

**Supplementary Table S1.** Number of patients with each PKU severity by centre.

	Centre A	Centre B	Centre C	Centre D	Centre E	Centre F	Centre G	Centre H	Centre I
<b>HPA</b> (number of patients; % of total number of patients in that centre)	97; 30%	NA	17; 16%	19; 20%	27; 23%	6; 10%	68; 22%	10; 16%	81; 51%
<b>Mild PKU</b> (number of patients; % of total number of patients in that centre)	69; 22%	38; 39%	46; 46%	21; 22%	54; 47%	8; 14%	82; 26%	13; 21%	26; 16%
<b>Classical PKU</b> (number of patients; % of total number of patients in that centre)	153; 48%	56; 58%	33; 33%	56; 58%	32; 28%	45; 76%	160; 51%	39; 63%	51; 32%
<b>Unknown</b> (number of patients; % of total number of patients in that centre)	1; 0.3%	3; 3%	5; 5%	0; 0%	3; 3%	0; 0%	4; 1%	0; 0%	0; 0%
<b>Total number patients</b>	320	97	101	96	116	59	314	62	158

**Abbreviations:** PKU, Phenylketonuria; HPA, Hyperphenylalaninemia; NA, not applicable. Severity of patients was defined by variants or diagnostic Phenylalanine levels depending availability.