Assignment 1: Compute Blood Types

AI-2 Systems Project (Summer Semester 2023) Jan Frederik Schaefer

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Topic:	Bayesian networks
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1 Task summary

Using Bayesian networks, compute the probability that someone has a particular blood type given some test results for the blood types of their relatives. The assignment repository [AR] contains example problems for you to solve.

Didactic objectives

- 1. Learn how to model a problem as a Bayesian Network,
- 2. gain experience working with graphs,
- 3. use complex conditional probability tables,
- 4. find and use a suitable library for Bayesian inference (many exist and finding good libraries is an important skill),
- 5. get to know the JSON format (in case you haven't used it before).

Prerequisites and useful methods

- 1. Basics of representing graphs in code and working with them,
- 2. Bayesian networks (taught in the AI lecture).

2 Background: The ABO blood group system

The **ABO** blood group system [ABO] is used to describe the presence/absence of A and B antigens on the red blood cells in humans. We distinguish 4 different (ABO) blood types: A (if only the A antigens are present), B (if only the B antigens are present), AB

(if both are present) and \mathbf{O} (if neither are present). Knowing the blood type is in particular important for blood transfusions as a transfusion with an incompatible blood type can be lethal. Another important antigen for blood transfusions is Rh, which we will ignore in this assignment.

2.1 Alleles

The ABO blood type is determined by the **ABO gene**, which comes in three variants, called **alleles**: the **A allele**, the **B allele** and the **O allele**. Humans have chromosome pairs and therefore two versions of the **ABO gene** – one taken from each parent. The A allele and B allele are co-dominant: if at least one ABO gene has the A allele, the person has A antigens, and if at least one ABO gene has the B allele, the person has B antigens. Humans with both the A and B allele have both antigens and therefore blood type AB and only humans with two O alleles have neither antigens and therefore blood type O. In the following, we will sometimes write XY to denote that someone has one X allele and one Y allele.

2.2 Inheritance

A child randomly gets one allele from each parent. For example, if the father has an A allele and an O allele (short AO) and the mother has an A allele and a B allele (short AB), then the child could have one of the following allele combinations:

- 1. AA (blood type A)
- 2. OA (blood type A)
- 3. AB (blood type AB)
- 4. OB (blood type B)

Each of the four combinations is equally likely.

3 Detailed problem description

The assignment repository [AR] contains problem files and example solutions encoded in the JSON format. Listing 1 shows an example problem file. The family-tree field specifies the relationships between family members: Omar (subject) is the father (relation) of Rory (object) and Samantha is the mother of Rory. Every family member is uniquely identified by their name. The country field specifies the country where the family lives. This is relevant because the distribution of alleles depends on the country (see Section 3.2 for more details).

```
{
    "family-tree": [
        {"relation": "father-of", "subject": "Omar", "object": "Rory"},
        {"relation": "mother-of", "subject": "Samantha", "object": "Rory"}
],
    "country": "North<sub>u</sub>Wumponia",
    "test-results": [{"type": "bloodtype-test", "person": "Samantha", "result": "O"}],
    "queries": [{"type": "bloodtype", "person": "Rory"}]
}
```

Listing 1: Example problem file.

Country	A allele	B allele	O allele
North Wumponia	25%	10%	65%
East Wumponia	35%	5~%	60%

Figure 1: Prior probabilities that the ABO gene has a particular allele by country.

If no country is specified, every country is equally likely. The test-results field lists the results of different tests. In this case, we know that Samantha has blood type O. At last, the queries field specifies what we should query for. In this case, we are interested in the blood type of Rory. Section 3.4 describes the solution format.

3.1 Details on the family tree specification

As mentioned above, the family tree is described via relationhips between people. The following types of relationship are used: mother-of, father-of, sister-of, brother-of. Note that some relationships may only be implicit. For example, if A is a parent of B, and C is a sibling of B, then clearly A is also a parent of C – even though it might not be explicitly stated. We also assume that everyone has exactly one male and one female parent.

3.2 Distribution of ABO alleles

To solve the assignment, you need to know the distribution of the ABO alleles in the population, which actually varies significantly across the world. In this assignment, we use imaginary countries with different allele distributions. Figure 1 shows the corresponding distributions. We also assume that the alleles of a person are independent, so e.g. the prior probability of having blood type AB in North Wumponia is $2 \cdot 25\% \cdot 10\% = 5\%$ (both the AB and the BA allele pair result in the AB blood type).

3.3 Details on test results

You get some information about the family from tests. For example, the bloodtype-test tells you the blood type of a family member (see Listing 1 for an example). We assume that these tests are accurate. But there are also *cheap* tests (prefixed with cheap-). For cheap tests, there is a 30% chance that the test center instead returned the test results from a different person (in the same country).

3.4 Solution format

The solution to a problem file should also be stored in a JSON file. It should list answers to all the queries. For example, the solution to the example problem in Listing 1 should be represented as

```
[
    {
        "type": "bloodtype",
        "person": "Rory",
        "distribution": {
            "O": 0.65,
            "A": 0.25,
            "B": 0.1,
            "AB": 0.0
        }
    }
]
```

meaning that e.g. Rory has blood type O with a probability of 65%.

The assignment repository contains more example problems and solutions that you can use for comparison. Furthermore, it provides a script for comparing your solutions to the example problem with the provided solutions.

4 What to submit

Your solution should be submitted to your team's repository. It should contain:

- 1. all your code,
- 2. a README file explaining how to run your code to solve other problem files (including how to install dependencies),
- 3. a brief summary of how you solved the problem either as a PDF file (≈ 1 page) or as part of your README (in particular, please describe what random variables you used in the Bayesian network),
- 4. a file solution-[LETTER]-[NUMBER].json for every problem-[LETTER]-[NUMBER].json that you managed to solve.

5 A few tips

- 1. The problems vary in difficulty (see Section 6). It might be a good idea to start with variations that work well for you. In particular, problem-a-*.json problems have a minimal family (father, mother, child). If you have difficulties getting started, you could try to solve such a problem on paper first and then try to use a library for Bayesian networks to do the computations for you.
- 2. Part of the problem is to find a suitable library for Bayesian inference (inference through variable elimination should be efficient enough). It might make sense to try out the library with a minimal example before integrating it with the rest of the code to make sure it actually works as expected.
- 3. Getting the encoding as a Bayesian network right is somewhat tricky. Make sure that you correctly distinguish between the ABO allele pairs of a person and their observed blood type (state vs evidence variables).
- Computing the family tree from the relations is error prone (at least for the more difficult problems). It might help with debugging if visualize the generated family trees.
- 5. For the more difficult problems, you might have to add people to the family tree that were not explicitly mentioned in the problem file.

Problems	Family relations	Test types	Countries	
problem-a-*.json	mother-of	hla a du	North Wumponia	
	father-of	bloodtype-test		
problem-b-*.json	mother-of	blaadtura taat	North Wumponia	
	father-of	bloodtype-test	North wumpoma	
problem-c-*.json	mother-of	bloodtype-test	North Wumponia	
	father-of	cheap-bloodtype-test	North Wumpoma	
problem-d-*.json	mother-of	h	North Wumponia	
	father-of	bloodtype-test	East Wumponia	
problem-e-*.json	mother-of	hla a du	Unknown	
	father-of	bloodtype-test		
nuchlans f i ison	mother-of	bloodtype-test	Unknown	
problem-i-*.json	father-of	cheap-bloodtype-test	Olikilowii	
problem-g-*.json	mother-of			
	father-of	bloodture test	North Wumponia	
	brother-of	bloodtype-test	East Wumponia	
	sister-of			
problem-h-*.json	mother-of			
	father-of	bloodtype-test	Unknown	
	brother-of	cheap-bloodtype-test		
	sister-of			

Figure 2: Overview of the different problem files. The first problems (problem-a-*.json) only use a minimal family (two parents and their child).

6 Points

You get 1 point for every correctly solved problem file. The problem files vary in their difficulty (Figure 2 provides an overview). As there are 80 problem files, that means that you can get up to 80 points for your solutions. Assuming you have at least a partial solution, you can additionally get up to 20 points for the quality of the submission (README, explanation, ...). The maximum number of points is therefore 100. If the grading scheme doesn't seem to work well, we might adjust it later on (likely in your favor).

References

- [ABO] ABO blood group system. URL: https://en.wikipedia.org/wiki/ABO_blood_ group_system (visited on 06/01/2022).
- [AR] Repository for Assignment 1: Compute Blood Types. URL: https://gitlab.rrze. fau.de/wrv/AISysProj/ss23/a2.1-compute-blood-types/assignment.