

Estimation of health-state utility values in phenylketonuria, via clinical-expert proxy assessment with a generic measure (SF-12[®]v2) of patient-reported impacts from a disease-specific measure (PKU-QOL): Interim results

Thomas O'Connell,¹ Suzanne Hollander,² Suresh Vijay,³ Nicola Longo,⁴ Roberto Zori,⁵ Jonathan J. Woolley,¹ Ioannis Tomazos⁶

¹Medicus Economics, Boston, MA, USA ²Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA, USA, ³Birmingham Children's Hospital, Birmingham, UK, ⁴Department of Human Genetics, University of California, Los Angeles, Los Angeles, CA, USA, ⁵Pediatric Genetics and Metabolism, University of Florida, Gainesville, FL, USA, ⁶PTC Therapeutics, Inc, Warren, New Jersey, USA

Supplementary materials

Individual-level patient data (N=16)

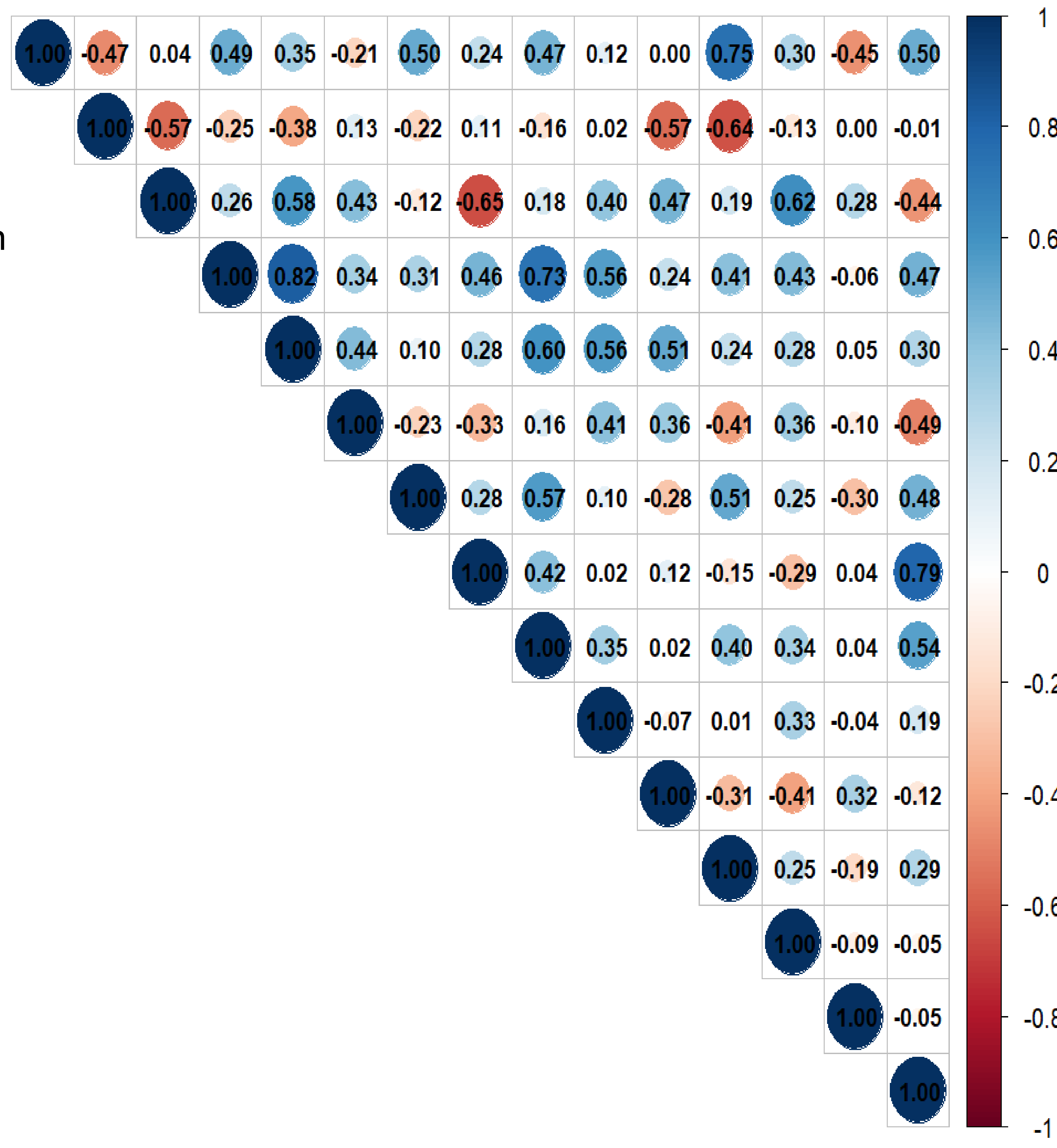
Patients 7, 12, 13, and 14 informed the definition of health state A

Patient	Country	Age (years) at BL	Controlled blood Phe at BL*	Dietary Phe challenge in MD-004	Max. follow-up in MD-004 (months)	Blood Phe levels (µmol/L)			Total daily protein equivalent consumed (mg/kg/day)			Supplemental (Phe-free substitute) % of total protein			EQ-5D VAS			PKU-QOL overall impact score		
						BL	Last visit	Diff.	BL	Last visit	Diff.	BL	Last visit	Diff.	BL	Last visit	Diff.	BL	Last visit	Diff.
1	TUR	12-17	Y	N	26	376	726	350	0.13	0.16	0.04	80%	81%	1%	50	80	30	43	64	21
2	TUR	12-17	Y	N	20	556	503	-53	0.49	1.03	0.54	61%	0%	-61%	75	85	10	33	48	15
3	BRA	12-17	Y	Y	26	454	115	-339	0.38	1.00	0.62	49%	0%	-49%	55	90	35	40	33	-8
4	TUR	12-17	Y	Y	26	507	285	-222	0.57	0.37	-0.20	64%	33%	-31%	60	65	5	48	33	-15
5	PRT	12-17	Y	Y	26	515	177	-338	0.29	1.32	1.02	61%	0%	-61%	10	95	85	58	40	-18
6	PRT	12-17	Y	Y	20	566	582	16	0.34	0.59	0.24	68%	46%	-22%	70	95	25	25	43	18
7	AUS	12-17	N	N	1	883	993	110	0.25	0.28	0.04	70%	47%	-23%	60	70	10	45	50	5
8	PRT	≥18	Y	N	14	500	322	-178	0.62	0.47	-0.15	64%	70%	6%	60	95	35	19	21	2
9	AUS	≥18	Y	Y	26	406	57	-349	0.15	0.49	0.34	86%	56%	-30%	50	70	20	65	42	-23
10	BRA	≥18	Y	Y	1	442	107	-335	0.11	0.36	0.25	0%	0%	-	75	80	5	15	48	33
11	CAN	≥18	Y	Y	26	566	337	-229	0.25	0.26	0.02	71%	69%	-2%	75	70	-5	58	67	9
12	AUS	≥18	N	N	20	652	1060	408	0.05	0.05	0.00	92%	92%	0%	70	NA	NA	38	44	5
13	CAN	≥18	N	N	20	717	99	-618	0.31	0.42	0.10	72%	66%	-6%	60	95	35	19	6	-13
14	DEU	≥18	N	N	1	869	855	-14	0.34	0.36	0.02	64%	61%	-3%	75	80	5	27	23	-4
15	BRA	≥18	N	Y	26	632	107	-525	0.52	1.01	0.49	0%	0%	-	75	85	10	25	10	-15
16	TUR	≥18	N	Y	20	1195	407	-788	0.40	1.12	0.71	0%	0%	-	60	70	10	35	29	-6

*Baseline blood Phe <600 for ages ≥12 years. AUS, Australia; BL, baseline; BRA, Brazil; CAN, Canada; DEU, Germany; Diff, difference; Phe, phenylalanine; PKU-QOL, Phenylketonuria Quality of Life; PRT, Portugal; TUR, Türkiye; VAS, visual analog scale

Bivariate associations

1. Blood-Phe concentration (µmol/L)
2. Daily protein-equivalent consumption (g/kg/day)
3. Amino-acid suppl. % of protein-equivalent consumption
4. Overall impact (domain score)
5. Social impact (domain score)
6. Emotional impact - Unfairness having PKU
7. Emotional impact - Self-esteem
8. Practical impact - Burden of care
9. Practical impact - Burden of visits
10. Social impact - Explain situation to others
11. Social impact - Difficulty making friends
12. Admin. of supplements - Guilt if poor adherence
13. Admin. of supplements - Taste of supplements
14. Dietary protein restriction - Food temptations
15. Dietary protein restriction - Food enjoyment



Mapping of SF-12[®]v2 item responses to SF-6Dv2 dimensions

Item responses on the SF-12[®]v2 were mapped to the SF-6Dv2 as follows:

Physical functioning:

- PF02 ("moderate activities") response of "Yes, limited a lot" = 4, response of "Yes, limited a little" = 3
- PF04 ("climbing several flights of stairs") assumed to correspond to "vigorous activity", and response of "Yes, limited a little" = 2; otherwise = 1.

Note: Possible score of 5 on the SF-6Dv2 dimensions (corresponding to "Limited in bathing and dressing a lot") cannot be inferred from the SF-12[®]v2.

Role functioning:

- Maximum of reversed responses (as per Mulhern et al. Appendix Table 1) of RP02 and RE02 (both originally ordered from "All of the time" = 1 to "None of the time" = 5).
- RP02 reflects "accomplished less than they would like as a result of their physical health", while RE02 reflects "accomplished less than they would like as a result of any emotional problems (such as feeling depressed or anxious)". The maximum of either response is used, as the SF-6Dv2 role-functioning dimension reflect "accomplished less than you would like" regardless of the cause.

Social functioning:

- Reversed responses (as per Mulhern et al. Appendix Table 1) of SF02 (originally ordered from "All of the time" = 1 to "None of the time" = 5).

Bodily pain:

- Mapped directly from responses to BP02.

Mental health:

- Reversed responses (as per Mulhern et al. Appendix Table 1) of MH04 (originally ordered from "All of the time" = 1 to "None of the time" = 5).

Vitality:

- Mapped directly from responses to VT02.



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Permission to use the SF-12[®]v2 (SF-12[®]v2 Health Survey) was granted by IQVIA Inc., and all licensing and permission requests should be directed to IQVIA Inc. at COAsolutions@iqvia.com.

CONTACT INFORMATION: ytomazos@ptcbio.com

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