**Patient S1563427:**

* 12 year old male who has experienced three episodes of muscle pain followed by myoglobinuria
* It was noted that these episodes appeared to follow intense exercise
* Family history:
  + Patient has a sister aged 8 who has not shown any symptoms
  + Mother aged 34, father aged 35. Both appear healthy
  + Mother has a sister, aged 28, who is married and has a daughter aged 4, and a brother aged 31 who is married with two sons, aged 10 and 8
  + Father has two brothers, aged 33 and 32, and a sister aged 29
  + Youngest brother is married with twin daughters aged 9
  + Sister is married with a 3 year old son
  + Maternal grandmother is 55 and grandfather is 60
  + Paternal grandmother aged 55 and grandfather aged 57
  + All other family members appear healthy
  + Paternal grandmother and maternal grandfather are brother and sister
* Referred to genetics for testing of genes associated with metabolic myopathies
* Testing revealed a homozygous mutation in the *CPT2* gene: c.338C>T, p.Ser113Leu
* Use transcript NM\_000098.2/NP\_000089.1 in your analysis. UniProt accession:   
  P23786

**From:**

https://goo.gl/forms/HM9LjajY4OTwNGbu1