	Mutations	Genetic	Country	Severity	kinky hair/ giant axons	References
Exon 1	V7Fs (ins)	ho (csg)	Tunisia	classical	yes/yes	(Bomont et al., 2000)
	V7Fs (del)	He^{L}	Italy	mild	no/yes	(Bruno et al., 2004)
	R15S	ho (csg)	Tunisia	mild	no/yes	(Bomont et al., 2000)
	H33P	He^{w}	China	mild	no/yes	(Wang et al., 2014)
	V35F	ho (csg)	Israël	mild	no/ni	(Aharoni et al., 2016)
	Q44X	He^{t}	?	mild	no/ni	(Roth et al., 2013)
	A49E	ho	USA	classical	yes/yes	(Boizot et al., 2014)
	A49T	He^{k}	Italy	classical	no/yes	(Bruno et al., 2004)
	A51P	ho (csg)	Pakistan	classical	yes/yes	(Houlden et al., 2007)
	S52G	He^{d}	USA	classical	yes/yes	(Bomont et al., 2000)
	S52N	ho (csg)	Algeria	classical	yes/ni	(Echaniz-Laguna et al., 2020)
	P53L	He^{w}	China	mild	no/yes	(Wang et al., 2014)
	Δex1-11	He^{v}	?	?	yes/yes	(Mohammad et al., 2014)
Intron	IVS1-1 G>A	ho (csg)	Algeria	classical	yes/yes	(Koop et al., 2007)
Exon 2	Y71X	ho	Scotland	classical	no/yes	(Houlden et al., 2007)
	L75H	He^{q}	China	classical	yes/yes	(Zhang and Zou, 2009)
	S79L	He^{e}	Morocco	classical	yes/yes	(Bomont et al., 2000)
	«	He^{ab}	China	classical	yes/yes	(Xu et al., 2020)
	V82F	He^{a}	France	classical	yes/yes	(Bomont et al., 2000)
	I86F	He^{g}	Germany	classical	yes/yes	(Bomont et al., 2003)
	Y89C	He^{n}	Serbia	classical	yes/yes	(Koop et al., 2007)
	Δex2-9	ho (csg)	Algeria	classical	yes/ni	(Echaniz-Laguna et al., 2020)
	Δex2-11	He^{r}	Belgium	classical	yes/yes	(Buysse et al., 2010)
Intron						
Exon 3	Δ114-119	He^{g}	Germany	classical	yes/yes	(Bomont et al., 2003)
	F124C	ho (csg)	China	mild	no/yes	(Wang et al., 2014)
	R138H	ho	Algeria	mild	no/yes	(Bomont et al., 2000)
	R162X	ho (csg)	Japan	classical	yes/yes	(Akagi et al., 2012)
	«	ho (csg)	India	classical	yes/ni	(Garg et al., 2018)
	E169K	ho (csg)	Algeria	classical	no/yes	(Tazir et al., 2009)
	E180Fs	ho (csg)	India	classical	yes/ni	(Garg et al., 2018)
	V195F	He^{p}	Germany	classical	yes/yes	(Koop et al., 2007)
	R201X	He^{i}	Germany	classical	ni/ni	(Kuhlenbaumer et al., 2002)
Intron	IVS3+1G>T	He^{p}	Germany	classical	yes/yes	(Koop et al., 2007)
Exon 4	R242X	ho (csg)	Turkey	classical	yes/yes	(Bomont et al., 2003)
	«	ho	India	classical	yes/yes	(Boizot et al., 2014)
	I244Fs	He^{u}	?	classical	yes/ni	(Roth et al., 2013)
	R269Q	ho	Germany	classical	yes/yes	(Bomont et al., 2003)
	R269W	He^{s}	China	mild	yes/yes	(Xu et al., 2013)
	«	He^{u}	?	classical	yes/ni	(Roth et al., 2013)
	G270S	He^{x}	Japan	mild	no/yes	(Koichihara et al., 2016)
Intron						
Exon 5	R293X	ho (csg)	Turkey	classical	yes/yes	(Demir et al., 2005)
	«	He^{c}	Turkey	classical	yes/yes	(Bomont et al., 2000)
	Y299C	He^{L}	Italy	mild	no/yes	(Bruno et al., 2004)
	L309R	ho (csg)	Tunisia	classical	yes/yes	(Bomont et al., 2000)
	P315L	He^{k}	Italy	classical	no/yes	(Bruno et al., 2004)
	«	He^{m}	England	classical	yes/yes	(Houlden et al., 2007)
	A324V	He^{h}	New Zeland	mild	no/ni	(Boizot et al., 2014)
	E325K	ho (csg)	Israël	classical	yes/yes	(Abu-Rashid et al., 2013)
Intron						
Exon 6	G332R	ho (csg)	North Africa	classical	yes/yes	(Boizot et al., 2014)
	K338X	He^{f}	France	classical	yes/ni	(Bomont et al., 2003)
	E362E*	ho (csg)	France	classical	yes/ni	(Echaniz-Laguna et al., 2020)
	Δex6-8	He^{f}	France	classical	yes/yes	(Bomont et al., 2003)
Intron	IVS6+1G>C	He^{j}	Italy	classical	yes/yes	(Bruno et al., 2004)

Exon 7	E392K	He[aa]	China	classical	yes/yes	(Cai et al., 2018)
	G368R	ho	Sri Lanka	classical	yes/yes	(Bomont et al., 2003)
	«	He[n]	Serbia	classical	yes/yes	(Koop et al., 2007)
	C393X	He[d]	USA	classical	yes/yes	(Bomont et al., 2000)
	W401X	He[b]	France	classical	yes/yes	(Bomont et al., 2000)
Intron	IVS7-1 G>A	ho	Japan	classical	yes/ni	(Miyatake et al., 2015)
8	I423T	He[i]	German	classical	ni/ni	(Kuhlenbaumer et al., 2002)
	W448L	ho (csg)	China	classical	no/yes	(Wang et al., 2014)
	R458W	ho (csg)	Algeria	classical	yes/ni	(Echaniz-Laguna et al., 2020)
Intron						
Exon 9	C464Y	He[h]	New Zeland	mild	no/ni	(Boizot et al., 2014)
	G474R	He[o]	Spain	classical	no/yes	(Koop et al., 2007)
	«	He[t]	?	mild	no/ni	(Roth et al., 2013)
	R477X	ho (csg)	Algeria	classical	yes/yes	(Bomont et al., 2003)
	«	ho (csg)	Algeria	classical	no/yes	(Bomont et al., 2003)
	«	He[o]	Spain	classical	no/yes	(Koop et al., 2007)
	«	ho (csg)	Algeria	classical	yes/yes	(Tazir et al., 2009)
	«	ho (csg)	Algeria	classical	yes/yes	(Tazir et al., 2009)
	«	ho (csg)	Algeria	classical	no/yes	(Tazir et al., 2009)
	«	ho (csg)	Algeria	classical	yes/yes	(Tazir et al., 2009)
	A461V	He[y]	Mexico	classical	yes/yes	(Normendez-Martinez et al., 2018)
	Q483X	ho (csg)	Tunisia	classical	yes/yes	(Bomont et al., 2000)
	E486K	ho (csg)	Tunisia	classical	yes/yes	(Bomont et al., 2000)
	«	He[b]	France	classical	yes/yes	(Bomont et al., 2000)
	«	He[r]	Belgium	classical	yes/yes	(Buysse et al., 2010)
	T489S	He[ab]	China	classical	yes/yes	(Xu et al., 2020)
	E493K	He[z]	India	classical	yes/ni	(Garg et al., 2018)
	«	ho (csg)	France	classical	yes/ni	(Echaniz-Laguna et al., 2020)
Intron	IVS9+1G>T	ho (csg)	Turkey	classical	yes/ni	(Demir et al., 2005)
	«	ho (csg)	Turkey	classical	yes/ni	(Demir et al., 2005)
	«	ho (csg)	Turkey	classical	yes/yes	(Incecik et al., 2015)
	«	ho (csg)	Turkey	classical	yes/yes	(Incecik et al., 2015)
	«	ho (csg)	Turkey	classical	ni/yes	(Edem et al., 2019)
10	W502X	ho (csg)	Pakistan	classical	yes/yes	(Houlden et al., 2007)
	L510X	He[j]	Italy	classical	yes/yes	(Bruno et al., 2004)
	F518Fs	He[m]	England	classical	yes/yes	(Houlden et al., 2007)
	Δex10-11	ho	USA	classical	yes/yes	(Boizot et al., 2014)
Intron						
Exon 11	R545C	ho	France	classical	yes/yes	(Bomont et al., 2000)
	R545H	ho	Finland	classical	yes/yes	(Koop et al., 2007)
	«	He[q]	China	classical	yes/yes	(Zhang and Zou, 2009)
	«	He[s]	China	mild	yes/yes	(Xu et al., 2013)
	«	He[aa]	China	classical	yes/yes	(Cai et al., 2018)
	R545L	He[y]	Mexico	classical	yes/yes	(Normendez-Martinez et al., 2018)
	T553Fs	ho (csg)	India	classical	yes/yes	(Nalini et al., 2008)
	P562A	ho	Italy	mild	yes/yes	(Bruno et al., 2004)
	R570Y	He[c]	Turkey	classical	yes/yes	(Bomont et al., 2000)
	A576E	He[x]	Japan	mild	no/yes	(Koichihara et al., 2016)
	«	He[z]	India	classical	yes/ni	(Garg et al., 2018)

Supplementary Table 1. Mutations identified in the *GAN* gene.

ho: homozygous mutation; csg: consanguineous family; He: heterozygous mutation (compound mutations for the same patient are indicated by letter in [exponent]); E362E*: silent mutation (at the last amino acid of exon 6) affects splicing and leads to premature stop codon; ni: non investigated. PS: we apologize to authors but 3 publications were excluded from the table for the following reasons: Leung *et al.,* BMC genet 2007 (absence of clinical data, no screen in controls); Wang *et al*., Zhonghua Yi Xue Yi Chuan Xue Za Zhi 2016 (article in chinese); Almeida *et al.,* An Bras Dermatol 2016 (incomplete data). Also, in Koop et al., Neuromscul Disord 2007: variant found in the promotor in family 9 was not included (uncertain & other allele not identified).

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