

# Causal Discovery for Risk Factors and Disease

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Data

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Graph Results

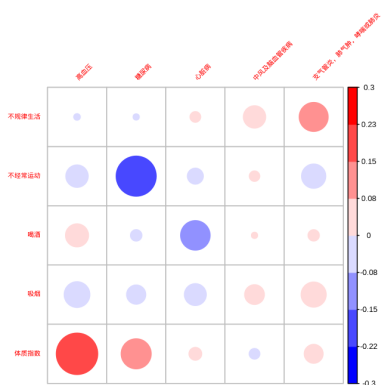
Comparison Between Methods

Discussion

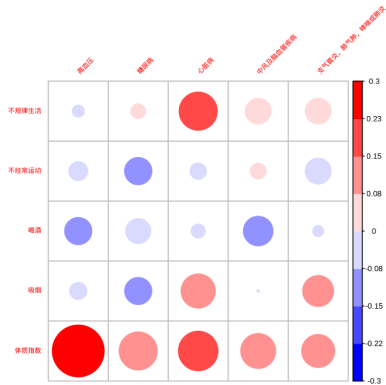
- **Background:** A cross-sectional dataset aiming to study the ageing process was collected by several hospitals using questionnaires and fitness tests, including over 15000 people of different ages.
- **Questions:**
  - ① Is there any causal relationship between risk factors and noncommunicable diseases?
  - ② If such causation exists, how it changes through the ageing process?
- **Risk Factors:** Drinking, Smoking, Lack of exercise, Irregular lifestyle, Body Mass Index(BMI).
- **Disease:** Hypertension, Heart disease, Diabetes, Apoplexy, Respiratory system diseases

# Brief Review on EDA

## Overall Correlation



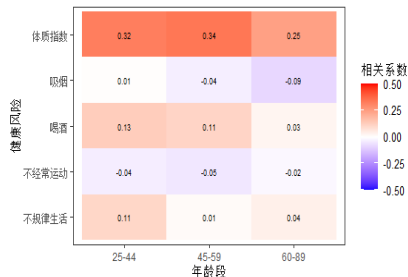
(a) Male



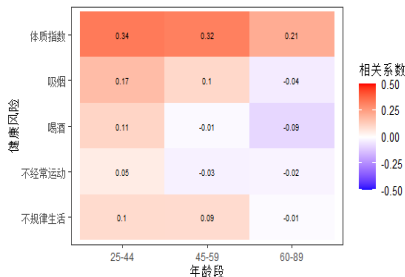
(b) Female

# Brief Review on EDA

## Correlation with Hypertension across different ages



(a) Male



(b) Female

# Directed Graphical Causal Model

## Introduction

**Idea:** Using directed acyclic graphs(DAG) to model the joint distribution of all variables and discover the causal relationship between them. We will write the joint distribution as

$$\Pr(X_1, \dots, X_m) = \prod_{i=1}^m \Pr(X_i | pa(X_i))$$

**Example:**

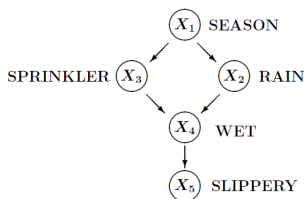


Figure 1: A Bayesian network representing causal influences among five variables.

# Directed Graphical Causal Model

## Introduction

### Assumptions

**Local Markov Condition:** Every  $X_i$  in DAG is independent of its non-descendants conditional on its parents(d-separation property).

**Faithfulness Assumption:** DAG demonstrates all conditional independence relations for the population probability distribution.

### Troubles caused by Assumptions

- Different DAGs may share same Markov conditions. Such collections for those DAGs are called **Markov Equivalent Class**.
- By Faithfulness, **confounders** are not allowed in the model.

# Structure Learning Methods

- Constrained-Based Methods
- Score-Based Methods
- Hybrid Methods
- Functional Causal Models



# Constrained-Based

## Ideas:

- ① Construct undirected graph by independence tests and conditional independence tests.
- ② Directions can be constructed by v-structure.

## Algorithms:

- PC algorithm (Spirtes, 2001; Implemented by Colombo, 2014)
- iAMB algorithm (Yaramakala, 2005)
- FCI algorithm (Spirtes, 2001)

## Advantages:

- Convenient interpretation for causal relationships.

## Disadvantages:

- Multiple Testing.
- May exist undirected edges.

## **Ideas:**

Each candidate DAG is assigned a network score reflecting its goodness of fit, which the algorithm then attempts to maximise.

## **Scores:**

- AIC, BIC
- NML, fNML, qNML (Shtarkov, 1987; Silander et al., 2008; Silander et al., 2018)

## **Advantages:**

- An optimized DAG which fits the distribution of the data will be produced.

## **Disadvantages:**

- Hard to give explanation on causal relationship.

## Ideas:

- 1 Use constrained-based methods to find the skeleton(undirected graph) of the Bayesian network.
- 2 Use score-based methods to direct the edges.

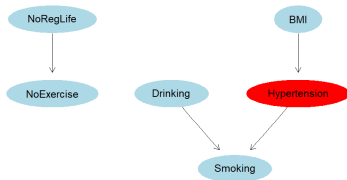
## Algorithm:

- MMHC (Tsamardinos et al.,2006)
- Rsmx2 (Scutari et al. 2014)
- H2PC (M. Gasse et al.2014)
- FRITL (Chen et al. 2021)

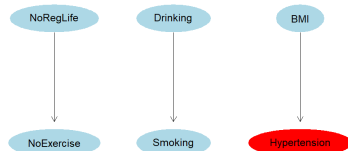
# Our Model

- Hybrid method is used for formal modeling.
- Constrained-based and score-based methods are used for comparison.
- R package bnlearn is used for modeling.

# Risk Factors v.s. Hypertension



(a) Male



(b) Female

Figure: Risk Factors and Hypertension across all age groups

# Risk Factors v.s. Hypertension

## Different Age Groups

Figure: Male



(a) 25-44



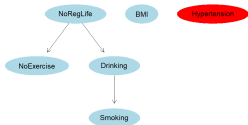
(b) 45-59



(c) 60-89



Figure: Female



(a) 25-44



(b) 45-59

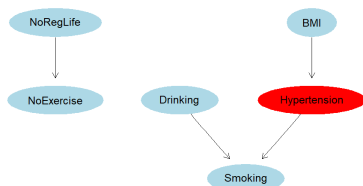


(c) 60-89

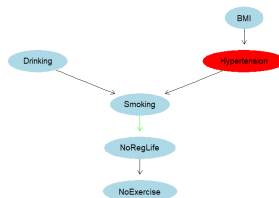


# Comparison Between Methods

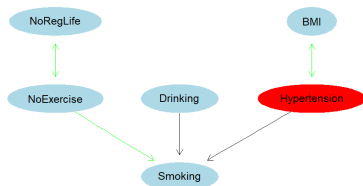
## Visual Comparison



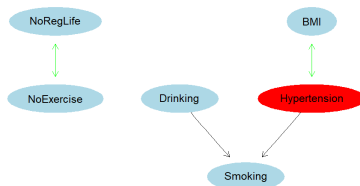
(a) Hybrid



(b) Score-based, qnml



(c) Constrained-based, PC,  $\alpha = 0.05$



(d) Constrained-based, PC,  $\alpha = 0.001$

# Comparison Between Methods

## Score Comparison

5-fold cross-validation was used to compute the log-likelihood loss.

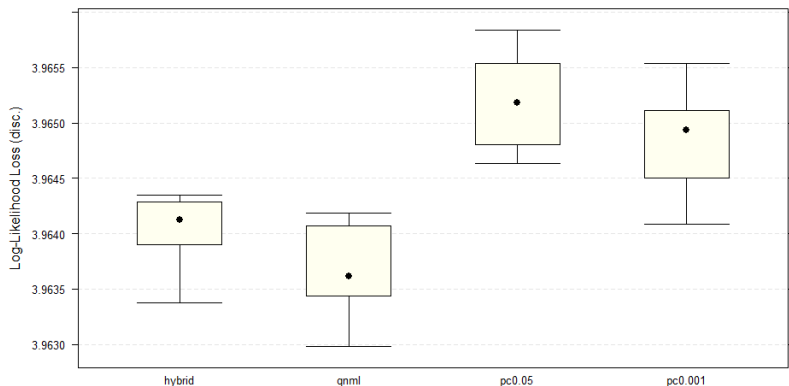


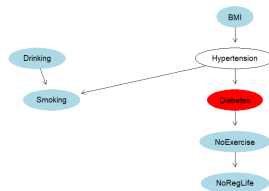
Figure: Log-likelihood Loss Comparison



# Confounder



(a) Diabetes without confounder



(b) Diabetes with Hypertension as confounder

- Different patterns were shown between genders and age groups.
- BMI may be a causal for hypertension, which meets our common sense.
- For male, hypertension may have influence on smoking. However, people may care more about the reverse direction. At least, our result showed that they have relationship between each other.

- Confounders should be considered, using FCI to fit such models.
- We can not figure out the influence is positive or negative by causal discovery. Further works may be focused on this subject.

# The End

Questions? Comments?