Expanding the Guthrie Test: The Implications of Adding Predictive Diagnostics Using Neonatal miRNA Profiling for Cardiovascular Disease Risk Assessment

OBSERVATIONS

The Guthrie test, which is designed to screen newborns for 27 inherited disease that require immediate intervention as a cornerstone of preventive healthcare.

The Dutch Ministery of Health has emphasized the importance of preventive healthcare in aleviating the national healthcare costs

Cardiovascular disease (CVD) remains a leading cause of mortality, attributing to 27% of all mortalities in the





Cardiovascular disease-related total costs per capita, adjusted for price differentials (Luengo-Fernández et al., 2023)

Netherlands

Recent advances made in biomedical research highlight the heritable nature of one's risk for CVD risk, with deregulated microRNA playing a key role

DEDUCTION & OBJECTIVES

Incorporating miRNA biomarkers into neonatal screening aligns with the Dutch preventive healthcare strategy which can potentially identify individuals for an elevated risk for cardiovascular disease.

"Our objective is to propose an expansion of the Guthrie test for neonatal cardiovascular disease screening by utilizing miRNA biomarkers to identify increased risks"

Neonatal MicroRNA Profiling as a Novel Predictive Diagnostic Tool

WHAT: MicroRNAs (miRNAs) are small DNA transcripts that do not code for proteins – the building blocks of our body – but rather act as inhibitors for the production of proteins; a bodily process known as RNA interference (RNAi)

miRNA are seen to play determining roles in cardiovascular health that revolve around regulating processes like cardiac muscle development & remodeling, fibrosis, and apoptosis.

Dysregulated miRNAs like miR–1, miR–21, miR–208, miR–409, and miR–133 are specially implicated in cardiovascular disease by research

HOW: Neonatal miRNA profiles may predict future cardiovascular disease risks due to their heritable and stable nature if quantified by PCR and detected through extraction methods such as column-based purification or precipitation methods

The Guthrie test, known for detecting diseases in newborns, could incorporate these miRNA biomarkers for cardiovascular risk assessment by deploying the abovementioned methods

CHALLENGES AHEAD include validating miRNA models in humans, correlating & establishing miRNA levels with disease risk, and conducting stringent clinical trials to validate false negatives and postives

Attitudes and Interests

Legal Implications

Bodily integrity

Privacy

Safeguarded by the GDPR Parental consent Art. 6 (1) (a) jo. 8 (1) **GDPR** No infringement

Physical and mental integrity

A justifiable infringement? Discussion amongst scholars

Applied to this proposal Individual vs parental autonomy

Costs and Benefits

Benefits would arise from the prevented loss of productivity and decreased treatment costs

N=~77(76)



I would want my parents to do this test on me

Would you want this test to be performed on your children?

I would be motivated to make lifestyle changes if I were aware of a high likelihood of developing cardiovascular disease

WHY

Early awareness, immediate action, early habit learning, frequent screening, taking precautions

However

Results might lead to an unnecessary psychological burden for both the parent and neonate & possibly highten the level of anxiety and fear

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Costs are composed of the **testing procedure**, **test kits &** laboratory analysis



Costs per year per child for lab analysis of the Guthrie test accounting for 76% of total costs

In the years of 2021, 2022 and 2023 diseases were added to the Guthrie programme

On this basis we estimate the costs to increase by approximately €6,76 per child

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