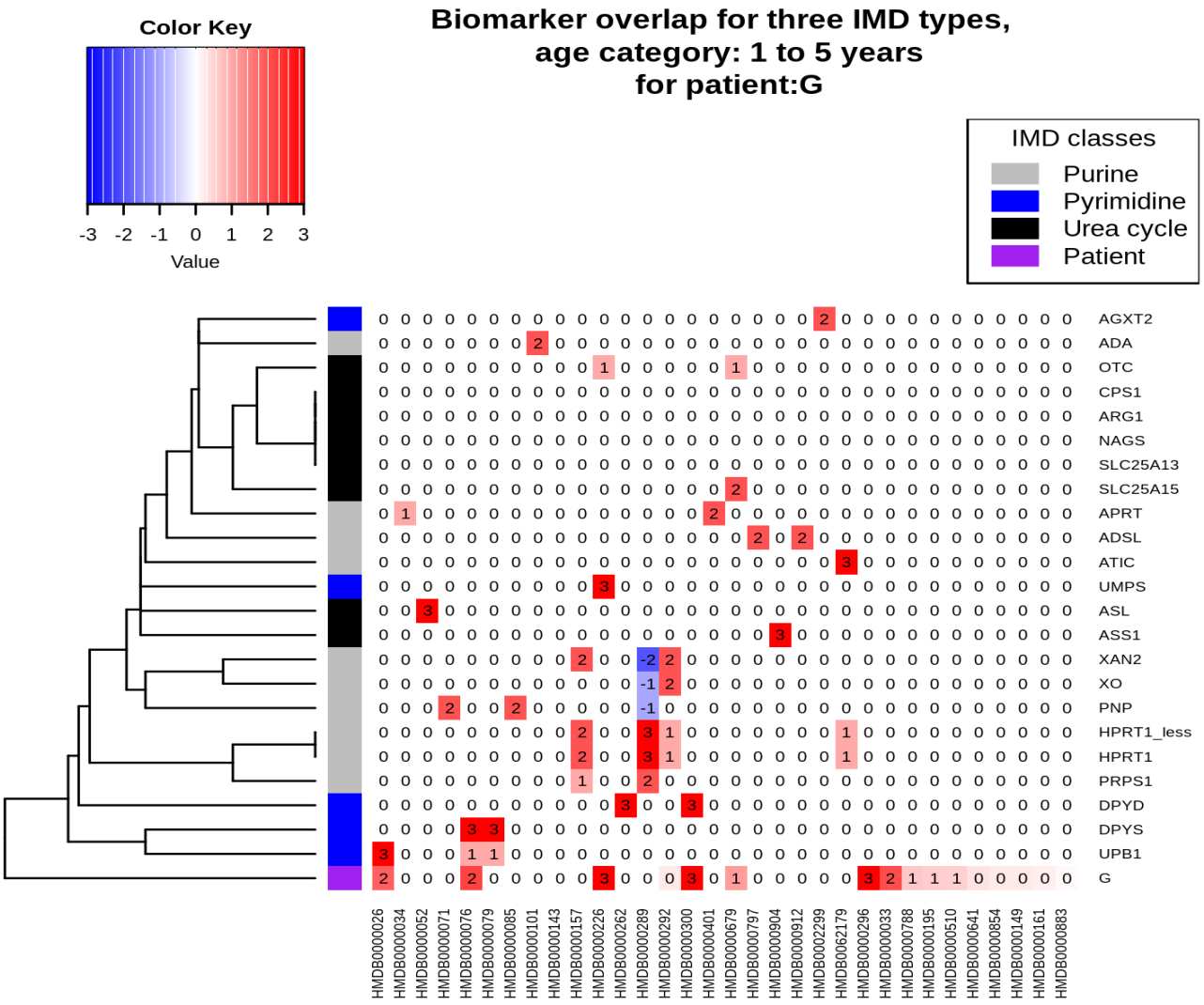
Selected Patient ID is: G , age is between: 1 to 5 years old

Relevant Biomarker data:

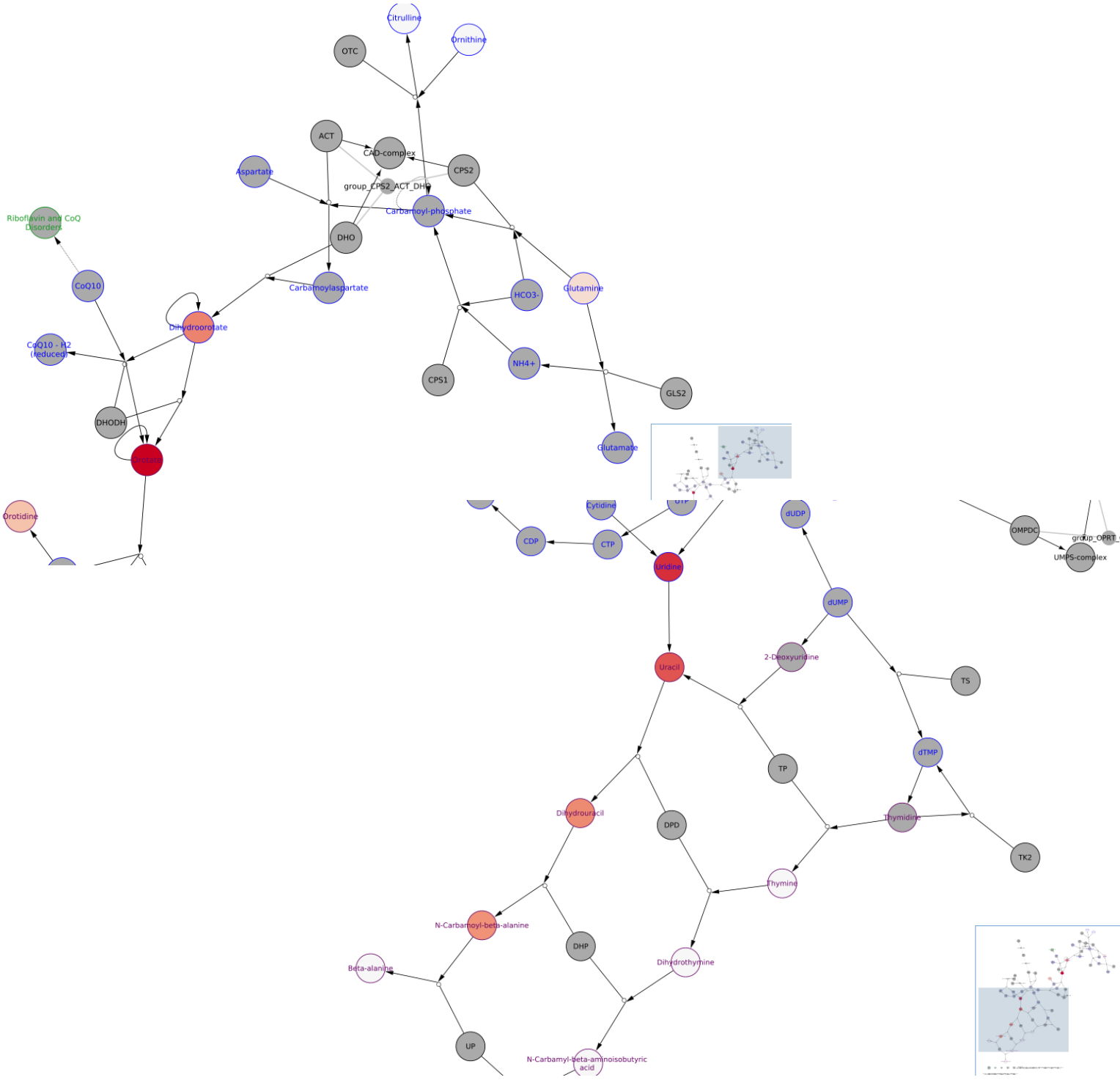
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
orootzuur	Orotate	CHEBI:30839	8.54
uridine	Uridine	CHEBI:16704	6.45
uracil	Uracil	CHEBI:17568	4.66
dihydro-orootzuur	Dihydroorotate	CHEBI:30864	2.54
dihydro-uracil	Dihydrouracil	CHEBI:15901	2.20
carnosine	Carnosine	CHEBI:15727	2.08
N-carbamyl-bala	N-Carbamoyl-beta-alanine	CHEBI:11892	1.86
alfa-aminoboterzuur	AABA	CHEBI:35621	1.42
homocitrulline	Homocitrulline	CHEBI:58148	1.13
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	1.03

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
orotidine	Orotidine	CHEBI:25722	0.65
inosine	Inosine	CHEBI:17596	0.58
alfa-amino-adipinezuur	2-Aminoadipic acid	CHEBI:37024	0.54
glutamine	Glutamine	CHEBI:58359	0.32
formimino-L-glutaminezuur	Formiminoglutamic acid	CHEBI:7274	0.25
ethanolamine	Monoethanolamine	CHEBI:16000	0.24
xanthine	Xanthine	CHEBI:17712	0.23
alanine	Alanine	CHEBI:16977	0.17
valine	Valine	CHEBI:16414	0.06

[1] "These biomarkers (as ChEBI IDs) are not in a pathway: 35621, 27596; with the following Database names: AABA, 3-Methyl-histidine"



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>
1	WP4225	Pyrimidine metabolism and related diseases	8
2	WP4584	Biomarkers for pyrimidine metabolism disorders	7
3	WP4595	Urea cycle and associated pathways	6
4	WP4022	Pyrimidine metabolism	4
5	WP4224	Purine metabolism and related disorders	3



Interpretation 1: OTC
Interpretation 2: hyperammonaemia, no apparent UCD

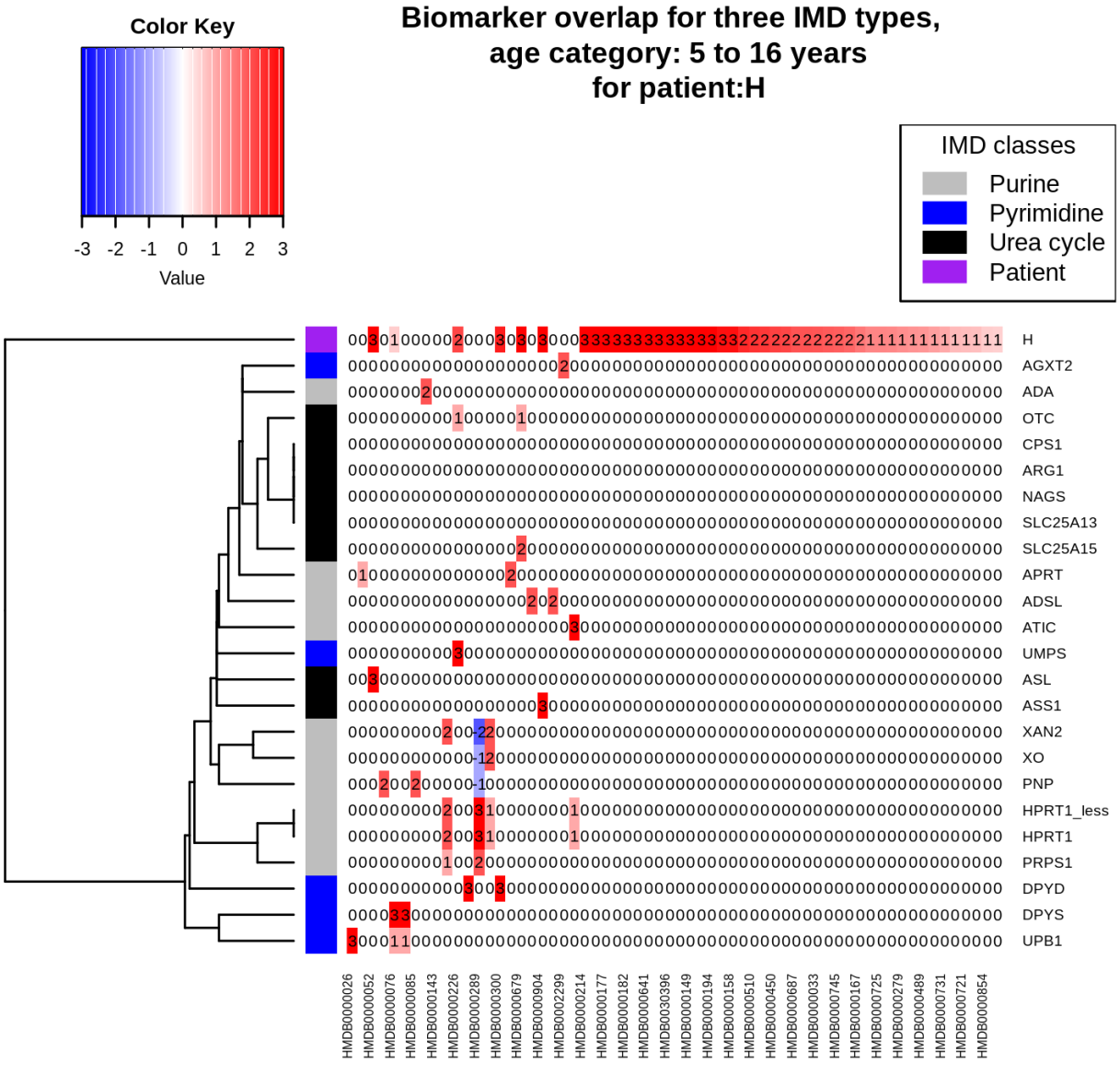
Final conclusion: Visualization is indicative of testing for OTC.

Selected Patient ID is: H , age is between: 5 to 16 years old

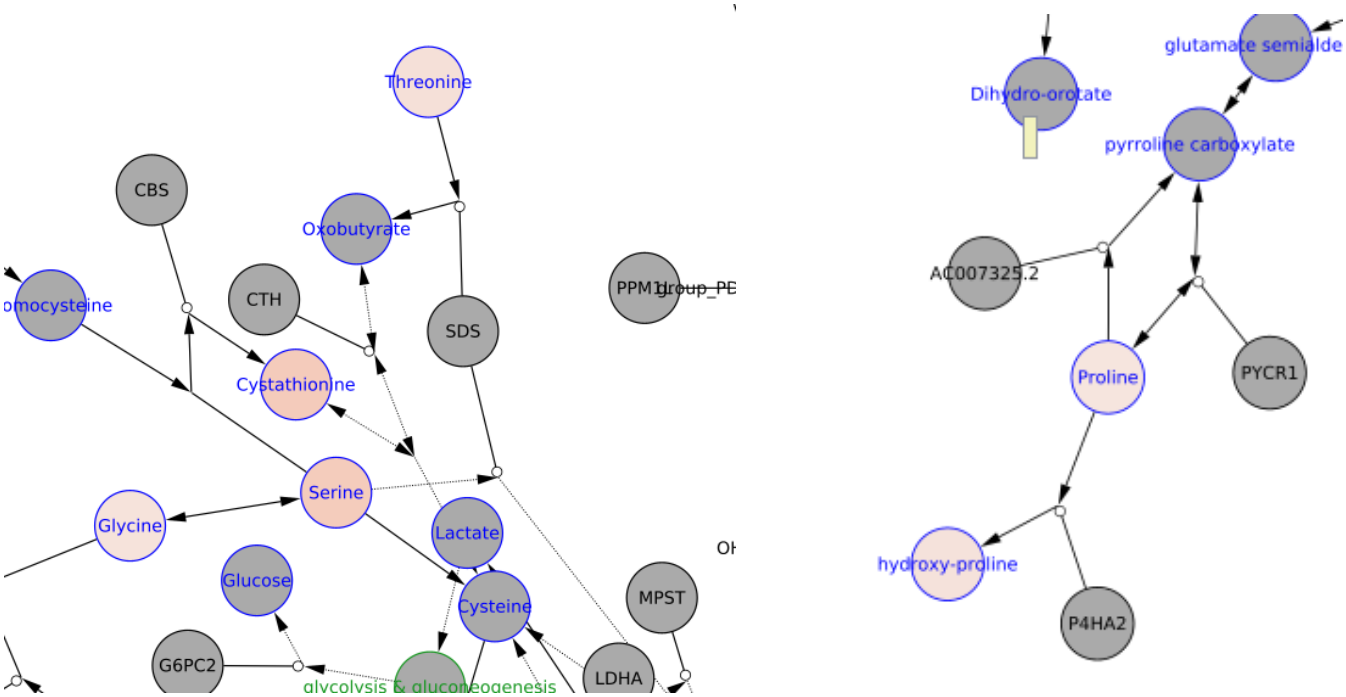
Relevant Biomarker Data (48 in total)

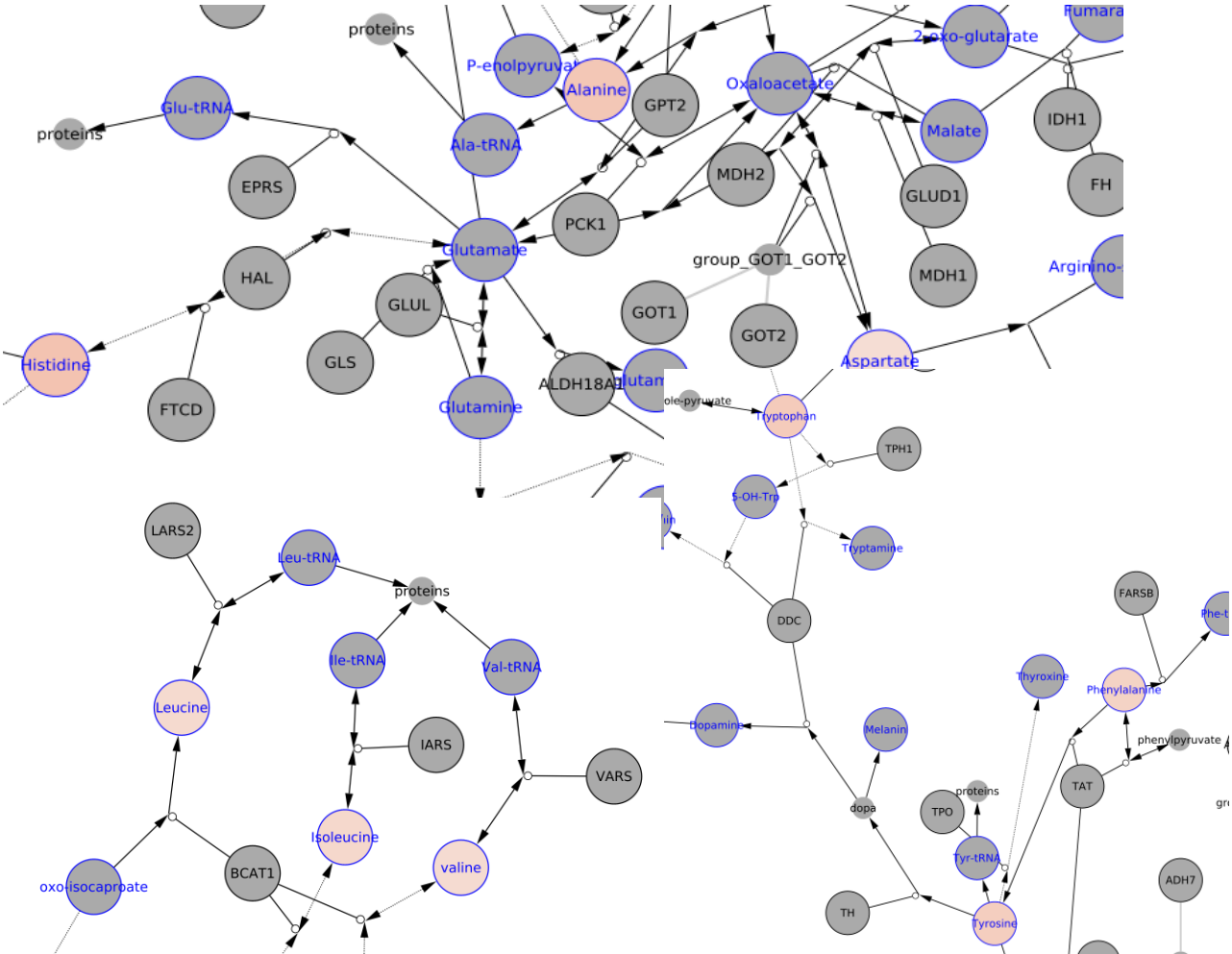
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
citrulline	Citrulline	CHEBI:57743	7.51
ornithine	Ornithine	CHEBI:46911	5.69
arginine	Arginine	CHEBI:32682	5.24
homocitrulline	Homocitrulline	CHEBI:58148	4.10
histidine	Histidine	CHEBI:15971	3.55
alanine	Alanine	CHEBI:16977	3.29
lysine	Lysine	CHEBI:18019	3.24
asparagine	Asparagine	CHEBI:17196	3.19
argininobarnsteenzuur	Argininosuccinate	CHEBI:57472	3.19
glutamine	Glutamine	CHEBI:58359	3.15

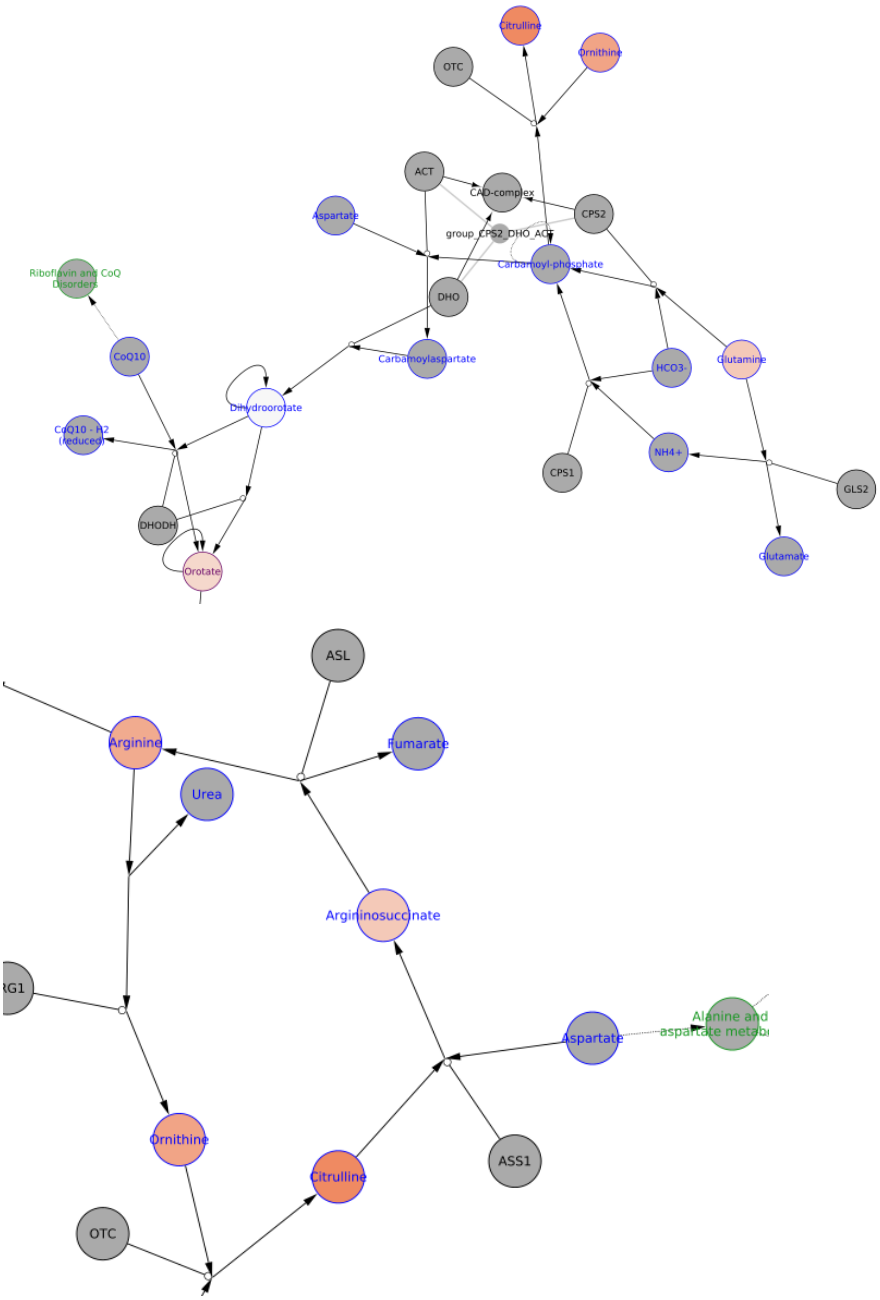
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	3.05
cystathionine	Cystathionine	CHEBI:17482	3.04
tryptofaan	Tryptophan	CHEBI:16828	3.04
sarcosine	Sarcosine	CHEBI:15611	3.00
ethanolamine	Monoethanolamine	CHEBI:16000	2.99
serine	Serine	CHEBI:17115	2.98
anserine	Anserine	CHEBI:18323	2.96
glutaminezuur	Glutamic acid	CHEBI:18237	2.81
tyrosine	Tyrosine	CHEBI:17895	2.81
uracil	Uracil	CHEBI:17568	2.64



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>
1	WP3925	Amino acid metabolism	16
2	WP4595	Urea cycle and associated pathways	10
3	WP4583	Biomarkers for urea cycle disorders	9
4	WP4225	Pyrimidine metabolism and related diseases	8
5	WP661	Glucose homeostasis	6







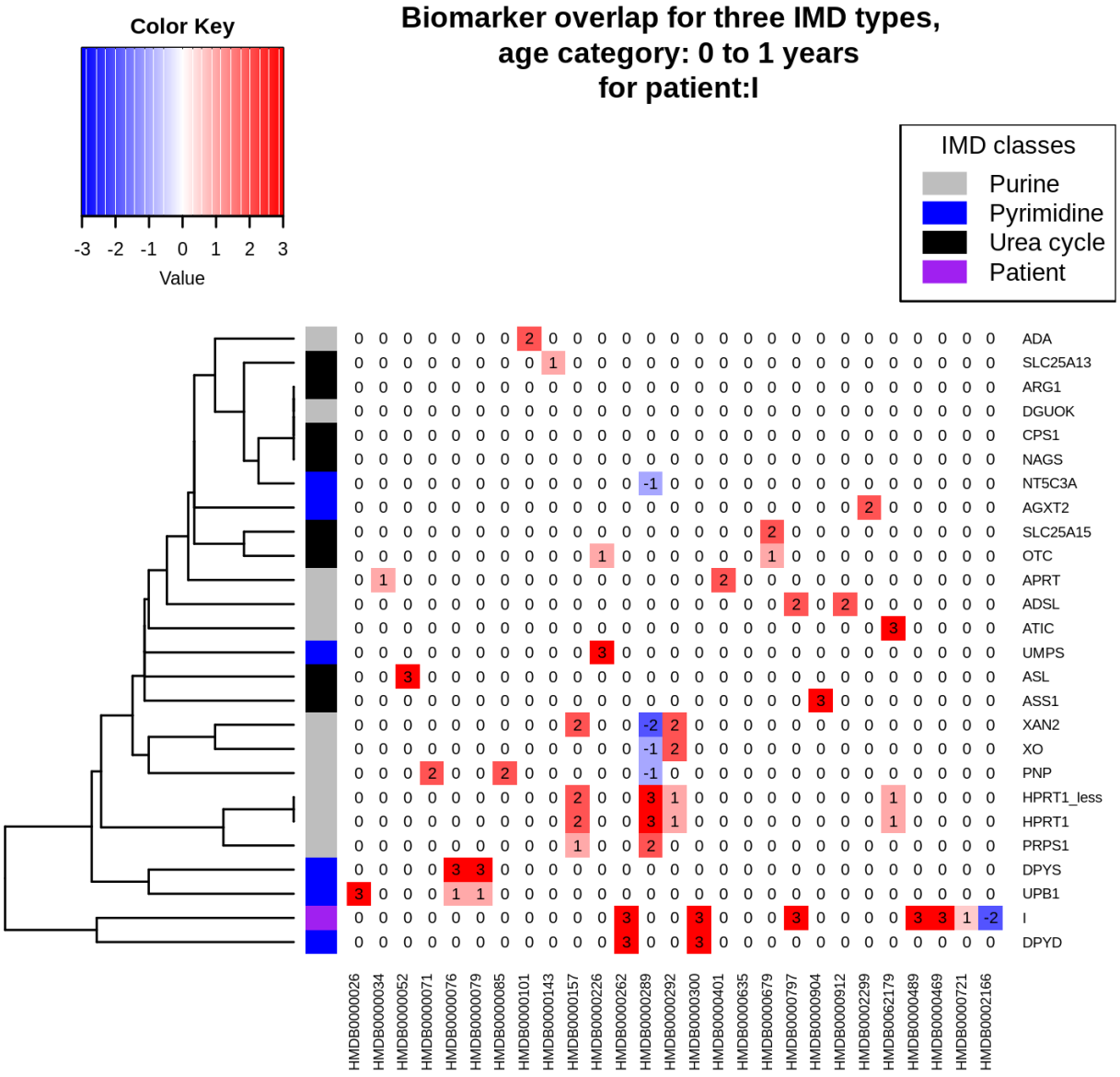
Interpretation 1: ASL
Interpretation 2: citrullinaemia type I or II

Final conclusions: Argininosuccinate is normally only elevated in ASL, so this is an unexpected finding. Example for “blind spot”. Citrulline was added as treatment for this patient, which made this case difficult to diagnose.

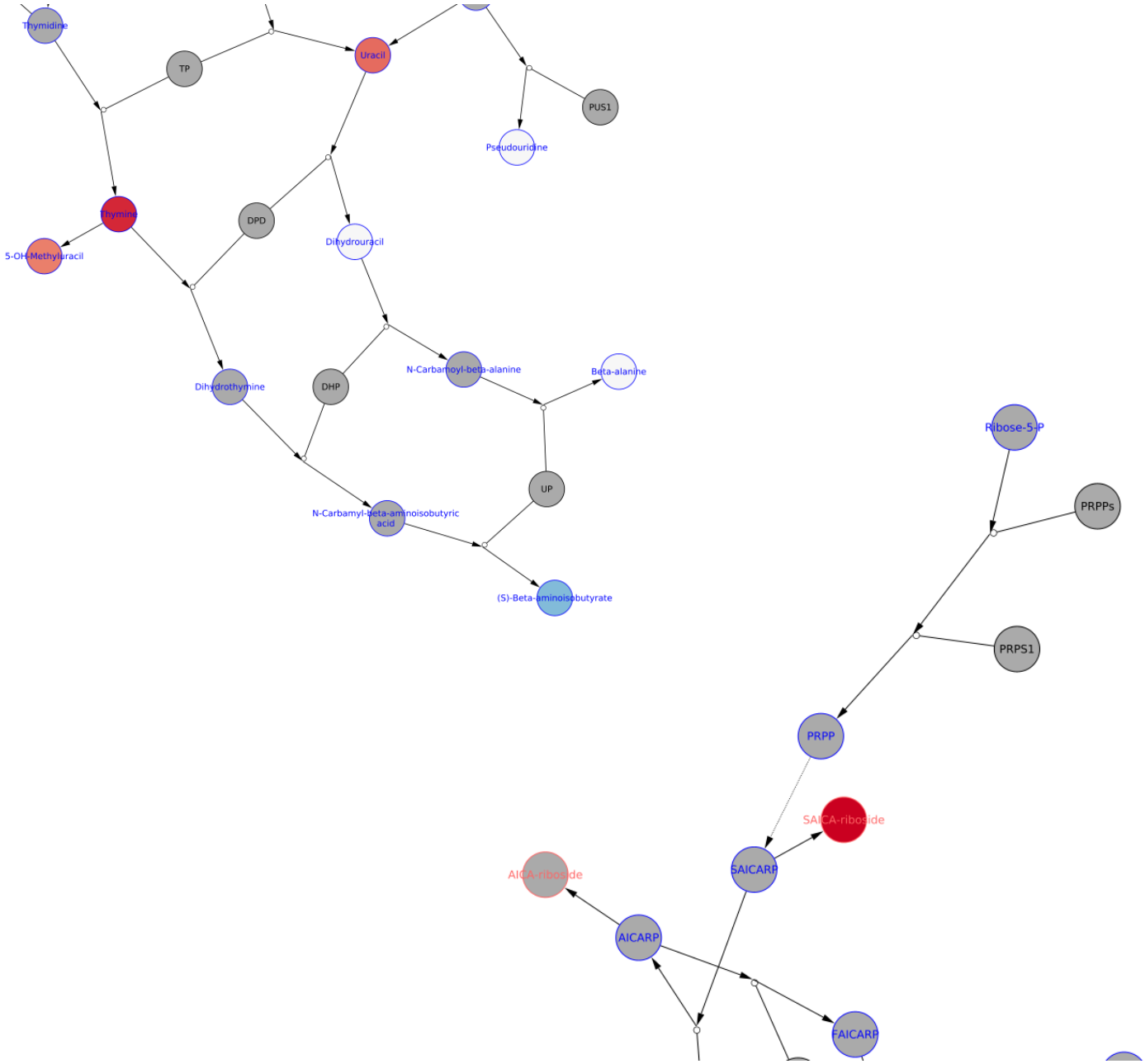
Selected Patient ID is: I , age is between: 0 to 1 years old

Relevant biomarkers

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
SAICAR	SAICARP	CHEBI:18319	9.77
thymine	Thymine	CHEBI:17821	7.68
N-aspartyl-glucosamine	N-Aspartylglucosamine	CHEBI:17261	5.75
uracil	Uracil	CHEBI:17568	4.09
5-hydroxy methyl uracil	5-(Hydroxymethyl)uracil	CHEBI:16964	3.04
gly-pro	Gly-pro	CHEBI:70744	0.61
beta-amino-isoboterzuur	(S)-Beta-aminoisobutyrate	CHEBI:57731	-2.00



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>	includedCHEBIs <chr>
1	WP4584	Biomarkers for pyrimidine metabolism disorders	4	16964 17568 17821 57731
2	WP4022	Pyrimidine metabolism	3	17568 17821 57731
3	WP4225	Pyrimidine metabolism and related diseases	3	17568 17821 57731
4	WP4157	GABA metabolism (aka GHB)	1	17568
5	WP4583	Biomarkers for urea cycle disorders	1	57731



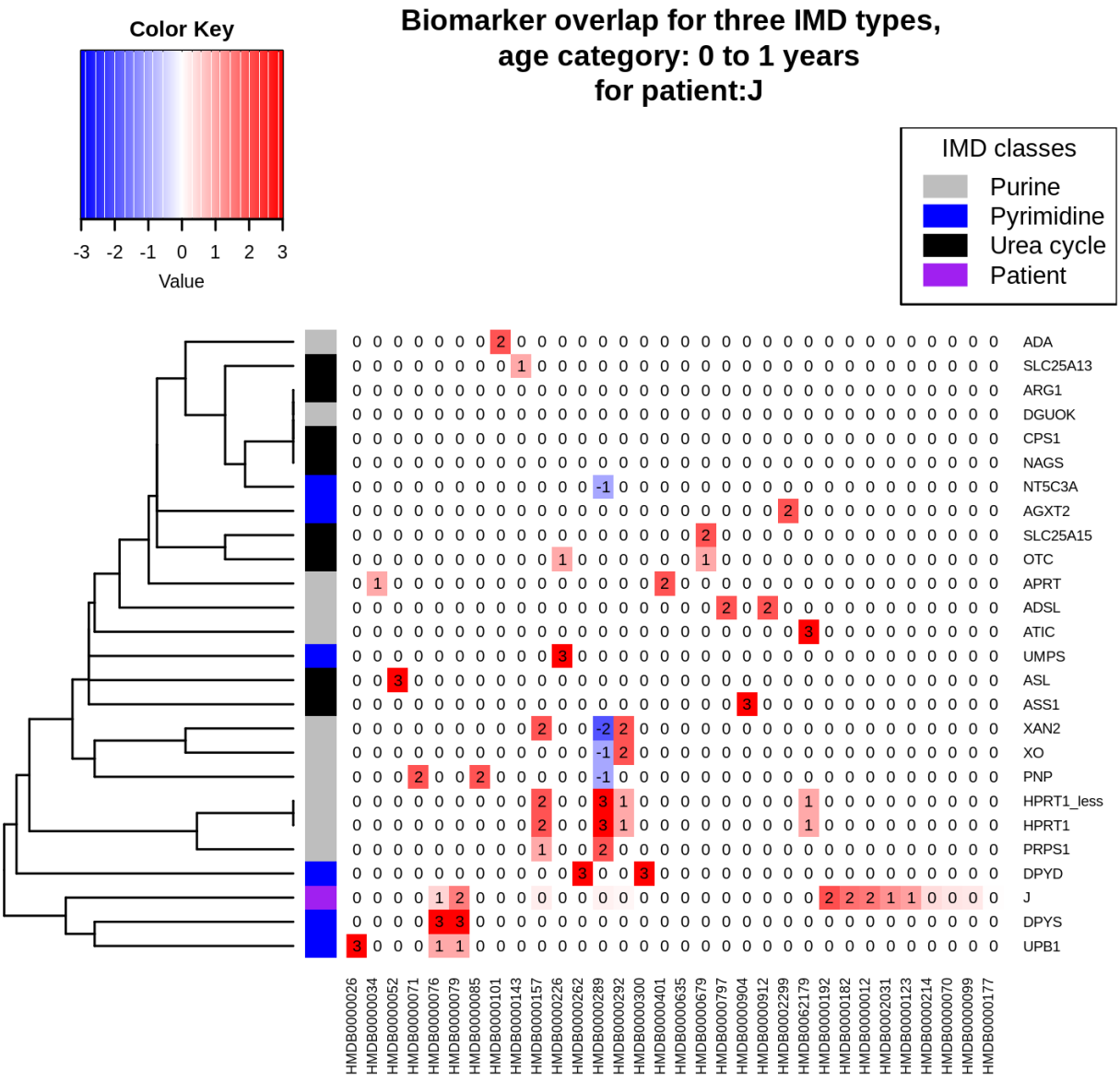
Interpretation 1: DPYD
Interpretation 2: DPYD, Interpretation: Thymine, Uracil and 5-OH-Me-Uracil are suggestive of dihydropyrimidine dehydrogenase deficiency. SAICAriboside does not make sense and by itself maybe suggests immaturity. Without SAdo or AICAriboside no indication of ADSL or ATIC deficiency.

Selected Patient ID is: J , age is between: 0 to 1 years old

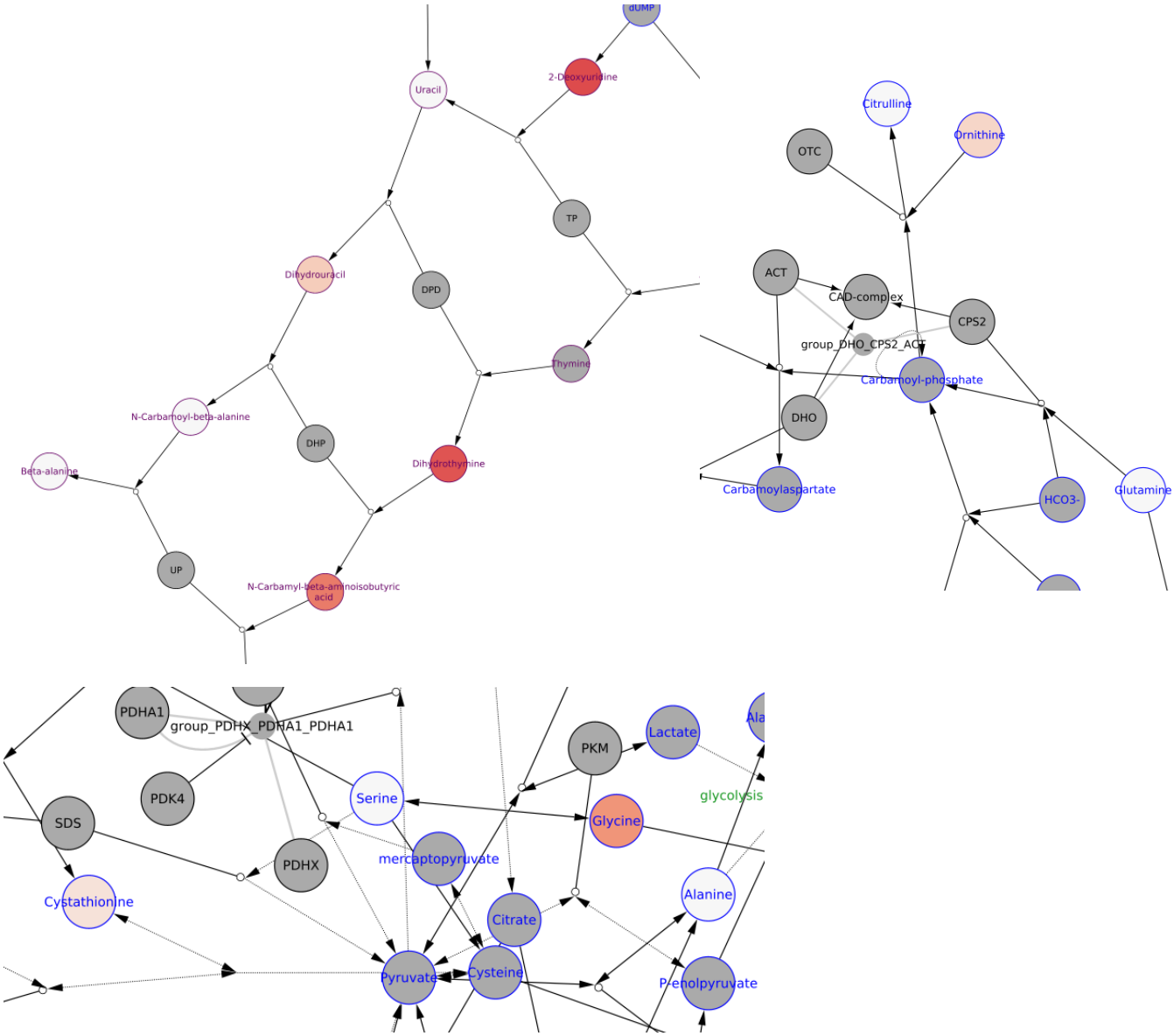
Relevant Biomarkers:

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
cystine	Cystine	CHEBI:16283	2.06
lysine	Lysine	CHEBI:18019	1.68
deoxyuridine	2-Deoxyuridine	CHEBI:16450	1.58
dihydrothymine	Dihydrothymine	CHEBI:27468	1.52
N-carbamyl-beta-AIBA	N-Carbamyl-beta-aminoisobutyric acid	CHEBI:74414	1.27
glycine	Glycine	CHEBI:15428	1.10
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	0.70
dihydro-uracil	Dihydrouracil	CHEBI:15901	0.51
ornithine	Ornithine	CHEBI:46911	0.40
pipecolinezuur	Pipecolic acid	CHEBI:30913	0.31

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
cystathionine	Cystathionine	CHEBI:17482	0.26
hypoxanthine	Hypoxanthine	CHEBI:17368	0.20
urinezuur	Urate	CHEBI:17775	0.16
xanthine	Xanthine	CHEBI:17712	0.10
histidine	Histidine	CHEBI:15971	0.03



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>	includedCHEBIs <chr>
1	WP4225	Pyrimidine metabolism and related diseases	5	15901 16450 27468 46911 74414
2	WP4584	Biomarkers for pyrimidine metabolism disorders	4	15901 16450 27468 74414
3	WP15	Selenium micronutrient network	4	17368 17482 17712 17775
4	WP3925	Amino acid metabolism	3	15428 15971 17482
5	WP661	Glucose homeostasis	3	15971 17368 18019



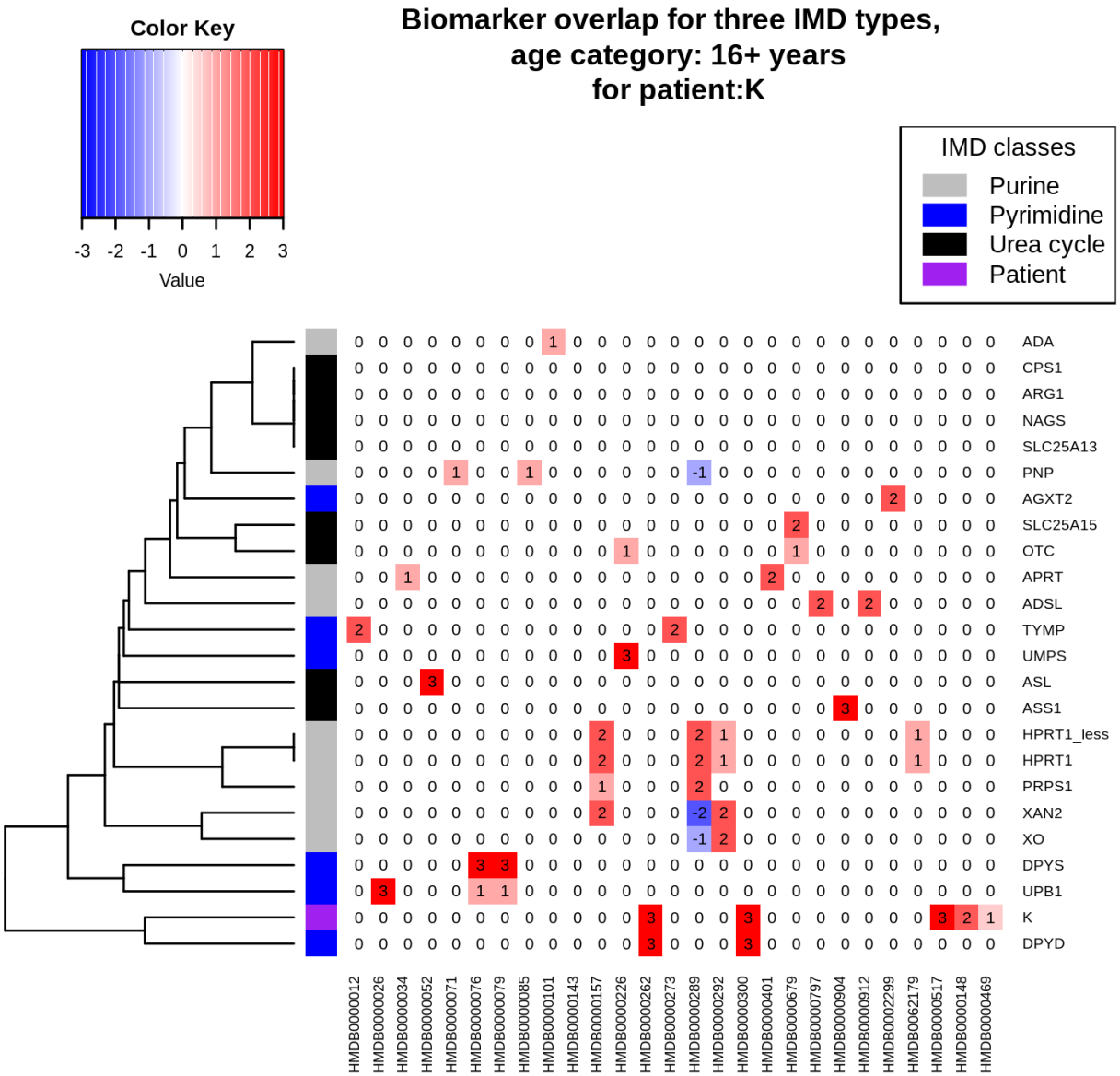
Interpretation 1: Cystinuria?
Interpretation 2: Interpretation: no diagnosis. Cystine and lysine suggest immaturity. Thymine degradation prominent, but pattern does not fit beta-ureidoprionase deficiency. Probably very young infant.

Final conclusions: Difficult sample to diagnose.

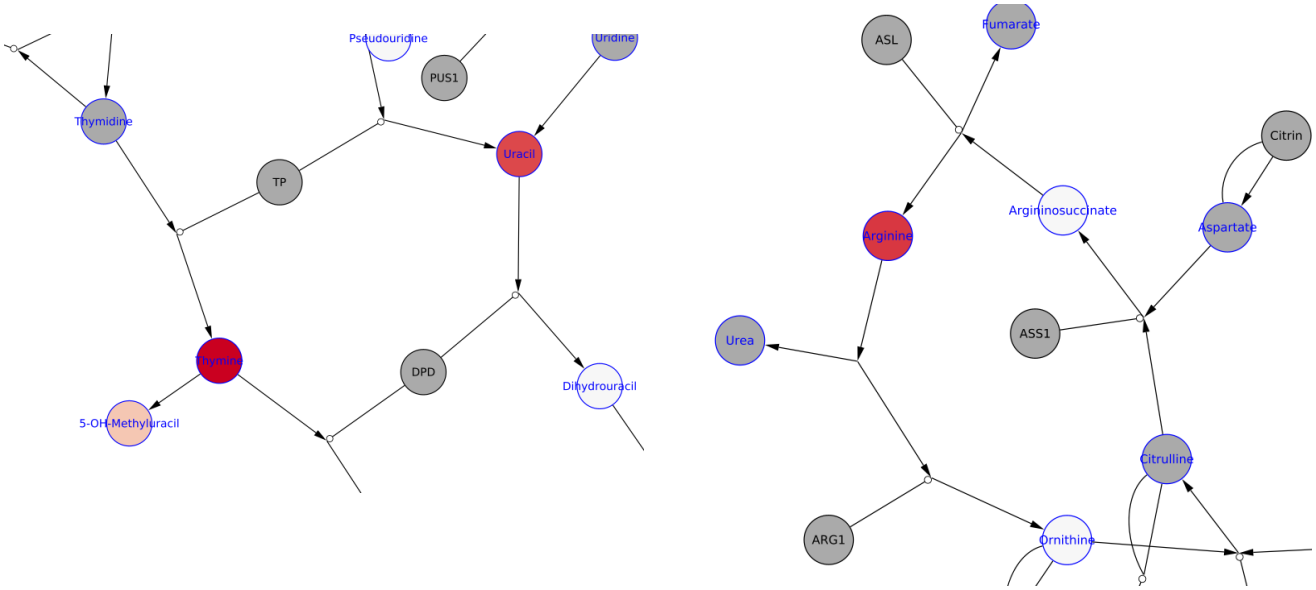
Selected Patient ID is: K , age is between: 16+ years old

Relevant Biomarkers:

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
thymine	Thymine	CHEBI:17821	4.88
arginine	Arginine	CHEBI:32682	3.58
uracil	Uracil	CHEBI:17568	3.18
glutaminezuur	Glutamic acid	CHEBI:18237	1.93
5-hydroxy methyl uracil	5-(Hydroxymethyl)uracil	CHEBI:16964	0.58



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>	includedCHEBIs <chr>
1	WP4584	Biomarkers for pyrimidine metabolism disorders	3	16964 17568 17821
2	WP4022	Pyrimidine metabolism	2	17568 17821
3	WP4225	Pyrimidine metabolism and related diseases	2	17568 17821
4	WP4595	Urea cycle and associated pathways	2	17568 32682
5	WP2636	Common pathways underlying drug addiction	1	18237



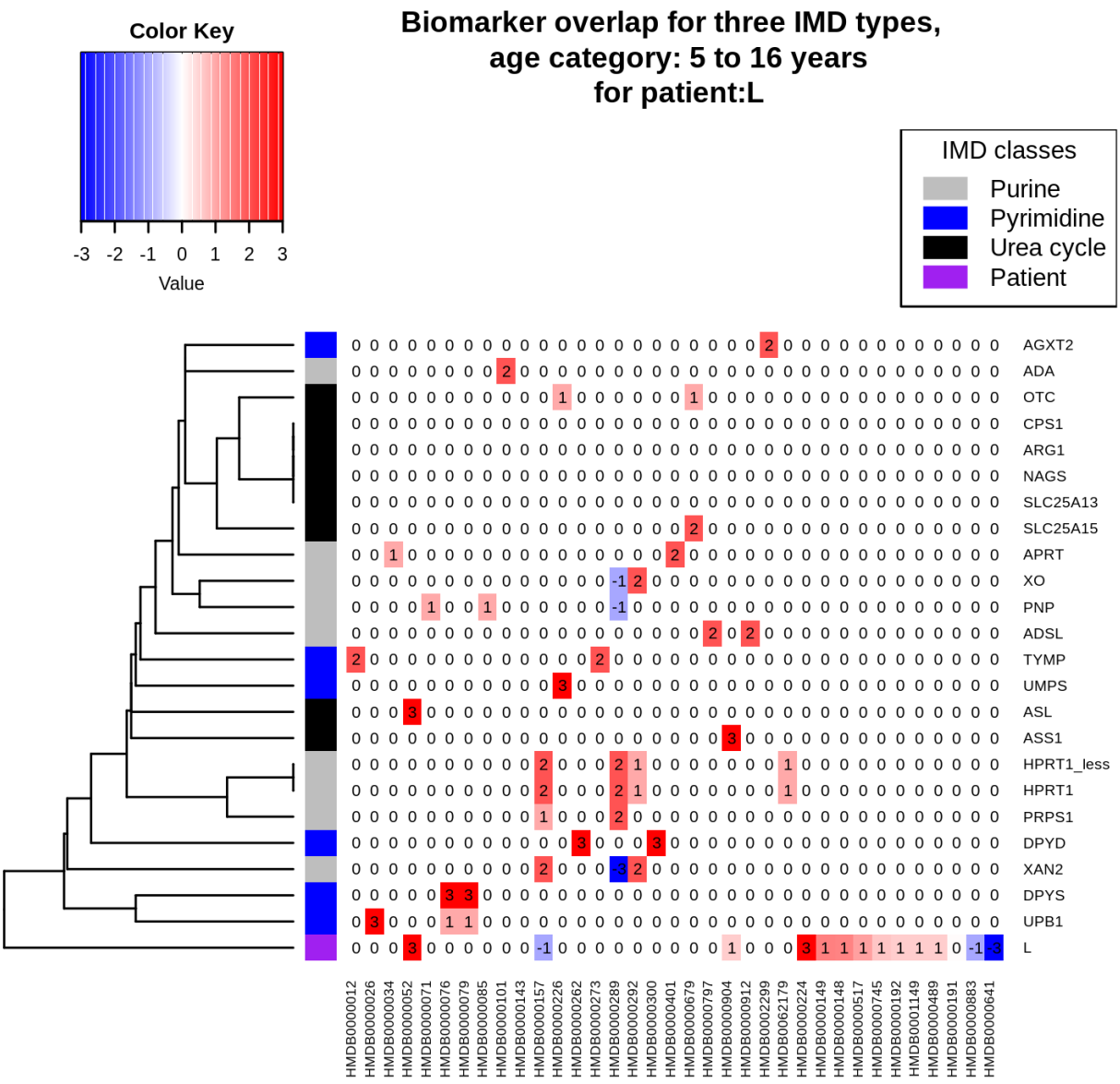
Interpretation 1: DPYD
Interpretation 2: DPYD, Interpretation: Thymine, Uracil and 5-OH-Me-Uracil are suggestive of dihydropyrimidine dehydrogenase deficiency. 5-OH-Me-Uracil is significantly lower than in patient I. Arginine is elevated, I cannot explain well why.

Selected Patient ID is: L , age is between: 5 to 16 years old

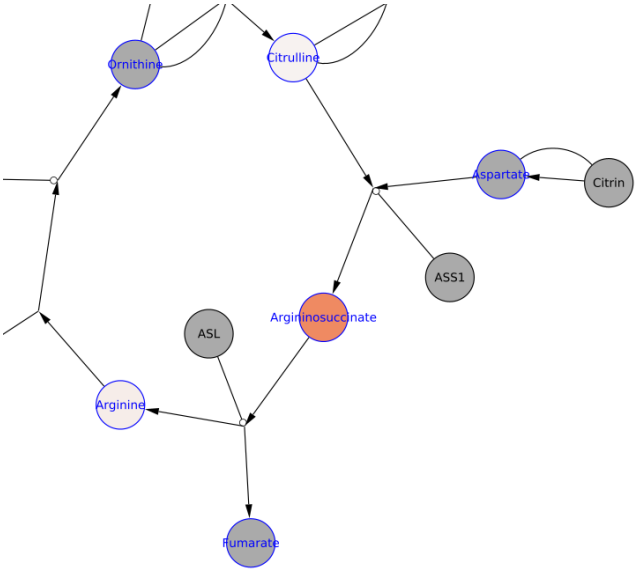
Relevant Biomarkers:

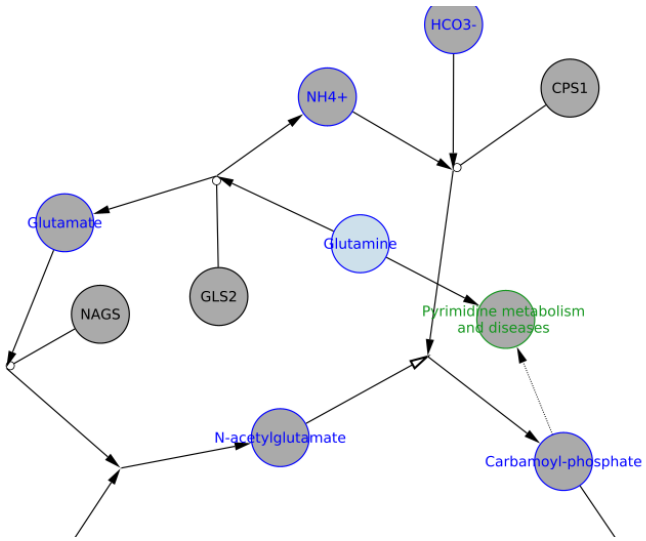
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
argininobarnsteenzuur	Argininosuccinate	CHEBI:57472	11.56
fosfoethanolamine	Phosphoethanolamine	CHEBI:17553	5.21
ethanolamine	Monoethanolamine	CHEBI:16000	1.48
glutaminezuur	Glutamic acid	CHEBI:18237	1.44
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	1.23
arginine	Arginine	CHEBI:32682	1.10
homocarnosine	Homocarnosine	CHEBI:85981	0.66
citrulline	Citrulline	CHEBI:57743	0.58
cystine	Cystine	CHEBI:16283	0.58
delta-aminolevulinezuur	Aminolevulinic acid	CHEBI:17549	0.58

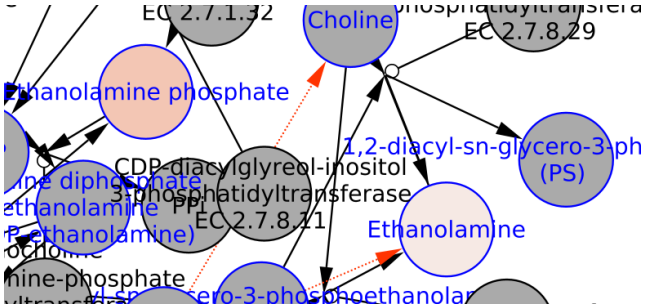
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
N-aspartyl-glucosamine	N-Aspartylglucosamine	CHEBI:17261	0.58
asparaginezuur	Aspartic Acid	CHEBI:17053	0.04
hypoxanthine	Hypoxanthine	CHEBI:17368	-1.00
valine	Valine	CHEBI:16414	-1.00
glutamine	Glutamine	CHEBI:58359	-3.39



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>	includedCHEBIs <chr>
1	WP4571	Urea cycle and related diseases	4	32682 57472 57743 58359
2	WP4583	Biomarkers for urea cycle disorders	4	32682 57472 57743 58359
3	WP4595	Urea cycle and associated pathways	4	32682 57472 57743 58359
4	WP4224	Purine metabolism and related disorders	3	17053 17368 58359
5	WP2533	Glycerophospholipid biosynthetic pathway	2	16000 17553





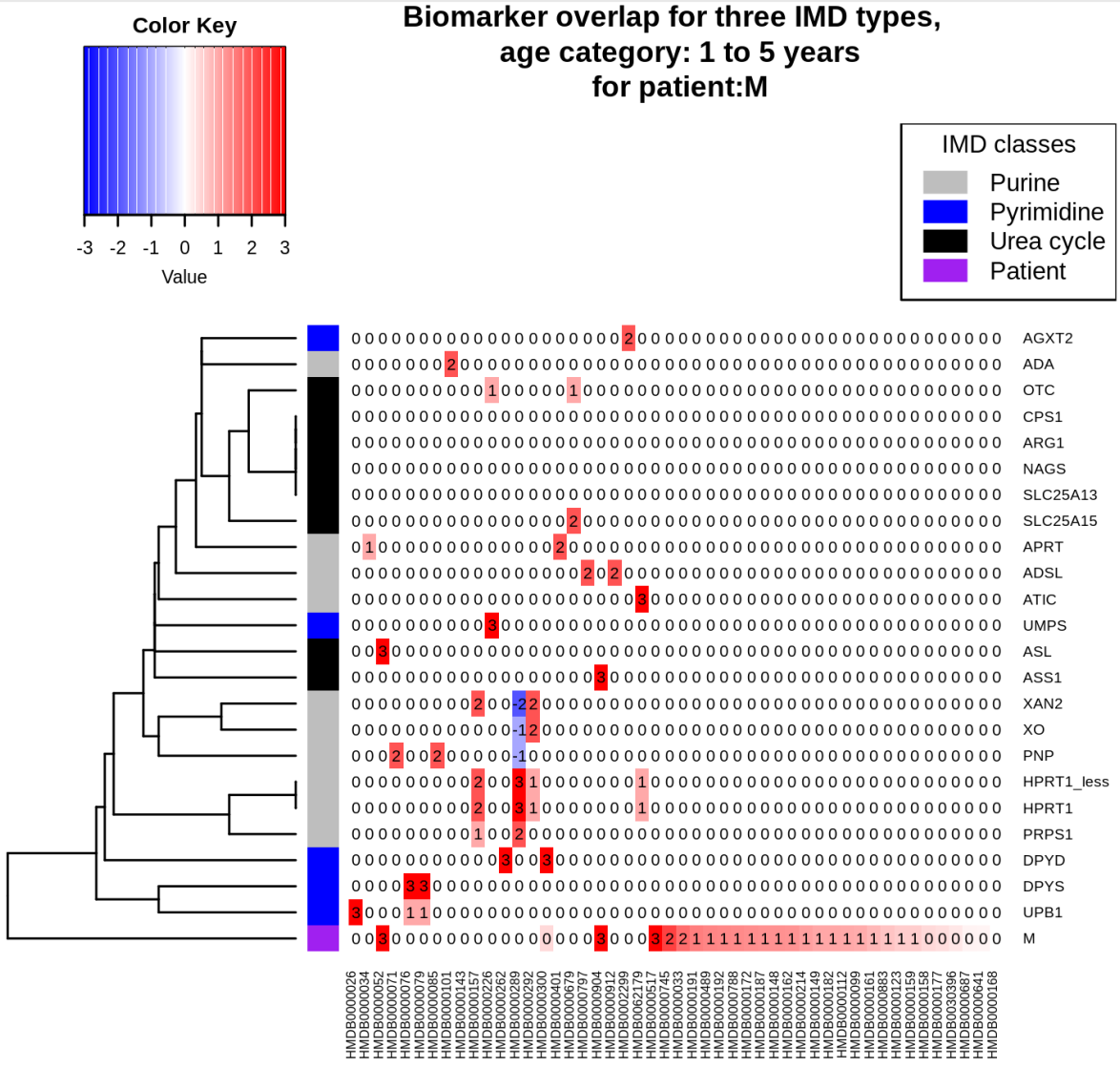


Interpretation 1: ASL
Interpretation 2: ASL

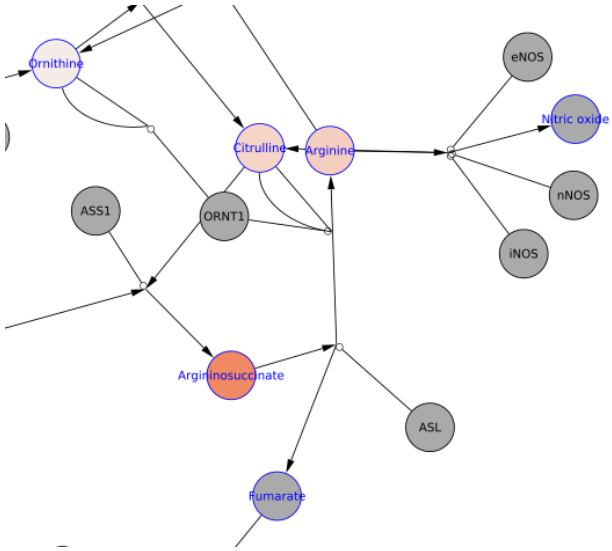
Selected Patient ID is: M , age is between: 1 to 5 years old

Relevant Biomarkers (out of 30):

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
argininobarnsteenzuur	Argininosuccinate	CHEBI:57472	10.62
arginine	Arginine	CHEBI:32682	4.04
citrulline	Citrulline	CHEBI:57743	3.32
homocarnosine	Homocarnosine	CHEBI:85981	2.22
carnosine	Carnosine	CHEBI:15727	1.70
asparaginezuur	Aspartic Acid	CHEBI:17053	1.49
N-aspartyl-glucosamine	N-Aspartylglucosamine	CHEBI:17261	1.42
cystine	Cystine	CHEBI:16283	1.38
orotidine	Orotidine	CHEBI:25722	1.36
isoleucine	Isoleucine	CHEBI:17191	1.32
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	1.32
serine	Serine	CHEBI:17115	1.27
glutaminezuur	Glutamic acid	CHEBI:18237	1.27
proline	Proline	CHEBI:17203	1.26
ornithine	Ornithine	CHEBI:46911	1.12
ethanolamine	Monoethanolamine	CHEBI:16000	1.02
lysine	Lysine	CHEBI:18019	1.01
gamma-aminoboterzuur	GABA	CHEBI:16865	1.00
cystathionine	Cystathionine	CHEBI:17482	0.96
alanine	Alanine	CHEBI:16977	0.83



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>
1	WP3925	Amino acid metabolism	14
2	WP4595	Urea cycle and associated pathways	9
3	WP661	Glucose homeostasis	6
4	WP4583	Biomarkers for urea cycle disorders	6
5	WP4571	Urea cycle and related diseases	5



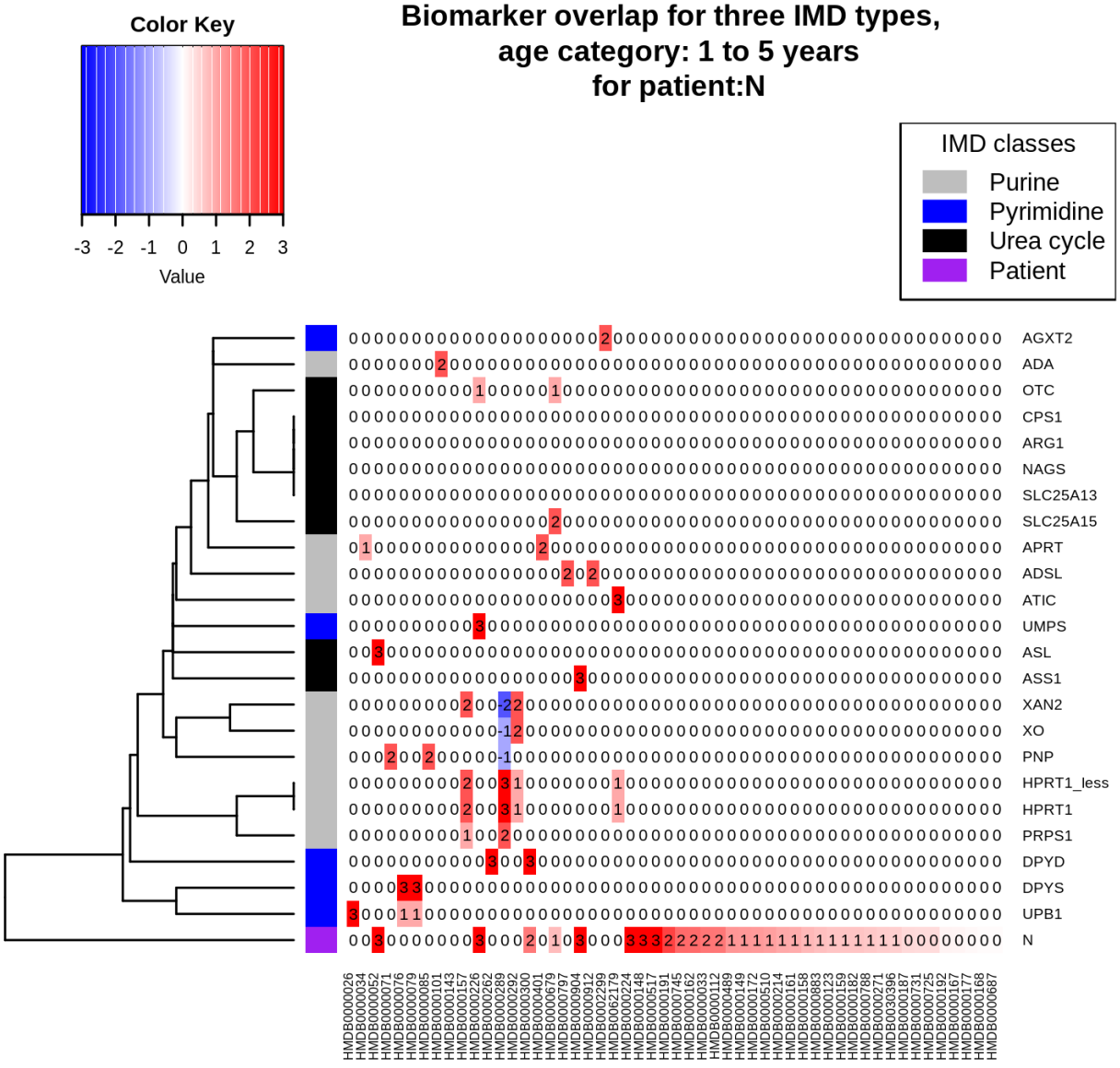
Interpretation 1: ASL
Interpretation 2: ASL

Selected Patient ID is: N , age is between: 1 to 5 years old

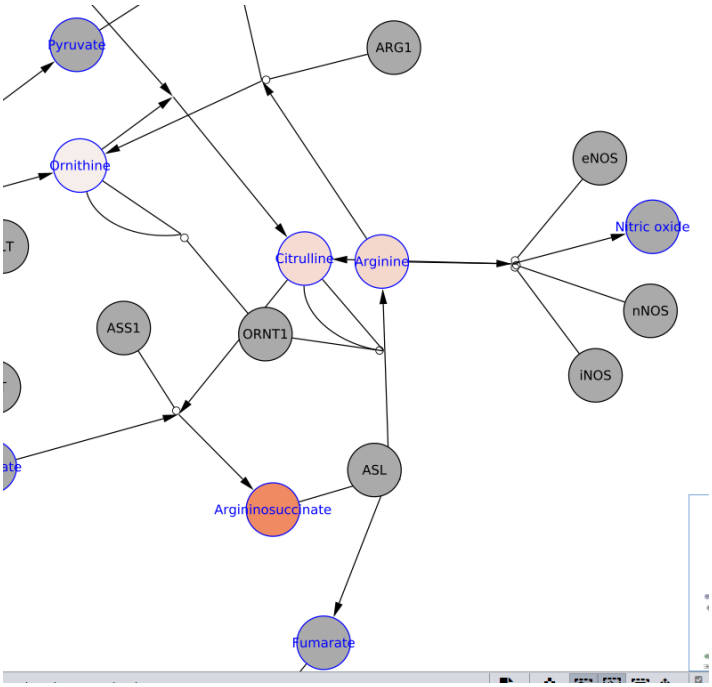
Relevant Biomarkers (out of 36):

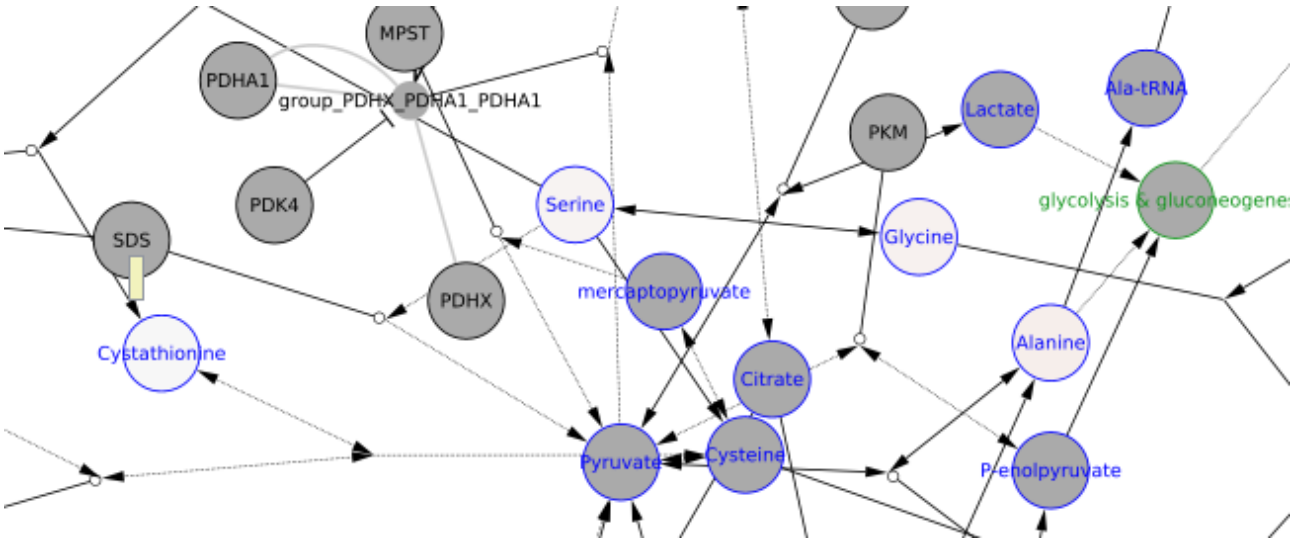
Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
argininobarnsteenzuur	Argininosuccinate	CHEBI:57472	12.43
fosfoethanolamine	Phosphoethanolamine	CHEBI:17553	5.80
orootzuur	Orotate	CHEBI:30839	5.01
glutaminezuur	Glutamic acid	CHEBI:18237	3.86
arginine	Arginine	CHEBI:32682	3.57
citrulline	Citrulline	CHEBI:57743	3.15
asparaginezuur	Aspartic Acid	CHEBI:17053	2.20
homocarnosine	Homocarnosine	CHEBI:85981	1.74
proline	Proline	CHEBI:17203	1.72
3-methyl-histidine	3-Methyl-histidine	CHEBI:27596	1.65

Dutch.name <chr>	Database.name <chr>	ID <chr>	log.Change <dbl>
carnosine	Carnosine	CHEBI:15727	1.60
gamma-aminoboterzuur	GABA	CHEBI:16865	1.58
uracil	Uracil	CHEBI:17568	1.56
N-aspartyl-glucosamine	N-Aspartylglucosamine	CHEBI:17261	1.22
ethanolamine	Monoethanolamine	CHEBI:16000	1.20
isoleucine	Isoleucine	CHEBI:17191	1.17
alfa-amino-adipinezuur	2-Aminoadipic acid	CHEBI:37024	1.11
ornithine	Ornithine	CHEBI:46911	1.00
alanine	Alanine	CHEBI:16977	0.97
tyrosine	Tyrosine	CHEBI:17895	0.83



	pathway <chr>	pathwayTitle <chr>	CHEBIsInPWs <int>
1	WP3925	Amino acid metabolism	15
2	WP4595	Urea cycle and associated pathways	9
3	WP4583	Biomarkers for urea cycle disorders	7
4	WP661	Glucose homeostasis	6
5	WP4225	Pyrimidine metabolism and related diseases	5





Interpretation 1: ASL
Interpretation 2: ASL