Background Research: AI-Based Early Detection of Type 2 Diabetes Using Lifestyle and Genomic Data

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1. Introduction

Type 2 Diabetes Mellitus (T2DM) is a chronic metabolic disorder characterized by insulin resistance and progressive β-cell dysfunction, leading to sustained hyperglycemia. With prevalence rising globally due to obesity, sedentary behaviour, and poor dietary patterns, T2DM has become a major public health concern. Traditional detection methods often identify individuals only after the onset of significant complications. Integrating lifestyle variables and genomic markers with artificial intelligence (AI) methods offers a promising strategy for early identification of high-risk individuals, enabling preventive interventions and reducing long term disease burden.

2. Commonly Used Lifestyle Variables

Lifestyle data provide critical insights into modifiable risk factors for T2DM. Large scale datasets such as the National Health and Nutrition Examination Survey (NHANES) and the UK Biobank are widely used in predictive modelling. These datasets capture behavioural and clinical markers that strongly influence diabetes risk.

1. **Body Mass Index (BMI):** A well-established predictor, with obesity significantly elevating T2DM risk (Mainous et al., 2016).
2. **Waist:** Indicators of central obesity, more strongly associated with insulin resistance than BMI alone (Zhang et al., 2021).
3. **Physical activity:** Measured via questionnaires or accelerometer data, with inactivity independently raising T2DM risk (Wang, 2025).
4. **Dietary intake:** Food frequency questionnaires assess intake of refined carbohydrates, fats and fiber, highlighting correlations between diet quality and metabolic risk (Jansink et al., 2012).
5. **Smoking and alcohol use:** Behavioural factors that worsen metabolic health (Chen et al., 2023).
6. **Metabolic biomarkers:** Variables such as fasting glucose, HbA1c, and lipid levels, often included alongside lifestyle indicators (Jarvandi et al., 2012).

Together, these variables capture the environmental and behavioural factors driving T2DM, and their integration into AI models enhances predictive accuracy.

3. Genomic Risk Markers

Genomic data illuminate the heritable components of T2DM risk. Repositories such as dbGaP, DIAGRAM, UK Biobank, and FinnGen have been instrumental in identifying genetic variants associated with T2DM susceptibility (Enya et al., 2014; Qi et al., 2010; Grarup et al., 2018).

Key genetic markers include:

1. **TCF7L2:** The strongest and most consistently replicated locus, influencing β-cell function and hepatic gluconeogenesis via the Wnt signaling pathway (Pitkänen et al., 2016).
2. **SLC30A8:** Encodes a zinc transporter critical for insulin granule formation, variants impair insulin synthesis and secretion (Pecioska et al., 2010).
3. **FTO:** Associated with obesity related T2DM risk by influencing appetite regulation and adiposity (Pecioska et al., 2010).
4. **KCNJ11:** Encodes a potassium channel subunit in β-cells, SNPs affect glucose stimulated insulin release (Pecioska et al., 2010).
5. **HHEX:** Linked to pancreatic islet development and glucose homeostasis, influencing insulin production capacity (Pecioska et al., 2010).

These markers highlight the polygenic nature of T2DM, underscoring the need to integrate genetic data with lifestyle variables for more personalized risk assessment.

4. Challenges

Despite its promise, the integration of lifestyle and genomic data for AI based T2DM detection presents several challenges:

1. **Privacy and Security:** Genomic and health data are highly sensitive. Breaches can undermine patient trust, while existing regulations like HIPAA may not fully address the complexities of AI driven data use (Gore & Dove, 2024).
2. **Ethical Concerns:** Underrepresentation of minority groups in training datasets risks producing biased algorithms that fail to generalize across populations (Ingol et al., 2020). Informed consent remains challenging due to the complexity of AI applications.
3. **Class Imbalance:** Datasets often overrepresent European populations while underrepresenting minorities, leading to models that achieve high accuracy overall but perform poorly in specific subgroups (Lim et al., 2023).
4. **Bias in AI Models:** Algorithmic design can inadvertently reinforce existing health disparities if socioeconomic and cultural contexts are not considered (Gore & Dove, 2024).
5. **Technical Barriers:**
    - Missing data reduces model robustness and requires imputation, which may introduce bias (Mumpuningtias et al., 2023).
    - Multimodal integration of lifestyle and genomic data requires harmonization across heterogeneous formats (Ingol et al., 2020).
    - Model interpretability remains a hurdle, particularly with deep learning approaches, reducing clinician trust in AI outputs (Gore & Dove, 2024).

Addressing these challenges is critical to ensure AI systems are equitable, secure, and clinically reliable.

5. Conclusion

AI based early detection of T2DM has the potential to transform diabetes prevention by integrating lifestyle and genomic data into predictive models. While significant progress has been made, challenges in data privacy, ethical representation, class imbalance, and technical implementation must be resolved to realize this potential. Future work should focus on developing transparent, fair, and secure AI systems that benefit diverse populations, advancing precision medicine in diabetes care.

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