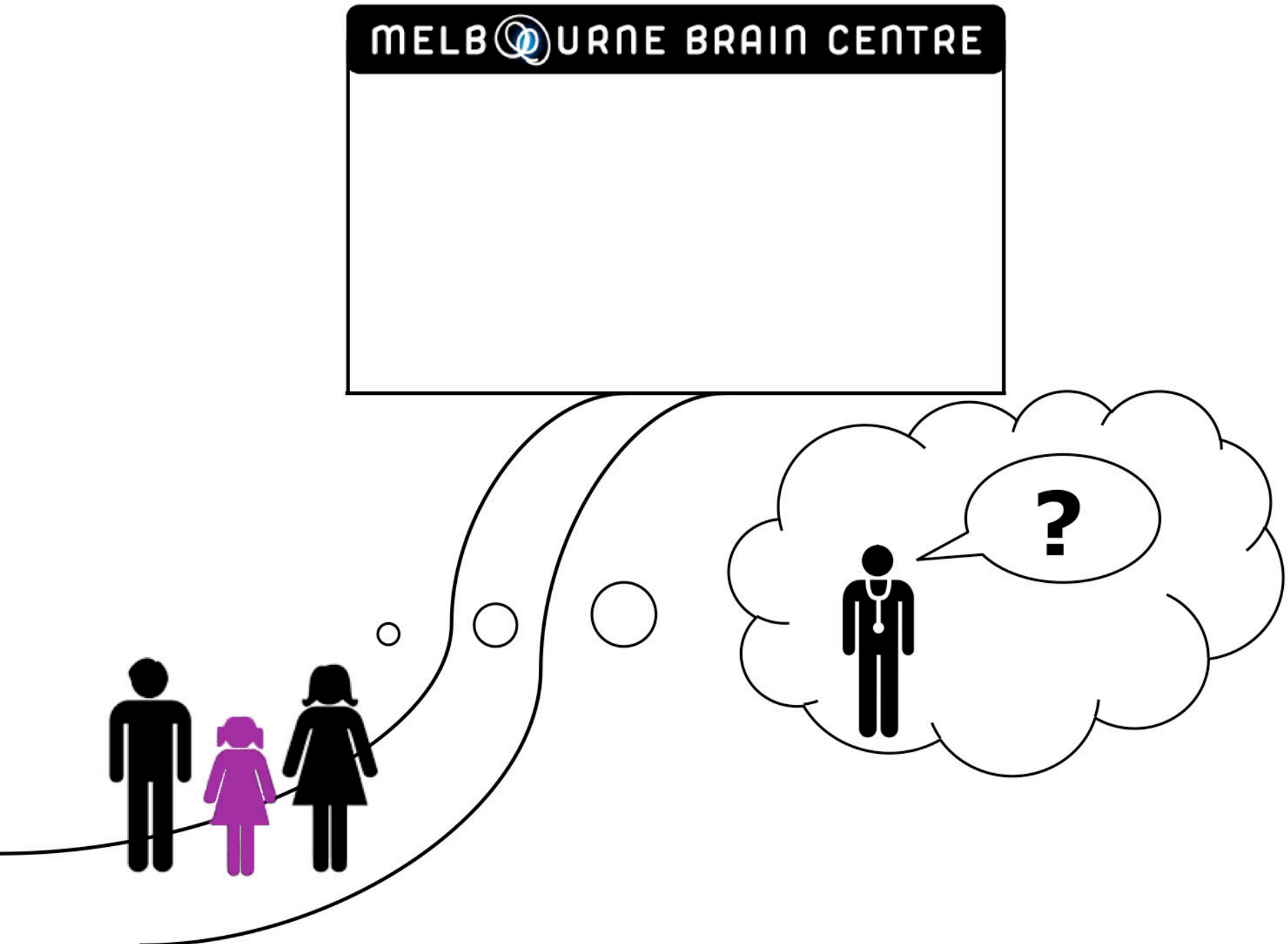
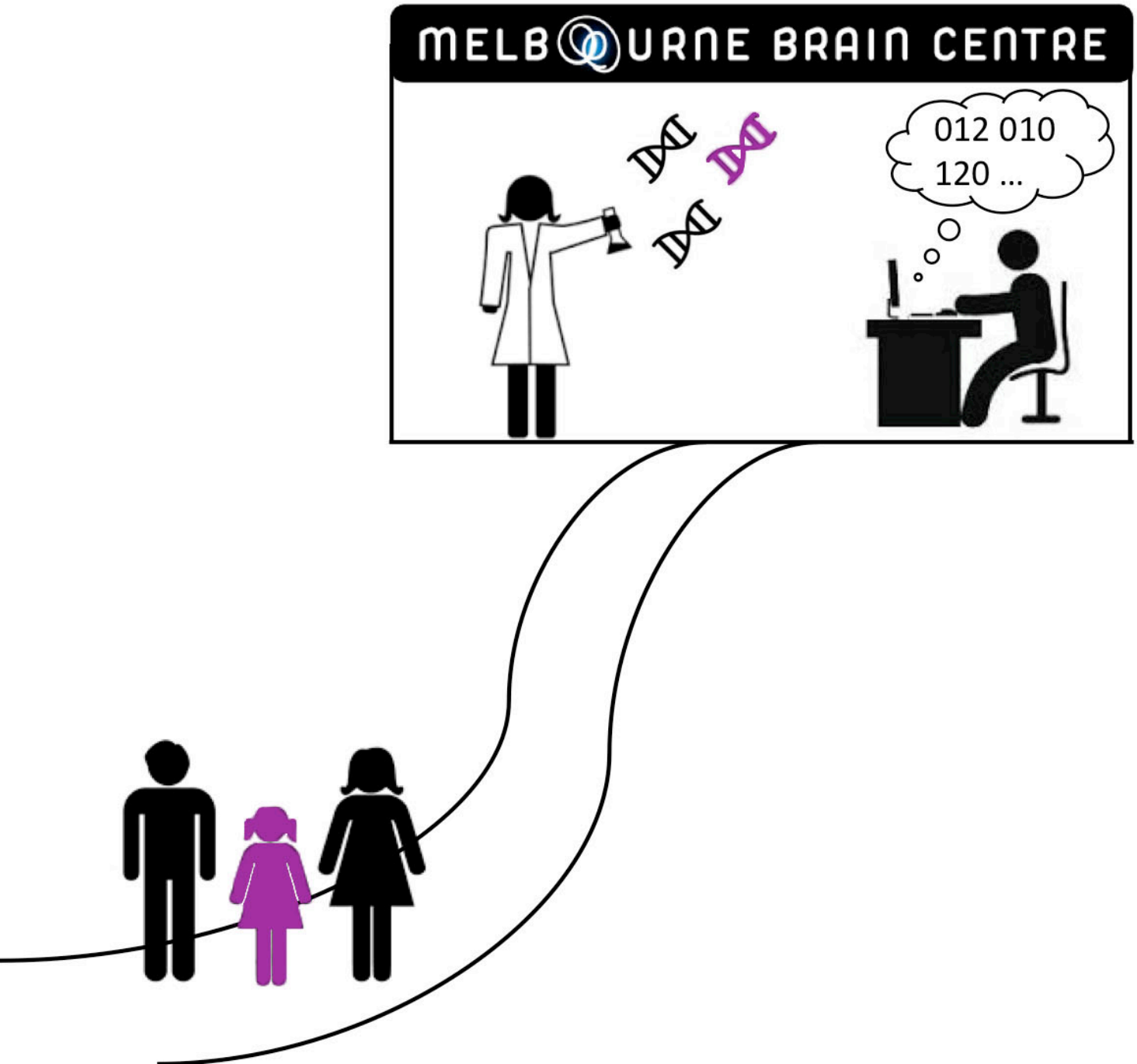
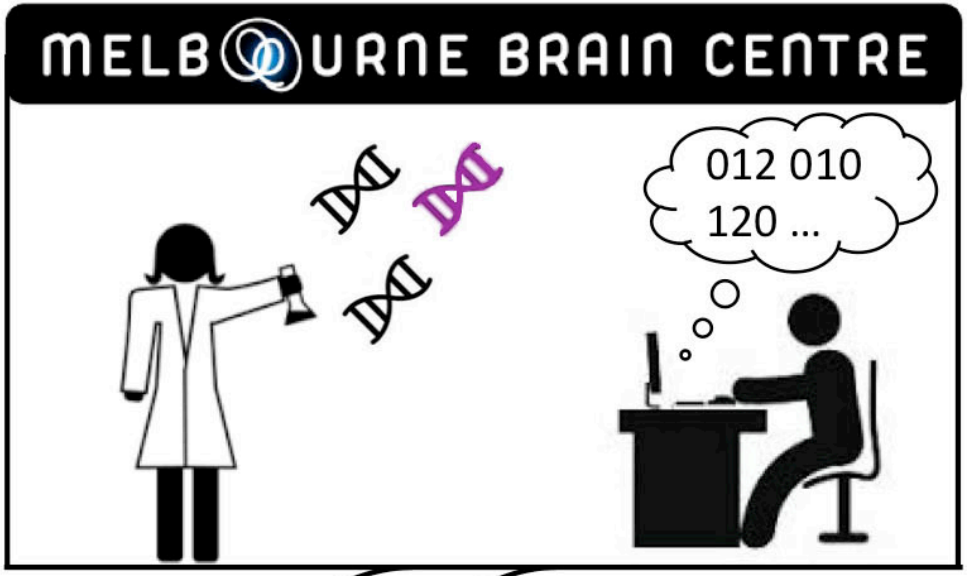
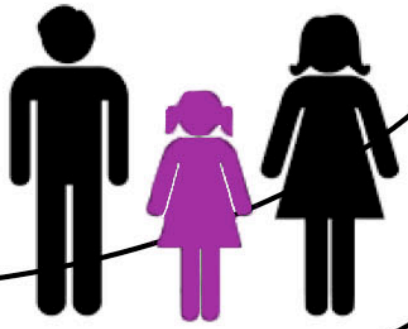

MELBOURNE BRAIN CENTRE
?


MELBOURNE BRAIN CENTRE
012 010
120 ...

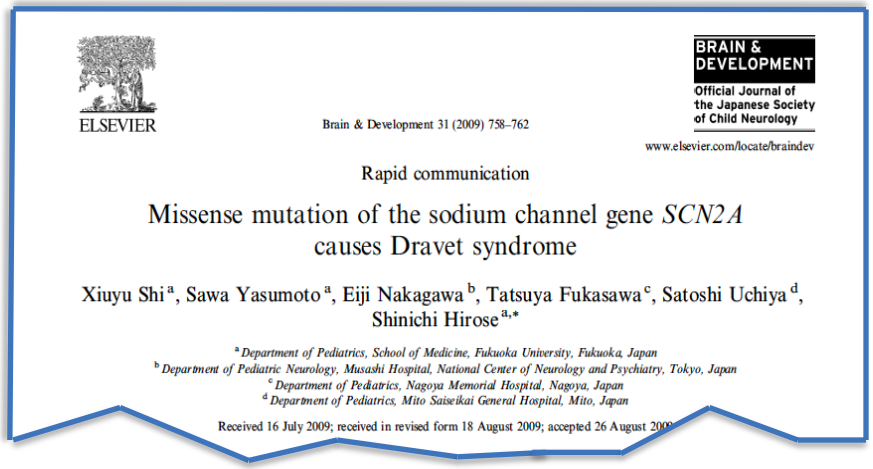




Taxing

Time-Consuming

6 Datasets



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Brain & Development 31 (2009) 758–762

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

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Rapid communication

Missense mutation of the sodium channel gene *SCN2A* causes Dravet syndrome

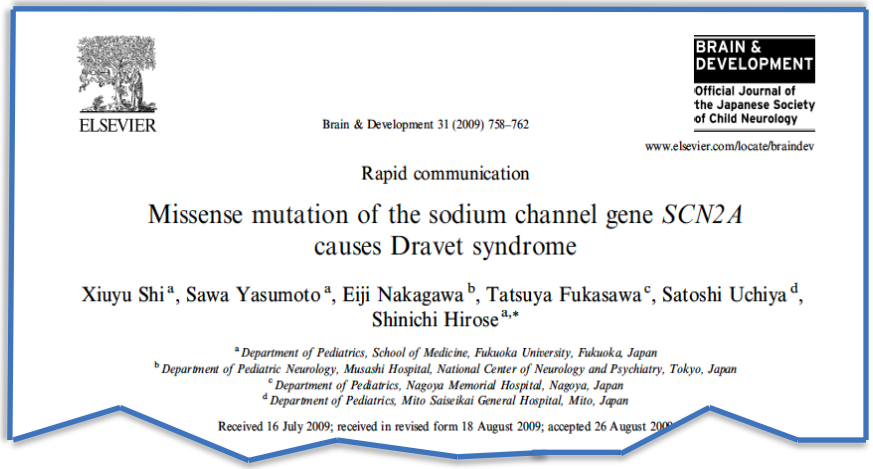
Xiuyu Shi [a], Sawa Yasumoto [a], Eiji Nakagawa [b], Tatsuya Fukasawa [c], Satoshi Uchiya [d], Shinichi Hirose [a,*]

[a] Department of Pediatrics, School of Medicine, Fukuoka University, Fukuoka, Japan
[b] Department of Pediatric Neurology, Musashi Hospital, National Center of Neurology and Psychiatry, Tokyo, Japan
[c] Department of Pediatrics, Nagoya Memorial Hospital, Nagoya, Japan
[d] Department of Pediatrics, Mito Saiseikai General Hospital, Mito, Japan

Received 16 July 2009; received in revised form 18 August 2009; accepted 26 August 2009

6 Datasets

Known Disease Genes



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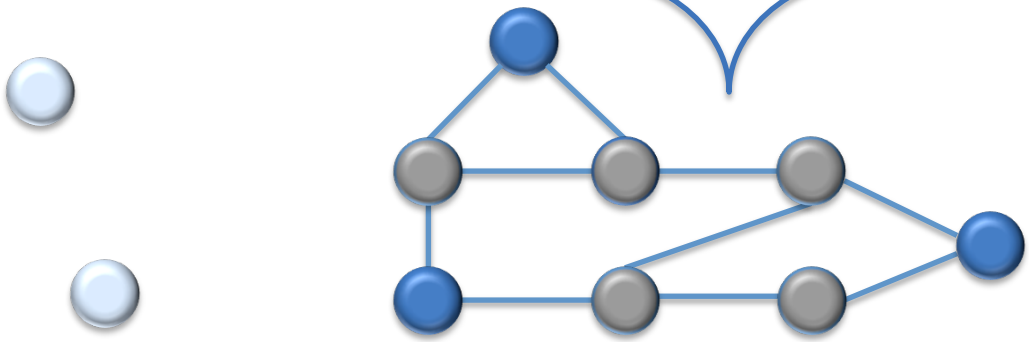


Known

True Candidates

Wrong Candidates

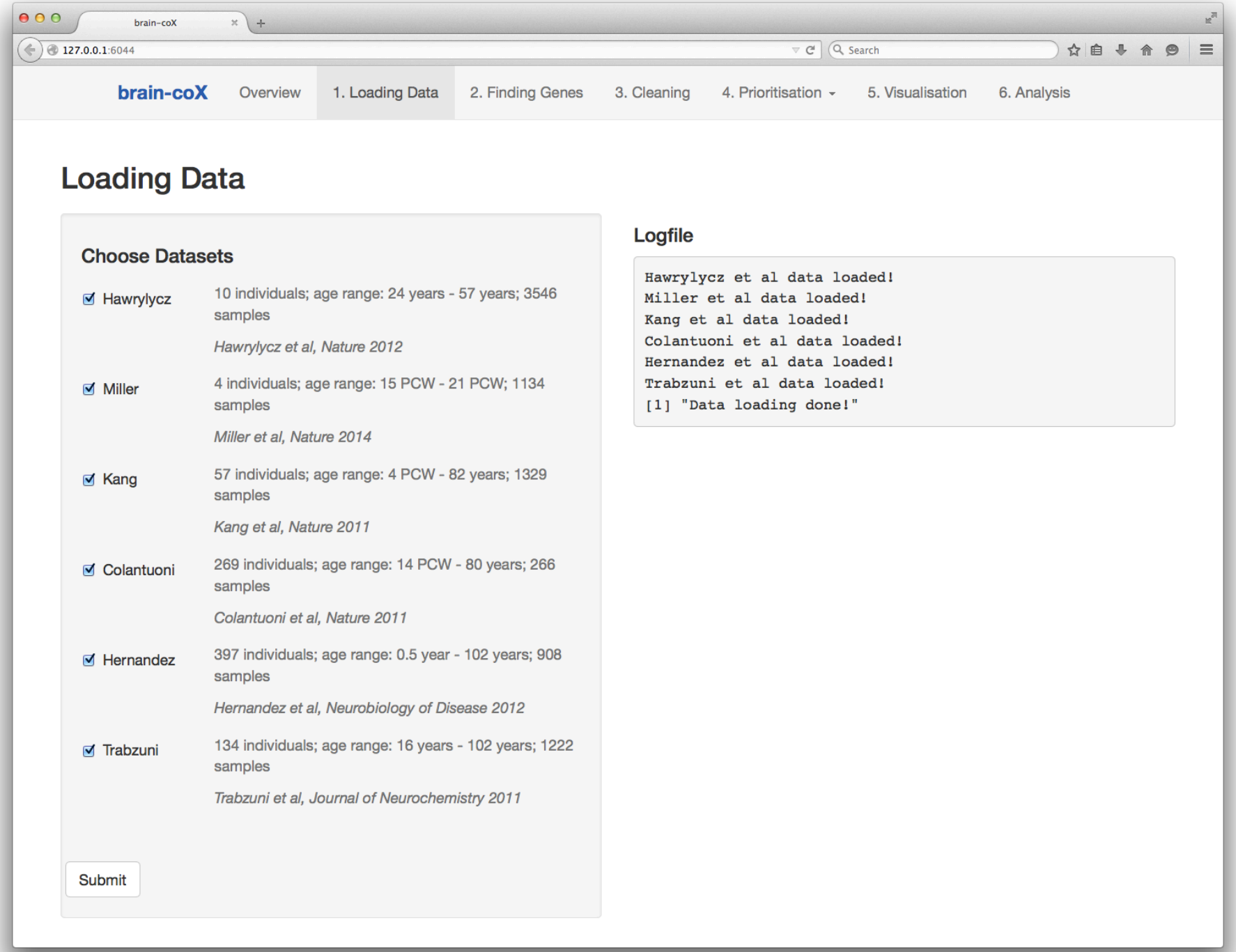
brain-coX

Data Cleaning

Use By Non-Bioinformaticians

Bioconductor
OPEN SOURCE SOFTWARE FOR BIOINFORMATICS

RUVcorr


brain-coX
127.0.0.1:6044
Search
brain-coX
Overview
1. Loading Data
2. Finding Genes
3. Cleaning
4. Prioritisation
5. Visualisation
6. Analysis
Loading Data
Choose Datasets
Hawrylycz
10 individuals; age range: 24 years - 57 years; 3546 samples
Hawrylycz et al, Nature 2012
Miller
4 individuals; age range: 15 PCW - 21 PCW; 1134 samples
Miller et al, Nature 2014
Kang
57 individuals; age range: 4 PCW - 82 years; 1329 samples
Kang et al, Nature 2011
Colantuoni
269 individuals; age range: 14 PCW - 80 years; 266 samples
Colantuoni et al, Nature 2011
Hernandez
397 individuals; age range: 0.5 year - 102 years; 908 samples
Hernandez et al, Neurobiology of Disease 2012
Trabzuni
134 individuals; age range: 16 years - 102 years; 1222 samples
Trabzuni et al, Journal of Neurochemistry 2011
Submit
Logfile
Hawrylycz et al data loaded!
Miller et al data loaded!
Kang et al data loaded!
Colantuoni et al data loaded!
Hernandez et al data loaded!
Trabzuni et al data loaded!
[1] "Data loading done!"

Finding Genes

1

Input Known Disease Genes

File input

Browse... No file selected.

Text input

DLL1, LFNG, NOTCH3, RBPJ,

Text input is ignored when file input is provided.

Input Candidate Disease Genes

File input

Browse... No file selected.

Text input

DLL4, JAG1, DVL2, DVL1, DTX

Text input is ignored when file

Logfile

```
Checking compatibility of known disease genes:
Checking compatibility of candidate disease genes:
Checking compatibility of disease related genes:
Log Hawrylycz:
Could not find the following known diseases genes: DAM17
Could not find the following candidate diseases genes:
Could not find the following related diseases genes:
Log Miller:
Could not find the following known diseases genes: DAM17
Could not find the following candidate diseases genes:
Could not find the following related diseases genes:
Log Kang:
Could not find the following known diseases genes: DAM17
Could not find the following candidate diseases genes:
Could not find the following related diseases genes: RFNG
```

Data Cleaning

2

Choose Negative Control Genes

- (•) Housekeeping Genes
- ( ) Empirically Chosen Genes

Number of negative controls

3000

Number of negative controls is ignored when housekeeping genes are selected.

Submit

Logfile

```
Log Hawrylycz:
Using 3622 housekeeping genes as negative controls!
Log Miller:
Using 3646 housekeeping genes as negative controls!
Log Kang:
Using 3551 housekeeping genes as negative controls!
Log Colantuoni:
Using 3137 housekeeping genes as negative controls!
Log Hernandez:
Using 3292 housekeeping genes as negative controls!
Log Trabzuni:
Using 3551 housekeeping genes as negative controls!
[1] "Data cleaning done!"
```

Gene Prioritisation

Logfile

```
[1] "All thresholds calculated!"
```

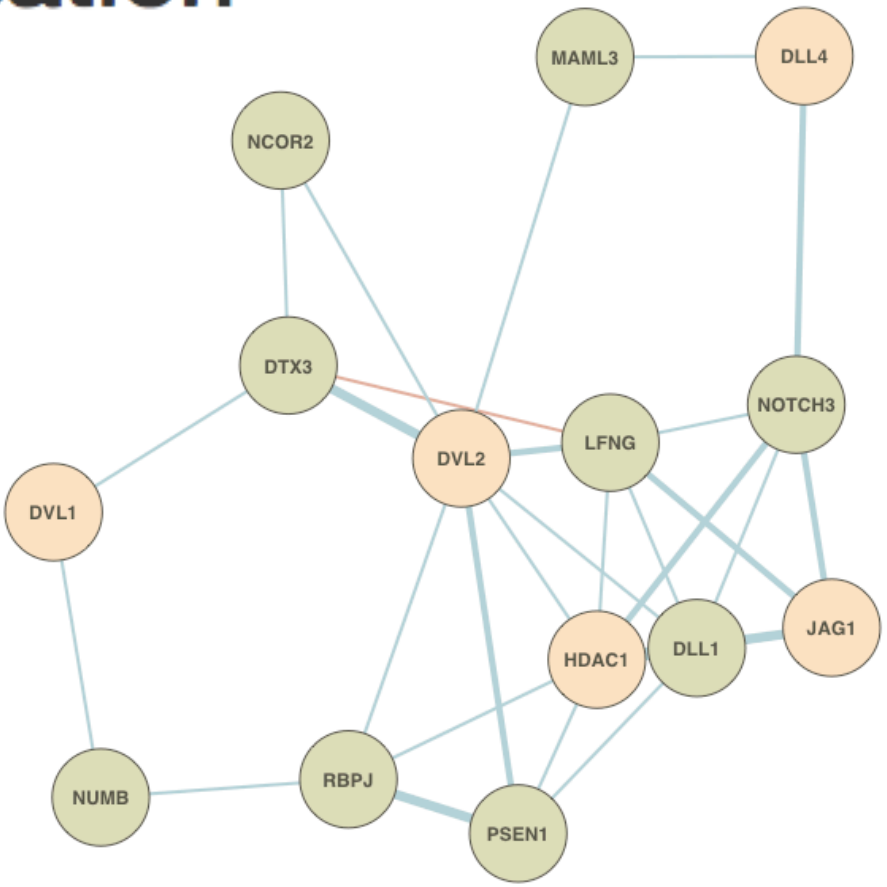
Results

	Hawrylycz	Miller	Kang	Colantuoni	Hernandez	Trabzuni	Number of Times Prioritised	Number of Times Seen
HDAC1	1.00	1.00	1.00	1.00	1.00	1.00	6.00	6.00

Significance

There is 1 candidate gene that is prioritised in all selected datasets compared to an expected number of 0.258 (Standard Deviation: 0.464).

Network Visualisation

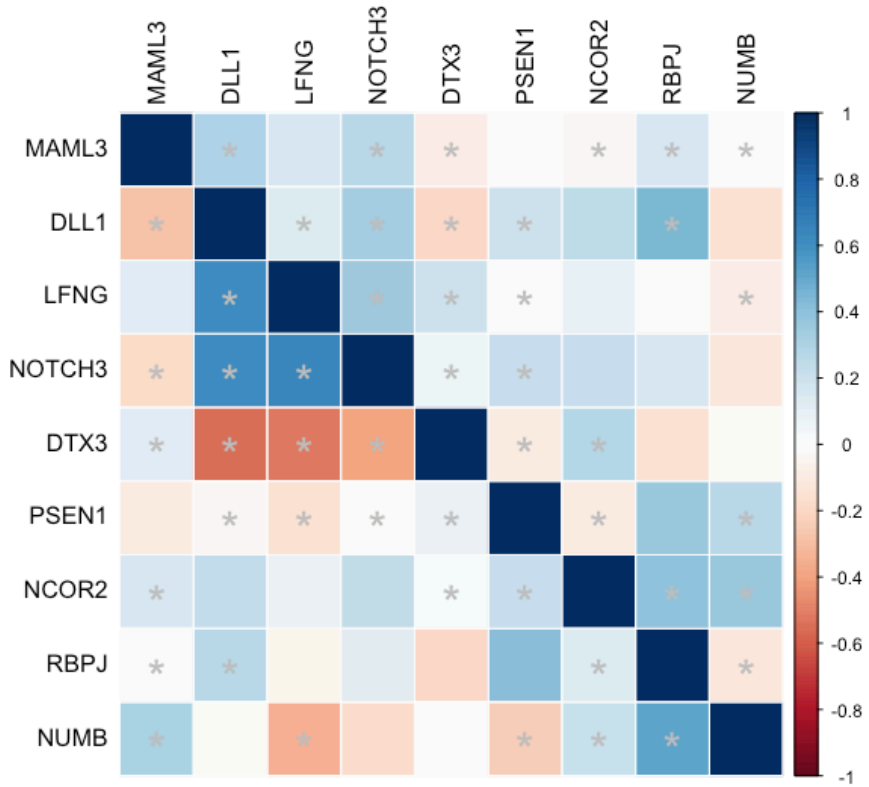


Inhibition
Activation
No Consensus

Seen in 1 Dataset
Seen in 2 Datasets
Seen in 3 Datasets
Seen in 4 Datasets
Seen in 5 Datasets
Seen in 6 Datasets

Known Gene
Candidate Gene

Developmental Analysis

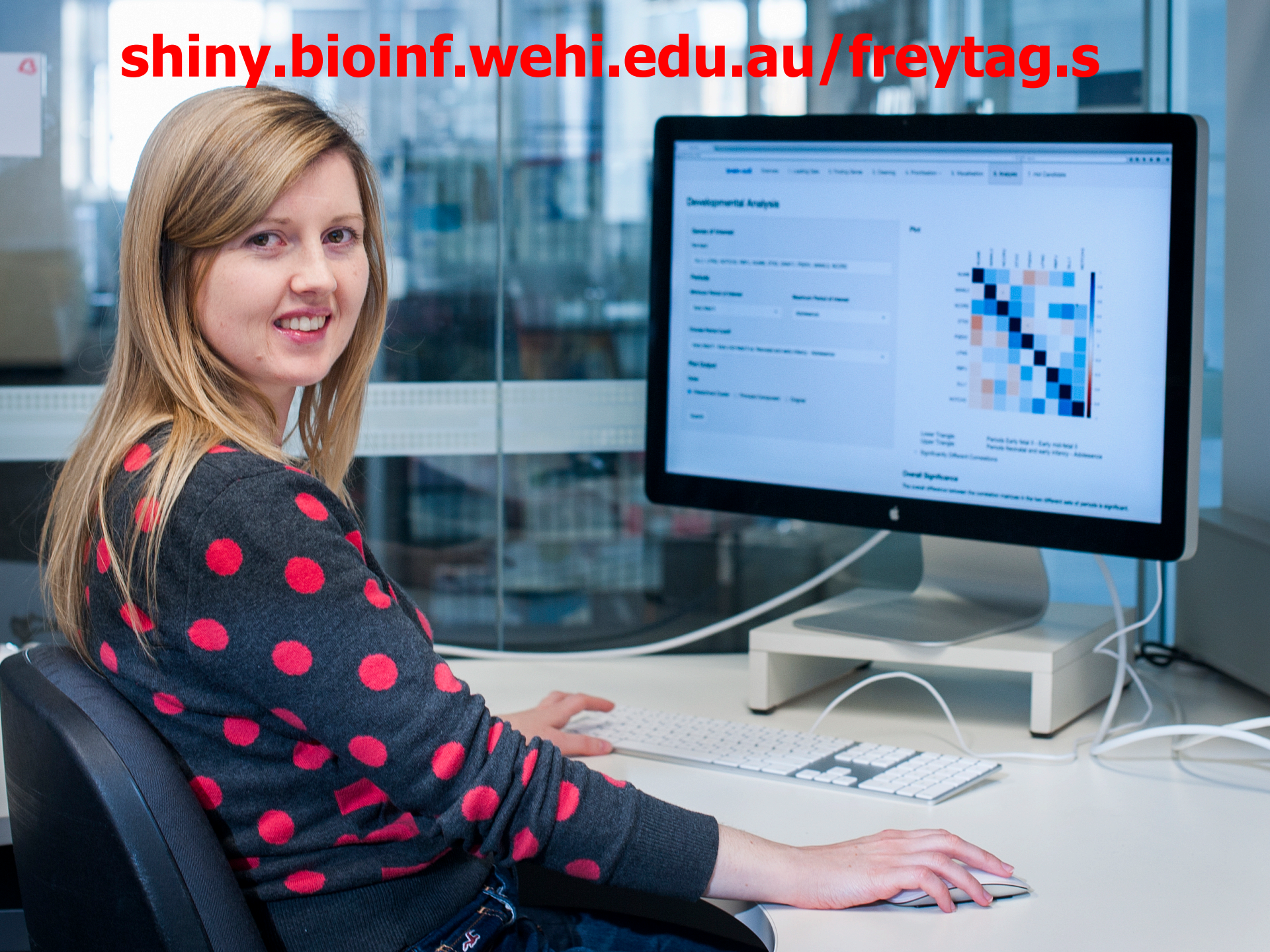


Lower Triangle: Periods Embryonic - Late fetal
Upper Triangle: Periods Neonatal and early infancy - Adolesence

* Significantly Different Correlations

Overall Significance

The overall difference between the correlation matrices in the two different sets of periods is significant.


shiny.bioinf.wehi.edu.au/freytag.s