**Patient S1598078:**

* 12 year old male who is experiencing stiffness and weakness in his legs
* This has been getting progressively worse since age 5 and he now requires a walking frame
* Family history:
  + Mother, aged 42, requires walking aids. Father, aged 47 is healthy
  + One other sibling, a sister aged 8 who is not showing any symptoms
  + Mother is the younger of two sisters. The sister is 44 and has no mobility issues
  + Maternal grandmother is 65 and is healthy, maternal grandfather is 70 and has used a walking stick since having a hip replacement at 67
  + Paternal grandmother is 67 and appears healthy. Paternal grandfather died aged 65 from a myocardial infarction
* Referred to genetics for testing of hereditary spastic paraplegia genes
* Testing revealed a heterozygous mutation in the *ATL1* gene: c.715C>T, p.Arg239Cys
* Use transcript NM\_015915.4/NP\_056999.2 in your analysis. UniProt accession:   
  Q8WXF7

**Form:**

https://goo.gl/forms/s9rfivTuEmtyizqD3