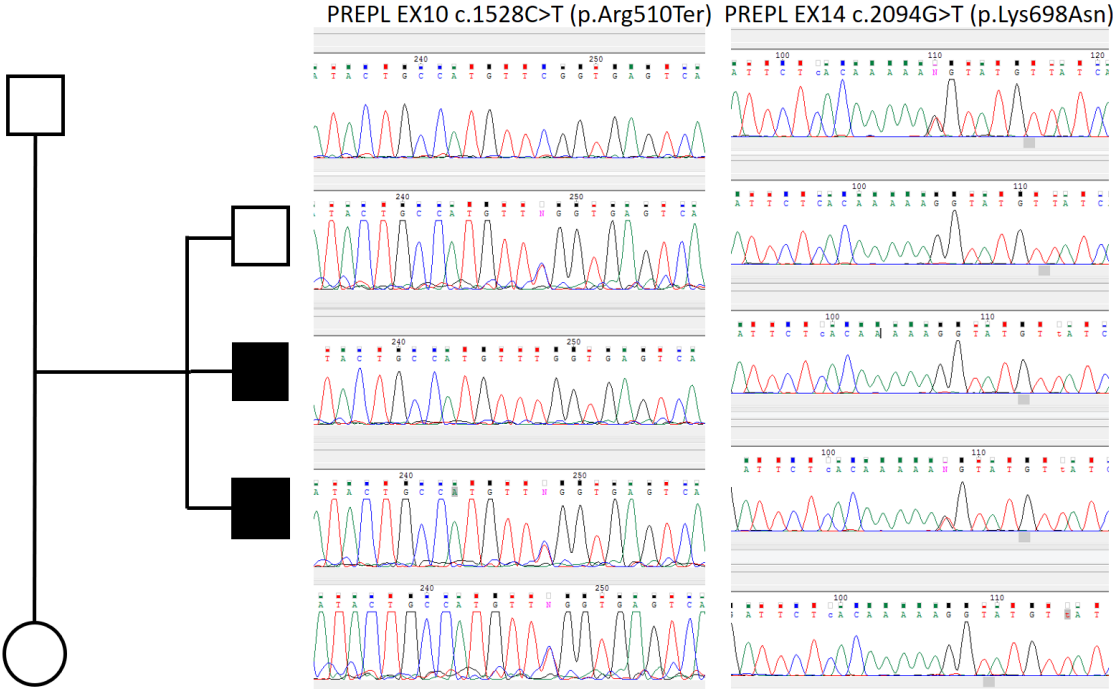


Supplementary Materials:

Figure S1:



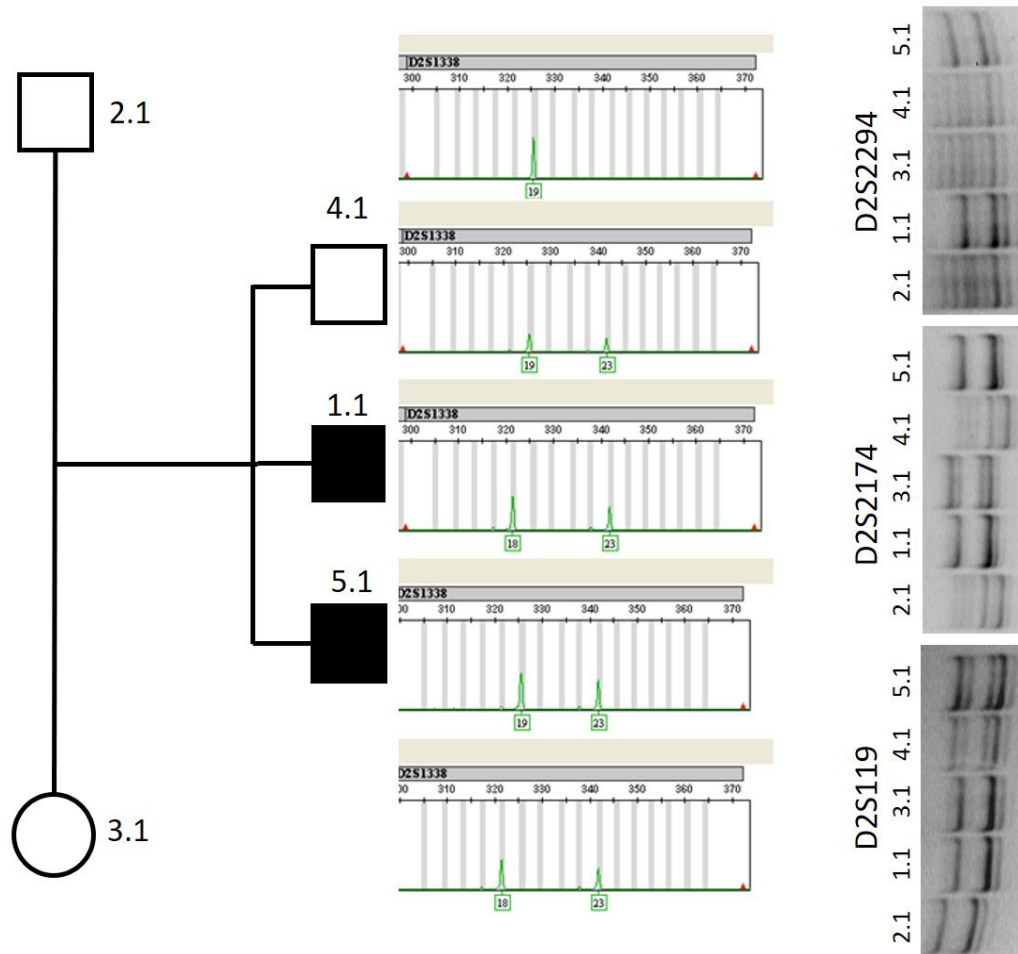
Sanger sequencing of *PREPL* exons 10 and 14.

Figure S2

		Probe target info	AllSamples								
			CYS 1.1	CYS 2.1	CYS 3.1	CYS 4.1	CYS 5.1	CYS 953	CYS 954	CYS 956	
FRSS (n=7)	n/a		90%	100%	100%	100%	90%	90%	90%	90%	
CAS (n=5)	FMRS	n/a	100%	100%	100%	100%	100%	100%	100%	100%	
	PSLP - Relati...	n/a	OK	OK	OK	OK	OK	OK	OK	OK	
	FSLP - Relati...	n/a	OK	OK	OK	OK	OK	OK	OK	OK	
	RSQ - Referenc...	n/a	Warning	Warning	Warning	Warning	Warning	Warning	Warning	Warning	
	RPQ - Referenc...	n/a	OK	OK	Warning	Warning	OK	OK	OK	OK	
02p (n=24)	PPM1B-6	02-044.313084	1.02	1.1	1.06	1.24	0.98	0.95	0.97	0.93	
	SLC3A1-1	02-044.356105	0.96	0.96	0.95	0.75	1.04	1.19	1.15	1.15	
	SLC3A1-2	02-044.361489	0.92	1.15	0.78	0.96	1.17	1.05	1.11	0.93	
	SLC3A1-3	02-044.362145	1.06	1.01	0.95	1	1.1	1	1	0.95	
	SLC3A1-4	02-044.366525	0.92	0.73	1.02	1.03	0.86	1.05	1	1	
	SLC3A1-5	02-044.380661	0.97	0.82	0.93	1	1.05	1.01	1	1.04	
	SLC3A1-6	02-044.381780	0.97	1	0.9	0.93	1.06	1.06	1.01	1	
	SLC3A1-7	02-044.384776	1	1.01	1.05	0.92	0.94	0.99	0.94	1.01	
	SLC3A1-8	02-044.393290	0.93	0.97	1.08	0.91	0.93	0.96	0.93	1	
	SLC3A1-9	02-044.394440	0.97	1.06	0.9	1.01	1.1	1.01	0.99	0.99	
	SLC3A1-10	02-044.400839	1.05	1.02	1.09	1.04	0.96	0.88	0.95	0.91	
	PREPL-15	02-044.401981	1.03	1.01	1.06	1.12	0.97	0.95	0.96	0.96	
	PREPL-13	02-044.403245	0.95	0.96	0.98	0.9	1.03	1.02	1.05	1.06	
	PREPL-12	02-044.403850	1	0.96	0.98	1.11	1.03	1	1	0.97	
	PREPL-11	02-044.407402	1	0.92	0.97	0.97	1.01	1	1	1.04	
	PREPL-10	02-044.409594	1.06	0.93	1.06	1.24	0.95	0.95	0.92	0.92	
	PREPL-9	02-044.413101	1.02	1.01	0.98	1.08	1.07	0.87	0.88	0.81	
	PREPL-7	02-044.419726	0.93	1.09	1.05	0.77	0.96	1.18	1.13	1.18	
	PREPL-6	02-044.423103	1.01	1.06	1.03	0.99	0.97	0.99	1.01	0.98	
	PREPL-5	02-044.424426	1.02	1.01	1.06	1.13	0.98	0.94	0.92	0.94	
	PREPL-4	02-044.428188	0.88	0.91	0.97	1.01	1.03	1.1	1.15	1.06	
	PREPL-2	02-044.440319	1.04	1.05	0.96	1.22	1.05	0.91	0.96	0.91	
	PREPL-1	02-044.442205	1.01	1.11	0.98	0.88	1.11	0.99	1.04	1.03	
	CAMKMT-2	02-044.453469	1	1.23	1.02	1.1	1	0.91	0.93	0.9	
19q (n=15)	TDRD12-13	19-037.973452	1.04	1.04	0.96	1.12	1.04	0.93	0.93	0.93	
	SLC7A9-13	19-038.013357	1.07	1.06	1.07	1.17	0.94	0.95	0.93	0.95	
	SLC7A9-12	19-038.015951	0.99	0.9	1.02	1.02	0.94	0.99	1.02	1.02	
	SLC7A9-11	19-038.024875	1.02	0.71	1.02	0.99	0.95	1	1	1.05	
	SLC7A9-10	19-038.026617	1.02	1.1	1.01	1.09	0.98	0.98	0.95	0.98	
	SLC7A9-9	19-038.041216	1.04	1.11	1.09	1.24	0.93	0.94	0.96	0.91	
	SLC7A9-8	19-038.042576	1.01	0.93	1.05	1.07	0.95	1.02	0.96	0.99	
	SLC7A9-7	19-038.043255	0.96	0.77	0.84	0.86	1.04	1.06	1.08	1.06	
	SLC7A9-6	19-038.044903	0.96	1.04	0.91	0.77	1.05	1.24	1.16	1.24	
	SLC7A9-5	19-038.045101	0.99	1.05	0.96	0.78	1.08	1.04	1.01	1.04	
	SLC7A9-4	19-038.047010	0.98	1.08	1.02	0.82	0.95	1.15	1.12	1.19	
	SLC7A9-3	19-038.047524	1.07	1.22	1.04	1.21	0.96	0.9	0.87	0.91	
	SLC7A9-2	19-038.051169	1	1.04	1	0.99	1.01	0.98	0.98	1.01	
	SLC7A9-1	19-038.052498	0.99	0.71	0.89	0.89	1.01	1.12	1.11	1.08	
	CEP89-19	19-038.061925	1.05	1.29	1.07	1.05	0.95	0.87	0.91	0.92	
	References...	Reference*	01-214.565404	0.96	0.88	1.05	0.95	1	1.01	1	1.01
		Reference*	04-053.940141	0.97	1.04	0.89	0.9	1.04	1	1.02	1
		Reference*	07-075.448405	1.04	0.95	1.07	1.21	0.91	0.99	0.96	1.01
		Reference*	11-006.369211	1	1.17	0.88	0.97	1.05	1.02	1	0.95
Reference*		12-013.797533	1.04	1.25	0.99	1.07	1.01	0.96	0.95	0.96	
Reference*		13-024.357452	0.95	0.91	0.9	0.94	1.06	1.23	1.21	1.16	
Reference*		16-055.463765	1	1.05	0.93	0.91	1	1.11	1.09	1.1	
Reference*		17-041.405030	1.04	0.92	1.11	1.1	0.97	1	1	1	
Reference*		18-045.817181	1.02	0.81	1.02	0.95	0.9	1.03	1.05	0.99	

Results of quantitative analysis using SALSA MLPA Probemix P426 Cystinuria (MRC-Holland). 1.1 – Patient 1; 5.1 – Patient 2; 4.1 – healthy sibling; 3.1 – mother; 2.1 – father; CYS 953, CYS 954, CYS 956 - control samples of healthy adults

Figure S3



Genotyping results of chromosome 2 markers. 1.1 – Patient 1; 5.1 – Patient 2; 4.1 – healthy sibling; 3.1 – mother; 2.1 – father.