

## Supporting Information

### **Appendix A. Example of association rule mining output**

Let us consider 3 symptoms and 2 skeletal Dysplasias defined in the following:

- A1 = Short stature
- A2 = Macrocephaly
- A3 = Osteoarthritis
- D1 = Hajdu-Cheney syndrome
- D2 = Diastrophic dysplasia

Let us also consider the following 4 patient cases:

- Patient 1 - {A1, A2, D1}
- Patient 2 - {A1, A2, D1}
- Patient 3 - {A1, A3, D2}
- Patient 4 - {A1, A3, D2}

Assuming minimum support 50% and minimum confidence 80%, the rule extraction algorithm presented in the paper will generate the following rules:

{A2} -> {D1} with confidence 1

{A3} -> {D2} with confidence 1

{A1, A2} -> {D1} with confidence 1

{A1, A3} -> {D2} with confidence 1

## **Appendix B. Example of Basic Belief Assignment and Belief computation in Dempster-Shafer theory.**

Let us consider a skeletal dysplasia patient examined by two clinicians. Let us assume the first clinician assigns a 70% certainty for the patient to have PSACH or SEDC and the second clinician assigns a 50% certainty for the patient to have SEDC or MED . The frame of discernment will hence be:  $Y = \{PSACH, SEDC, MED\}$ .

The assumption of the first clinician leads to a BBA  $m_1$  of 0.7 given the focal element  $\{PSACH, SEDC\}$ , i.e.,  $m_1(\{PSACH, SEDC\}) = 0.7$ . Provided that we have no additional about the remaining probability, this is allocated to the whole frame of the discernment, i.e.,  $m_1(\{PSACH, SEDC, MED\}) = 0.3$ . Similarly, the assumption of the second clinician assumption leads to a BBA  $m_2(\{SEDC, MED\}) = 0.5$ , the remaining probability allocated to the whole the frame of the discernment  $m_2(\{PSACH, SEDC, MED\}) = 0.5$ .

We can combine the BBA's from these two different sources to express our overall belief using Dempster's rule of combination. The result of combining two BBAs of sets B and C, where  $m_1(\{B\}) = M1$  and  $m_1(\{C\}) = M2$  is the BBA of the intersection of sets B and C. This BBA value is the product of M1 and M2 as the value of K is zero due to the fact that there is no set where the intersection is null.

$$m_3(\{SEDC, MED\}) = m_1(\{PSACH, SEDC, MED\}) \times m_2(\{SEDC, MED\}) = 0.3 \times 0.5 = 0.15$$

	$m_1(\{PSACH, SEDC\})=0.7$	$m_1(\{PSACH, SEDC, MED\})=0.3$
$m_2(\{SEDC, MED\})=0.5$	$m_3(\{SEDC\})=0.35$	$m_3(\{SEDC, MED\})=0.15$
$m_2(\{PSACH, SEDC, MED\})=0.5$	$m_3(\{PSACH, SEDC\})=0.35$	$m_3(\{PSACH, SEDC, MED\})=0.15$

**Combination of the two evidences**

From the combination of the two evidences we can express our overall belief in any set. The belief in any set is calculated by adding all BBAs of all the subsets of that set. For example:

$$\begin{aligned} \text{Bel}(\{PSACH, SEDC\}) &= m_3(\{PSACH\}) + m_3(\{SEDC\}) + m_3(\{PSACH, SEDC\}) \\ &= 0 + 0.35 + 0.35 = 0.7 \end{aligned}$$

$$\begin{aligned} \text{Bel}(\{SEDC, MED\}) &= m_3(\{SEDC\}) + m_3(\{MED\}) + m_3(\{SEDC, MED\}) \\ &= 0.35 + 0 + 0.15 = 0.5 \end{aligned}$$

## **Appendix C. Example of Constrained Belief computation**

**Aim:** Find the diagnosis for an undiagnosed skeletal dysplasia patient with following symptoms: Short Stature ( $A_1$ ) and Osteoarthritis ( $A_2$ ).

Let us consider the following set of hypotheses and conclusions for the dysplasia domain:

- $A_1$  = Short stature
- $A_2$  = Osteoarthritis
- $A_3$  = Short legs
- $A_4$  = Short arms
- $A_5$  = Large head
- $D_1$  = Diastrophic dysplasia
- $D_2$  = Achondroplasia

Let's consider the following evidences in the rule set:

$\{A_1, A_3\} \rightarrow D_1$  with confidence 0.7

$\{A_2, A_4\} \rightarrow D_1$  with confidence 0.8

$\{A_1, A_3\} \rightarrow D_2$  with confidence 0.7

$\{A_2, A_4\} \rightarrow D_2$  with confidence 0.6

$\{A_3, A_4\} \rightarrow D_2$  with confidence 0.65

$\{A_3, A_5\} \rightarrow D_2$  with confidence 0.3

$\{A_4, A_5\} \rightarrow D_2$  with confidence 0.2

### **Solution**

**Step 1:** Select the suitable evidences based on the symptoms of the patient ( $A_1, A_2$ ):

$\{A_1, A_3\} \rightarrow D_1$  with confidence 0.7 (1)

$\{A_2, A_4\} \rightarrow D_1$  with confidence 0.8 (2)

$\{A_1, A_3\} \rightarrow D_2$  with confidence 0.7 (3)

$\{A_2, A_4\} \rightarrow D_2$  with confidence 0.6 (4)

**Step 2:** We select the candidate hypothesis based on the selected rules. These are:  $D_1, D_2$ . Hence the propositions of interest in the diagnosis of the patient will be:

- $\{A_1, A_2, D_1\}$
- $\{A_1, A_2, D_2\}$

**Step 3:** We perform belief value calculation for each candidate hypothesis.

#### **Belief value calculation for $D_1$ :**

The hypothesis elements of the patient for conclusion  $D_1$ :  $A_1, A_2, A_3, A_4, D_1$ .

From (1),  $m_1(\{A_1, A_3, D_1\}) = 0.7$  (from this, we get  $m_1(\{A_1, A_2, A_3, A_4, D_1\}) = 0.3$ )

From (2),  $m_2(\{A_2, A_4, D_1\}) = 0.8$  (from this, we get  $m_2(\{A_1, A_2, A_3, A_4, D_1\}) = 0.2$ )

Combination of two pieces of evidence ( $m_1, m_2$ ) through the Dempster's rule of combination and here  $K=0$  as there is no set where the intersection is null:

	$m_2\{A_2, A_4, D_1\} = 0.8$	$m_2\{A_1, A_2, A_3, A_4, D_1\} = 0.2$
$m_1\{A_1, A_3, D_1\} = 0.7$	$m_3\{D_1\} = 0.56$	$m_3\{A_1, A_3, D_1\} = 0.14$
$m_1\{A_1, A_2, A_3, A_4, D_1\} = 0.3$	$m_3\{A_2, A_4, D_1\} = 0.24$	$m_3\{A_1, A_2, A_3, A_4, D_1\} = 0.06$

From the combination of these evidences, we can calculate overall belief associated with Dysplasia  $D_1$ .

$$\begin{aligned}
 \text{Bel}(\{A_1, A_2, D_1\}) &= m_3(\{D_1\}) + m_3(\{A_1, D_1\}) + m_3(\{A_2, D_1\}) + m_3(\{A_1, A_2, D_1\}) \\
 &= 0.56 + 0 + 0 + 0 \\
 &= 0.56
 \end{aligned}$$

#### **Belief value calculation for $D_2$ :**

The hypothesis elements of the patient for conclusion  $D_2$ :  $A_1, A_2, A_3, A_4, D_2$ .

From (3),  $m_1\{A_1, A_3, D_2\} = 0.7$  (from this, we get  $m_1\{A_1, A_2, A_3, A_4, D_2\} = 0.3$ )

From (4),  $m_2\{A_2, A_4, D_2\} = 0.6$  (from this, we get  $m_2\{A_1, A_2, A_3, A_4, D_2\} = 0.4$ )

Combination of two pieces of evidence ( $m_1, m_2$ ) through the Dempster's rule of combination and here  $K=0$  as well.

	$m_2\{A_2, A_4, D_2\} = 0.6$	$m_2\{A_1, A_2, A_3, A_4, D_2\} = 0.4$
$m_1\{A_1, A_3, D_2\} = 0.7$	$m_4\{D_2\} = 0.42$	$m_4\{A_1, A_3, D_2\} = 0.28$
$m_1\{A_1, A_2, A_3, A_4, D_2\} = 0.3$	$m_4\{A_2, A_4, D_2\} = 0.18$	$m_4\{A_1, A_2, A_3, A_4, D_2\} = 0.12$

$$\begin{aligned}
 \text{Bel}(\{A_1, A_2, D_2\}) &= m_4(\{D_2\}) + m_4(\{A_1, D_2\}) + m_4(\{A_2, D_2\}) + m_4(\{A_1, A_2, D_2\}) \\
 &= 0.42 + 0 + 0 + 0 \\
 &= 0.42
 \end{aligned}$$

**Step 4:** We rank the belief values and choose the highest as the best candidate. Considering the calculations above, the ranking of the belief values is:  $D_1, D_2$ .