

Figure S1. Conformational changes of the SERAC1 protein structure. (A) The 3-dimensional structure of the wild-type SERAC1 protein and (B) accessibility of its binding site at position 339 with glycine, which demonstrates marked conformational changes where glycine is substituted with arginine. The arrows indicate the site where the mutation occurs. Serine-lipase domain controls intracellular cholesterol trafficking and the mutation is shown to be located near the domain at the border of exon 10-intron 10. SERAC1, serine active site-containing 1.

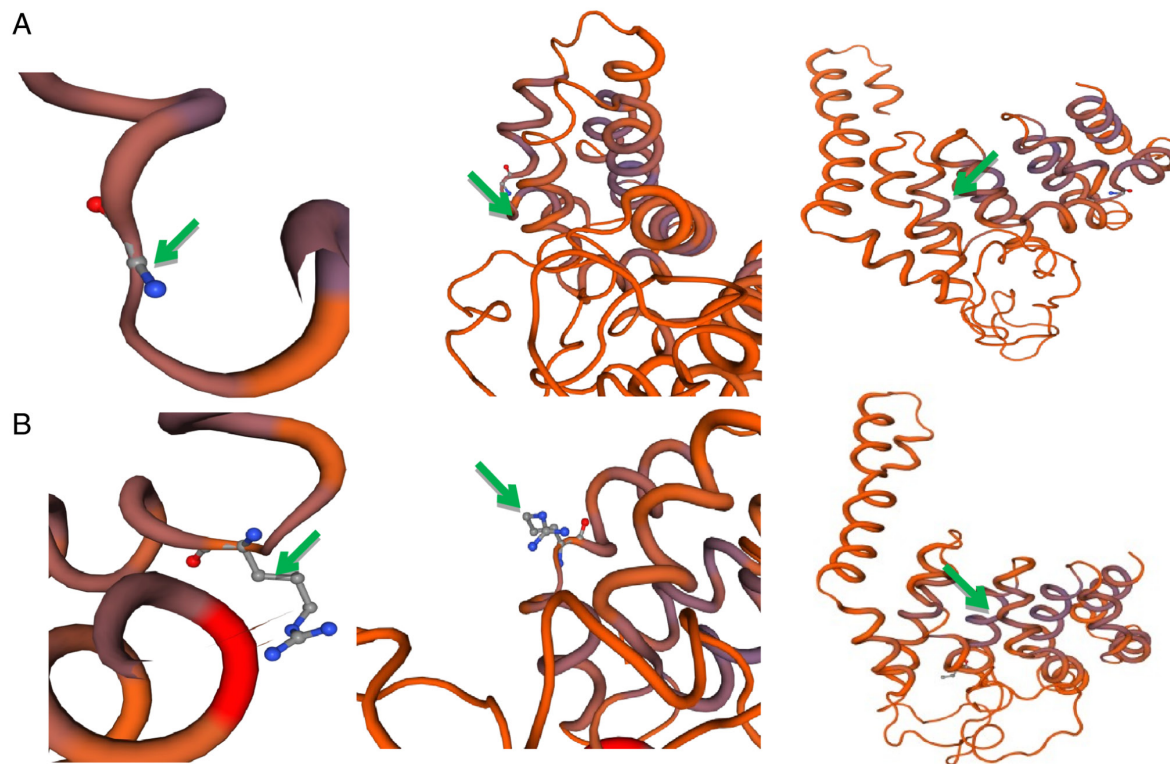


Table SI. Biochemical analysis of the patient with 3-methylglutaconic aciduria with deafness, encephalopathy and Leigh-like syndrome.

Biochemical test	Patient's result	Reported patients	Normal range
Range of 3-methylglutaconate in urine (mmol/mol creatinine)	35-78	16-196	<20
Range of 3-methylglutarate in urine (mmol/mol creatinine)	45-170	Not reported	<15
Plasma lactate	Increased	Increased	Normal
Plasma alanine	Increased	Increased	Normal
Plasma cholesterol	Low	Low	Normal