

Table S3. Plasma amino acid levels from members of family 1426.

Amino Acid	1426-12 (U)	1426-6 (A)	Normal range (2-18 years old)
Glycine	226.1	215.9	127-341
Ornithine	22.7	15.1	10-163
Arginine	0.9	0.5	10-140
Citrulline	10.2	11.7	1-46
Alanine	601.1	579.3	152-547
Serine	189.4	202.2	69-187
Valine	219.1	164.0	74-321
Threonine	48.7	50.4	35-226
Leucine+Isoleucine	189.3	148.8	N.A.
Methionine	20.1	18.6	7-47
Histidine	71.5	46.0	41-125
Phenylalanine	71.9	58.4	N.A.
Tyrosine	57.6	51.0	24-115
Aspartate	7.7	23.0	1-24
Glutamate	59.7	131.1	5-150

Plasma amino acids were quantified on bloodspot cards by FIA-MS or LC-MS/MS at the Mayo Clinic (Novarino et al. 2012). U, unaffected; A, affected. Values are expressed in nmol/ml.