

**Table S1.** Genetic variants affecting the ‘dynactin subunit 1’ gene reported in Perry syndrome.

<b>Phenotype</b>	<b>Transcript</b>	<b>HGVS nucleotide</b>	<b>HGVS protein</b>	<b>Country of the patient (ref.)</b>
Perry syndrome	NM_004082	c.121C>T	Arg41Ter	Japan (1)
		c.133G>C	Ala45Pro	India (2)
		c.156T>G	Phe52Leu	Japan(3), Japan (4), Portuguese (5)
		c.200G>A	Gly67Asp	UK (6), Republic of Korea (7), Argentina (8)
		c.202A>G	Lys68Glu	Japan (9)
		c.202A>G	Gly71Val	Japan (9)
		c.212G>C	Gly71Ala	USA (10), England (10), Japan (10), USA (11)
		c.212G>A	Gly71Glu	Mexico (present study), France (10), Poland (12), France (13), Canada(14), Poland (14)
		c.211G>A	Gly71Arg	Canada (10), Turkey (10), USA (15), Colombian (15), UK (16), Republic of Korea (7), USA (11), UK (17)
		c.212G>T	Gly71Val	Japan (9)
		c.214A>C	Thr72Pro	USA (10)
		c.221A>C	Gln74Pro	Japan (10)
		c.233A>G	Tyr78Cys	New Zealand (15), New Zealand (18), Republic of Korea (7)
		c.232T>C	Tyr78His	USA (19)

HGVS, Human Genome Variation Society.

**Table III.** Clinical characteristics of patients with Perry syndrome with Gly71Glu (G71E) reported in various countries.

<b>Characteristic</b>	<b>Mexico (present study)</b>	<b>Poland - Konno <i>et al</i> (17), 2017</b>	<b>France - Caroppo <i>et al</i> (16), 2013</b>	<b>France - Lechevalier <i>et al</i> (20), 1992</b>	<b>Canada - Purdy <i>et al</i> (21), 1979</b>
Number of affected members	4	4	8	5	3
Age of proband at onset, years	49	42	39-59	NA	46
Age of proband at death, years	57	NA	43-64	NA	48
Cause of death	Respiratory failure	NA	Respiratory failure	Respiratory failure	Respiratory failure
Parkinsonism	+	+	+	+	+
Weight loss	+	+	+	+	+
Depression/ apathy	- <sup>a</sup>	+	+	+	+
Hypoventilation	+	+	+	+	+
Additional clinical features	NA	NA	Slow vertical saccade, blepharospasm	NA	NA

<sup>a</sup>Depression was not reported by the patient or his wife, but a formal psychiatric evaluation was not performed in the patient. Farrer *et al* 2008 (10) detected a family in France but did not report the clinical data. NA, information not available; +, positive/present; -, negative.