Supplementary Figure, Tables and Note for:

Mutations in *GNAL* cause primary torsion dystonia

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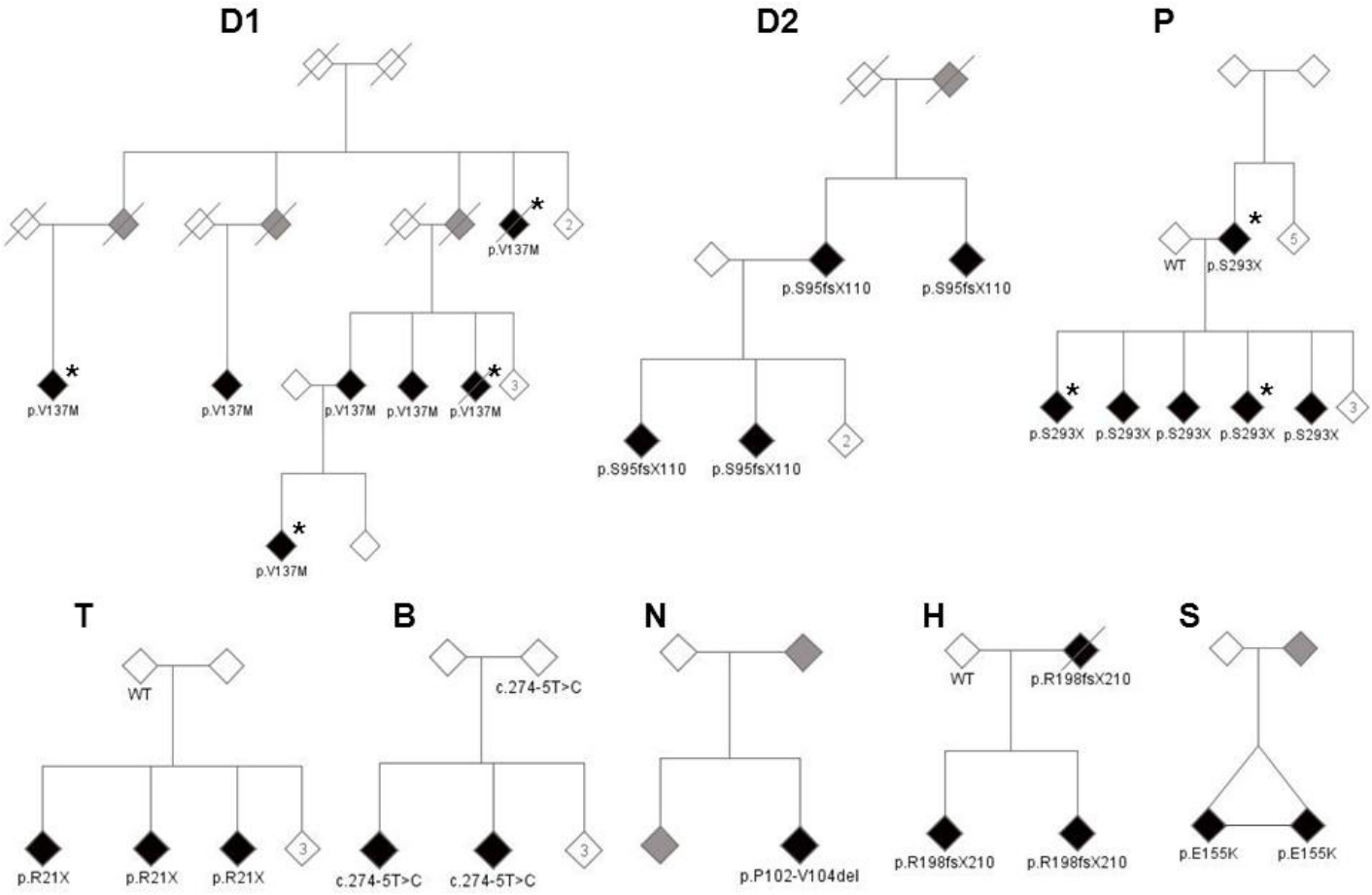
Figure 1



Supplementary Figure 1. Pedigrees of families with *GNAL* mutations. Filled in black symbols represent affected individuals, filled in grey symbols represent affected, known by history, diamond symbols with numbers represent additional sibs. WT indicates no mutation. Asterisks indicate the samples that were exome sequenced. Mutation status is indicated below the symbols for individuals whose DNA was available for testing. The pedigrees have been modified to protect the identities of the families.

Supplementary Table 1. Sites Involved for 28 GNAL Patients from 8 Families.

	SITES INVOLVED													
	upper face	lower face	neck	larynx	pharynx	tongue	jaw	right arm	left arm	right leg	left leg	trunk	Dystonia distribution	Site of onset
*FAMILY D1**														
1	•	•	•			•						•	S	Neck
2	•	•	•	•		•	•	•	•			•	S	Neck,Larynx,Trunk
3	•	•	•	•									S	Neck
4^			•					•	•	•	•	•	G	Legs
5	•		•									•	S	Neck
6		•	•					•					S	Neck
7	•	•	•			•	•					•	S	Neck, Face
*FAMILY P **														
1			•										F	Neck
2			•					•	•	•	•	•	G	Neck
3			•										F	Neck
4^	•		•					•	•		•	•	G	Leg
5			•					•					S	Neck
6			•										F	Neck
*FAMILY S**														
1^			•										F	Neck
2		•	•				•						S	Neck
FAMILY D2														
1	•	•	•	•			•	•				•	S	Neck
2^						•	•						S	Jaw
3	•	•	•		•	•	•	•	•				S	Neck
4						•							F	Tongue
FAMILY H														
1			•										F	Neck
2^			•										F	Neck
3			•										F	Neck
FAMILY B														
1			•										F	Neck
2^			•	•				•					S	Neck
FAMILY T														
1^			•										F	Neck
2			•										F	Neck
3		•	•	•			•						S	Larynx
FAMILY N														
1^			•										F	Neck

Dystonia distribution: F – focal, S – segmental, M – multifocal, G – generalized
^ Denoted probands
* reported previously and updated here: Fam D1 in[13,14,60], Fam P in[14] and[15], Fam S in[61]. The following are the corresponding identifiers from[13] for the individuals in D1: 1=individual 207, 2=302, 3=307, 4=304, 5=317, 6=315, 7=403.

Supplementary Table 2. Single Nucleotide Variants indentified in Exome Sequencing.

Family P Variants			Shared HZ	Novel against dbSNP132	Annotation			
113	10831	10835			MS	NS	UTRs	Others
57,663	56,435	58,354	11,124	458	68	1	13	376

Family D1 Variants				Shared HZ	Novel against dbSNP132	Annotation			
10081	10097	10547	11484			MS	NS	UTRs	Others
57,185	59,975	62,412	58,148	4,578	208	20	0	6	182

Supplementary Table 3. Indels identified in Family 1 exome sequencing.

Family P Indel Variants			Shared HZ	Novel against dbSNP132	In coding regions
113	10831	10835			
11,861	11,535	13,207	5,482	1,644	79

Supplementary Table 4. Summary of Clinical features in *GNAL* mutation carriers.

	Mutation positive (n=28)
Women (n,%)	14 (50%)
Mean age of onset[1] (years ±SD, range)	31.32± 12.39(7-54)
Mean age at final exam (years ±SD, range)	49.28 ± 13.30 (19-75)
Site of onset	
Arm	0 (0%)
Leg	2 (7.14%)

Cranial	5 (17.86%)
Face	1 (3.57%)
Jaw/Tongue	2 (7.14%)
Larynx	2 (7.14%)
Cervical	23 (82.14%)
Sites at final examination	
Arm	9 (32.14%)
Leg	3 (10.71%)
Cranial	16 (57.14%)
Face[1]	13 (48.15%)
Jaw/Tongue[1]	10 (35.71%)
Larynx	5 (17.86%)
Cervical	26 (92.86%)
Speech[2]	12 (44.44%)

[1]Unratable facial, speech and final distribution in 1 *GNAL* carrier

Supplementary Table 5. Primers used in GNAL exon amplification.

Exon	Forward Primer	Reverse Primer	Annealing Temp.
NM_182978_5UTR	GGCTCAGACGGCATTATTTACGGT	TCTCCTTCGGCTTGTCTGCTTT	59
NM_182978_ex1	ATGGGTCTGTGCTACAGTCTGC	AGAATCAGCCCGTAGTGTCCTC	59
NM_001142339_5UTR	AGAAGCTGAGCAGAACAAAGGC	AATAGAGAGTTGGAGACGTGTGCG	58
NM_001142339_ex1-2	AAACGCCTGCTCTGAATCGGAA	ACRTCTGGGAGAGAGACGAAGTTT	59
ex3-4	AACCTTTGCRGATGTCTTGG	CCTCTTATGCATTTAGAGAGCTGG	55
ex5	TCACACTAAACATAGAGTSGGTGCAT	TTATTCATTCCTTGCACTCAAG	57
ex6	ACTCACTTCAGCTTCTTGGCCT	ATGACCAATCCACRCACACACA	59
ex7	ATGCTCGGCCAATGTTGGTT	TGGGCCATGCAGCTCTTAACAA	58
ex8	ATGTGTGAACGCTGGAACCT	GCAGTGCTGAGTGTTAGAATTCAC	56
ex9	TGCAGGCTGKTCTGTGACT	AGAGATACTTCGTACGGTTCTGG	56
ex10	ACAGATGGAATGAGGATACTGGTG	GGGATCCKATTACAAATAGGACCTTG	56
ex11	GCTGGGAATATACAGCAGTCTAACR	TGAAGTCTAGCCCATCCAAG	57
ex12	TGCATTGAGACCATTCCTGCCT	GGGAGGCYGTATTCAATGACAACA	59

Supplementary Note:

Human research subjects were recruited through advertisement or referral from movement disorder physicians. All families chosen for the study were multiplex of Northern European ancestry. All study subjects gave informed consent prior to participation, and the study was approved by all institutional review boards. Videotaped examinations and determination of affected status was undertaken as previously published[11].

11. Bressman, S.B. et al. Idiopathic dystonia among Ashkenazi Jews: evidence for autosomal dominant inheritance. Ann Neurol 26, 612-20 (1989).