

A Ground-truth Causal Relations

In this paper, we focus on the neuropathic pain caused by discoligamentous injuries and radiculopathies. Table 11 shows all the ground-truth causal relations which are used for establishing the ground-truth causal graph for our simulator. Figure 5 shows the whole causal graph of neuropathic pain diagnose.

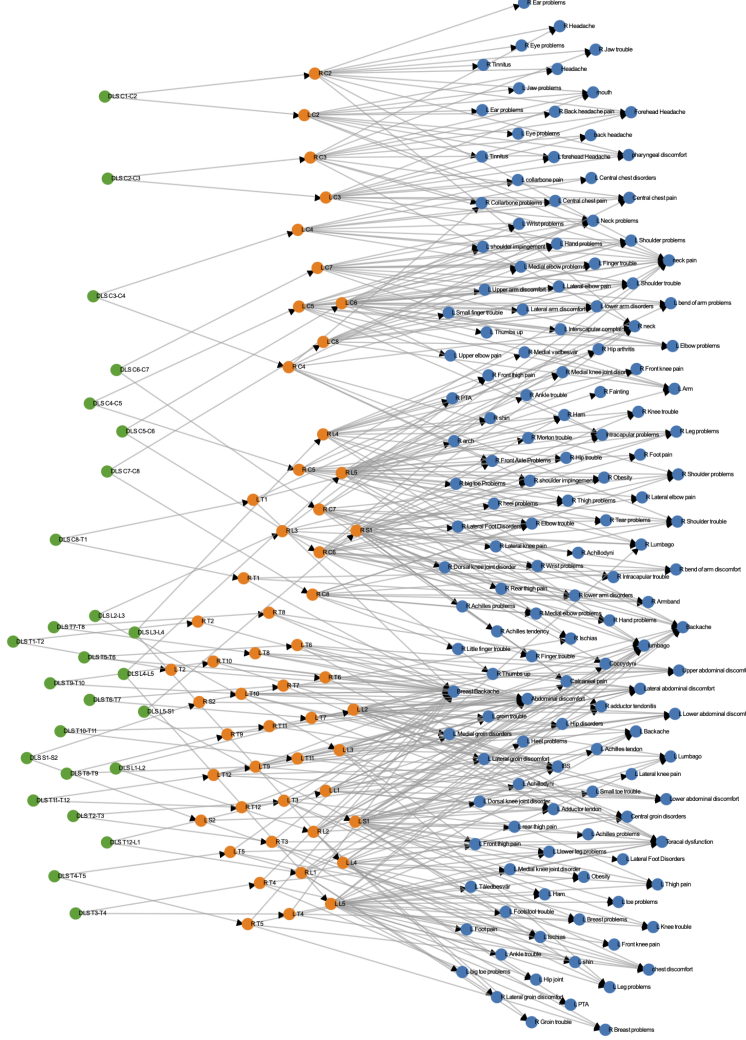


Figure 5: The causal graph of neuropathic pain diagnoses.

B Simulation Quality Evaluation

We show several simulated patient diagnostic records and the corresponding physician’s evaluation scores in Table 6. Moreover, we show the correlation matrices of the real dataset and the simulated dataset in Figure 6. We can clearly see the pattern of the correlation matrix of the simulated dataset, which is similar to the pattern of the correlation matrix of the real dataset. The correlation matrix of the simulated dataset contains many white lines. When all values of a variable are zero in the simulated dataset, the row and the column of the variable in the correlation matrix are white lines. Since our simulator cancels out many correlations between variables which might be introduced by unknown confounders and selection bias, the simulated dataset with a small sample size might have many variables with only zero value. The number of full-zero variables can be controlled by introducing different levels of random noise in the data generation process.

Table 6: A part of the physician’s evaluation results.

Pathophysiology diagnosis	Pattern diagnosis	Symptom diagnosis	Score
DLI L3-L4, DLI T11-T12	L L4 Radiculopathy, R L4 Radiculopathy	L Front thigh pain, R Front thigh pain, R shin	4
DLI C3-C4, DLI L4-L5, DLI L5-S1	L C4 Radiculopathy , R C4 Radiculopathy, L L5 Radiculopathy, R L5 Radiculopathy, L S1 Radiculopathy, R S1 Radiculopathy	L Neck problems, R neck, L collarbone pain, R Front Axle Problems, L shoulder impingement, L Shoulder trouble, R Shoulder problems, R Shoulder trouble, L PTA, L Front knee pain, R Front knee pain, R arch, L Obesity, R Ham, R Tear problems, R heel problems, L Heel problems, L rear thigh pain	4
DLI C2-C3	L C3 Radiculopathy, R C3 Radiculopathy	L Neck problems, R neck	3
DLI C1-C2, DLI C5-C6, DLI C6-C7, DLI S1-S2	L C2 Radiculopathy, R C2 Radiculopathy, L C6 Radiculopathy, R C6 Radiculopathy’, L C7 Radiculopathy, R C7 Radiculopathy	neck pain, L Eye problems, R Eye problems, L Jaw problems, L forehead Headache, R Jaw trouble, L Shoulder problems, R Shoulder problems, L Thumbs up, L Hand problems, R Armband, L Medial elbow problems, R Medial elbow problems	3
DLI L4-L5, DLI L5-S1	L L5 Radiculopathy, R L5 Radiculopathy, L S1 Radiculopathy, R S1 Radiculopathy	R adductor tendonitis , lumbago, R Lumbago, L Hip joint, R Hip arthritis, L Medial knee joint disorder, R shin, R Knee trouble, R Tear problems, L Lateral Foot Disorders, L Heel problems, R Achilles tendency	2
DLI L4-L5, DLI L5-S1	L L5 Radiculopathy, R L5 Radiculopathy, R S1 Radiculopathy	lumbago, L Hip joint, L Ankle trouble, L Footstool trouble, R Dorsal knee joint disorder, Coccydyni, R Rear thigh pain, R Achilles tendency	2

C Experiment Details

In this section, we mainly show recall, precision, and SHD results of causal discovery algorithms under different experiment settings. Table 7, Table 8, Table 9, and Table 10 are the results of the experiments designed for evaluating causal discovery algorithms in the presence of different sample sizes, unknown confounders, selection bias, and missing data.

For generating the datasets with confounders that are external variables, we choose the discoligamentous injury C2-C3, C3-C4, C4-C5, C5-C6, C6-C7, and C7-C8 as the direct effects of an unknown confounder. In this experiment, the confounder can be interpreted as the occupation that can easily damage the neck part of people during the work time. Thus, the chosen discoligamentous injuries are correlated with each other. Then, we use Bernoulli distribution as the marginal distribution of the confounder, and assign a default CPD to each chosen discoligamentous injury. In the end, we generate the data from our modified simulator and delete the data of the introduced confounder in the simulated dataset.

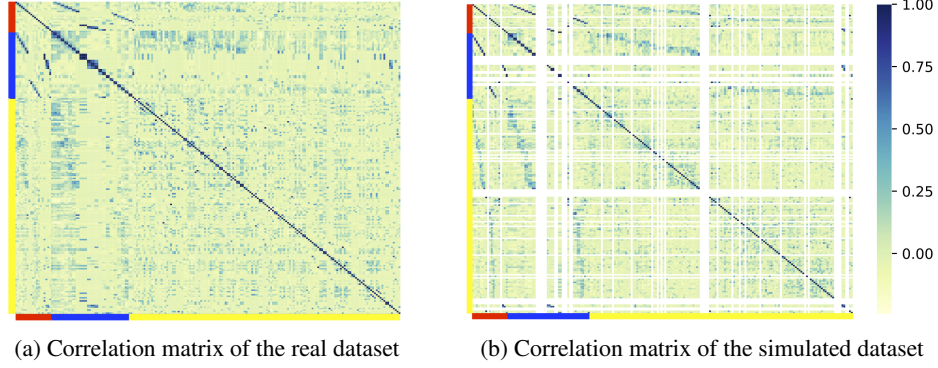


Figure 6: Comparison of the correlation matrices of the real and simulated datasets.

Table 7: Performance of different causal discovery algorithms on the datasets with different sample sizes. The SHD performance is better when it has smaller value. Precision, recall, causal accuracy and F1 score are better when they have larger value. Total SHD is 24642.

Sample size	128	256	512	1024	2048	4096	8192	16384
SHD_{PC}	794.5	809.0	830.0	834.5	826.5	822.5	801.5	802
SHD_{GES}	800.5	802.5	814.0	815.0	824.0	834.0	808.5	773
$Precision_{PC}$	0.143	0.141	0.069	0.138	0.221	0.301	0.413	0.438
$Precision_{GES}$	0.200	0.316	0.344	0.361	0.366	0.393	0.441	0.505
$Recall_{PC}$	0.010	0.0156	0.009	0.023	0.039	0.060	0.086	0.120
$Recall_{GES}$	0.023	0.048	0.073	0.095	0.113	0.149	0.185	0.239

Table 8: Performance of causal discovery methods in the presence of unknown confounders. Total SHD is 10082. The sample size is 1024.

	FCI	RFCI	GFCI	PC	GES
Cau_Acc	0.013	0.014	0.009	X	X
SHD	X	X	X	123.5	83.5

Table 11: Ground-truth causal relations. A capital letter with a number represents a radiculopathy. For example, "C2" represents "C2 radiculopathy". A radiculopathy as an effect in the table represents both sides of a radiculopathy. A radiculopathy as a cause in the table has the same side with its effect. We denote left as "L" and right as "R".

Effect	Cause
C2	DLS C1-C2
C3	DLS C2-C3
C4	DLS C3-C4
C5	DLS C4-C5
C6	DLS C5-C6
C7	DLS C6-C7
C8	DLS C7-C8
T1	DLS C8-T1
T2	DLS T1-T2
T3	DLS T2-T3
T4	DLS T3-T4
T5	DLS T4-T5
T6	DLS T5-T6
T7	DLS T6-T7
T8	DLS T7-T8
T9	DLS T8-T9
T10	DLS T9-T10
T11	DLS T10-T11

T12	DLS T11-T12
L1	DLS T12-L1
L2	DLS L1-L2
L3	DLS L2-L3
L4	DLS L3-L4
L5	DLS L4-L5
S1	DLS L5-S1
S2	DLS S1-S2
C2	craniocervical junction
C3	craniocervical junction
C4	craniocervical junction
Ibs	T10;T11;T12;L1;L2
L neck problems	C2;C3;C4;C5;C6;C7
Neck pain	C2;C3;C4;C5;C6;C7
R neck	C2;C3;C4;C5;C6;C7
L tinnitus	C2
L eye problems	C2
L ear problems	C2
R tinnitus	C2
R eye problems	C2
R ear problems	C2
Headache	C2
L jaw problems	C2
L forehead headache	C2;C3
Mouth	C2;C3
Forehead headache	C2;C3
R headache	C2;C3
R pta	L4;L5
Pharyngeal discomfort	C2;C3
R jaw trouble	C2;C3
Back headache	C3
R back headache pain	C3
L collarbone pain	C3;C4
R collarbone problems	C3;C4
Central chest pain	C3;C4
L central chest pain	C3;C4
L central chest disorders	C3;C4
R front axle problems	C4;C5;C6
L shoulder impingement	C4;C5;C6
R shoulder impingement	C4;C5;C6
L shoulder problems	C4;C5;C6;C7;C8
L shoulder trouble	C4;C5;C6;C7;C8
R shoulder problems	C4;C5;C6;C7;C8
R shoulder trouble	C4;C5;C6;C7;C8
L upper arm discomfort	C5
L upper elbow pain	C5
Intracapular problems	C5;C6
L interscapular complaints	C5;C6
R intracapular trouble	C5;C6
L lateral elbow pain	C5;C6
L lateral arm discomfort	C5;C6
R lateral elbow pain	C5;C6
L elbow problems	C5;C6;C7;C8
R elbow trouble	C5;C6;C7;C8
L arm	C5;C6;C7;C8;T1
L thumbs up	C6
R thumbs up	C6
L wrist problems	C6;C7
R wrist problems	C6;C7

L lower arm disorders	C6;C7;C8
R lower arm disorders	C6;C7;C8
L hand problems	C6;C7;C8
R hand problems	C6;C7;C8
L bend of arm problems	C6;C7;C8;T1
R armband	C6;C7;C8;T1
R bend of arm discomfort	C6;C7;C8;T1
L medial elbow problems	C7;C8
R medial elbow problems	C7;C8
L finger trouble	C7;C8
R finger trouble	C7;C8
L small finger trouble	C8
R little finger trouble	C8
L groin trouble	L1;L2
L medial groin disorders	L1;L2
L lateral groin discomfort	L1;L2
Central groin disorders	L1;L2
R lateral groin discomfort	L1;L2
R groin trouble	L1;L2
L adductor tendon	L1;L2;S1;S2
R adductor tendonitis	L1;L2;S1;S2
L hip disorders	L2;L3
L backache	L2;L3;L4;L5;S1
Backache	L2;L3;L4;L5;S1
L lumbago	L2;L3;L4;L5;S1
Lumbago	L2;L3;L4;L5;S1
R lumbago	L2;L3;L4;L5;S1
L front thigh pain	L3;L4
R front thigh pain	L3;L4
R thigh problems	L3;L4;L5;S1
L leg problems	L3;L4;L5;S1
L thigh pain	L3;L4;L5;S1
R leg problems	L3;L4;L5;S1
R medial vadbesvär	L4
L pta	L4;L5
L hip joint	L4;L5
R hip trouble	L4;L5
R hip arthritis	L4;L5
L medial knee joint disorder	L4;L5
L front knee pain	L4;L5
R medial knee joint disorder	L4;L5
R front knee pain	L4;L5
L shin	L4;L5
R shin	L4;L5
L llower leg problems	L4;L5;S1
L knee trouble	L4;L5;S1
R knee trouble	L4;L5;S1
L tåledbesvär	L5
L big toe problems	L5
R big toe problems	L5
L foot pain	L5
L ankle trouble	L5
R ankle trouble	L5
L footstool trouble	L5
R arch	L5
R morton trouble	L5
R fainting	L5
L ischias	L5;S1;S2
R ischias	L5;S1;S2

L ham	L5;S1
L obesity	L5;S1
R ham	L5;S1
L toe problems	L5;S1
R foot pain	L5;S1
R tear problems	L5;S1
R obesity	L5;S1
R dorsal knee joint disorder	S1
L dorsal knee joint disorder	S1
L lateral knee pain	S1
R lateral knee pain	S1
L small toe trouble	S1
L lateral foot disorders	S1
R lateral foot disorders	S1
R heel problems	S1
Calcaneal pain	S1
L heel problems	S1
Coccydyni	S1
L rear thigh pain	S1
R rear thigh pain	S1
L achilles problems	S1
L achilles tendon	S1
L achillodini	S1
R achilles problems	S1
R achilles tendency	S1
R achillodini	S1
Breast backache	T1;T2;T3;T4;T5;T6;T7;T8;T9;T10
Chest discomfort	T3;T4;T5
L breast problems	T3;T4;T5
R breast problems	T3;T4;T5
Toracal dysfunction	T3;T4;T5;T6;T7
Upper abdominal discomfort	T6;T7;T8
Lateral abdominal discomfort	T6;T7;T8;T9;T10;T11;T12;L1;L2
Abdominal discomfort	T6;T7;T8;T9;T10;T11;T12;L1;L2
L lower abdominal discomfort	T9;T10;T11;T12;L1;L2
Lower abdominal discomfort	T9;T10;T11;T12;L1;L2

Table 9: Performance of different causal discovery methods in the presence of selection bias.

	F1	F1 ref	Recall	Recall ref	Precision	Precision ref
PC	0.072	0.076	0.042	0.045	0.276	0.236
GES	0.193	0.185	0.126	0.125	0.416	0.356

Table 10: Performance of causal discovery methods in the presence of missing data.

	FCI	RFCI	PC	GES
CauAcc _{MNAR}	0.059	0.051	0.061	0.154
CauAcc _{MAR}	0.063	0.049	0.050	0.135
CauAcc _{MCAR}	0.066	0.055	0.067	0.161
CauAcc _{ref}	0.062	0.050	0.059	0.145
SHD _{MNAR}	X	X	806.5	812.0
SHD _{MAR}	X	X	806.5	778.5
SHD _{MCAR}	X	X	804.5	801.5
SHD _{ref}	X	X	795.0	765.5
Recall _{MNAR}	X	X	0.081	0.177
Recall _{MAR}	X	X	0.080	0.160
Recall _{MCAR}	X	X	0.086	0.181
Recall _{ref}	X	X	0.094	0.168
Precision _{MNAR}	X	X	0.376	0.435
Precision _{MAR}	X	X	0.389	0.490
Precision _{MCAR}	X	X	0.405	0.440
Precision _{ref}	X	X	0.462	0.514
F1 _{MNAR}	X	X	0.133	0.251
F1 _{MAR}	X	X	0.132	0.241
F1 _{MCAR}	X	X	0.141	0.256
F1 _{ref}	X	X	0.156	0.253