**S2_Table: D801Y and R756H associated phenotypes.** Summary of the clinical informations from the patients presenting the D801Y and the R756H mutations

| | **Patient presenting the D801Y mutation** |
|---|---|
| | Male, 32years old. |
| **Birth** | Normal |
| **Development** | Motor milestones and speech acquisition normal<br>At 14 months: episodes of fine tremor of the extremities |
| **Childhood and Adolescence** | Recurrent episodes of right or left sided hemiplegia, induced by excitement or fear, usually preceded by severe headaches and associated with dystonia,<br>Less frequent episodes of quadriplegia<br>No effect of Flunarizine nor Inderal<br><br>Between the spells: Ataxia, right hemiparesia, right sided athetosis not improved by Sinemet, fluctant dysarthria, abnormal eye movements<br><br>Mental development: IQ=123 at 4y3M and 92 at 12yo |
| **At 30 yo** | First seizure (Grand Mal) |
| **Last examination at 31 yo** | Decreased frequency of the hemiplegic/dystonic episodes<br>Dysarthria<br>Tremor between the spells<br>Mild ataxia and uncoordination but walks independently<br><br>Graduated from High School but decreased mental skills |

| | **Patients presenting the R756H mutation** | |
|---|---|---|
| | Female, 5.5 years old | Female, 11.5 years old |
| **Birth** | Normal | Normal |
| **Development** | Motor milestones and speech slightly delayed by 3-4 months | Motor milestones and speech acquisition normal |
| **Childhood** | Febrile seizure at 2.5 yo with residual persistent right sided hemiplegia and ataxia<br><br>Recurrence of seizure at 3 yo, followed by dysarthria, dysphagia. | Febrile seizure at 2 yo<br><br>First episode of hemiplegia or dystonia at 3.5 yo |
| **Last examination** | **at 3yo**<br>Right sided hemiplegia<br>Truncal ataxia needing assistance for walking and sitting<br>Poor speech | **at 9.5 yo**<br>Ataxia and dysmetria<br>Dysarthria<br><br>Learning disability |