A Causal Perspective on Challenges for AI in Precision Medicine

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TL;DR

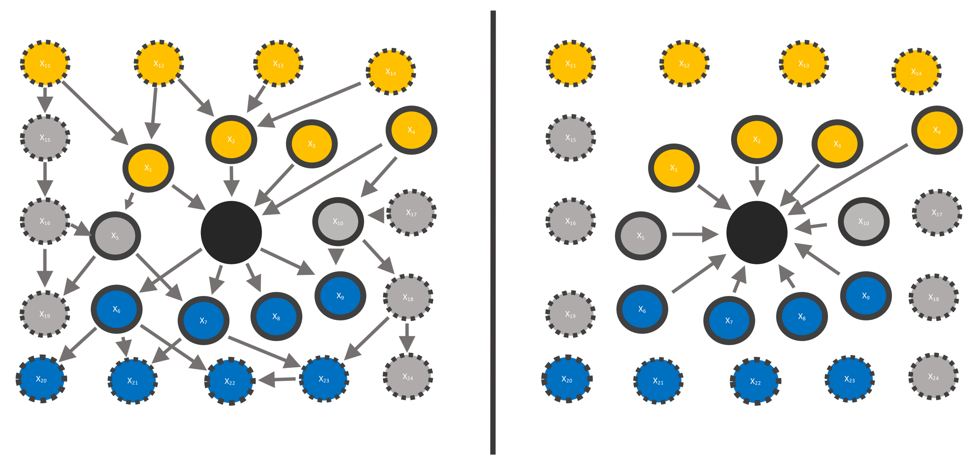
- We provide a classification of tasks in precision medicine.
- Many of them require answering causal questions.
- Many popular AI methods are not designed to consider causality.
- We provide causal interpretation rules.

Classification of Tasks

Non-Causal	Causal
- diagnosis	- risk factor identification
- genotyping/phenotyping	- treatment recommendation
- disease outbreak/ progression prediction	- genotype to phenotype mapping
	- treatment development
	- treatment effect prediction
	- understanding of disease mechanism

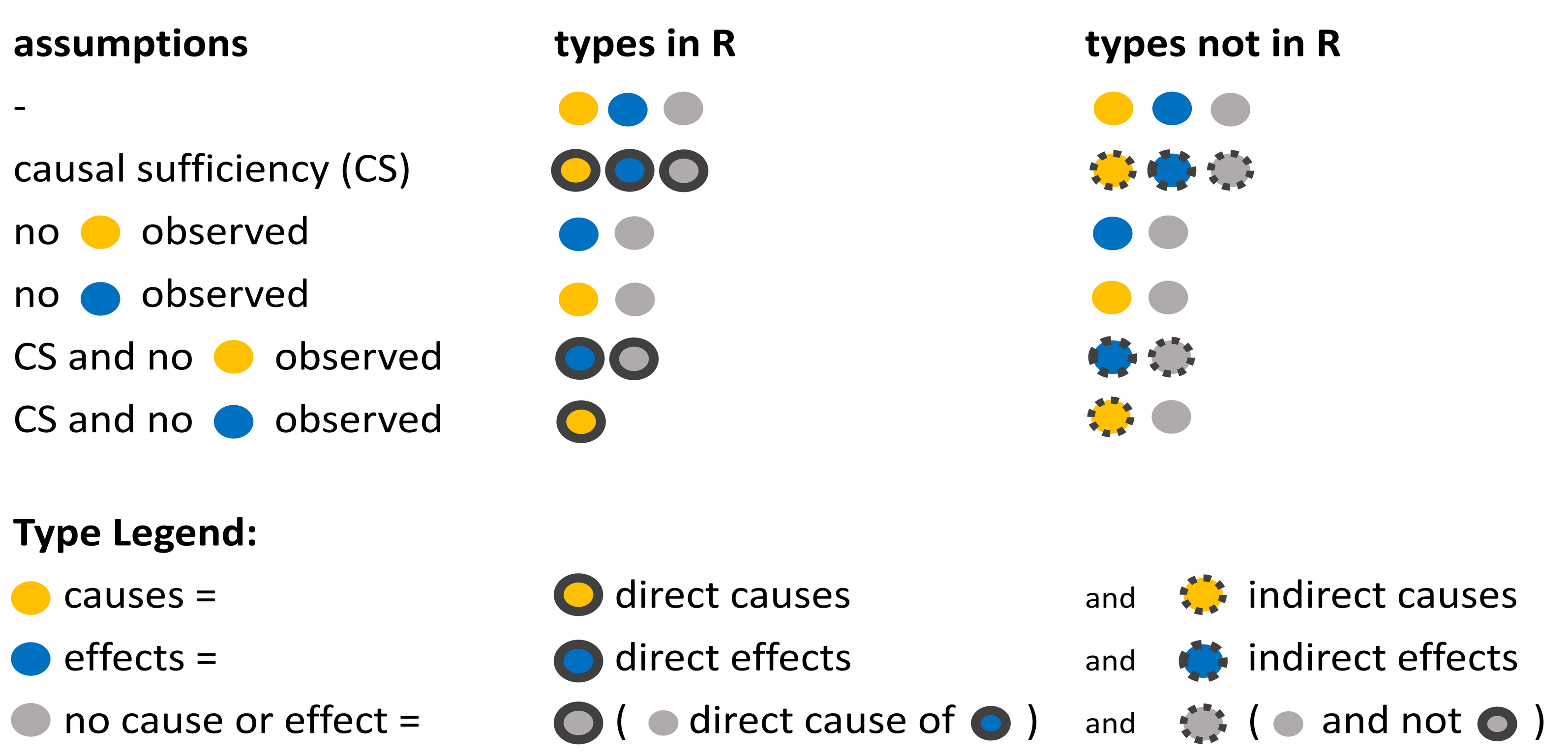
Prediction with AI

Supervised Machine Learning: Learn function $\hat{f} : X \to Y$ such that empirical risk $E[\mathcal{L}(\hat{f}(X), Y)]$ is minimal.
Minimal Bayes optimal set of features: Minimal set of features R such that all other variables $X_i \in \Omega' \backslash R$ become independent of Y ($Y \perp X_i | R$). This so-called *Markov Blanket* may not only include variables that cause Y. A change of $\hat{Y}$ (model) as a result of a change in X may therefore not correspond to a change in Y (real world).



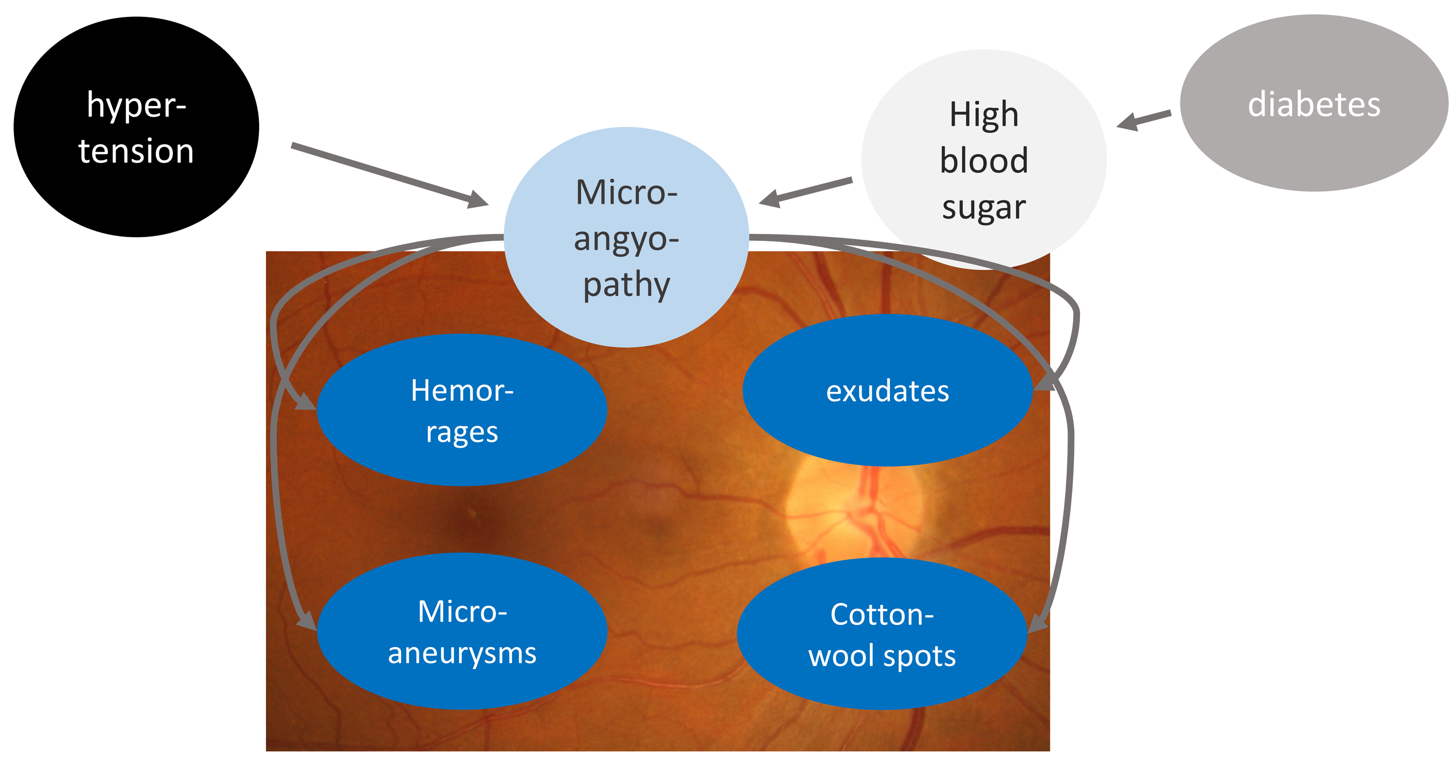
Left: Underlying causal graph. **Right:** Dependencies as perceived by the AI model.
Given a target Y (black) not only causes (yellow), but also effects (blue) and context variables (grey) are in R (here thick black outline).

Causal Interpretation Rules

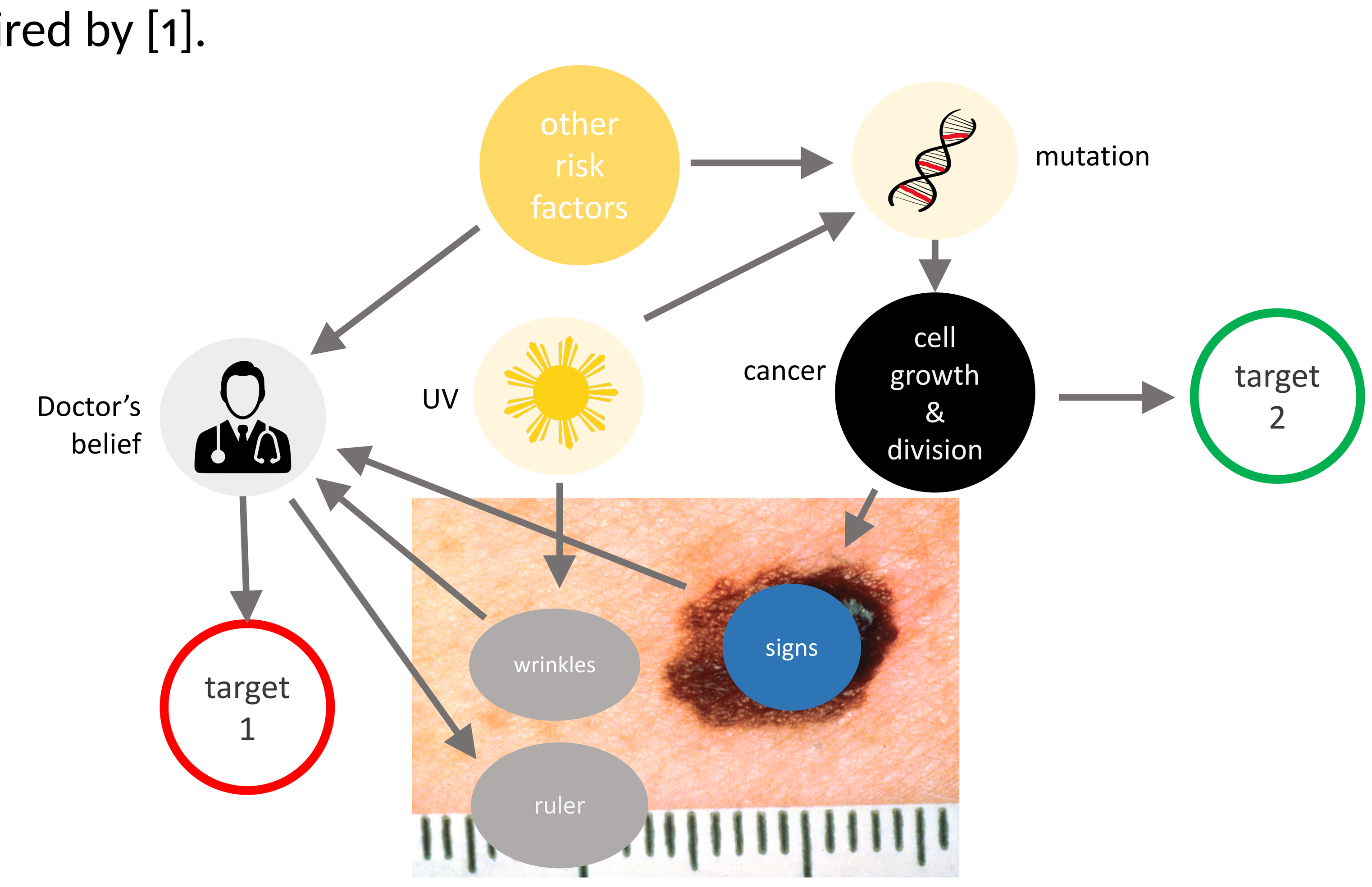


Based on [5]. *Causal sufficiency:* No variable that causes more than one observed variable is unobserved.

Examples



Collider/Differential diagnosis: When assessing the presence of hypertensive retinopathy, knowledge about whether the patient has diabetes helps distinguish the condition from diabetic retinopathy. Inspired by [1].



Hidden Confounding/Associated Signs: UV radiation causes both the ageing of skin (wrinkles) and genetic mutation that leads to skin cancer. Wrinkles may therefore be included in a skin cancer diagnosis model.
Target dependence: A medical doctor may place a ruler in photographs of tumors to indicate their size. Depending on whether our label reflects the doctors belief (target 1) or the actual condition (target 2) a model may rely on the presence of a ruler for its diagnosis. Inspired by [2, 3]. Photograph taken from [4].

References

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