्याय यह भी अन्वेषण करता है कि रोगी समानता और कॉलेबोरेटिव फ़िल्टरिंग विधियों की भूमिका क्या है, जिसके माध्यम से ऑटिज़म स्पेक्ट्रम विकल्पों के लिए सलाह देना और व्यक्तिगत इंटरवेंशन करना संभव है। यह अध्याय दो स्कोपिंग समीक्षाओं (अध्याय 3 और 6) का आयोजन करने के लिए प्रिस्मा मैथोडोलॉजी को शामिल करता है।

अध्याय 3 और 4 में स्कोपिंग समीक्षा और मानव क्रिया पहचान प्रयोगों का विवरण प्रस्तुत है। इन अध्यायों में मशीन लर्निंग मॉडल की क्षमताओं का मूल्यांकन किया जाता है, जो विभिन्न डेटा प्रकारों का उपयोग करके ऑटिज़म स्पेक्ट्रम विकल्प की पहचान, मूल्यांकन, और इंटरवेंशन में सहायक हैं।

अध्याय 5 में मशीन लर्निंग एल्गोरिदम का उपयोग करने वाले प्रयोगों का विवरण दिया गया है जो ऑटिज़म स्पेक्ट्रम विकल्प के लिए व्यक्तिगत इंटरवेंशन की सिफारिशें करते हैं। यह अध्याय रोगी समानता और कॉलेबोरेटिव फ़िल्टरिंग मशीन लर्निंग विधियों का विवरण करता है ताकि ABA (Applied Behavior Analysis) उपचार की सिफारिशें और व्यक्तिगत बनाई जा सके।

अध्याय 6 एएसडी के बच्चों के लिए रोबोट-संचालित इंटरवेंशन की भूमिका की मूल्यांकन करने के लिए एक स्कोपिंग समीक्षा को कवर करता है। विभिन्न रोबोटिक प्रौद्योगिकियों को उजागर किया गया है, जो रोबोट्स के द्वारा ऑटिज़म स्पेक्ट्रम विकल्पों के बच्चों के कौशल को बढ़ाने में बारह क्षेत्रों में विविध भूमिकाओं को दर्शाते हैं।

अध्याय 7 मशीन लर्निंग और इनफार्मेशन टेक्नोलॉजी का उपयोग करने पर स्टेकहोल्डरों के दृष्टिकोण को एएसडी और अन्य डिसऑर्डर की पहचान और इंटरवेंशन के लिए COVID-19 महामारी के दौरान और बाद में इकट्ठा किया गया है। अध्याय स्टेकहोल्डरों समुदाय में एक अनुभूति-विडम्बना का परिचय देती है, जिससे COVID-19 महामारी के दौरान उत्पन्न कारणों का ग़्यान होता है। इन कारणों में टेक्नो-तनाव, महामारी द्वारा स्वयं, केयर देने और पैरेंटशिप के चारों ओर स्थानांतरण की बहुमुखी भूमिकाओं, और तकनीक के विश्वास और साक्षरता से संबंधित चुनौतियों को शामिल किया गया है। अध्याय 8, आखिरी अध्याय, अनुसंधान का एक संपूर्ण संक्षिप्तीकरण प्रस्तुत करता है, जिसमें इसके योगदान, सीमाएँ, और संभावित भविष्य के दिशानिर्देशों को उजागर किया गया है। यह थीसिस मशीन

लर्निंग तकनीकों के क्षमताओं का प्रदर्शन करती है जो पहले डिटेक्शन, इंटरवेंशन की व्यक्तिगतकरण, और अंत में न्यूरोडेवलपमेंटल डिसऑर्डर्स वाले व्यक्तियों और परिवारों के लिए अनुकूल परिणाम, वित्तीयता, पहुंच, और देखभाल की गुणवत्ता में सुधार करने में सक्षम हैं। यह थीसिस साहित्य में महत्वपूर्ण योगदान करती है, पहचान और इंटरवेंशन के लिए एक अंत से अंत डिजिटल ढांचा विकसित करने की दिशा में मार्गदर्शन करती है, जो ग्रामीण और दूरस्थ स्थानों में परिवारों और चिकित्सकों की मदद सकता है।

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Abbreviations

ASD	A utism S pectrum D isorder
RMI	R obot M ediated I ntervention
CASP	C ritical A ppraisal S kills P rogram
NT	N euro T ypical
LMICs	L ow and M iddle I ncome C ountries
ADLs	A ctivities of D aily L ivings
ADHD	A ttention D eficit H yperactivity D isorder
AAC	A ugmentative and A lternative C ommunication
TOBY	T herapy O utcomes B y Y ou
EIBI	E arly I ntensive B ehavioural I nterventions
KASPAR	K inesics A nd S ynchronization in P ersonal A ssistant R obotics
ODD	O ther D evelopmental D isorders
RQ	R esearch Q uestions
EEG	E lectro E ncephalo G ram
MRI	M agnetic R esonance I maging
PRISMA	P referred R eporting I tems for S ystematic R eviews and M eta- A nalyses
NAO	NAO robot
RET	R obot E nhanced T herapy
IEEE	I nstitute of E lectrical and E lectronics E ngineers
ADOS	A utism D iagnostic O bservation S chedule
SHT	S tandard H uman T reatment
CBT	C ognitive B ehavioural T herapy
LTM	L east T o M ost
TD	T ypically D eveloping
ANOVA	A Nalysis O f V ariance
CARS	C hildhood A utism R ating S cale
WISC	W echsler I ntelligence S cales for C hildren
RCTs	R andomized C ontrolled T rials
EG	E xperimental G roup
CG	C ontrol G roup
ID	I ntellectual D isabilities
SLI	S pecific L anguage I mpairment

ODD	O ppositional D efiant D isorder
IQ	I ntelligence Q uotient
SCQ	S ocial C ommunication Q uestionnaire
DSM	D iagnostic and S tatistical M anual
MSEL	M ullen S cales of E arly L earning
SRS	S ocial R esponsiveness S cale
ADI	A utism D iagnostic I nterview
PEP-3	P sycho E ducational P rofile 3
WAIS	W echsler A dult I ntelligence S cale
GMDS	G riffiths M ental D evelopment S cale
K-ABC	K aufman A ssessment B attery for C hildren
TIQ	T otal I ntelligent Q uotient
BSID	B ayley S cales of I nfant D evelopment
MINI	M ini- I nternational N europsychaitric I nterview
DISCO	D iagnostic I nterview for S ocial and C ommunication D isorders
LSAS	L iebowitz S ocial A nxiety S cale
PDD	P ervasive D evelopmental D isorder
GAP	G lobal A ssessment F unctioning
VMI	V isual M otor I ntegration
ETCH	E valuation T ool of C hildren's H andwriting
THS-R	T est of H andwriting S kills R evised
SBIS	S tanford- B ineet I ntelligence S cale
WPPSI-3	W echsler P reschool and P rimary S cale of I ntelligence 3
SON-R	S nijders- O omen N onverbal R evised
GARS	G illiam A utism R ating S cale
WHO	W orld H ealth O rganization
EHRs	E lectronic H ealth R ecords
IMU	I nertial M easurement U nit
EDA	E lectro D ermal A ctivity
EMG	E lectro M yo G raphy
MARIA	M obile A utomatic R obot for I nteraction with A utistics
ML	M achine L earning
DL	D eep L earning
AI	A rtificial I ntelligence
CRI	C hild- R obot I nteraction
CNN	C onvolutional N eutral N etwork
CLNF	C onstrained L ocal N eural F ield
IJA	I nitiating J oint A ttention
RJA	R esponding J oint A ttention
ABA	A ppplied B ehavior A nalysis
FATE	F airness, A ccountability, T ransparency, and E thics

HIC	H igh- I ncome C ountries
HR	H igh- R isk
NICU	N ewborn I ntensive C are U nit
AE	A uto E ncoder
BLSTM	B idirectional L ong S hort T erm M emory networks
CE-CLM	C onvolutional E xperts C onstrained L ocal M odel
CVA	C omputer V ision A nalys
DA	D iscriminant A nalys
DS	D ecision S tump
FF	F ace to F ace
GBDT	G radient B oosted D ecision T ree
HM	H and M ovement
LR	L ogistics R egresssion
HOG	H istogram of O riented G radients
LSTM	L ong S hort- T erm M emory
MGOA	M odified G rasshopper O ptimization A lgorithm
PCA	P rincipal C omponent A nalys
PQ	P arental Q uestionnaire
SA	S tatistical A nalys
SF	S till- F ace
SVM	S upport V ector M achine
SVM-RFE	S upport V ector M achine - R ecursive F eature E limination
RF	R andom F orest
RGF	R egularized G reedy F orest
DT	D ecision T ree
FDR	F isher D iscriminant R atio
GLMRM	G eneralized L inear M ixed R egression M odel
LBP	L ocal B inary P attern
LMM	L inear M ixed M odel
NB	N aïve B ayes
OCR	O ptical C haracter R ead
RAT	R obot A ssisted T herapy
SVD	S ingular V alue D ecomposition
ABC	A utism B ehavioral C hecklist
ADI-R	A utism D iagnostic I nterview - R evised
ADOS-T	A utism D iagnostic O bservation S chedule for T oddlers
AOSI	A utism O bservation S cale for I nfants
ASQ	A utism S pectrum Q uotient
BEIQ	B est E stimate I ntellectual Q uotient
CBCL	C hild B ehavior C heck L ist
CCMD	C hinese C lassification and diagnostic criteria of M ental D isorders

CSBS-DP	C ommunication and S ymbolic B ehavior S cales - D evelopmental P rofile
DQ	D evelopment Q uotient
GDSQ	G esell D evelopmental S cale Q uotient
K-CARS	The K orean version of the C hildhood A utism R ating S cale
MCHAT-R	M odified C Heklist for A utism in T oddlers - R evised
MCHAT-R	M odified C Heklist for A utism in T oddlers - R evised with Follow-up
VABS	V ineland A daptive B ehavior S cale
WS	W est S yndrome
VIRSA	V ideo-referenced I nfant R ating S ystem for A utism
CI s	C ircumscribed I nterests
AOI	A rea O f I nterest
FACS	F acial A ction C oding S ystem
KST	K olmogorov- S mirnov T est
LDA	L atent D irichlet A llocation
SSI	S ub S et I nstance
MEMS	M icro- E lectro- M echanical S ystem
MOOCS	M assive O nline O pen C ourse S
POMDP	P artially O bservable M arkov D ecision P rocess
NDD	N euro D evelopmental D isorders
CV	C omputer V ision
HAR	H uman A ction R ecognition
NDCG	N ormal D iscounted C umulative G ain
RS	R ecommendation S ystems
SL	S upervised L earning
CF	C ollaborative F iltering
CTR	C linical T rial R egistry
PSM	P atient S imilarity M etric
ALS	A lternating L east S quare
TP	T rue P ositive
TN	T rue N egative
FP	F alse P ositive
FN	F alse N egative
TE	T reatment E ffectiveness
DTR	D ynamic T reatment R egime
ROC	R eciever O perating C haracteristic
VSMS	V inland S ocial M aturity S cale
ADIS	A nxiety D isorders I nterview S chedule
AP@k	A verage P recision a t k
AUC	A rea U nder the R OC C urve
BCBA	B oard C ertified B ehavior A nalyst
CDS	C linical D ecision S upport

CHR	C linical H igh- R isk
C3I	C omputer-mediated C aregiver- C hild I nteraction
CG@k	C umulative G ain a t k
DCG@k	D iscounted C umulative G ain a t k
EMR	E lectronic M edical R ecord
fMRI	F unctional M agnetic R esonance I maging
FPR	F alse P ositive R ate
GDD	G lobal D evelopment D elay
HR-ASD	H igh- R isk A utism S pectrum D isorder
IDCG@k	I deal D iscounted C umulative G ain a t k
IEP	I ndividualized E ducation P rogram
IJA	I nitiating J oint A ttention
INDT-ASD	I NCLEN D iagnostic T ool for A utism S pectrum D isorder
ISAA	I ndian S cale for identification of A utism
LSML	L ocally S upervised M etric L earning
MAP@k	M ean A verage P recision a t k
P@k	P recision a t k
RNN	R ecurrent N eural N etwork
T-scores	T otal scores
TPR	T rue P ositive R ate
VB-MAPP	V erbal B ehavior M ilestones A ssessment A nd P lacement P rogram
DNN	D eep N eural N etwork
DF	D ecision F orest
KNN	K - N earest N eighbor
J48	J48 T ree-based A lgorithm
KS	K olmogorov- S mirnov
Log OR	L og O dd R atio
LOSO	L eave O ne S ubject O ut
NN	N eural N etwork
BeDevel-I	B ehavior D evelopment S creening for T oddlers I nterview
BeDevel-P	B ehavior D evelopment S creening for T oddlers P lay
WSI	W echsler I ntelligence S cale
LR	L ow- R isk
SLC	S peech and L anguage C ondition
LD	L earning D isorder
ODD	O ther D evelopmental D isorders
M-CHAT-R/F	M odified C hecklist for A utism in T oddlers - R evised with F ollow-up
NLP	N atural L anguage P rocessing
SNA	S ocial N etwork A nalysis
ICT	I nformation and C ommunication T echnology
AUROC	A rea U nder the R eceiver O perating C haracteristics

AOI	A utomated O ptical I nspection
NDCG@k	N ormalized D iscounted C umulative G ain@ k
PRISMA-ScR	PRISMA Extension for S coping R eviews
DOI	D igital O bject I dentifiers
ANN	A rtificial N eural N etwork
EG	E xperimental G roup
GOA	G rasshopper O ptimization A lgorithm
EeGeMAPS	G eneva M inimalistic A coustic P arameter S et
DoF	D egree o f F reedom
ADL	A ctivity of D aily L iving
ASOTS	A utonomous S ocial T raining S ystem
ERGM	E xponential R andom G raph M odels
GRiST	G alatean R isk S creening T ool
GATA	G raph A ugmented T riplet A rchitecture
DBN	D ynamic B ayesian N etwork
CBOW	C ontinuous B ag- O f- W ords
OT	O ccupational T herapy
PT	P hysiotherapy

Symbols

L^2	Squared error loss function
λ	Regularization parameter
k	Relevant items
$P(k)$	Precision for k items
i	Items
u	Users
x_u^T	Vector associated with the user
y_i^T	Vector associated with the items
$\hat{R}$	User-item interaction matrix or rating matrix
$C_{implicit}$	Cost function for implicit feedback
c_{ui}	Degree of confidence in user-item interactions
p_{ui}	Predicted user-item interaction
α	Implicit model parameter tuned for data
C^i	Diagonal matrix with c_{ui}