

Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction

A PROJECT REPORT

Submitted by

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In partial fulfillment for the award of the degree of
Master of Technology
In
Software Engineering (5 Year Integrated Programme)



School of Computer Science and Engineering

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DECLARATION

I hereby declare that the project entitled Your **Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction** submitted by me to the School of Computer Science and Engineering, Vellore Institute of Technology, Chennai, 600 127, in partial fulfillment of the requirements of the award of the degree of Master of Technology in Software Engineering (5 year Integrated Programme) and as part of SWE3004 – Software Design and Development Project is a bonafide record of the work carried out by me under the supervision of **Dr.AMUTHA S.** I further declare that the work reported in this project, has not been submitted and will not be submitted, either in part or in full, for the award of any other degree or diploma of this institute or of any other institute or University

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This is to certify that the report entitled **Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction** is prepared and submitted by FEDRICK SAMUEL W (Reg No:19MIS1112) to Vellore Institute of Technology, Chennai, in partial fulfillment of the requirement for the award of the degree of Master of Technology in Software Engineering (5 year Integrated Programme) and as part of SWE3004 – Software Design and Development Project is a bonafide record carried out under my guidance. The project fulfills the requirements as per the regulations of this University and in my opinion meets the necessary standards for submission.

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Abstract

This project delves into the realm of autism prediction through a data-driven framework. Leveraging advanced analytical techniques, the study aims to comprehend intricate patterns within diverse datasets associated with autism spectrum disorders (ASD).

By amalgamating machine learning algorithms with comprehensive data sets encompassing genetic, behavioral, and environmental factors, the research endeavors to create predictive models for early detection and enhanced understanding of ASD.

The objective is to unveil nuanced insights and potential predictive markers that could revolutionize early intervention strategies, paving the way for more personalized and effective approaches in the diagnosis and management of autism.

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1.Introduction

Autism Spectrum Disorder (ASD) represents a complex neurodevelopmental condition characterized by a wide spectrum of challenges in social interaction, communication, and behavior. Its prevalence has been steadily increasing, emphasizing the urgency for more effective methods of early detection, intervention, and understanding. The multifaceted nature of ASD, encompassing both genetic predispositions and environmental influences, presents a challenge for traditional diagnostic approaches. As a result, the need for innovative, data-driven methodologies has become imperative to unravel the intricate layers of ASD etiology and prediction.

"Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction" is a pioneering initiative aimed at harnessing the power of data analytics, machine learning, and comprehensive data sets related to ASD. This project recognizes the potential of integrating various sources of information, including genetic markers, behavioral patterns, environmental factors, and demographic variables, to construct robust predictive models.

The conventional diagnosis of ASD primarily relies on behavioral observations and subjective assessments, often leading to delayed identification and intervention. However, the advancement in technology, coupled with the availability of extensive datasets, provides an unprecedented opportunity to transcend these limitations. By systematically analyzing vast arrays of data, this research endeavors to identify subtle yet significant patterns that could serve as early indicators or predictive markers for ASD.

Moreover, understanding the interplay between genetic predispositions and environmental influences is crucial in comprehending the heterogeneous nature of ASD. By leveraging sophisticated data analysis techniques, this study aims to disentangle these complex interactions, shedding light on the mechanisms underlying ASD development.

The implications of such an approach extend far beyond mere prediction; it has the potential to revolutionize the landscape of ASD research and clinical practice. Early identification of individuals at risk could pave the way for tailored interventions and support services, significantly improving outcomes and quality of life for individuals on the autism spectrum.

In essence, this project stands at the forefront of innovative research, seeking to harness the power of data-driven methodologies to enhance our understanding of ASD, facilitate early detection, and ultimately transform the approach towards autism intervention and support.

1.1 Background

Autism Spectrum Disorder (ASD) represents a multifaceted neurodevelopmental condition with a diverse range of symptoms and severities. Over the past few decades, there has been a significant increase in the prevalence of ASD, with estimates indicating higher rates than previously reported. This rise has sparked intensive research aimed at understanding the underlying factors contributing to ASD, including genetic predispositions, environmental influences, and their intricate interplay.

Traditional diagnostic methods for ASD rely heavily on behavioral observations and clinical evaluations. However, these approaches often face challenges in early identification, leading to delayed intervention and support for individuals on the autism spectrum. Furthermore, the heterogeneity of ASD presents a formidable obstacle in accurately diagnosing and predicting the condition based on singular criteria or observable traits.

The convergence of technological advancements, increased availability of vast and diverse datasets, and the emergence of sophisticated data analytics techniques has opened up unprecedented opportunities in the field of autism research. This convergence serves as the catalyst for your project, aiming to harness the potential of data-driven methodologies to analyze extensive datasets encompassing genetic, behavioral, environmental, and demographic information related to ASD.

The complexity of ASD necessitates a holistic approach, acknowledging the multifaceted nature of its etiology. Genetic predispositions coupled with environmental factors play pivotal roles in shaping the developmental trajectory of ASD. Therefore, your project seeks to delve into these complexities by employing advanced data analysis techniques to unravel patterns, correlations, and predictive markers within these diverse datasets.

The ultimate goal of your project is not only to develop predictive models for early detection but also to gain deeper insights into the underlying mechanisms driving ASD. By identifying potential predictive markers and understanding the interactions between genetic and environmental factors, this research aims to pave the way for more targeted and personalized interventions.

In essence, the background of your project underscores the pressing need for innovative approaches in ASD research and emphasizes the transformative potential of leveraging data-driven methodologies to enhance early detection, intervention strategies, and overall understanding of autism spectrum disorders.

1.2 Statement

"Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction" aims to pioneer a paradigm shift in the field of autism research by harnessing the power of comprehensive data analysis. This project seeks not only to develop predictive models for early detection but also to unravel the intricate interplay between genetic predispositions, behavioral patterns, and environmental influences contributing to Autism Spectrum Disorder (ASD). By leveraging innovative data-driven methodologies, the endeavor is to pave the way for more effective, personalized interventions and a deeper understanding of ASD, ultimately improving the lives of individuals on the autism spectrum and their families.

1.3 Motivation

The motivation behind "Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction" is deeply rooted in addressing critical challenges within the landscape of autism research and intervention.

Firstly, the increasing prevalence of Autism Spectrum Disorder (ASD) has highlighted the pressing need for early detection and personalized intervention strategies. Current diagnostic methods often encounter delays, hindering timely support and tailored interventions for individuals on the autism spectrum.

Secondly, the complexity and heterogeneity of ASD necessitate a more nuanced understanding of its etiology. Genetic predispositions, behavioral patterns, and environmental influences contribute to the development of ASD, forming an intricate web that traditional approaches struggle to decipher.

This project is driven by the transformative potential of data-driven methodologies. Leveraging advanced analytics and comprehensive datasets encompassing genetic, behavioral, environmental, and demographic information, the aim is to unearth subtle yet crucial patterns and predictive markers that could revolutionize early detection and intervention strategies for ASD.

The ultimate motivation lies in improving the quality of life for individuals on the autism spectrum and their families. By gaining deeper insights into the underlying mechanisms of ASD and developing predictive models, this research endeavors to pave the way for earlier identification, personalized interventions, and enhanced support services, thereby fostering better outcomes and empowering individuals with ASD to thrive in their unique ways.

1.4 Challenges

1. **Data Complexity and Integration:** One of the primary hurdles is the complexity of integrating diverse datasets. Collating information spanning genetics, behavioral patterns, environmental factors, and demographics poses challenges in data harmonization, normalization, and ensuring compatibility across various sources.

2. **Heterogeneity of ASD:** Autism Spectrum Disorder encompasses a wide spectrum of symptoms, behaviors, and genetic variations. This diversity poses a challenge in identifying consistent predictive markers or patterns, considering the individualistic nature of ASD manifestations.

3. **Data Quality and Accessibility:** Ensuring data quality and accessibility of comprehensive datasets can be a challenge. Variances in data collection methods, missing or incomplete data, and privacy concerns might limit the completeness and reliability of the datasets used for analysis.

4. **Interdisciplinary Collaboration:** Effective collaboration among multidisciplinary teams—spanning data scientists, clinicians, geneticists, psychologists, and more—is essential. Bridging the gap between these disciplines to interpret findings accurately and apply them practically can be a challenge.

5. **Ethical Considerations:** Balancing the utilization of sensitive, personal data with ethical guidelines and privacy concerns is crucial. Respecting the privacy and rights of individuals contributing to the datasets while conducting robust analyses is a significant challenge.

6. Translation to Clinical Practice: Developing predictive models is just the initial step. Translating these models into practical, usable tools for clinicians and healthcare providers involves addressing usability, validation, and integration into existing clinical workflows.

Addressing these challenges requires a comprehensive approach, involving robust data governance, advanced analytical techniques, collaboration across disciplines, adherence to ethical standards, and a keen focus on translating research outcomes into actionable insights for clinical and practical applications.

2. Planning & Requirements Specification

2.1 System Planning

1. Data Collection and Integration: Define data sources: Gather diverse datasets encompassing genetics, behavioral patterns, environmental factors, and demographics related to ASD. Ensure data quality: Implement protocols for data cleaning, normalization, and validation to enhance data quality. Establish data governance: Develop guidelines for data handling, privacy, and compliance with ethical standards.

2. Infrastructure and Tools: Set up robust infrastructure: Determine the computational resources required for data storage, processing, and analysis. Select analytical tools: Choose appropriate data analysis and machine learning algorithms suited for predictive modeling. Ensure scalability: Design the system to accommodate expanding datasets and evolving analytical needs.

3. Model Development and Validation: Model selection: Utilize machine learning techniques to develop predictive models considering the complexities of ASD. Cross-validation and testing: Validate models rigorously using cross-validation techniques and independent test datasets. Fine-tuning and optimization: Refine models for accuracy, sensitivity, and specificity while addressing overfitting or biases.

4. Interdisciplinary Collaboration:Foster collaboration: Facilitate communication and collaboration among data scientists, clinicians, researchers, and stakeholders.Interpretation and translation: Ensure clear communication of findings to clinicians and experts for validation and practical implementation.

5. Ethical Considerations and Compliance:Data privacy and security: Implement stringent measures to safeguard sensitive personal information and comply with privacy regulations.Ethical review: Seek ethical approvals and ensure adherence to ethical guidelines in data handling and analysis.

6. Implementation and User Interface:Practical application: Develop user-friendly interfaces or tools to facilitate the utilization of predictive models by clinicians or healthcare providers.Integration with existing systems: Ensure seamless integration into existing clinical workflows or diagnostic protocols.

7. Continuous Improvement and Evaluation:Monitor and update: Establish protocols for regular model evaluation, updating algorithms, and incorporating new data.Feedback loop: Encourage feedback from clinicians and stakeholders to refine and improve the system continually.A well-structured system plan addresses technical, ethical, and practical considerations, ensuring that the developed models are not only accurate but also usable and ethically sound for practical applications in the diagnosis and management of ASD.

2.2 Requirements

- Functional Requirements:

1. Data Acquisition and Integration:

Data Collection: Ability to gather and aggregate diverse datasets related to genetics, behavior, environment, and demographics associated with ASD.

Data Preprocessing: Capability to clean, normalize, and integrate disparate data sources for analysis.

- 2.Data Analysis and Modeling:

Predictive Modeling: Implement machine learning algorithms to develop predictive models for ASD detection and risk assessment.

Feature Selection: Ability to identify and prioritize relevant features or variables contributing to ASD prediction.

Model Evaluation: Conduct rigorous validation, including cross-validation techniques, to assess model performance and accuracy.

3. Scalability and Performance:

Scalable Infrastructure: Ensure the system can handle large volumes of data and scale to accommodate future expansions.

Computational Efficiency: Optimize algorithms and processing methods for efficient analysis of complex datasets.

4. User Interface and Accessibility:

User-Friendly Interface: Develop an intuitive interface for clinicians or researchers to interact with the system and interpret results.

Accessibility: Ensure accessibility compliance for users with diverse needs, considering usability standards.

5. Security and Privacy Measures:

Data Security: Implement robust security protocols to safeguard sensitive personal information and comply with privacy regulations.

Ethical Guidelines: Incorporate measures to ensure ethical handling of data and adherence to ethical guidelines and regulations.

6. Integration and Compatibility:

Compatibility with Existing Systems: Ensure compatibility and integration capabilities with existing healthcare or diagnostic systems used by clinicians.

Interoperability: Enable data exchange and interoperability with other relevant systems or databases.

7. Continuous Monitoring and Maintenance:

Monitoring and Updating: Establish mechanisms for continuous monitoring of model performance and regular updates to algorithms based on new data or research findings.

Maintenance and Support: Provide ongoing technical support and maintenance for the system's functionalities.

3. System Design

1. Data Collection and Integration Layer:

- Data Sources: Interface with diverse sources such as research databases, healthcare records, genetic repositories, and environmental datasets.
- Data Ingestion: Develop mechanisms for collecting, cleansing, and harmonizing data from different sources.
- Data Storage: Utilize secure and scalable databases or data warehouses to store integrated datasets.

2. Data Processing and Analysis Layer:

- Data Preprocessing: Implement tools and pipelines for cleaning, normalization, and feature extraction.
- Machine Learning Algorithms: Employ various algorithms for predictive modeling, including supervised learning methods suitable for ASD prediction.
- Model Training and Validation: Perform training, validation, and evaluation of models using cross-validation techniques and performance metrics.

3. User Interface and Interaction Layer:

- Dashboard or Interface: Design a user-friendly interface for clinicians, researchers, and authorized users to interact with the system.
- Visualization Tools: Incorporate visualizations and summary statistics to aid interpretation of model outputs and insights.
- Accessibility Features: Ensure accessibility compliance for users with diverse needs.

4. Security and Compliance Layer:

- Data Security Measures: Implement encryption, access controls, and protocols to safeguard sensitive information.
- Privacy Compliance: Ensure compliance with relevant privacy regulations and ethical guidelines in data handling and storage.

5. Scalability and Performance Layer:

- Infrastructure Scaling: Architect the system for scalability to handle growing datasets and increased computational demands.
- Optimization Techniques: Employ optimization methods to enhance computational efficiency and model performance.

6. Integration and Compatibility Layer:

- APIs and Interfaces: Develop APIs or integration points for interoperability with existing healthcare systems or databases.
- Standards Compliance: Ensure adherence to industry standards for data exchange and interoperability.

7. Monitoring and Maintenance Layer:

- Continuous Monitoring: Implement monitoring tools to track system performance, model accuracy, and data quality.
- Update and Maintenance: Establish procedures for regular updates, bug fixes, and maintenance of the system components.

4.Implementation

The implementation of "Seeing Beyond the Spectrum: A Data-Driven Approach to Autism Prediction" involves translating the system design into a functional and operational platform. Here's a roadmap for implementation:

1. Data Collection and Preparation:

- Identify and gather diverse datasets related to genetics, behavior, environment, and demographics associated with ASD.
- Clean, preprocess, and integrate these datasets into a unified format suitable for analysis.

2. Infrastructure Setup:

- Deploy the necessary computational infrastructure, including servers, databases, and storage solutions, considering scalability and security requirements.
- Install and configure software frameworks and tools for data processing, analysis, and modeling.

3. Algorithm Development and Model Building:

- Develop and implement machine learning algorithms suitable for predictive modeling of ASD based on the integrated datasets.
- Train, validate, and fine-tune the models using appropriate techniques and validation methodologies.

4. User Interface Development:

- Design and develop a user interface or dashboard that enables easy interaction and visualization of model outputs for clinicians and researchers.
- Incorporate user feedback to enhance usability and accessibility features.

5. Security and Compliance Implementation:

- Implement robust security measures to protect sensitive data, including encryption, access controls, and compliance with privacy regulations.
- Ensure ethical handling of data and adherence to relevant guidelines and standards.

6. Integration and Testing:

- Integrate the developed system with existing healthcare systems or databases through APIs or interfaces.
- Conduct thorough testing, including functionality, usability, and performance testing, to identify and rectify any issues.

7. Training and Adoption:

- Provide training sessions and documentation for users, including clinicians and researchers, on how to effectively utilize the system.
- Encourage adoption and gather feedback to iteratively improve the system based on user experiences.

8. Continuous Monitoring and Maintenance:

- Implement monitoring tools to track system performance, data quality, and model accuracy in real-time.
- Establish protocols for regular updates, bug fixes, and ongoing maintenance to ensure the system's optimal functioning.

5. Results & Discussion

1. **Model Performance:** Present the performance metrics of the developed predictive models, including accuracy, sensitivity, specificity, and area under the curve (AUC), showcasing their effectiveness in predicting ASD risk.
2. **Identified Predictive Markers:** Highlight specific genetic, behavioral, or environmental factors identified by the models as influential in ASD prediction, shedding light on potential early indicators or risk factors.
3. **Validation and Robustness:** Discuss the robustness of the models through cross-validation, testing on independent datasets, and handling diverse populations, demonstrating their reliability and generalizability.

Discussion:

1. **Clinical Implications:** Interpret the significance of the identified predictive markers and their potential impact on early detection and intervention strategies for ASD. Discuss how these findings could inform clinical practices and facilitate early support for individuals on the autism spectrum.
2. **Insights into ASD Etiology:** Explore the insights gained from the data-driven approach, elucidating the complex interplay between genetic predispositions and environmental influences in the development of ASD. Discuss how these insights contribute to a deeper understanding of ASD etiology.
3. **Challenges and Future Directions:** Address any limitations encountered during the study, such as data constraints or model complexities. Propose avenues for further research, including refining models, exploring additional data sources, or investigating specific subtypes of ASD.
4. **Ethical Considerations:** Discuss ethical considerations surrounding data usage, privacy, and the responsible application of predictive models in clinical settings. Emphasize the importance of ethical guidelines in implementing these models.
5. **Translation to Clinical Practice:** Outline practical steps for integrating the developed models into clinical workflows, ensuring usability, interpretability, and acceptance among healthcare professionals.

6. Conclusion and Future Work

- **Conclusion:**

1. **Summary of Findings:** Summarize the main outcomes, including the effectiveness of predictive models, identified markers, and their implications for ASD prediction and understanding.

2. **Impact and Significance:** Highlight the significance of the study in advancing the field of autism research, emphasizing its potential for early detection, personalized intervention, and deeper insights into ASD etiology.

3. **Clinical Relevance:** Emphasize the practical implications of the study for clinicians, healthcare providers, and individuals affected by ASD, focusing on how the developed models can enhance early support and tailored interventions.

- **Future Work:**

1. **Refinement of Models:** Discuss avenues for improving the predictive models by incorporating additional data sources, refining algorithms, and enhancing model interpretability and usability.

2. **Longitudinal Studies:** Propose conducting longitudinal studies to track the progression of ASD and validate the predictive markers identified, enabling a deeper understanding of the developmental trajectory.

3. **Subtype Analysis:** Explore further analysis to understand specific subtypes or variations within the autism spectrum, recognizing the heterogeneity of ASD and its implications for predictive modeling.

4. **Collaboration and Validation:** Advocate for collaboration with a broader network of researchers and clinicians for validation studies across diverse populations, ensuring the robustness and generalizability of the models.

5. Ethical and Social Considerations: Stress the importance of continued adherence to ethical guidelines and responsible data usage while addressing social implications and the impact on individuals and communities.

6. Integration into Healthcare Systems: Focus on the practical integration of predictive models into healthcare systems, emphasizing user training, system integration, and usability in clinical settings.

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