Table A2. Genes with pseudodeficiency alleles identified in our cohort, which were excluded from statistical analysis and reporting.

Gene	Pseudodeficiency alleles
ARSA	c.*96A>G (Non-coding)
FAH	c.1021C>T (p.Arg341Trp)
GAA	c.2065G>A (p.Glu689Lys)
GALC	c.550C>T (p.Arg184Cys)
	c.742G>A (p.Asp248Asn)
	c.1685T>C (p.Ile562Thr)
IDUA	c.246C>G (p.His82Gln)