Project Proposals for Doctoral Researcher Positions 2025

ID07: **Data-based outlier detection and risk validation for multi-class prediction in newborn screening (Vincent Heuveline, Stefan Kölker)**
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Newborn screening plays a crucial role in early detection of rare, inherited metabolic diseases, facilitating timely interventions and improved outcomes for affected infants and families. However, the rarity of these diseases results in highly imbalanced datasets with few positive screening results, which are often perceived as outliers. This project seeks to address these challenges by developing advanced outlier detection algorithms capable of identifying positive screening results accurately.

Additionally, certain conditions like methylmalonic acidemia, propionic acidemia, homocystinurias, remethylation disorders, and infantile vitamin B12 deficiency face issues with false-positive screening results. The project aims to develop a multi-class machine learning approach to effectively classify newborns with these conditions, thereby reducing false positives and minimizing unnecessary treatments.

Integrating machine learning into clinical decision-making requires transparency and risk assessment. Hence, the project will focus on developing explainable AI methods to interpret algorithm decisions and uncover underlying metabolic patterns. Furthermore, a data-driven risk management model will be developed to provide clinicians with predictive risk values associated with disease likelihood.

In summary, this project aims to advance state-of-the-art methods for data-driven outlier detection and risk prediction in newborn screening, ultimately improving diagnostic accuracy and reducing the impact of false positives on newborns and their families.

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