**Patient S1585674:**

* Four year old male who has shown global developmental delay and has had several seizures. The patient also has some abnormalities in his facial features including long palpebral fissures and long eyelashes.
* It is noted by the clinician that the child has an unsteady gait and mother adds that he is prone to falling
* Family history:
  + Mother aged 30 and father aged 37 are both healthy
  + Patient has a sister aged 7 who also appears healthy and met all developmental milestones as expected
  + Mother has a sister aged 32 and brother aged 28, both appear healthy. Sister is married to the maternal cousin of the patient’s father and they have a son who is 2 years old and has shown delay in some milestones.
  + Father has a brother, aged 40 who is healthy. He has a daughter aged 16 who is also healthy
  + Maternal grandmother aged 52 and grandfather aged 55. Both appear healthy
  + Paternal grandmother aged 63 and grandfather aged 65. Both appear healthy
* Referred to genetics for testing of genes associated with intellectual disability
* Testing revealed two mutations in the *KIF7* gene: c.2482G>A, p.Val828Met (which has been previously classified as class 3) and c.1635G>C, p.Trp545Cys
* Use transcript NM\_198525.2/ NP\_940927.2 in your analysis. UniProt accession: Q2M1P5

**Form:**

https://goo.gl/forms/xU2Jb32hTZv4mQDH3