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SUPPLEMENTARY MATERIAL**Table S1:** List of primers used to conduct genetic screening for the HMN-J mutation, cloning and qPCR analysis.

Name	Sequence	Description
SIGMAR1 seq	(F) tgggagtgcagacttcata	<i>SIGMAR1</i> Jerash dHMN mutation sequencing
	(R) gccaaagaggtaggtggtga	
SIGMAR1_pGFP-C3	(F) cggaattctgatgcagtgggccgtgggc	<i>SIGMAR1</i> cloning into pEGFP-C3 vector
	(R) cgcggatcctcaagggtcctggccaaaga	
SIGMAR1_pFlag	(F) cggaattcaatgcagtgggccgtgggc	<i>SIGMAR1</i> cloning into pFLAG-CMV2 vector
	(R) cgcggatcctcaagggtcctggccaaaga	
Flag-SIGMAR1	(F) ccgctcgagatggactacaagacgat	<i>Flag-SIGMAR1</i> cloning into pIRES2-EGFP vector
	(R) cgcggatcctcaagggtcctggccaaaga	
SIGMAR1 qPCR	(F) ggcgctactgggctgagat	<i>SIGMAR1</i> fragment qPCR amplification
	(R) gcccggtgactaccgtctcc	
L19	(F) gcggaagggtacagccaat	<i>L19</i> fragment qPCR amplification
	(R) gcccggtgactaccgtctcc	
Flag-SIGMAR1 qPCR	(F) atggactacaagacgatgacg	<i>Flag-SIGMAR1</i> fragment qPCR amplification
	(R) tatctcttcgcgctggaaga	
GFP qPCR	(F) aaggcatcgacttcaag	<i>GFP</i> fragment qPCR amplification
	(R) tgcttgctggccatgatatag	

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Table S2: Clinical features of five HMNJ patients and 7 non-affected members from their families.

Genotype	ID	Sex	Age (y)	Age of onset (y)		Motor signs and symptoms				Pyramidal signs			Sensory symptoms
				UL	LL	Muscle wasting		Muscle weakness		Knee R	Ankle R	Babinski	
						UL	LL	UL	LL				
<i>SIGMAR1</i> ^{HMNJ} Homozygous	B22	F	41	18	12	Mild	Mild	Mild	Mild	++	-	-	-
	A9	F	24	16	6	Moderate	Severe	Moderate	Severe	+++	+	-	Numbness
	C27	M	32	14	6	Mild	Mild	Mild	Mild	+++	+	-	Numbness
	C29	M	35	16	5	Moderate	Severe	Moderate	Severe	+++	-	-	Numbness
	A11	M	10	8	4	Moderate	Severe	Moderate	Severe	+++	++	+	-
<i>SIGMAR1</i> ^{HMNJ} <i>SIGMAR1</i> ^{WT} Heterozygous	B20	M	35	-	-	Absent	Absent	Absent	Absent	++	+	-	-
	A15	M	58	-	-	Absent	Absent	Absent	Absent	++	+	-	-
	C25	M	25	-	-	Absent	Absent	Mild weak grip	Absent	+++	++	-	-
	C33	M	37	-	-	Absent	Absent	Absent	Absent	+++	+	-	-
<i>SIGMAR1</i> ^{WT} Homozygous	B16	F	23	-	-	Absent	Absent	Absent	Absent	+++	++	-	-
	A8	M	21	-	-	Absent	Absent	Absent	Absent	+++	+	-	-
	C26	M	28	-	-	Absent	Absent	Absent	Absent	+++	++	-	Numbness

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Abbreviations: UL = upper limb; LL = lower limb; R = reflexes; Knee and ankle reflexes: absent = -, decreased = +; normal = ++, brisk = +++;
Babinski: absent = -, present = +;