

Supplemental Data Table S7. Comparison of metabolite level among groups

Biomarkers	Cutoff*	APC (n=59)		PCD (n=125)		PPC (n=85)		P
		N of samples	Median level (range)	N of samples	Median level (range)	N of samples	Median level (range)	
TSH	12	30	15 (12.1-94.1)	57	16.6 (12-94.1)	17	47.5 (12.9-94.1)	0.0044
FT4	0.8	10	0.35 (0.2-0.6)	34	0.5 (0.1-0.7)	19	0.5 (0.1-0.7)	0.0299
Total galactose	13	12	23.4 (16.5-40.4)	30	18.4 (13.5-40.4)	11	15.4 (13.1-32.2)	0.0810
Citrulline	55	4	238 (129-430)	1	61.5	0	NA	NA
Phenylalanine	130	1	142	0	NA	2	3.8 (2.6-4.9)	NA
C3	5	3	10 (9-10)	3	10 (9-11)	0	NA	NA
C5OH	0.6	3	1.1 (1.1-2.6)	2	1.8 (1.1-2.6)	2	4.1 (1.1-7.3)	NA

Nine samples showed multiple metabolite abnormalities. All cases were not validated using an independent Sanger sequencing. The Kruskal–Wallis test was performed to compare the metabolite levels among group with MedCalc version 11.5.1.0 (Mariakerke, Belgium). *P* values less than 0.05 were considered statistically significant.

*The units of TSH, FT4, total galactose, citrulline, Phe, C3, and C5OH were mU/L, ng/dL, μmol/L, μmol/L, μmol/L, μmol/L, respectively.

Abbreviations: APC, Association-Positive Cases; PCD, Positive Cases with Discrepancy; PPC, Presumptive Positive Cases; NA, not applicable.